

ASTUTE, ASSERTIVE, AND ALPHA-1: QUANTIFYING EMPOWERMENT IN A RARE
GENETIC COMMUNITY

By

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To John and Freddie Walsh; and to my beloved sons, Elijah and Sam.

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Abstract of Dissertation Presented to the Graduate School
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Chair: Peter Collings

Major: Anthropology

We investigated empowerment in the Alpha-1 Antitrypsin Deficiency (Alpha-1) community, a rare, genetic disease network in the United States. The research was motivated by nine years of observations in the community. After observing what seemed to be a heightened amount of activism among Alpha-1 community members, I had hypothesized that this community represents a model of empowerment, and designed a study to define and quantify empowerment from a community-based perspective.

Fieldwork was conducted from 2006 to 2008 to obtain the community's definition of empowerment. Focus group sessions and semi-structured interviews were held to elucidate specific domains of patient empowerment. The data from the interviews and focus group sessions was analyzed, and a codebook was developed of the primary themes. These themes were used to build a theoretical model of empowerment, and to develop research questions about the relations of the various domains in the construction of empowerment. Data from office visits and physician-nurse interviews were analyzed using an ethnography-of-speaking approach towards understanding the hegemony, or balance of power, between physician and patient.

Patient data was analyzed statistically to quantify the relations between components of empowerment.

The study confirmed the validity of a model of empowerment in the Alpha-1 community as a Guttman-like scale (Guttman, 1950). For, the components of empowerment identified in the community can be seen as a series of activities, and states of awareness, that evolved from simple to complex. Statistical analysis also confirmed hypothesized, and significant, relations between specific components of empowerment. However, the study did not demonstrate that empowerment indicated a loss of medical authority; rather empowerment was mastery of the language and knowledge of the medical domain. And, although, core components of empowerment are consistent among a range of Alphas, empowerment is best characterized as variable, even among this stable, closed population.

The study results could prove relevant to other types of patients, as core components of empowerment defined in the study are not unique to the Alpha-1 community. This includes the value of family support and networking, the importance of a community identity, and the positive impact an effective infrastructure provides.

CHAPTER 1
EMPOWERMENT IN THE MEDICAL DOMAIN

Finding an Appropriate Model

The aim of this study was to define and quantify patient empowerment in the Alpha-1 Antitrypsin Deficiency (Alpha-1) community, a rare disease network in the United States. Alpha-1 is a rare, genetic condition discovered in 1963, in Sweden, in patients with the lung disease emphysema (Laurell and Eriksson, 1963). Subsequent research in the United States led to a discovery of an association between Alpha-1 and liver disease (Sharp et al., 1969). This genetic condition can lead to severe liver disease in newborns, and is the fourth leading cause of liver transplantation in children in the United States (Teckman, 2005). However, the majority of individuals with a deficiency of Alpha-1 have progressive lung disease that generally begins to appear in the fourth decade of life (Anonymous, 2000). Individuals with Alpha-1 lung disease have increasing difficulty with breathing, may have to use portable oxygen, and, eventually die of advanced lung disease after years of illness. As lung diseases are now the 6th leading cause of death worldwide, studying how patients become empowered within the Alpha-1 community could facilitate healthcare efforts in what is fast becoming a major international health problem. For, those with Alpha-1 lung disease are like the canaries that miners sent ahead of mining crews to test air quality. The canary foretold the conditions in the mines for those who followed. Highlighting empowerment within the Alpha-1 community, and the positive way individuals deal with their genetic lung disease, might also serve as the forerunner, and model, of the potential role patients can play in their own lung disease management. It is a model that may have relevance for all types of lung patients in the world, or promote efforts by any type of patient to become empowered.

The study arose out of my involvement in the Alpha-1 community as a paid employee, and many years of participant observation of various stakeholders, or members, of the community. The stakeholders include diagnosed individuals, and family members, interacting through patient support groups, staff of national patient, and scientific, organizations, and a well defined research network consisting of research scientists, clinicians, and nursing staff (Figure 1-1, Table 1-1).

Over a period of nine years, I observed what seemed to be an extraordinary amount of activism and positivism among many of the individuals involved with the Alpha-1 community. The positive attitudes, and forms of activism I observed, were exhibited not only by the patients, but also by caregivers and family members, and by the medical professionals associated with the community. Based on my observations, and a review of the social and medical literature on empowerment, I hypothesized that this community is particularly empowered. To test this hypothesis, I developed the following research questions, and a study design, that would allow me to understand empowerment and begin to measure it among community members:

- **Research Question 1:** Is patient empowerment a visible sign of a shift in the hegemonic relation between physicians and patients?
- **Research Question 2:** Does business expertise, and organizational leader's participation in policy making and drug development, make these organizations, and their leaders, structurally powerful?
- **Research Question 3:** Are patients with access to resources in the Alpha-1 community more likely to become empowered on a personal level? And, what are the components of personal empowerment?
- **Research Question 4:** Do domains of empowerment form an unidimensional scale (Figure 1-2).

The development of the research questions and how they were operationalized into a research plan are described in further detail below (page 25-29).

Defining Empowerment

Empowerment is not a new concept – initial usage of the word is noted in the mid-17th century as a legal term meaning “to invest with authority, authorize”. A dictionary listing from the late 1970s maintains the connotation of delegation and authorization, but adds an extended meaning relating to enabling, (Funk and Wagnalls, 1978). More recent, primarily online dictionary listings define empowerment as “freedom to do something”, or that “makes you more confident and makes you feel you are in control of your life” (Cambridge Advanced Learner’s Dictionary, 2008), “give strength and confidence to” (www.askoxford.com), or that “promotes self-actualization” (Merriam Webster Online Dictionary Vers 10, 2008). A dictionary website that searches all web-available listings found 22 dictionaries with English definitions of empowerment including usage of the word in management, accounting, economics, computing, medicine, conflict resolution, environmental science, and in technologies such as construction and material physics (www.onelook.com).

In the medical domain, patient empowerment has been variously defined in terms of patient autonomy, the patient as consumer, or as representing numerous possibilities for social transformation (Church et al. 2002; Teff, 1994; Leps 1995, Clarke et al., 2003, Schlesinger 2002). Patient empowerment is more recently defined on a patient advocacy website as individuals who “need to: take responsibility, set goals, collaborate with others, gather evidence, be a smart healthcare consumer, stay safe in the healthcare environment”, and who “understand and support the tenets of patient advocacy” (Torrey, 2007). However, as my study shows, empowerment may also indicate the experience of caregivers, physicians, researchers, organizational leaders, and other stakeholders associated with a disease community.

Patient autonomy and patients thinking they share common attributes were observed in the Alpha-1 community. However, like the authors cited above, I consider empowerment as a

social process involving patients within the context of larger social relations. I, therefore, chose to move beyond a narrow focus on the patient's experience, and to situate an understanding of empowerment within the context of the medical domain (in relation to patient-physician dynamics), and within a larger societal context.

When examined in relation to various types of medical interactions, I agreed with Teff (1994) and Leps (1995), that patient empowerment may represent not only a rise in patient autonomy, but also a challenge to the traditional dominance of medical authority. I thought this an interesting research question that might prove true, and if so, would explain why the 'patient' empowerment I had observed in the Alpha-1 community seemed to affect patients, caregivers, and medical professionals. Testing this research question required broadening the scope of my research to include all affected individuals in the Alpha-1 community, including newly diagnosed individuals, and exploring other possible forms of empowerment besides patient empowerment.

I emphasize the context of empowerment in the medical domain, since empowerment, as defined above, is used to characterize other forms of social change in the United States. Since the focus of my study on empowerment is on a process situated within the medical domain, the definitions used throughout this study are based on the use of the term in medical settings. For this study, I define empowerment as disease management on both the personal and structural level, a condition that encompasses both individual and community-level attributes, and a condition that manifests among not only individuals diagnosed with a disease, but their caregivers and family members, health care team, and nationwide network of contacts.

Examining the Basic Premise

Empowerment in the Alpha-1 community is the premise, and basis, for my research, and led to research questions, and a study design, to define empowerment from an emic perspective. I

patterned the study design on social and medical research that successfully utilized both qualitative and quantitative analyses (Bernard and Ryan 2005, 1998; Carey et al. 1996; Chavez et al. 1995; Clark et al. 2004; Fries 1983; Goodacre et al. 2007; Gottschalk 1997; Grunfeld et al. 2008; Hajiro et al. 1998; Handwerker 2003; Handwerker and Wozniak 1997; Johnson 1995; Kaufert and O'Neil 1993; Ketalaars et al. 1986; Ryan and Martinez 1996; Stoller et al. 1994; Tu 2006; Weinfurt et al. 2003; Wyrwich et al. 2007, 2006). Many of these studies focused on techniques for narrative analysis of interview data; however the medical literature on Quality of Life (QOL), and measuring disability, were also influential on my study design. For the Alpha-1 empowerment study, this combined approach meant a qualitative assessment of empowerment, and a quantitative analysis of the relation between components of empowerment, and demographic characteristics of the study participants.

Before describing the development of my study design in more detail, I think it important to justify why I concluded the Alpha-1 community, and Alphas, are empowered, and why it became the basis for my study. Early in graduate school, I asserted in a paper that the Alpha-1 community is empowered, and I was challenged, not to prove the community is empowered, but, to define what I meant by empowerment. I thought it was clear what empowerment meant, but the professor noted she was referring to an anthropological approach that unpacks complex constructs. She suggested that I quantify empowerment in the Alpha-1 community, not just state that it exists.

However, my perception of empowerment did first arise from nine years of participant observation. Although, my employment did not overtly include research on the community, I deliberately, and consciously, undertook to observe the interaction of the scientists with each other, with diagnosed individuals, and with organizational leaders, and compared my experience

with previous experience of scientists. Over time, I also began to assess aspects of the patient and caregiver experiences, and began to read literature on empowerment. My training, and interest, in anthropology underlay my perceptions, and led to a valid, and carefully constructed, assessment of basic dynamics in the community. To justify my evaluation, I could provide numerous examples from my years in the community, and cite the many professionals who share my opinion. I could also point to the similarities between the definitions of empowerment in the literature, and common attributes in the Alpha-1 community. Instead, I will relate two stories that are typical, and representative, of both the personal and structural forms of empowerment and, that I think demonstrate the extent of empowerment I witnessed in this community. Instead, I will relate two stories that are typical, and representative, and that I think demonstrate both the structural and personal forms of empowerment I witnessed in the Alpha-1 community. I also include a particularly pertinent quote from Dr. Groft, whose perspective, as Director of the Office of Rare Disease Research at the National Institutes of Health, is particularly relevant.

The first example comes from my initial exposure to the Alpha-1 community. On my first day of work with the Alpha-1 Foundation (Foundation), I was told that in a month the Foundation would be convening its first scientific workshop, in Bethesda, MD, and that I would be organizing, and managing, this event. I had done this type of work before at the University of Miami, but it was challenging to organize people, and an event, without knowing anyone. So, the first thing I did was to assemble all the scraps of paper I was handed about the workshop, and prepare a proper attendees list, complete with contact information, titles, and affiliations. I noticed there seemed to be equal numbers of academic researchers, government people, and industry representatives. And, not just government drones, but the heads of agencies, like the Deputy Director of the United States Food and Drug Administration (FDA), Jack McCormick,

key researchers working under him in the Center for Biological Evaluation and Research (CBER) and Center for Drug Evaluation and Research (CDER), and several of the Division Directors of the National Institutes of Health (NIH), including those from the lung and liver divisions, genomic research, and office of rare disease research. The drug company representatives, according to their titles, were also individuals clearly high up in their company's chain of command. The meeting itself was a revelation to me, and my first indication of the power of the Alpha-1 community. To understand why, I need to put the experience in context.

After many years as an administrator in a dean's office at a local university, I went to work for a newly formed, not-for-profit organization, the Alpha-1 Foundation. The Foundation was established by three diagnosed individuals, and the President and CEO, John Walsh, my direct supervisor, is an Alpha with chronic and severe lung disease. The Foundation's office is located in Coconut Grove, FL. Coconut Grove, as opposed to downtown Miami's business district, and is known as a more casual business environment. The Foundation office was staffed, at that time, by only four other people, and although John and the CFO wore suits, the accountant wore a short sleeve shirt, and the secretary wore sandals, and very casual outfits to work. As opposed to my university experience, where the institutional structures and policies (like dress codes) are very formalized, I thought of the Foundation as a small, grass roots organization, with a visionary set of goals, but basically a small number of people advocating for a very rare, genetic condition. The Foundation office was a step above sitting around a kitchen table stapling newsletters, but, at first, it did not seem as professional, or organized, as a university or successful business operation.

My impression of a small organization flew out the window the minute I arrived at the hotel for the workshop. It was a four-star hotel, and our accommodations for the meeting were

luxurious. My boss, John Walsh, had told me ahead of time to dress in a suit, and, when the invitees arrived for the workshop, all of them were similarly dressed in formal business attire - the men in suits and ties, the women in dresses, or suits, and high heeled shoes. The workshop itself was conducted in a very formal and professional manner, with a chair, session heads, and designated speakers. There was, however, a strong sense of conviviality in the room, and during the breaks, and at lunch, it was obvious that this group of approximately 60 people knew each other well, enjoyed each other's company, and were going to work together on this problem of Alpha-1 drug development. During the breaks, and at lunch, I was introduced to many of the participants. Without exception, every person I was introduced to praised John Walsh, and the Alpha-1 Foundation. I thought it a bit odd at the time, but it was an ongoing experience throughout my nine years in the community, and a consistent theme in the positive relations of the Foundation with industry, and government.

The purpose of the workshop was to stimulate faster development of new aerosol drugs for Alpha-1 lung patients. Since available therapy was limited, at that time, concurrent development of several promising, new therapies was a central goal of the Alpha-1 community. After all the presentations were given explaining the delays in drug development, and during the discussion period when specific plans for drug development were being made, Jack McCormick, Deputy Director of the United States FDA, jumped up, and announced in loud voice (just like a game show host), "let's make a deal". Dr. Brantly, who served as chair of that workshop, remembers McCormick saying, "don't let perfection be the enemy of progress", which he interpreted as a comment on the laborious, and precise, process of drug approval. Given my unfamiliarity with the scientific content of the workshop, in those first weeks on the job, it is understandable that my memory of this incident is of the theatrical nature of McCormick's pronouncement.

However, Dr. Brantly, John Walsh, and I all remember, and agree, that McCormick provided the impetus, during that workshop, for the various stakeholders in drug development to move beyond a roadblock in the approval process that was holding up much-needed Alpha-1 therapy.

For, McCormick offered, during the workshop, the unprecedented option to competing pharmaceutical companies to concurrently develop aerosol drugs for Alpha-1. Traditionally, one company has exclusive rights to develop a certain drug, and it is only after the original license runs out that other companies can develop their versions of the same chemical compound. But, in the Alpha-1 community, there was a dangerous shortage of drugs that year, because of contaminated blood supplies used to develop the drug. It was critical that more companies start producing more drugs to meet the growing demand. Jack promised that the FDA would permit the fast-tracking of more than one application to develop aerosol therapy for Alpha-1. It is a promise that he kept, and it led to more drugs available to the Alpha-1 community in a shorter timeframe.

The workshop was a complete success in that it provided an understanding of the state of drug development at that time, and because all of the important stakeholders in drug development participated. But, the workshop also led to a change in how the FDA worked with rare disease groups, a successful outcome not originally envisioned as a goal of the meeting. In addition, the workshop was important to the newly organized Alpha-1 Foundation, as it validated the Foundation's role in negotiating deals between researchers, industry, and government. Another unique aspect of the workshop was that it brought researchers and industry people together, and talking, in a way that promoted working together to develop approvable drugs. Before that time, the Alpha-1 researchers had submitted drug trial proposals to industry that were returned over and over for changes; since that workshop the Alpha-1 researchers have been able

to coordinate with FDA employees during development of their projects, and work out potential problems, so that a faster approval is more often assured. According to Dr. McCormick, there were no other Voluntary Health Organizations at that time that had achieved this type, or level, of success in fast-tracking rare disease drug development (McCormick, Pers. Comm., 2002). In recognition of the unique role the Foundation, and John Walsh, play in drug development, the FDA awarded John a special citation in 2002.

Over nine years, I became better acquainted with Jack McCormick, who attended all the Foundation's scientific conferences and workshops. He told me, on more than one occasion, that John Walsh, and the research network created around the Alpha-1 Foundation, was the reason that the process of drug development changed, at least for rare diseases. Jack was wont to announce, during lavish awards ceremonies that the Foundation always organized during conferences, that "John Walsh is the smartest man I know." I always understood that compliment as Jack's way to praise the Foundation, and as his recognition of the power of a single patient. The kudos was especially meaningful, in the early years of the Foundation's growth, as our immediate organizational goal at that time was to establish scientific credentials. Jack was acknowledging that we were achieving the positive relation we desired with the scientific, and regulatory, communities. Jack was a key player in government approval of drugs at that time, and his opinion of John, and the Foundation, may help explain why I began to perceive this community as empowered, and why I began to wonder how Alphas, and the community, became that way.

My second story is about Marta. Marta asked that I talk about her as a way to explain how empowered a person can become in the Alpha-1 community. Marta is in her late fifties, and has served as an AlphaNet coordinator for many years. Although her educational background is

limited (high school), and her diagnosis of Alpha-1 was initially “overwhelming and scary”, she is very knowledgeable about the disease in lay terms, and plays an important role in cultivating the patient support network. She is, in her own words, “totally dedicated” to reaching out to newly diagnosed individuals, and spends many hours a day on the phone counseling other Alphas, and accelerating their process towards empowerment. She also served as a key informant for this study. But, Marta is not only Alpha-1 Antitrypsin Deficient; she has had breast cancer to deal with as well.

I spoke to Marta in July, 2008, as I had been told she has a re-occurrence of her cancer. Unfortunately, despite her complaints and requests for help from her physicians, and because of insurance issues, she was not diagnosed with the re-occurring cancer until it had already metastasized. She is in pain, and tired from her chemotherapy. She is also having her usual Alpha-1 related breathing problems, so talking to her was difficult - she kept gasping for breath while explaining about her cancer. When I asked her how long she had to stay on chemotherapy, she said, “the rest of my life”. But she did not sound sorry for herself, and although she acknowledged she had some pain, she did not complain.

When I mentioned that I needed to strengthen my argument about why I thought the Alpha-1 community is so empowered, Marta was incredulous. She asked me, “Did you tell them about the coordinators....Alphas taking care of each other....how many hours a week the coordinators work? Did you tell them about the Registry.... the educational days?” When I said all those things were in the study, Marta told me,

Write about my situation. Explain that [despite having Alpha-1 and breast cancer], I continue to FIGHT every day of my life, and for a cure for all Alphas. My strength comes in doing something about what I have. I’m not a victim. I’m lucky to be part of this community.

Although Marta pointed to the structural components of empowerment in the community, ultimately it is her individual choice to be empowered rather than be a victim, and in that sense, she does represent the majority of Alphas I have met and interviewed. However, she is an AlphaNet coordinator, and as such, is one of those who have made a very strong commitment to community involvement. She also represents someone diagnosed many years earlier and further along in awareness, than perhaps the majority of Alphas in the United States. Whether Marta represents all Alphas, or just the high end of the empowerment scale in Alpha-1, may be answered in future studies. What is true, is that she has an empowered attitude, is a visible representative of the Alpha-1 community, and her strength, despite her condition, is similar to what I observed among the majority of Alphas I met. Meeting people like Marta and John Walsh made me want to explain what made individuals in this community so empowered.

I should also note several other attributes of the Alpha-1 community that are unique, for a voluntary disease community, and I identified as variables of empowerment. These attributes are, 1) extremely effective (and charismatic) leadership; 2) the use of a profitable business model for a not-for-profit organization; 3) the amount and quality of resources available in the community for both scientists and patients; 4) an effective advocacy program and numerous, notable accomplishments in that area; 5) the effective networking between patients, organizations, government, and industry; and, 6) a very well defined community identity complete with nickname (Alphas), color (purple), and ritualized, annual events and meetings. Each of these attributes is described in more detail in Chapter 2: Leader of the Pack: Ethnography of Alpha-1 and the Alpha-1 Community in the United States. Taken together, the structural and personal forms of empowerment I observed in this community, assessed against the literature on empowerment, led me to my belief that empowerment in the Alpha-1 community is more

pronounced, and visible, than other rare and genetic disease communities with comparable numbers of diagnosed individuals.

My opinion about empowerment in the Alpha-1 community is not unique, or overstated, and there are, as noted earlier, numerous examples that I could provide to demonstrate that this opinion is a widely-held view. I, therefore, conclude this argument by quoting from a lengthy email that I received on September 10, 2008, from Stephen C. Groft, Pharm. D., Director, Office of Rare Diseases, National Institutes of Health. When asked if he could confirm my impression of empowerment in Alpha-1, he wrote:

Thank you for the opportunity to discuss the success of the Alpha -1 Foundation. Without any doubts, I can say that the Foundation under the leadership of John Walsh is one of the truly outstanding patient advocacy organizations in the United States.

John has developed the Foundation to be responsive to the informational and psychological needs of the patients and their families. Information for patients and researchers has been developed and is readily available on the Foundation website on topics such as standards for diagnosis and treatment, education opportunities, and ways that you can help. Presence and participation of the patients and the families at the annual meeting is remarkable.

I have seen the Foundation grow in numbers and in impact upon government agencies as their voices are now being heard by program officials who recognize the Foundation as a truly successful and active Voluntary Health Organization with a commitment to research and the discovery of treatments for Alpha-1 Antitrypsin deficiency.

The Foundation also makes available direct service to health care providers. This information is essential to the effective treatment of patients. Not every patient organization is able to provide this extensive amount of information.

The foundation is viewed as a model for other patient advocacy groups to follow. I have referred numerous newly established patient advocacy organizations to John Walsh the President and CEO of the Foundation to learn how to establish an organization and expand the activities to enable an increase of research emphasis and to provide needed services to patients and families. John has also presented numerous speeches about the functions and activities of the Foundation to national medical and professional associations as well as to national patient organizations.

I hope this helps explain the significant contributions that the Foundation provides to the rare diseases community affected by Alpha- 1 Antitrypsin Deficiency and the level of accomplishment of the Foundation. Please let me know if additional information is needed.

Dr. Groft's comments are typical of the complimentary attitude government officials, and industry representatives, have always shown towards John Walsh, the Alpha-1 Foundation, and, by extension, the Alpha-1 community. His willingness to provide this information is also typical, and a further indication of the level of influence John Walsh has created for all members of the community, including myself. And, Dr. Groft's remarks may put my own impressions of the Alpha-1 community, and assessments of my qualitative data, into proper perspective as unique among the rare disease communities.

Developing the Research Questions

This empowerment study is envisioned as the first in a series of studies relating to quantifying, and measuring, empowerment in the Alpha-1 community. This long term research plan is motivated by a series of questions that arose during my years of working with the Alpha-1 community. My doctoral research explores the initial question, "What constitutes empowerment in the Alpha-1 community?" But, to answer that question I needed to be able to quantify, and measure empowerment, so I expanded the initial question to explore broader topics of inquiry, i.e., the underlying methodological and theoretical implications of my research. I considered the diverse ways of measuring qualitative data, the challenges of recruitment, how much community participation affects study outcomes, and the translational aspects of mixed methods. These led to additional questions beyond my initial question of, "What constitutes empowerment in the Alpha-1 community?"

The study design addresses both the initial five questions, and the additional issues that arose. This includes questions like, how could a complex social construct like empowerment be quantified (i.e., how do I operationalize this research question into a study design)? Which definitions of empowerment should I use, and who should provide those definitions? What is the best method to obtain a community-derived definition of empowerment? How do I recruit, who

do I recruit, and would the recruitment method result in a representative sample? How do I turn the qualitatively derived data into a useful format for statistical analysis? Which statistical approaches would be appropriate for measuring the data? How should I interpret the data – what is the best way to derive meaning from the numbers? And, since a statistical analysis may either confirm or refute the qualitative assessments, how should I evaluate any discrepancies between the qualitative and quantitative results?

To answer these questions I began by reviewing relevant literature, and refining my questions about empowerment in the Alpha-1 community. The development of these research questions required unpacking the social construct of empowerment into distinct components. Unpacking the concept empowerment also involved operationalizing such components as awareness into distinct stages of learning, and defining networking as the quantity, and quality, of relationships, in order to measure them (Handwerker, 2003). .

This research is modeled, in part, on scientific studies, and is based on several research questions, my years of observation, and a review of literature. I had developed preliminary research goals, during my years of participant observation that I modified after my literature review. However, for a workable study design, I had to further refine the research goals, and turn them into questions that were testable, and that would answer specific questions about empowerment in the Alpha-1 community. Further changes to the research questions followed early focus group data analysis, and continued throughout the study. This evolution of the questions, and research goals, was appropriate within the paradigm of a grounded theory approach, and gave me the freedom to adjust the theoretical parameters of the study as I gained more information about the community (Charmaz, 1994; Glaser and Strauss, 1967; Glaser, 1992; Irurita, 1996; Kearney et al. 1995; Maijala et al. 2003; Strauss, 1992). I applied this approach as

it fit the situation – empowerment had been defined many times in the literature, but a definition relating specifically to a rare disease community needed to be developed as a reiterative process between me and members of the community. I recognize that there are theoretical debates about grounded theory; however, my focus is on the utility of the techniques developed in grounded theory, and their usefulness for my study.

A description of the basis for each research question, and implementation strategies for each research goal, follows.

Research Question 1: Is patient empowerment a visible sign of a shift in the hegemonic relation between physicians and patients? Key elements in a shifting hegemony in the construction of knowledge in Alpha-1 relate to the increased use of the Internet, the institutionalization of patient empowerment as a social movement, and the increased effectiveness of patient advocacy groups for specific genetic disease communities. This question was explored through interview questions about responsibility and control of chronic disease, the quality of physician-patient interaction and power relations, and the patient's level of trust in the knowledge their medical providers possess.

Research Question 2: Does business expertise, and organizational leader's ability to garner a seat at the table in policy making and accelerated drug development, make these organizations, and their leaders, structurally powerful? Patient-driven organizations in the Alpha-1 community are led by particularly effective and knowledgeable leaders. This question was operationalized as interview questions about involvement of Alphas in policy making, membership in organizations, advocacy activities, network relations (number of Alphas they know), and attitudes about assisting, representing, or leading other Alphas.

Research Question 3: Are patients with access to resources in the Alpha-1 community more likely to become empowered on a personal level? And, are the components of personal empowerment effective disease self-management, increased awareness of the condition, and effective communication with medical staff? The earliest signs of empowerment in Alpha-1 are the evolution of awareness about the condition, which may depend on good communication with medical staff, followed by improved disease management. These research questions were explored through interview questions about the level of disease self-management, scaled responses on understanding of condition, and evaluation of the quality of interaction, and communication, with health professionals.

Research Question 4: Do domains of empowerment form a unidimensional scale (Figure 1-1). There is a qualitative difference between Alpha leaders and the general Alpha population, however, structural empowerment of a few benefits the entire community. This question was explored through a statistical analysis of the components of empowerment, to verify whether or not certain forms of empowerment preceded other forms.

Choosing the Alpha-1 Community

A primary motivation to study the Alpha-1 community was my access to, and familiarity with, this closed population. I had already learned the local lingo, (the medical terminology associated with Alpha-1), knew people from all segments of the community, and had played a visible, and central, role in the community that facilitated my recruitment efforts.

A secondary motivation was the role the Alpha-1 community has played among other disease communities in the United States. I perceive it is a model community - it represents the ideal of an effective, and productive, network that serves both the patient and biomedical communities. Answering questions about empowerment in Alpha-1 might be of great benefit to

other communities as they develop their resources and infrastructures, and give members of the Alpha-1 community an additional source of pride and positive identity.

However, like many other researchers, I am drawn to a particular research topic because of its relevance to me intellectually and personally. I was interested in studying the Alpha-1 community when I entered school, but after readings for a medical anthropology graduate course I had a clearer understanding of how to approach an ethnographic study of a disease group, and how to tell such a complex story. There were several medical anthropology studies that particularly stimulated my interest in creating an ethnography of the Alpha-1 community, such as Cohen's study of Alzheimer's in India (1998), and Lock's study of menopause in Japan. But, it was the approach used by Mol, in *The Body Multiple* (2002), to describe atherosclerosis, that I found the most intriguing. Her study of the variety of enactments and locations of atherosclerosis strongly influenced my thinking about the Alpha-1 community, and gave me a framework for how best to characterize such a variable condition as Alpha-1.

Mol takes the reader through a number of different locations in a hospital relating to the experience of atherosclerosis, and gives a detailed description of the enactments, or types of activities, that occur in the patient's life, and in the medical setting leading up to a diagnosis and treatment. Atherosclerosis, like Alpha-1, does not reside in one place in the body, but is a condition affecting many different organs and systems. Diagnosing atherosclerosis, and Alpha-1, requires discerning a particular set of markers (measurements of body function) as indicative of a condition, ordering a test, interpreting the test results, communicating with the patient and their family members, treating the condition, and together with the patient, trying to achieve a cure or lessen the negative impacts of disease progression. These events occur in different places, and involve a variety of scientists, clinicians, nursing staff, and lay individuals, each with their own

way of describing the experience of atherosclerosis, or Alpha-1. Mol's book provided me with a model for understanding a complex medical condition: first, unpack the various enactments and locations, describe the different languages used to describe the enactments, and then reassemble the elements into one story. Although I already understood Alpha-1 as a complex condition, I felt Mol's book provided the most useful model for how I could explain Alpha-1, and the many people involved in the Alpha-1 community.

On a more personal level, I was drawn to studying a lung disease community because my mother, at that point in my life, had advanced lung disease, and I seemed to be developing some lung-related problems. Although my mother was diagnosed with Chronic Obstructive Pulmonary Disease (COPD), not Alpha-1, I knew that through my involvement in the Alpha-1 community I would discover more about lung biology, hear about the latest therapies and treatments, and learn how I could manage our conditions. Lawrence Cohen, in his study on Aging in India, best sums up my personal motivation for studying a lung disease community. Although his book focuses on the aging process, I saw myself in his comment, "The natives of the country of the old, for those of us who visit as tourists, are our own future selves" (Cohen, 1998). In a community of individuals grappling with fatal lung disease, I could become more aware, and empowered, about my own family's health issues.

I also aspire, as an applied anthropologist, to intervene in the lives of my study subjects through my research, and contribute to the body of knowledge in anthropology and medicine relating to the social aspects of disease management. The interventional potential of my research became a stronger motivation as I read literature on empowerment, medical anthropology, and Science and Technology Studies (STS). Like many before me, I was drawn with fascination into the world of science and technology, and hope my research on existing, and shifting, hegemonies

in a rare, genetic disease community might contribute to a larger body of knowledge in medical anthropology and linguistic anthropology.

Summary of Research

The following chapters describe the fieldwork methods, types of analyses, and the results of the research. However, the research results are prefaced by an ethnographic description of the condition Alpha-1, and the Alpha-1 community, in Chapter 2, Ethnography of the Alpha-1 Community. The ethnography is essential background information for understanding the variability of the genetic condition Alpha-1, and, as a framework for my research questions about the empowerment of community members. The ethnography includes details about specific enactments of Alpha-1, such as the patient's perspective of illness, the structure of laboratory culture, and the complexities of laboratory procedures. The ethnographic information also covers the Alpha-1 community, its evolution and current structure, and the characteristics, interactions, and relations among the stakeholders in the community.

Chapter 3, Theoretical Framework, describes the theories used as the context for the study, the anthropological fieldwork methods, and the analytic techniques employed in the linguistic phase of the study. I describe recruitment procedures, the format of focus group sessions and interviews, my experience of hiring in, and provide a self-assessment of the level of communicative competence I had gained in the community. Through examination of the philosophical and pragmatic framework underlying my research, I seek to situate my study within the larger body of knowledge in anthropology and linguistics, and to acknowledge the influence of previous research on the design of my study.

Chapter 4, Narrative Analysis, presents the results of the analysis of interview data, and focus group sessions, and a preliminary definition, and model, of empowerment. The analytic techniques that are described led to development of a codebook that includes themes, and

overarching meta-themes, relating to the experience of Alpha-1. These thematic components, in turn, became the basis for the design of a statistical analysis.

Chapter 5, *Linguistic Analysis of Medical Encounters*, describes the fieldwork conducted in medical offices, and the linguistic analysis of those encounters. The chapter also explains the purpose, and format, of the ethnography-of-speaking approach, and concludes with the results of the analysis. This phase of the research also included an assessment of the hegemonic relations between physicians and patients, based on observations of their verbal and non verbal interactions during medical office visits.

The statistical analysis is detailed in Chapter 6, *Quantifying Empowerment*. This chapter includes description of how qualitative components were translated into quantitative data, the statistical approach used to analyze the data, and concludes with a discussion of the statistical significance identified between specific components of empowerment, and how quantitative results were correlated with the qualitative results.

Chapter 7, *Empowerment is Simple, Empowerment is Complex*, presents the conclusions of the study, the relevance of the findings for the Alpha-1 community, and for other rare disease communities, and the possible benefit the study results may have for chronic lung disease patients. Chapter 7 also includes a description of future research studies that will be based on the results of this research.

The study resulted in recommendations that could promote many different types of empowerment, and a view of empowerment as both an individual experience and an experience that is shared by communities. These recommendations are based on identified, and effective, components of empowerment in the Alpha-1 community, and could assist this community, and

other patient groups, to achieve maximal levels of empowerment, or augment current efforts at empowerment into the most effective channels.

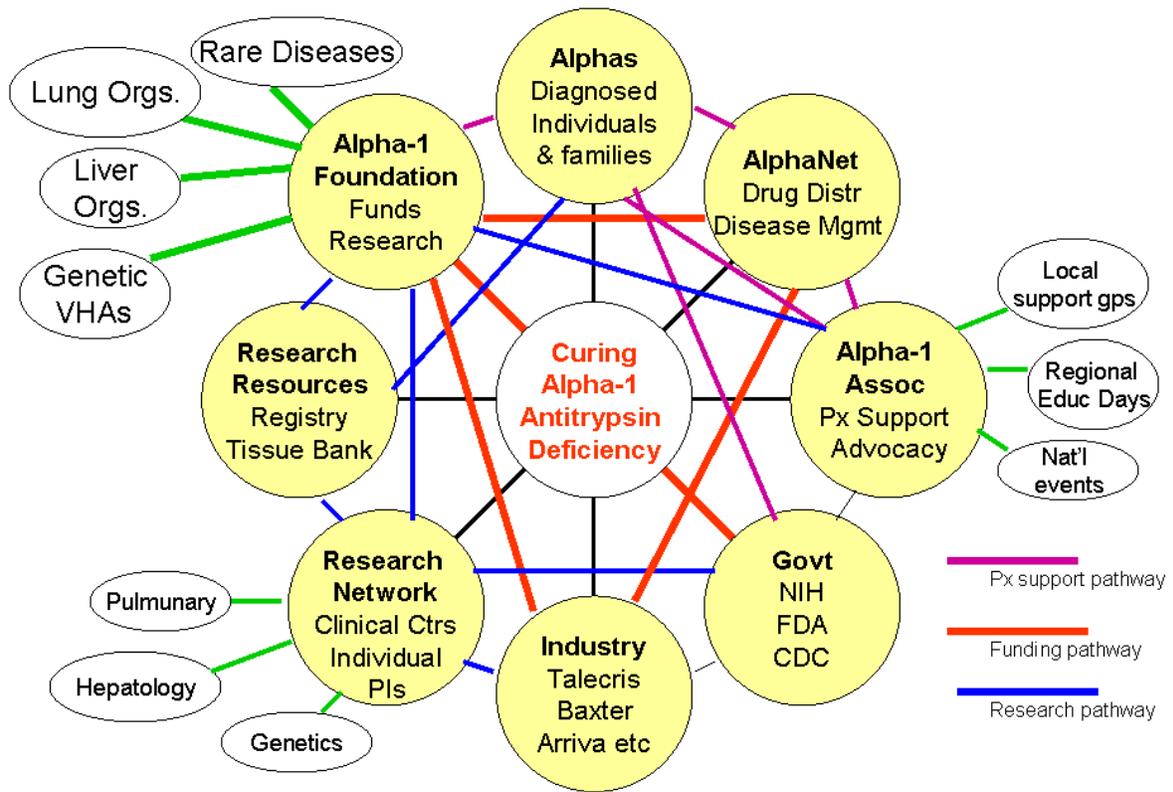


Figure 1-1. Network relations in the Alpha-1 community

Table 1-1. Stakeholders in the Alpha-1 community

User Group	Characteristics	Activities	Relation to Alpha-1
Diagnosed individuals	Newborn liver patients (with jaundice)	Illness, failure to thrive, hospitalization	Patient
	Pediatric liver patients	Illness, transplantation	Patient
	Adult liver patients	Illness, transplantation, research, advocacy	Empowered person or disabled patient
	Pediatric lung patients (with lung disease)	Illness, hospitalization	Patient
	Adult lung patients (dyspnea, lung disease)	Illness, disease mgmt, advocacy, research	Empowered person or disabled patient
Alpha family members	Spouses, Partners	Provide care giving, emotional & economic support or desert family	Marital responsibility
	Children, Grandchildren	Support, test for Alpha-1 or denial	Familial involvement Genetics
	Siblings	Support, test for Alpha-1 or denial	Familial involvement Genetics
	Parents	Care giving, emotional & economic support or desertion	Parental responsibility, Genetics
	Cousins, Nieces, Nephews	Support, test for Alpha-1 or denial	Familial involvement Genetics
Caregivers	Family member	Care giving, economic responsibilities	Familial responsibility
	Professional caregiver	Care giving, physical support, chores	Commercial
	Friend	Care giving, emotional support, chores	Social
Voluntary Health Associations (VHA)	Lung Organization staff	Provide support, education, advocacy, fund research	Scientific and Educational
	Liver VHA staff	Provide support, education, advocacy, fund research	Scientific and Educational
	Genetic VHA staff	Provide education, advocacy, networking	Scientific and Educational
Medical Professionals	Bioethics VHA staff	Provide education, advocacy	Philosophical
	Liver and Lung Doctors	Diagnosis, medical treatment, education	Biomedical and Educational
	Nurses, PAs	Medical treatment, education	Biomedical and Educational
	Respiratory Therapists	Therapy, education	Biomedical and Educational
	X-ray technicians, physicians	Diagnoses	Biomedical
	Transplant surgeons	Surgery	Biomedical

Table 1-1. Continued

User Group	Characteristics	Activities	Relation to Alpha-1
Researchers	Pulmonology/ Allergy	Study lung, upper airways	Biomedical
	Hepatology/Gastroenterology	Study liver related conditions	Biomedical
	Basic Science: biology, pathology, chemistry	Study molecular & chemical processes	Academic - Scientific
	Genetics	Study genes, gene therapies	Academic - Scientific
	Bioethics	Study ethical issues related to Alpha-1	Academic - Philosophical
	Social/behavioral science, psychology, anthropology	Study social impacts of Alpha-1	Academic- Social
	Public Health: epidemiology, statistics, economics	Investigate prevalence, costs of Alpha-1	Academic - Numerical
Government Employees	Laboratory personnel	Diagnoses, research, tissue and DNA banking	Biomedical
	National Institutes of Health	Support, prioritize and manage research	Scientific
	Centers for Disease Control	Conduct research	Biomedical/Scientific
	US Food & Drug Administration	Oversee safety and use of drugs	Biomedical/Scientific
	Presidential Advisors Legislators	Advise executive branch Prioritize needs, appropriate funds	Political Political
Business Interests	Lawyers and Judges	Defend patient rights, rulings	Legal
	Pharmaceutical personnel	Research and manufacture drugs	Biomedical and Commercial
	Medical Devices personnel	Research and manufacture devices (portable O2, aerosol delivery devices)	Biomedical and Commercial
	Health Management personnel	Distribute drugs, nursing services	Biomedical and Commercial

	ACC RES	AWA	DSM	RES PAR	ORG MEM	ORG LEA	PX ADV	MSO	POL
	Personal Empowerment					Structural Empowerment			
a	1	1	1	1	1	1	1	1	1
b	1	1	1	1	1	1	1	1	0
c	1	1	1	1	1	1	1	0	0
d	1	1	1	1	1	1	0	0	0
e	1	1	1	1	1	0	0	0	0
f	1	1	1	1	0	0	0	0	0
g	1	1	1	0	0	0	0	0	0
h	1	1	0	0	0	0	0	0	0
i	1	0	0	0	0	0	0	0	0

Figure 1-2. Personal and structural empowerment as a unidimensional scale. 1 = Yes, 0 = No; ACC RES = Access to resources; AWA = Awareness; DSM = Disease Self-management; RES PAR = Research participation; ORG MEM = Organizational membership; ORG LEA = Organizational leadership; PX ADV = Patient Advocacy; MSO = Membership in supra-organization; POL = Policy maker. This figure is a visualization of the temporal progression from personal forms of empowerment, such as access to resources, to structural forms of empowerment, like being an organizational leader. Using a Guttman scale implies that empowerment is not a unitary condition that can be described or answered with one descriptor, but is an evolutionary process. The scale also demonstrates which forms of empowerment form the dimension, personal empowerment, and which components form the second dimension, structural forms of empowerment.

CHAPTER 2 ETHNOGRAPHY OF THE ALPHA-1 COMMUNITY

What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin Deficiency (hereafter referred to as Alpha-1) is a rare, genetic condition that is estimated to affect 100,000 individuals in North America (Abboud et al. 2001; Campbell, 2000; Campos et al. 2005, Crystal, 1989; Crystal et al. 1989; deSerres et al., 2006a, 2006b, 2003; deSerres, 2002; Eriksson, 1963, 1996, 1999; Hersh et al., 2004a,b; Hutchinson, 1988; Luisetti and Seersholm, 2004; Miravittles et al., 1998; Silverman et al. 1989). The deficiency primarily affects soft tissue, like the lungs, and the majority of those diagnosed are lung affected. However, Alpha-1 can also affect the liver, (and more rarely, the skin or eyes), and a smaller proportion of those diagnosed have actual liver disease. Of the 3,376 diagnosed individuals enrolled in the Alpha-1 Research Registry, 68.08% have one or more lung symptoms, but only 328, or 9.92%, have one or more liver symptoms.¹

The lung disease associated with Alpha-1 appears as chronic bronchitis, asthma, severe allergies, or recurrent pneumonia, and the patient is often misdiagnosed as having one of these other lung, or allergy, conditions (Stoller et al. 2005, 2007a). Adults, with liver disease from Alpha-1, may show elevated liver enzymes, but their liver disease is considered a Non-alcoholic Steatotic Condition, or NASH, and their deficiency of Alpha-1 is not confirmed until a biopsy is conducted, and cirrhotic liver tissue found (Teckman, 2007; Teckman and Lindblad, 2006). Adult liver patients are often not diagnosed as Alpha-1 until after death.

Alpha-1 is usually diagnosed when an individual is symptomatic for lung or liver disease, and in some rare cases has an eye or skin condition, or is a family member of a diagnosed

¹ Symptoms are self-reported; individuals may list more than one symptom; some provided no symptoms. (Streng, 2008, Alpha-1 Research Registry Quarterly Report, Medical University of South Carolina).

individual (Campos et al. 2005; Celedon et al. 2000; Dahl et al. 2001; deSerres 2003; Eriksson 1987, 1999; Fleming et al. 2001; Luisetti et al. 1999; Piitulainen et al. 1997; Piitulainen and Sveger 1998; Navickis and Wilkes 2004; Stoller et al. 2005, 2007a; Sveger and Mazodier, 1979; Sveger and Thelin, 2000; Wencker, 2000). Symptoms are variable: newborn infants or children may develop severe jaundice; adults may have unexplained liver disease not caused by alcohol; or children and adults may have chronic and increasing lung and upper airway diseases (Abbott, 1988; Anonymous, 1997; Anonymous, 2003; Buist, 1990; Chappell et al. 2008; Cox, 1978; Eriksson, 1987; Fischer et al. 2000; Knebel et al. 2000; Needham and Stockley, 2004; Piitulainen and Eriksson 1999; Piitulainen and Sveger 2002; Roberts et al. 1984; Sandhaus, 2006, 2004; Seersholm and Kok-Jensen 1998; Stoller 2004; Teckman 2007; Teckman and Lindblad 2006; Tanash et al. 2008; Vreim et al. 1998; Zhou and Fischer, 1998, 2000; Fregene and Stolk, 2008; Hogarth and Rachelefsky, 2008). As noted above, since the pulmonary symptoms are outwardly similar to other conditions, Alpha-1 is frequently misdiagnosed as asthma, Chronic Obstructive Pulmonary Disease (COPD), or allergies (Stoller et al. 2007a, 2005, 2004; McElvaney et al. 1997; Mulgrew et al. 2007). Since it is a rare condition many primary care physicians and pulmonologists do not consider Alpha-1 as a possible cause of their patients' lung disease, or they are reluctant to order a test for a condition with no known cure. Liver specialists, on the other hand, are more apt to order a test for Alpha-1, especially among their pediatric patients (Chappell et al. 2008; Teckman, 2007; Bakula et al. 2007).

Diagnosis is done through a variety of modalities: the preliminary test is a simple blood test to determine the level of Alpha-1 in the bloodstream (lower levels indicating deficiency), followed by genotyping and phenotype to determine the particular form of Alpha-1 (the phenotypic variant), lung x-rays and CT scans to determine the extent, and type, of lung damage,

and liver enzyme tests and liver biopsies to evaluate liver function, and confirm the presence of cirrhosis (Brantly, 2006; Carpenter et al. 2007; Costa et al. 2000; Eriksson, 1987; Guest and Hansell, 1992; Kaczor et al. 2007; Ljujic et al. 2008; Ortiz-Pallardo et al. 2000a, b; Pittschieler 2001; Shaker et al. 2004; Stockley and Campbell, 2001; Stoller et al. 2007a, 2005; Parr et al. 2008; Ferrarotti et al. 2007, 2008). Lower levels of Alpha-1 in the blood, and certain phenotypic variants, are known to be higher risk factors for developing either the lung or liver disease associated with Alpha-1 (Fregonese and Stolk, 2008; Brantly 2006; Sandhaus, 2006, 2004; Stolk et al. 2006; Teckman 2007; Teckman and Lindblad 2006). However, the precise development of the disease is variable, and some with the highest risk phenotype are not sick whereas others, with a lower risk, are quite ill (Black and Kueppers, 1978; Coakley and Taggart, 2001; Dahl et al. 2001, 2002, 2005; Hersh et al. 2004a, b; DeMeo and Silverman, 2004, 2003; Scott and Enger, 2006; Seersholm et al. 2000; Zhou and Fischer, 1998, 2000; Sandford et al. 2001, 1999; Graziadei et al. 1998; Hutchinson, 1988; Pelham et al. 1985; Shigeoka et al. 1976). Although over 3,000 individuals have enrolled in the Alpha-1 Registry, it is estimated that approximately 5,000 individuals in the United States, and an equal number in Europe and the Middle East, have been diagnosed with Alpha-1 (deSerres et al. 2007; Luisetti and Seersholm, 2004).

Biomedical Descriptions

Since its discovery in 1963 (Laurell and Eriksson 1963; Eriksson, 1990), descriptions of Alpha-1 Antitrypsin Deficiency in the biomedical literature usually begins with the date of its discovery, and notation of its genetic position at locus 14 among a class of proteins called SERPINS (Altay, 1973; Blank and Brantly, 1994; Brantly, 1997; Brantly et al. 1988a, 1988b; Byth et al. 1994; Carrell 2004; Carrell and Lomas, 1997; Cox 1997; Cox and Mansfield 1987; Cox et al. 1985; Crystal et al 1989; DeMeo and Silverman 2004, 2003; Gooptu and Lomas, 2008; Lieberman et al. 1986; Liu et al. 1998; Lomas, 2007; Lomas and Parfrey 2004; Lomas and

Silverman et al. 2001, 2000, 1992; Silverman and Palmer 2000; Marcus et al. 1998; McElvaney et al. 1997; Mulgrew et al. 2007; Seixas et al. 2001; Wood and Stockley 2007). Many articles characterize Alpha-1 as monogenetic, as a metabolic condition, or as a genetic cause of emphysema, and there is usually some discussion of its genetic transmission, and inheritance patterns (Axelsson and Laurell, 1965; Bamforth and Kalsheker, 1988; Barnes, 1999; Cox 1978; Cox et al. 1985; DeMeo and Silverman 2003, 2004; Elzouki et al. 1997; Fagerhol, 1968; Flint and Mott, 2001; Greene et al. 2008; Kalsheker, 1994; Lomas and Silverman, 2001, Sharp et al. 1969; Silverman et al. 2001).

Secondarily, Alpha-1 is characterized epidemiologically. Prevalence statistics are cited, and most articles note the higher penetrance, or number of people identified with the condition, among North European, Caucasian, or 'white', populations, and the rarity of Alpha-1 among Asians or 'blacks' (Stoller et al. 2007; Campos et al. 2005, Balbi et al. 2007; Stockley et al. 2007, 2002; deSerres et al., 2007, 2006a, 2006b, 2003; Stolk et al. 2006; Navickis and Wilkes, 2004; Ioachimescu and Stoller, 2005; Corbo et al. 2003; Miravittles et al. 2002; deSerres, 2002; Takabe et al. 1992; Seyama et al. 1995; Silverman et al. 1989). Occasionally, one hears of the origins of Alpha 1 in romantic terms: such as the oft-repeated story that Alpha-1 was spread via Viking explorations (deSerres, Pers. Comm. 2002). The scientific articles are much more constrained in their theories about the genetic patterns, and, primarily have focused on identification of specific haplotypes in Europe and South Africa, or describe the slight genetic differences in prevalence from the North to the South of Spain, or between 'blacks' and 'whites' in South Africa (Byth et al. 1994; deSerres, 2003, 2002; Seixas et al. 2001; Gaillard et al. 2004; Smith, 1994).

The pathogenesis, or development of disease in Alpha-1, is then generally described. The development of different types of disease is detailed, and a description of the particular symptoms relating to the lung or liver disease is provided. Articles on disease development also describe current treatments, the development of new therapies, and, generally include some mention of family risk factors, and recommendations for targeted testing of family members and symptomatic individuals (Anonymous, 1997; Anonymous, 2003; Barker et al. 1994; Dirksen et al. 1999; Eriksson 1983; Gildea et al 2003; Gottlieb et al. 2000; Hill et al. 1999a, b; Hubbard and Crystal 1990a, b; Fregene and Stolk, 2008; Hogarth and Rachelefsky, 2008; Knebel et al. 2000; Köhnlein and Welte 2008; Lieberman 2000; Miravittles et al. 1994; Needham and Stockley, 2004; Perlmutter et al. 2007; Sandhaus, 2006, 2004; Seersholm et al. 1997; Silverman and Speizer 1996; Snider, 1989; Stocks et al. 2006; Stockley et al. 2002; Stolk 2006; Stoller 2001; Stoller and Aboussouan 2004; Stoller et al. 2007b, 2002; Stone et al. 1995; Taraseviciene-Stewart and Voeklel 2008; Teckman 2007; Teckman and Lindblad 2006; Tanash et al. 2008; Turino, 1991; Viskum and Kok-Jensen, 1990; Vogelmeier et al. 1990; Wencker et al. 2001, 1998; Wewers et al. 1987).

There is a separate body of scientific literature dealing with the basic processes involved in Alpha-1 production, transport, and molecular function (Zhou and Fischer 1998, 2000; Song et al. 1998, 2000, 2001; Sifers et al. 1992, 1990 1989a, b; Lomas et al. 1992; Lomas, 1994; Aldonyte et al. 2008; Lin et al. 1998; Janciauskiene et al. 2002; Crystal et al. 1989; Mallya et al. 2007; Termine et al. 2005; Gooptu and Lomas, 2008; Wencker and Brantly, 2005). To molecular scientists, Alpha-1 is considered a conformational disease, and classifies it as one of several genetic conditions that are caused by a misformation of a protein (Carrell and Lomas, 1997; Carrell, 1990). Other conformational diseases include Alzheimer's disease, Parkinson's disease,

and Cystic Fibrosis. Many of the basic studies in Alpha-1 have applicability to these other genetic conditions, which is a fruitful approach during an era of diminished funding for science.

However, a great deal is still unknown about both the basic science relating to Alpha-1, and the pathogenesis of disease. I asked Robert A. (Sandy) Sandhaus, MD, PhD, a recognized expert in Alpha-1 research and clinical care, to articulate some of these gray areas, the unknowns, about Alpha-1. Dr. Sandhaus, or Dr. Sandy as he is called in the community, is Medical Director of AlphaNet, and one of his responsibilities is to give orientation sessions about Alpha-1 to new employees, and newly diagnosed individuals. He is also one of the first individuals in the United States to study Alpha-1, and the only person, until recently, whose medical dissertation was about Alpha-1. He seems to love talking about Alpha-1, and we have had many conversations through the years about the science of Alpha-1.

He readily agreed to an interview, and I prepared by reviewing a lay article in an Alpha-1 magazine, that describes how the lung is damaged (Alpha-1 to One, Summer, 2004):

“Scientists know how Alpha-1 affects the lungs only in the most obvious way. It causes lung damage known as emphysema. But to develop treatments that work, they need to know more...In the lungs, the body produces proteases, which attack invading microbes and serve other important functions. The body also produces anti-proteases such as AAT, which supposedly protects tissues from collateral damage caused by proteases’ ‘friendly fire’. According to conventional wisdom, lung damage occurs when the body produces too little anti-protease to provide sufficient protection”.

The article quotes Dr. Trapnell, the Scientific Director of the Alpha-1 Foundation, who notes in the article that effective treatments will not be developed until the scientists “understand exactly what is occurring on the molecular level” (Trapnell, 2004). To achieve that understanding, Alpha-1 pulmonary researchers are focusing on three basic questions: what precise part of the lung is attacked? Which protease does the attacking? And where and why does inflammation occur?

Based on the information provided in the article, I prepared questions for Dr. Sandhaus, and arranged a time for our phone interview. Portions of the interview follow.

SF: “Sandy, I read Trapnell’s article, and have a bunch of questions based on specific things he says in the article. Like, are proteases produced in other parts of the body?”

It occurred to me that a therapy that altered the protease/antiprotease balance for the sake of improving the health of Alpha-1 patients might unintentionally change other protease interactions, but this proved to be a typical lay misunderstanding of a broadly stated scientific fact. Dr. Sandhaus clarified:

SS: “Proteases are a broad sub-set of proteins called enzymes, and all enzymes facilitate chemical reactions, like the activity of amylase in saliva in breaking down starches into sugars. Proteases are a special class of chemical reactions that break down proteins into amino acids. Each protease has an active site, a target group that it likes, that it breaks apart. There are approximately 1,000 proteases and every cell, not just lung cells, has proteases.

SF: “Do we know yet which protease does the attacking in the lungs? Is it proteases from white cells, from macrophages, or from both?”

SS: Research has shown that serine proteases in neutrophils, and macrophages were involved.

SF: What about the cause of inflammation in the lungs. Is it from the protease/antiprotease balance, polymerized AAT in the lungs, or abnormal AAT produced in epithelial cells?

SS: This is still unknown, but David Lomas and Sabina and Ewa Marszal [researchers I know in Sweden, England and in Washington, D.C.] have shown that similar polymers were found in blood vessels, and the lungs (Lomas and Parfrey, 2004; Gooptu and Lomas, 2008; Janciauskiene et al. 2002; Marszal et al. 2003).

But we do know which part of the lung is attacked.

SF: Who’s working on that?

SS: Steve Shapiro. His studies have focused on different areas of damage including the wall of the air sac (Shapiro, 2007; Shapiro et al. 2003; Churg et al. 2002), and Rubin Tudor is looking at the underlying tissue scaffolding and interstitial areas (Cantor and Turino, 2004; Snider, 2004; Turino, 2002). There’s also some research on the blood vessels that feed the scaffolding. I don’t remember who, but you can look them up (Voelkel et al. 2007; Tudor and Yun, 2008; Yoshida and Tudor, 2007).

Dr. Sandhaus then veered off into a half hour discussion of the “DNA central dogma” in Alpha-1, intoning “unfolding, transcription, translation and processing” as if it were a mantra. I felt a bit lost with his detailed description of the molecular processes. However, it was obvious that Dr. Sandhaus’ long explanation about genetics was intended to address my questions, but many of the details he provided were deeply embedded in an understanding of biology that I still do not possess. What did become clear to me, during the interview, is that an understanding of the relevant molecular processes in Alpha-1, and the development of Alpha-1 disease, are still areas requiring a great deal of research before effective therapies are developed.

The interview with Dr. Sandhaus also highlighted a common frustration many Alphas had told me about, and that I also experience – the scientists are clearly excited about what they know, and the implication of this knowledge for Alpha-1 patients, but are unable to convey exactly what is so exciting, or put it into a context that the patients can grasp. In addition, the scientists might be appropriately excited about pivotal discoveries, but these discoveries may only be a way to facilitate the next set of experiments, and will not lead to a new drug, or a cure, for Alpha-1. The gap is wide between the priorities of scientists, and the priorities of patients.

Psychosocial Aspects

In addition to the medical literature on Alpha-1, there is also a smaller body of literature dealing with the psychosocial impacts of a diagnosis or, as it is referred to in the Alpha-1 community, the Ethical, Legal, and Social Implications (ELSI) issues. Bioethical considerations have always been central in the Alpha-1 community, and, on several occasions, important research has been stimulated by specific events in the community. These events included the perception of a breach of confidentiality, and the development of research programs requiring long term informed consent. Some of the ELSI research deals with the ethics of genetic testing for a condition with variable expression (Stoller et al. 2007; Strange et al. 2006; Rachelefsky and

Hogarth, 2008; Wilcke et al. 1990; Wilfond and Fost, 1990). Several of the articles focus on ethical issues related to transplantation, such as the fairness of organ distribution, or the commercialization of body parts (Lynch et al. 2006; Zamberlan 1992, Sharp, 2006). Other bioethical studies have explored informed consent, the legal, and social, ramifications of genetic discrimination, privacy, and confidentiality, and community participation in research (Botkin, 2005; Pope et al. 2003; Foster and Sharp, 2006; Fuller and Ip, 2001; Fuller and Jeffries, 2001; Fuller, 1999; Guy et al. 1999; Greely, 2005; Hunter and Caporaso 1997; Nelson et al. 2001; Sharp and Foster, 2002, 2006, 2007; Sharp et al. 2004; Weinfert et al. 2003; Schutta and Burnett, 2000; Pentz et al. 2002; Moore, 2001; Meropol et al. 2003; Hutchinson, 1998; Roberts 1998; Halpern, 2003; Grant et al. 2000; Cox and McGarry, 2003; Comis et al. 2003; Cheng et al. 2000; Albrecht et al. 2003; Wilcke et al. 1990, 1999; Wilcke, 1998; Widdershoven and Verheggen 1999; Wulfsberg et al. 1994; Lappe, 1998; Stoller et al. 1994; Yarborough and Sharp 2007). There is also research on participation in clinical trials, which has been conducted for a variety of conditions. I include this research in a listing of literature on Alpha-1, as these articles are relevant to the experience of Alpha-1 patients. Participation in clinical trials is generally a more onerous process than participating in a research survey, for example, and I use such participation as an indicator of a greater level of commitment on the part of the patient, and a marker of empowerment (Albrecht et al. 2003; Anonymous 2004; Cheng et al. 2000; Comis et al. 2003; Cox and McGarry 2003; Dickersin and Rennie, 2003; Mannheimer and Anderson, 2002; Meropol et al. 2003; Schneider et al. 2003; Weinfurt et al. 2003; Zwitter, 1997).

There is also some work about the formation of the Alpha-1 Research Registry, DNA & Tissue Bank, and the Alpha-1 Foundation (Fleming et al. 2001; Stoller et al. 2000; Walsh et al. 2006; Strange et al. 2004, 2006; Baker et al. 2002). Unfortunately, there is still a limited amount

of social research specifically about Alpha-1, and some of the work cited here does not deal with Alpha-1. I reference the articles, as they provide me with insight into broader social issues that are also important to the Alpha-1 community, such as participation in clinical trials, privacy, confidentiality, and ownership of genetic material (Merz et al. 2002; Merz and Cho, 2005; Beskow et al. 2004; Moyer et al. 2008; Botkin, 2005).

Another field of interest in the Alpha-1 community has been pharmoeconomics. Several analyses of the costs associated with respiratory and pharmaceutical therapies, genetic testing, and transplantation among Alpha-1 patients have been conducted over the past two decades (Mullins et al. 2003, 2001; Alkins, 2000; Hay and Robin, 1991; Welch and Burke, 1998; Gildea et al., 2003; Shermock et al. 2005; Schoonmaker et al. 2000; Weinstein, 1991; McNeil et al. 1988). However, it is the articles written about the familial and psychological impacts of Alpha-1 on family members, and spouses, that I found the most relevant for this study on empowerment (Fanos and Strange, 2004; Lappe, 1998; McNeil et al. 1985, 1986a, b; Nelson et al. 2001; Stoller et al. 1994; Sveger et al., 1999, 1997; Thelin et al., 1985a, 1985b; Rachelefsky and Hogarth, 2008; Coors et al. 2008; Wilcke, 1998; Wilcke and Seersholm 1999; Wilcke et al. 1990, 1999; Knebel et al. 1998, 1999). These articles highlight the personal and familial context affected by Alpha-1, and suggest that family members, and caregivers, may be the stakeholders in the community who are not so empowered.

The psychosocial literature also includes a number of studies relating to Quality of Life (QOL) in Alpha-1 and other lung diseases, and in liver disease (Dawson et al. 1999; Gunzareth et al. 2001; Jones et al. 1991, 1992; Ketelaars et al. 1996; Knebel et al. 1998, 1999; Wijkstra and Jones, 1998; Spencer et al. 2001; McNeil et al. 1988, 1986a, b; Seemungal et al. 1998; Stoller et al. 1994; Jones et al. 1991, 1992; Barley and Jones, 1999; Thelin et al., 1985a, b; Zamberlan,

1992; Celli, 2008; Renwick and Connolly, 1996; Okubadejo et al. 1996; Quirk et al. 1991; Quirk and Jones, 1990). Although, my doctoral research does not specifically relate to QOL, my future research aim is to correlate the extent of empowerment with QOL in the Alpha-1 community. I, therefore, used QOL as a reference, and framework, for some of my questions about empowerment. I also reviewed studies of QOL in the medical literature, as a way to understand how to scale disability, and construct effective measurements of qualitative data (Wyrwich et al., 2007, 2006; Tsukino et al. 2002; Reardon et al. 2006; Gunzerath et al. 2001; Gutierrez et al. 2007; Hajiro et al. 1998, 2000; Jones et al. 1991, 1992; Katsura et al, 2007; Beddington, 1977; Fries et al. 2006; Goodacre et al. 2006; Rat et al. 2007; Swigris et al. 2005; Fries, 1983; Grunfeld et al. 2008).

Anthropological Approach

Despite the broad range of topics relating to Alpha-1 that is covered by the literature, descriptions of the condition remain centered within a biomedical framework, as if Alpha-1 was a solitary, well-defined entity. It is, however, a genetic condition that may or may not progress to disease, the disease may be in the lungs or in the liver (or both), the condition may effect siblings and other relatives, or not, and an adult may be disease free, but have children with Alpha-1. In addition to its variability of expression (including being asymptomatic), Alpha-1 may represent a protease-antiprotease imbalance on the molecular level, a deficiency of a protective enzyme that causes cumulative destruction of soft tissue, a neurodegenerative condition like Alzheimer's Disease, a gain of function or loss of function problem for the liver and lungs, an irritant to the liver because it polymerizes, programmed for cell death because of miscues from the messenger RNA, none of the above, or all of the above. Describing it as a genetic flaw, located in a specific place on the genome, may be one of the only uncontested facts about Alpha-1, but it is not a summary of what it means to live with it, to research what causes it, or to cure it.

How then to describe the condition Alpha-1 Antitrypsin Deficiency? Mol's study of atherosclerosis in a Dutch hospital had convinced me that diseases or genetic conditions, such as Alpha-1 Antitrypsin Deficiency, should not be explained as solitary conditions. Instead, a complex condition like Alpha-1 might best be described as an interlocking set of experiences ranging over a variety of enactments and domains - from the frightening experience a patient has of breathlessness to the complicated scientific procedures conducted in a laboratory. Following Mol's model approach with atherosclerosis, I examined Alpha-1 through multiple narratives and viewpoints, and present the experience of Alpha-1 as a "body multiple", ranging over time, locations, knowledge, and experiences (Mol, 2002).

Locations and Enactments of Alpha-1

The condition known as Alpha-1 Antitrypsin Deficiency affects a wide range of stakeholders (Table 1-1), and is best understood through its many enactments and locations, rather than as a genetic entity around which all other experiences of Alpha-1 revolve. By removing the centrality of the biomedical description, Alpha-1 is shown to be equally involved in social and familial experiences, philosophical and scientific inquiries, commercial interests, and as a set of issues that policy makers and legislatures grapple with. The condition Alpha-1 is the hub, or *raison d'être* of the Alpha-1 community, surrounded by groups of stakeholders who all have an interest, or involvement, in Alpha-1 (Figure 1-1). These stakeholders include diagnosed individuals, patients and their families, caregivers, support group members; organizational leaders and staff, professional lobbyists; medical professionals, bioethicists and social scientists; government employees; scientific organizational staff; and biomedical industry employees. All of these groups, and individuals, have a common purpose, curing Alpha-1, and have, over time, created a group, or community, identity. This group refers to itself as the Alpha-

1 community and as the Alpha-1 Research Network (see pages 62-71 for a detailed explanation of the community infrastructure).

Each of the stakeholder groups in the Alpha-1 community is situated in a distinct location where Alpha-1 is enacted, or played out. This includes academic offices in medical departments such as pulmonary, hepatology, gastroenterology, pathology, molecular biology, radiology and biochemistry, social science departments such as bioethics, psychology, anthropology, epidemiology, and public health, respiratory therapy clinics, and different types of diagnostic, and clinical, research laboratories. The locations also include patient and scientific organizational offices, government offices, scientific conferences, policy workshops, national and international assemblies, print shops, website companies, pharmaceutical laboratories and factories, local patient support group meetings, regional patient educational days, legislative offices, doctors offices, and in the homes, and workplaces, of individuals diagnosed with Alpha-1. All of these locations, and the activities that occur in each place, together make up a picture of the culture, and world, of the Alpha-1 community.

Alpha-1 is important to each of these stakeholders, and in each of these locations. However, I perceive the primary enactment of Alpha-1 as the body of an individual with the condition, and the experience of illness. By emphasizing the centrality of the lived experience, Alpha-1 may cease to be a disembodied enactment described primarily in biomedical or genetic terms.

Path to Diagnosis and Treatment

Using a schema of Alpha-1 as centered in the patient, rather than from a medical point of view, I start with what happens first, the individual's experience of illness. The stories of lung affected Alphas usually begin with noticing increasing breathlessness, or chronic illness that seemed to increase in severity, and frequency, when the individual is in their forties.

BC (female, 57 years old): “it felt like I was smothering to death.

CD (female, 57 years old): “I had had a couple bouts with pneumonia. They were really long lived illnesses. I remember the first time – I think my doctor sent me for an x-ray in January but then I had to stay on antibiotics until June until the infection cleared. Then the following year I got pneumonia again. After I had the pneumonia I felt as though I never really recovered in the sense of breathing well again. Shortness of breath continued on and never really improved, or not much. Finally, after a while, I agreed to see a pulmonologist. I was struggling and getting worse. It wasn’t curiosity, I needed more treatment. This was when I was 45; I first had pneumonia at 37. Prior to that I was in that, “I’m ashamed I can’t breathe” mode. Thinking back to childhood, I realize I was very susceptible to all childhood illnesses, more than my siblings.

Others remembered a turning point, when illness began to dominate their life:

HI (female, 55 years old): “I was in such bad shape; struggling to breathe, my heart going so hard. No treatment was working. I was taking lots of Prednisone, having exacerbation after exacerbation and being hit with odors from perfume or hand cream.

QR (male, 66 years old): “I was a construction boss; it was my crew who noticed my wheezing. I hadn’t realized how much my health was deteriorating. After their comments, I began to notice how much difficulty I had walking upstairs or doing roof work, and when I couldn’t get rid of a cold I finally went to the doctor.

QR had gone for testing, and been diagnosed, after comments by her pharmacist, who noticed that in addition to high doses of medicine, she was pallid, and often breathless.

For several Alphas, however, the story began in their childhood.

JK (male, 62 years old): “I was ill throughout my childhood – for me, the diagnosis was a relief because it represented a reasonable explanation for some bad memories of childhood. I was home, stuck in bed when other kids were out playing. I really resented it.”

For the majority of Alphas, the story begins on a flight of stairs, when carrying bags of groceries, or on a bicycle, doubled over and fighting for breath, and progresses to an unwanted familiarity with their doctor, a hospital bed, and an array of medicines. But, the frequent bouts with pneumonia, the inability to control allergies, or COPD, or asthma, and the increasing feeling of breathlessness, led many to question their doctors, and ask for a better answer about their condition. Pushing for better answers, and the motivation to get a better handle on their condition, are for many Alphas the first steps toward empowerment.

Diagnosis is a milestone experience in the life of an Alpha, and the study participants seemed eager to relate stories about their diagnosis; unfortunately, almost half of those surveyed had seen a series of doctors before finally being diagnosed correctly. For, even when a patient had heard of Alpha-1, being diagnosed was often a long, drawn out, and frustrating process of trying to convince their primary physician, or pulmonologist, to test them for Alpha-1. Others asked for the test but their physician either had not heard about it, thought it too expensive or believed Alpha-1 was so rare that it probably was not the cause.

KL (female, 57 years old): “I felt let down when the chief of the pulmonary department didn’t know to test me [for Alpha-1]”.

FG (male, 58 years old): “When I was 46 years old I saw an allergist; I had sinus problems and had that surgery, but then my breathing got worse even though I was on an inhaler. I started getting sick in the morning and had some blood tests and was sent to a pulmonologist who diagnosed me with early stage COPD.

The first 2 pulmonologists I saw didn’t seem to know what was going on. One I lost confidence in their knowledge – they didn’t seem to know what my problem was. The 3rd did a liver enzyme test and followed me every 3 months. He sent me to a hepatologist. The GE [gastroenterologist] diagnosed me with AAT through a liver biopsy.

The hepatologist told me I needed a liver transplant in 3-5 years (this was when I was 47 years old). The summer after that I went to Dr. B [a recognized Alpha-1 expert] for a clinical trial of that sheep thing inhalant. After the third visit Dr. B. told me to worry about my liver. The focus had shifted and my interest in the liver was elevated.

My diagnosis came 2 years after first being seen for asthma.

Such anecdotal evidence is common in the Alpha-1 community, and led to heightened awareness campaigns by Alpha-1 organizations directed at primary physicians, pulmonologists, and allergists, and that recommended testing for symptomatic patients. This type of story also motivated Dr. Stoller, and colleagues, to conduct several reviews of demographic data on Alpha-1 patients, to establish the average length of time from first symptoms of illness until diagnosis. In their first review, Stoller, and colleagues, found it took an average of seven years for a correct diagnosis; however, in 2004, their second review found it took an average of four years to be

correctly diagnosed with Alpha-1 (Stoller et al., 2004, 1998). The diminishing length of time to correct diagnosis may be attributed to the awareness campaigns, the more regular inclusion of information on Alpha-1 in medical curricula, and the growing availability, and ease, of testing, such as self-administered finger-prick tests, in the intervening years (Ferrarotti et al. 2007; Rodriguez et al. 2002; Costa et al. 2000).

Unfortunately, Alphas' problems do not end with diagnosis, for, once a primary physician has agreed to a blood test for Alpha-1, they frequently do not know how to interpret the results, or how to treat the condition (Stoller et al., 2004; Anonymous, 1989, 2003). In addition, almost half of the Alphas surveyed (15/34) were told by a primary physician, or pulmonologist unfamiliar with Alpha-1, that they would die within five years of their diagnosis, or be totally incapacitated, and on oxygen. This type of fatalistic slant on the diagnosis became the second component of negative diagnosis in study data. And, makes it understandable why many remembered their diagnosis as a "death knell".

After a diagnosis the majority of Alphas told of going through an emotional reaction ranging from fear, despair, and overwhelming confusion to anger. Being told one has a fatal condition is hard to assimilate right away, and despite previous symptoms and unexplained illnesses, for most individuals it seemed to be unexpected news.

HI (female, 55 years old): "not at any point was I prepared for a catastrophe illness.

LM (female, 52 years old): I went on a grieving trip.

OP (female, 59 years old): I felt devastated, floating around in Neverland.

NO (female, 67 years old): I felt instant isolation upon diagnosis.

Many remarked on periods of denial, and refusal to learn more about their condition; for these individuals not knowing was better than accepting they might die within five years, or be totally disabled, and on oxygen. Others faced families in denial ("you're not really sick"), or,

worse, family members who were indifferent. But for some individuals, diagnosis was just another stage in the frustrating process of finding an expert who could help them deal with their condition.

After a diagnosis, each diagnosed individual faces a key intersection in the process of empowerment – either they have family support AND become aware of resources about Alpha-1, and experts in the field, or, they continue to feel overwhelmed by their diagnosis. Family support seems a vital component of empowerment, but without access to resources, and experts, many Alphas continue to be (mis)treated by primary physicians, or specialists, unfamiliar with standards of care for Alpha-1. If, there is access to resources, however, Alphas can find information that can positively impact the course of their treatment, and their feelings about being diagnosed. RS (male, 58 years old) was “lucky to have neighbors”, one of whom an anesthesiologist, another a pathologist, who knew about Alpha-1; ST (female) had a friend who worked in a hospital and recommended contacting the pulmonary rehabilitation department for help; PP (female, 62 years old), went online, and immediately found websites about Alpha-1, and referrals to local resources and experts. But, for many Alphas, the process takes longer until they find the expertise, and help, needed to manage their condition, get on a transplant list, or find social support. One key informant remembers the process of getting involved with the community:

GH (male, 43 years old): “I go to support group meetings once in a blue moon. Attend an education day? No, that was part of my denial – I don’t want to live in disease but in the solution. And, I didn’t want to be stared at.

But [going to an education day] not as bad as I thought. The second time it was beneficial – I learned a lot, had a few laughs. I found out if there is something wrong there is someone I can call. The first time I went I thought it would harm me but it didn’t and I was amazed to see the variety of ages. One girl was only one year older than me, although others were much older; it gave me hope.

For liver affected Alphas, the process of diagnosing Alpha-1 may not be as difficult as it is for lung affected Alphas. This is particularly true of the pediatric cases, as Alpha-1 is the third leading cause for liver transplantation in neonates and children, and many more physicians, and liver specialists (gastroenterologists and hepatologists), are aware of the condition (Taddei et al. 2008; Teckman, 2007; Teckman and Lindblad, 2006). Alpha-1 liver disease is classified as one of several unexplained, nonalcoholic liver diseases, and most pediatric liver physicians know to order a test for Alpha-1 when certain symptoms present (Teckman, 2007; Perlmutter, 2004). However, the process of identifying Alpha-1 in infants and children involves parents, and the emotional and physical burdens are different than when an individual is dealing with their own health (McNeil et al. 1985a, b; Sveger et al. 1999, 1997; Thelin et al. 1985a, b). One mother knew right away something was terribly wrong when her infant's eyes turned bright yellow. However, her doctor knew to test the baby for liver conditions that may have caused the jaundice, and although he had not encountered the rare condition previously, the infant was correctly diagnosed right away. Other parents have noted they recognized problems when their child did not thrive (grow at the expected pace for a child that age), was listless, or had an unusually distended stomach; this usually occurred in young children, not infants.

For many parents, a diagnosis of Alpha-1 in their child leads to years of uncertainty and guilt – would the condition clear up, would their child need a transplant, would their child survive? Since, Alpha-1 is a recognized co-morbidity with several other liver diseases, for other parents a diagnosis of Alpha-1 is just one more piece of a complex puzzle of illnesses that make their child suffer. For these latter children, their early lives may consist of a series of surgeries and hospital visits; but for the majority of Alpha-1 children, transplantation is a viable, and effective, treatment for the condition, and in some cases, the condition clears up as they age

(Teckman, 2007; Teckman and Lindblad, 2006; Perlmutter, 2004). Although it is rarer, there are also adult liver-affected Alphas. For most of them, the diagnosis is associated with transplantation, but, in several cases I heard about, the diagnosis of Alpha-1 was obtained post-mortem. During my years in the community, I met, and spent time with, several liver-affected Alphas, and Alpha parents/spouses, as when we exhibited at scientific liver conferences. This included two of the most active patient advocates in the Alpha-1 liver community, both widows of Alpha-1 patients. Although neither is Alpha-1, they were inspired by the deaths of their spouses to get involved with the community, and remain committed to increasing awareness of Alpha-1 diagnosis among all liver patients years after their spouses' deaths. One of these individuals spent many years building up a telephone, and Internet-based, Children's Liver Network that provides support, information, and referral to experts for parents with Alpha-1 children awaiting transplantation. Another is a key employee of an Alpha-1 organization, and continues her leadership role in the Alpha-1 liver community. For these individuals, care giving has an extended meaning, and helping those with Alpha-1 has become their mission in life, and a way to overcome the loss of their spouses.

For both lung affected and liver affected Alphas, the process of diagnosis may figure prominently into how quickly they become empowered, and the individual's optimism, or pessimism, about survivability. As noted earlier, another key factor seems to be the amount, and quality, of support available to a newly diagnosed individual. Unfortunately, for some Alpha-1 spouses and parents, the challenges they face, after a diagnosis of Alpha-1, proves too stressful for their particular family. I met three people in the course of the study, who had been deserted by their spouse, or divorced because of their diagnosis. One of these individuals I met while attending a patient convention - her husband had simply been unable to cope with their

daughter's diagnosis. But, she was accompanied by her mother, and spent time happily relating the process of discovery she and her physicians were going through, to understand more about Alpha-1, and treat her daughter properly. It seemed likely she would find the wider community support, information, and access to resources she needed to become fully empowered, despite her marital situation. However, I also know three Alphas who got married after their diagnosis - for these Alphas, family support had been chosen with foresight, and was, in all three cases, willingly provided. Even though the experiences among those interviewed was so variable, family support became a useful characteristic to quantify for this study, and a reliable predictor of progress towards empowerment. However, since 'family support' represents a range of experiences, it seems a fruitful topic to unpack in future studies. Ethnographic interviews about family support can help to identify the specific components of such support that leads to empowerment.

The centrality of the patient experience of illness may be the primary enactment, but there is a multiplicity of key enactments that center on the condition Alpha-1. Once an individual becomes ill, and is diagnosed with Alpha-1, the enactments begin to shift from the personal realm to the medical and scientific arenas. The second key enactment of Alpha-1 is, therefore, the medical office visit, and interactions with the doctor, nurse, and other medical staff. These interactions are described in detail in Chapter 5, which is an anthropological linguistic analysis of the medical office visit. The third key enactment involves scientific research activities, and is described below.

Research Setting

The scientific enactments of Alpha-1 occur primarily in research laboratories (hereafter referred to as labs), that are part of a worldwide network of researchers in the field of Alpha-1 Antitrypsin Deficiency. This research network is a small, bounded community within the larger

biomedical and research communities, and, consists of approximately 275 researchers recognized, and funded, for their work since 1972. The majority of these researchers conduct research that may apply to Alpha-1, but a good number also investigate multiple phenomena that represent broader areas of science. A molecular biologist, for example, may do research of Alpha-1 molecular processes, such as protein folding, but this basic research is also applicable to Alzheimer's Disease and other conditions (Carrell and Lomas, 1997) ; a pulmonologist may conduct research on Alpha-1, but also be involved in Cystic Fibrosis (CF) research, lung inflammation, or COPD studies (Wilfond and Fost, 1990; Sandford et al. 1999; Turino, 1991; Voelkel et al. 2007); or, a geneticist may be researching RNA transcription, that uses Alpha-1 as the model, but, applies to a wider range of genetic conditions (Termine and Sifers, 2005). However, there are a few individuals worldwide who are known for their focus on, and dedication to, Alpha-1 research. Many of these individuals have established Alpha-1 Centers of Excellence. These centers are comprised of labs and offices at academic institutions, or hospitals, where the Alpha-1 experts have assembled research teams, conduct research studies and clinical trials, mentor the next generation of scientists, and publish extensively on their research. Most of these experts also treat patients, serve as advisors to the patient organizations, assist with advocacy efforts at the State and Federal level, serve as scientific reviewers and advisors for the National Institutes of Health (NIH) and US Food & Drug Administration (FDA), and lecture at patient and scientific conferences about their research, and clinical care of Alpha-1 patients.

During my years with the Alpha-1 Foundation, I had the opportunity to visit many of these Alpha-1 Centers of Excellence in the United States, and Europe, including labs in Cambridge, England, Dublin, Ireland, and in South Carolina, North Carolina, New York, Texas, Oregon, Colorado, and Florida, in the United States. My reason for visiting was twofold – to see

firsthand the progress at programs funded by the Alpha-1 Foundation, and to write lay descriptions of the processes I witnessed as a way to help the patient community understand progress in Alpha-1 research. My experience was often overwhelming – the labs themselves are labyrinths of equipment and computers, and the processes explained to me were complex and time-consuming, and expressed almost entirely in biotechnical terms. But, it is this very complexity that needs explaining, and, given the amount of community feedback on this topic, I think it critical to convey why it is taking so long to find a cure for the condition, and why some of the research seems to have little to do with finding specific therapies for patients.

Many study participants expressed similar frustrations about the time lag between a researcher’s statement of progress, and the length of time until patients receive a new therapy or a cure for their condition. Others went further and expressed a desire to be part of the research process:

MN (female, 60 years old): “Patients should have some voice [in prioritizing research], the funding process is flawed.

PQ (male, 57 years old): “Research is a two-way street and communication [between researcher and patient] should be a two-way street

MM (female, 45 years old): “Who decides between future needs and pressing needs?

Understanding the length, and complexity, of scientific processes may address some of the miscommunications related to research progress, and priorities, in the community. So, although I often find the explanations go well beyond my current understanding of biology, I continue to read scientific articles, visit labs, interview lab personnel, ask lots of questions, and work at accurate lay descriptions of the research being done in Alpha-1. Over many years, I have come to understand enough of the basic processes to describe some of them in this chapter; however, my understanding is as a lay person, and my interest remains centered on the social

characteristics of laboratory culture, and the physicality of lab procedures, rather than the scientific basis of the biotechnical processes.

Laboratory culture

I am most familiar with the activities of one particular Alpha-1 center, and have visited this lab many times over a period of 9 years. During my visits, I have been shown processes, such as nephelometry, gene scanning, tissue banking, and genotyping, but as an anthropologist, I found the culture of the lab equally interesting.

Laboratory culture is strictly hierarchal, and based on academic credentials. The lead researcher, known as the Principle Investigator (PI), is at the top of the pyramid, and has at a minimum a medical degree (MD), an MD/PhD, a pharmacy degree (PharmD), or a Doctor of Nursing Science (DNSc). The PI applies for research funding many times throughout the year, and, when funded, is able to hire administrative and technical staff to conduct the research projects and clinical trials. There were often as many as 30 people working in the lab, all of them dependent on the skill of the PI to obtain funding, and direct their research. The lab manager is usually either a clinical nurse, or is in possession of a Masters degree in a scientific discipline. Nurses, lab technicians, administrative staff, and database staff are the other members of the hierarchy, each with precisely defined responsibilities, designated authority, and appropriate credentials for their job.

In this one particular lab, the infrastructure, and interactions, is precisely defined in writing, and there are shelves of Standard Operating Procedures (SOPs) binders in each office and lab. The lab must demonstrate compliance with these standards, and with a variety of supervisory entities. This includes compliance with University regulations and procedures, such as hiring and firing, Internal Review Board (IRB) requirements, and purchasing procedures; State and Federal regulations of laboratory procedures, cleanliness, safety, and security;

legislative regulations, like HIPAA (Health Insurance Portability and Protection Act), that cover use of human subjects in research and confidentiality; the legal requirements of grant contracts, and research agreements, with funding agencies, pharmaceutical companies, and private organizations.

When someone joins the staff in this lab, they go through a series of orientations; this includes University staff orientation, certification classes for each procedure in the lab, and instruction in the handling of equipment. New staff, in this particular lab, must also sign a confidentiality agreement that covers both the identity of patients, and procedures used in the lab.

This lab is among the few in the world that identifies rare Alpha-1 alleles (phenotypic variants), and it has the largest standard of rare alleles in the world, over 100 variants. The lab conducts not only the basic blood level diagnostic procedure, but also genotypes, and phenotypes, blood samples for a precise diagnosis. Other activities conducted at this lab include, tissue banking, Bronchial Alveolar Lavage (BAL) research, clinical trials on new drugs, research studies of lung inflammation and the genetic characteristics of Alpha-1, and the development of an Alpha-1 liver cell line, that is shared with Alpha-1 researchers worldwide.

There are 6 distinct teams among the staff in the lab, although some employees serve on more than one team. The teams include, a quality assurance team that receives, and records, each sample, an “evaluative sample”, or quality control, team that prepares each sample for testing, a diagnostic team that tests, and reports on all samples, a database team that enters data from the test reports, a reporting team that communicates with physicians, and patients, about the test results, and the supervisory team, comprised of the PI and lab manager, who ensure the quality of all data from the lab. All employees participate in multiple meetings: every Monday morning a general lab meeting is held to discuss progress, plan the weekly activities, and deal with any

personnel issues. In addition, a clinical detection meeting is held each week, and each team has its own meetings to discuss the specifics of that team's activities. This lab also holds several annual parties, as well as monthly birthday parties for staff during office hours. Unlike offices where everyone stops working and gathers to sing happy birthday and eat cake, in the lab people drift by in between lab procedures to get a piece of cake. The priority of timed experiments takes precedence over socializing. The annual parties, on the other hand, are held outside of work. The summer party is usually held on a Sunday, at the PI's home, with a barbecue, and swimming. The winter party is held in a restaurant, the PI's home, or the home of another staff member, and is a potlatch dinner. Since, the lab personnel come from countries around the world, this dinner is an ethnic treat, and typically includes Russian, Spanish, Middle Eastern, and Chinese dishes. The research team is treated at all times like a family. The lab manager remarked that "professionalism and getting along leads to productivity" and I agree – the conviviality in this lab seems to serve a useful purpose, and helps make for a dedicated staff.

In fact, during each visit to this lab, I had noted the amount of conversation and interaction between the lab staff, interspersed with highly concentrated activities with equipment and samples. It occurred to me that the popular image of the lone scientist bent over their microscope was just that, an image. And, when I queried the lab manager about the ideal characteristics of a lab technician, he agreed with my assessment, and noted they discouraged the solitary worker. The qualities he looked for when interviewing new staff were "common sense, instincts, and organization (which includes accuracy and precision)". He also noted, that the PI had always told him to "hire minds not hands" for the lab work. This statement seemed to recognize the value of each employee to contribute to the research. Perhaps, it is the overt valuation of staff that makes this lab such a conducive, and productive, environment for research.

My final observation relates to the potential for empowerment among the researchers in the Alpha-1 community. A key characteristic I had noted in this Alpha-1 lab was the amount of interaction with patients. Lab staff regularly participate in patient education days, where they meet many diagnosed Alphas, and their families; lab staff also frequently assist in recruitment efforts for research studies, which also brings them into contact with the patient community. But, in addition, patients and donors are given the opportunity to visit many of the Alpha-1 labs, take conducted tours of the pharmaceutical production facilities, or attend scientific conferences, and advisory meetings, organized by the Alpha-1 organizations. One scientist noted, after his first Alpha-1 scientific conference that having the patients attend “put a face on the disease” and ultimately made his laboratory research much more meaningful (Carrell, pers. comm.... 2000).

Scientific procedures

My observations in a laboratory, and interviews of lab technicians, are usually prefaced by a review of the type of research being conducted in that lab. One group, based in a pathology department of a large teaching hospital, had proposed the following description, in one of their research studies:

“The immediate goal within the context of this proposal is to incorporate the use of stably-transfected cells that express natural and genetically engineered human alpha-1-antitrypsin variants for the molecular, biochemical and ultrastructural analysis of this degradative pathway. (Termine and Sifers, 2005).

This particular research team has been studying the degradation pathway in the endoplasmic reticulum, in the liver, where the AAT is processed before passing into the bloodstream. Through biochemical analyses, this research team had established that cell signaling sets off a chain of chemical reactions that lead to cell death, and the disposal of the misformed AAT proteins. Each of this research team’s sequential experiments sought to describe a particular stage in the complex chain of processes of cell signaling, biochemical degrading of

cells leading to cell death, and disposal of dead cell material. The majority of studies in this particular lab use Alpha-1 as the model for describing degradative processes. Although, I had had many conversations through the years with the PI of this laboratory, and a basic understanding of the aims of this type of research, I sought permission to interview a laboratory technician on the project, for a greater understanding of the specific techniques used to achieve their aims.

Before giving me permission, the PI stressed the fact that many of the processes, and materials, used in their lab are proprietary, and had been developed by him as the only way to answer the research questions being asked. This PI is one of my better friends in the community, and had often told me stories of another well-known researcher who stole his ideas, and processes, and then took credit for the ideas. Because of these experiences, this particular laboratory maintains levels of secrecy that may be unusually high. I believe I was given access to the secret procedures, and materials, based on two factors – the PI knows I can be trusted to be discrete, and he knows I understand details of the biological and technical explanations only in general terms, and despite my pages of notes, would not be able to divulge critical information about specific reagents, or the sequence of processes.

I arranged to interview the principal lab technician over the phone. I began the interview by noting that many of the terms used in the laboratory were familiar to me, but that I still did not know exactly how these terms are ‘enacted’. I asked the lab technician to describe, as an example, how one transfects a cell.

DT: “That’s a process that takes a few days to complete.

SF: What do you do first?

DT: Well, I start by taking some DNA from embryonic kidney cells and coating it with a specific reagent.

SF: Where do the embryonic kidney cells come from, and where do you get the specific reagents?

DT: The embryonic cells were developed by the PI, and the reagents are purchased from a pharmaceutical company. Next I put the cells in a 10 cm petri dish and incubate in a freezer at 37 F degrees for 5 hours.

SF: How can you incubate something in a freezer?

DT: Well, in science incubate connotes growth.

SF: Oh, it's not like how we associate it [in lay terms] with warmth, like chicks hatching?

DT: No! (laughs). After 5 hours, I remove the media....

SF: What's that?

DT: The reagent used in the first step. So, I remove it with a pipette, replace it with fresh media and put it back in the freezer overnight.

SF: What does it look like at that point? What color is it?

DT: The media is red with white flecks, [pause] I was never asked that.

I was aware that for the technician each term represents a complex biotechnical process that is packaged, or summed up, by a single word or phrase. I had to keep reminding him that I did not know what simple terms like "expressing genes" looked like in a lab, or what it involved in terms of time and effort.

SF: What do you do for the 5 hours when the material was incubating?

DT: I work on several different experiments and, like the other technicians in the lab, I'm working on different procedures throughout the day. Next I perform a pulse chase, or metabolic radio labeling. In this step, I add a radioactive label to the clear liquid on the petri dish. This radioactive label gives off a particular wave length which lets me follow the progression of the labeled protein throughout the series of processes in this experiment. After the pulse chase phase, I harvest the plates, and add a neutral buffer with detergent. The buffer helps to break down the membranes in the cells. Then there are several more incubations at different temperatures and for different times (after 30 minutes, and 60 minutes on ice).

The buffer had been added to make the cells rupture, which in turn allows the isolation of the protein of interest, in this case Alpha-1 Antitrypsin (AAT). After isolating the AAT, I put the material in a tube and spin it slowly on a rotator in a 4 degree incubator. I add an

antibody for AAT, made from rabbits, to the mixture. This antibody creates a slurry-like mixture where the protein binds to the antibody.

SF: Why keep incubating, how do you know if the process worked?

DT: The cells are happy. I mean they grow and proliferate; they thrive and do what they need to do.

The vocabulary seemed to be anthropomorphizing the chemical reactions, but the technician became self-conscious when I noted the use of the word happy, and although I assured him that it was an interesting, and useful, remark, he seemed abashed that he had expressed something so unscientifically.

DT: After creating the slurry, I put the material in a centrifuge and spin it down, and the heavier cells go to the bottom. I then suck off the liquid at the top. The cells at the bottom of the centrifuge are then washed with a salt concentration buffer to get rid of the 'background'.

At this point the interview had already lasted over 45 minutes, and we were barely through the first technical term in the research description, so I did not stop and ask him to explain the term "background".

DT: After the salt buffer is added, which we call a disruption buffer, the AAT protein is released from the antibody mixture. I then remove a portion of the released AAT with a pipette, and load it onto a polychromatic gel 14" tall and 14" wide. I apply a specified amount of voltage to this gel, which causes linear patterns.

SF: Why does it form patterns?

DT: Proteins have a negative charge and are attracted to the positive charge in the gel. They flow down the gel at different rates and a pattern emerges. After that, I place the gel in an acid buffer and gel dryer machine for 2 ½ hours to enhance the signal of the radioactive protein. We also use an x-ray film on top of the gel, which also gives off a radioactive signal, which forms an image of a band. The darker bands signify intensity. The final step is to compare the molecular weight of the AAT with standard degradation rates.

Transfection seems a long drawn out process for isolating and weighing the AAT protein, but, is just the first of several biochemical processes, each lasting several days or weeks, that are performed to achieve the aims of the project. Moreover, the aim of the research project is not to

produce transfected cells but, to use them to analyze the molecular, biochemical, and structural processes involved in a degradative pathway. Given the number of steps and time involved just to prepare the basic materials for the experiment, it began to make sense to me why research of this type requires a series of two year projects. Each step of the process has to be carefully documented, measured, and reproduced, and the results need to use consistent, and well established, measurements for the data to be meaningful. In addition, laboratories must constantly calibrate their equipment, and test their experimental results, against established standards.

Length of time, however, is not the only issue patients have with research. A primary concern that many patients and their families expressed was ‘what does basic scientific research have to do with my breathing problem, or my survivability after transplantation?’ One answer is that by conducting a basic scientific experiment, this particular PI is more likely to receive funding, as the research is applicable to a number of disease states, not just to Alpha-1, a rare condition. But, the answers the PI is seeking ARE applicable to Alpha-1, and, by framing his research in broader scientific terms, he is more likely to receive ongoing funding, and generate translational results that will benefit the Alpha-1 community. By translational I mean that his basic science experiments, and a fuller understanding of the degradative process, has the potential to lead to, or translate into, therapeutic approaches that target the liver, but, that will address both the liver and lung conditions associated with Alpha-1.

The Alpha-1 Community

Three highly successful organizations were founded by Alphas in the 1980s, and 1990s: the Alpha-1 Foundation (Foundation), the Alpha-1 Association (Association) and AlphaNet. Each of these organizations has a distinct, but complementary, mission. The Alpha-1 Foundation provides support for research, the Alpha-1 Association provides support, and education, for

Alphas nationwide, and, AlphaNet provides low cost drug distribution and disease management. Together with the physicians, nurses, and other health professionals who care for Alpha-1 individuals, these three organizations, and the thousands of Alphas they represent, comprise the Alpha-1 community.

Alpha-1 Organizations: Models of Entrepreneurship

In the 1990s, Alphas around the United States began to organize local and regional support networks for themselves, and their families, and founded the Alpha-1 Association. This organization's primary mission is to provide support to patients, information about Alpha-1, and to unify common concerns into advocacy efforts. The Association holds regional educational days throughout the United States, and an annual educational conference (the 2008 conference had over 350 attendees). The Association also focuses on advocacy efforts in Washington, D.C., and maintains a website with downloadable resources for patients, family members, and caregivers (www.alpha1.org).

The more prescient of the patients involved with the Association recognized needs that go beyond patient support, such as the importance of funding research of Alpha-1, and the need for a strong organizational infrastructure to support such research. These patients recognized the potential for Alphas to contribute to their quality of life, and even their own cure, by supporting research, becoming more informed about Alpha-1, and interacting closely with the research community towards finding a cure (Stoller, 2000, Kicklighter and Sharp, 2005; Sharp and Foster, 2007, 2006, 2002; Yarborough and Sharp 2007).

A number of Alphas had participated in NIH studies of Alpha-1, and, based on this experience, identified their mission, or goal, as finding a cure for Alpha-1. This goal led to the establishment of two other Alpha-1 organizations, the Alpha-1 Foundation and AlphaNet.

The history of these organizations began in late 1994. Following an Association board meeting, some of the members met to discuss an innovative idea – to create a not-for-profit organization governed by Alpha-1 patients, and employing patients to care for other Alpha-1 patients. Their motto was, and is, “Alphas serving Alphas”. The board members involved in this discussion envisioned the profits, derived from an Alpha-1 care network, as a means of supporting Alpha-1 research. By the summer of 1995, three Alphas, John W. Walsh, Susan Stanley, and Sandy Lindsey, had established two separate, not-for-profit organizations, the Alpha-1 Foundation and AlphaNet. The Foundation was established to focus on stimulating research, and AlphaNet to fund the research. John Walsh had set up a relationship with two local pharmacies, that allowed AlphaNet to dispense the only available therapy for Alpha-1, Prolastin. Relationships with other pharmacies were established, and diagnosed Alphas were trained as coordinators to serve wide geographic networks of Alpha-1 patients. By 1999, Bayer (now Talecris Biologics, Inc.) established a direct distribution program with AlphaNet. The direct distribution model allocates product directly to the patient, and according to Robert C. Barrett, the CEO of AlphaNet, represented the “single most important business breakthrough [that] spurred [AlphaNet’s] dramatic growth”. AlphaNet is a highly successful business operation, contributing an average of \$2 million per year to the Alpha-1 Foundation’s research efforts, with a total contribution of \$18 million in ten years (www.alphaone.org; www.alphanet.org).

Research Network

One of the first community-wide research programs developed by the Foundation was a Research Registry. The NIH had compiled a registry of Alpha patients, but by 1996 their program was nearing completion (The Alpha-1 Research Registry Study Group 1998; Baker et al. 2002; Eden et al. 2003; McElvaney et al. 1997; Strange et al. 2004, 2006; Turino et al. 1996). James Stocks, a researcher involved in the NIH project, remembered how it “became clear that

future Alpha-1 drug investigations would require maintenance of the professional expertise represented by the 37 Registry sites, as well as a cohort of Alpha-1 patients”. The Alpha-1 Foundation made a commitment to continue this important research network, and established the Alpha-1 Research Registry at the University of Miami, School of Epidemiology and Public Health (Fleming et al. 2001; Stoller et al. 2000). The Registry is currently housed at the Medical University of South Carolina, and numbers over 3,298 enrollees (Figure 2-1). Enrollment statistics (Figure 2-2) are a clear demonstration of the effectiveness of the infrastructure of the Alpha-1 community, and how each organization serves the missions of the other two organizations. The Registry provides Alphas the opportunity to participate in research, but, it also serves the scientific community, providing demographic and medical data on Alphas for analysis, and a large enough cohort for statistically significant survey results (Strange et al. 2004, 2006; Baker et al. 2002). Over 25 research studies have utilized the Registry cohort, or data, since 1998, including this doctoral research.

Other research programs were established by the Foundation since 1998, and include a DNA and Tissue Bank, a Coded Testing Study, that provides confidential testing for Alpha-1, a national detection program that targets symptomatic lung and liver patients, a formalized network of Clinical Resource Centers, a peer-reviewed grants program, and a cadre of advisory groups led by the Medical and Scientific Advisory Committee, known by its acronym, MASAC. The MASAC is an advisory body comprised of the leading experts in the various disciplines related to Alpha-1, and includes physicians who treat lung and liver patients, researchers, nurses, bioethicists, lawyers, epidemiologists, and importantly, patient representatives. The MASAC meets twice a year to deliberate pressing issues in the community, and to make specific recommendations to the Board of the Alpha-1 Foundation.

The Foundation also established a number of working groups, including an Educational Materials Working Group that prepares, and validates, printed materials about Alpha-1, an Ethical, Legal and Social Issues (ELSI) working group that considers the ethical, legal, and social issues in the community, an Epidemiology/Registry working group, and the newly formed Transplant Issues and Public Policy Working Groups. The Chairs of the Working Groups are members of the MASAC, and provide individual recommendations, and action items, to the MASAC to consider, before passing on these recommendations to the Board of Directors.

My role at the Alpha-1 Foundation was to staff all of the advisory committees, assist with the development of protocols for the research programs, and prepare the written reports, recommendations, and policy initiatives for the Board of Directors' meetings. I was also responsible for providing program templates and start up information on the Registry, Tissue Bank, and Grants Program to other rare disease groups, for them to use in developing their own research infrastructures.

It is a badge of honor among both the scientists, and the patients, to be appointed for a two-year term on the MASAC. The meetings are generally held in five-star hotels in Miami, where the Foundation is based, and although they are fast-paced and productive meetings, they also include a strong element of socializing among the organizational staff, patient leaders in the community, and the scientific network. Dr. Stoller, former Chair of MASAC, wrote an article about the importance of the Foundation's advisory committees, in which he noted, "Communication is the element that holds disparate parts of the community together" (Stoller, 2005). The interaction and conviviality of the biannual meetings has clearly helped maintain the commitment of the scientific community, and a sense among the individuals who serve on the Foundation's advisory bodies that promote, "I know that I am making a difference" (Stoller,

2005). In addition, the contacts between the Alpha community and the scientific network at scientific meetings, in laboratories, during clinical trials, at the MASAC and working group meetings, have given the scientists a better sense of the role patients play in advancing Alpha-1 research. James Stocks, one of the original NIH Alpha-1 researchers and an active member of the Alpha-1 community, articulated a the common attitude among the physicians and researchers when referring to progress in the community,

“Clinical investigators have used the resources of the Foundation’s Research Registry (for clinical trials). But even more critical have been the Alphas themselves. They have stepped forward and volunteered from across the continent and subjected themselves to long-distance travel, numerous medical exams, and procedures like bronchoscopy and liver biopsies. Some singular few have even volunteered for multiple studies, their only compensation being “thanks”. I have long held these volunteers to be heroic examples of our Alpha-1 community. (Alpha-1: A Ten Year Retrospective, 2005).

Other researchers have remarked on the courage, and commitment, of the patient community in their interviews, and in their articles for the Foundation’s 10 year retrospective publication. Dr. Brantly has been particularly complimentary, and encouraging, to the patients he knows. In his article for the Foundation’s 10 year retrospective he noted,

“None of the (Alpha-1) center’s programs would be possible without the courage and sacrifice of Alphas everywhere. The research center belongs to them. Their active, willing participation makes the ongoing search for a cure possible and makes my work so rewarding. (Alpha-1: A Ten Year Retrospective, 2005).

Alphas have, in turn, often expressed their appreciation for the commitment of the scientists to their cause. One grateful transplant recipient wrote a book about his experience, entitled, Heroes of My Transplant (Russell, 2001); others have expressed their appreciation by donating to research, raising funds for research, or expressing their thanks through awards for, and recognition of, individual researchers. There is a great deal of mutual respect and admiration flowing between stakeholders in the community, which over time has strengthened other stakeholders, and promoted a sense of purpose, and accomplishment, that all share in.

Public Policy

A key element in the success of the Alpha-1 community is its advocacy program. In 1998, following a shortage of Prolastin (which is a drug one injects into the bloodstream), John Walsh was appointed to the President's Council on Blood Safety and Availability. At these meetings, he met Miriam O'Day, who, at that time, served as a professional lobbyist for another rare disease organization. She is knowledgeable about Beltway politics, appropriately aggressive, and has carved out a niche, among the rare disease communities, as an effective professional and advocate. John recruited Miriam to work as Public Policy Director for the Alpha-1 Foundation, and Alpha-1 Association, positions she holds to this day.

Among the many successes achieved by the community, under Miriam's tutelage, are Medicare and Medicaid 'carve-outs' for Alpha-1 therapy, a voluntary blood safety program, that notifies patients immediately when blood-derived products are withdrawn or recalled, legislatively-mandated funding for Alpha-1 research at the NIH, creation of a Congressional committee on COPD and Alpha-1, and, more recently, a concerted effort to convince the FAA to allow use of portable oxygen containers on commercial airlines.

Miriam accomplished the Medicaid 'carve-out' by organizing grass-roots efforts in 2002, and again in 2003, to lobby Congress to recognize augmentation therapy under a special category of orphan drugs. Her efforts included organizing patient groups to visit Congressmen, rehearsing with the scientists who would provide testimony on the issues in Washington, and drafting the language used by Congress to create the carve-out. By rallying community members to a common cause, Miriam resolved a major barrier to access to care for hundreds of eligible Alphas. She should be proud of her achievements in the public policy arena, but, like many others associated with the Alpha-1 community, expressed her personal accomplishments in relation to the goals of the community:

“I feel profoundly grateful to John Walsh and the Foundation for giving me the opportunity to work in a dream job. As one of its many rewards, I’ve learned that one person really can make a difference”. (Alpha-1: A Ten Year Retrospective, 2005).

Vision for the Future

On the ten year anniversary of the founding of the Alpha-1 organizations, leaders in the community published a retrospective report. This publication includes chapters on the founding of the organizations, the formation of medical and scientific advisory groups, clinical research in the community, public policy efforts, descriptions of scientific and patient meetings, and key patient related resources, like disease management, and drug distribution. Each chapter is written by the person who developed that aspect of the Alpha-1 infrastructure, and is a remarkable record of achievements, and progress. It is also a highly personal set of accounts, and demonstrates the level of commitment and dedication to a common cause that exists among Alpha-1 community members.

The Alpha-1 retrospect begins, fittingly, with “A Vision for the Next Decade” written by John W. Walsh. John is a very visible Alpha-1 leader – he serves as President and CEO of the Alpha-1 Foundation, is President of the COPD Foundation, and was a co-founder of both AlphaNet and the two Foundations. He has been President of the National Health Council, been recognized by the FDA for his role in accelerating drug development for Alpha-1, and currently serves on the Public Advisory Committees of the American Thoracic Society, and the National Institutes of Health. John is an Alpha, as is his twin brother Fred, and two other siblings in his family.

Since 1999, I have worked closely with John, and have come to appreciate the extent of his leadership abilities, and his charisma. Many time, I observed his ability to grasp the implications of various types of research, and to negotiate at the highest levels of government, and industry, on behalf of the Alpha-1 community. He, and his wife, Diane, remain instrumental in the success

of the community and, in the opinion of many of us, are the driving force that keeps scientific, government, and industry leaders committed to finding a cure for Alpha-1.

One board member wrote about the early days of the Foundation when “some [board] members had a truly ambitious, entrepreneurial vision and a sense of urgency. Others saw the need to move at a more deliberate, methodical pace”. The comment about ambition and entrepreneurial vision was obviously referring to John, who, according to his sister-in-law, Pam Walsh, has always driven “in the fast lane” (Pers. Comm., 2006).

In his essay John notes the accomplishments of the first 10 years, and his vision for the future of the Alpha-1 community.

“In just ten years, we’ve created a research infrastructure, including the Alpha-1 Research Registry, DNA and Tissue Bank, Alpha-1 Research Program at the University of Florida, and an extensive Clinical Resource Network around the country to support the international investigator community.

We’ve forged strategic partnerships with the American Thoracic Society, American Association for the Study of Liver Diseases, the Genetic Alliance, the National Health Council, FasterCures, the American Association for Respiratory Care, and other professional organizations and patient groups. We’ve piqued the interest of the pharmaceutical industry to develop new treatments; we’ve brought industry regulators together with government regulators to make those treatments available faster. And we’ve worked with Congress and the Centers for Disease Control to launch a National Targeted Detection Program for Alpha-1.

The article is also an excellent articulation of empowerment in Alpha-1:

“Some things about the Alpha-1 community won’t change in the coming decade - namely, its unity and sense of purpose. By rallying to the Alpha-1 Foundation and AlphaNet over the last decade, we Alphas have demonstrated our commitment to taking our destiny into our own hands. Not content to sit by passively, we’ve found a way to come together, apply our collective effort and imagination, and support pioneering research, even as we support one another.

It is hard to convey just how central, and important, John is to the Alpha-1 community, and how much of the successes through the years have depended on his vision. He has demonstrated remarkable business savvy, which has enabled large amounts of research funding to be

generated. And, he is primarily responsible for creating the strategic alliances among the stakeholders in the community, and with government, industry, and donors. He is a true leader. The only problem, for me, in researching empowerment has been to recognize the uniqueness of John's attributes. When I first went out into the community, and met other Alphas, I was surprised how little some of them knew, until I realized I was comparing everyone to John. This realization altered my thinking about empowerment, and I began to conceive of empowerment as an evolution, or staged process. This conception of empowerment, as an evolution, would more accurately reflect the forms of empowerment individual Alphas could achieve on the personal level, as well as the community-wide, or structural forms of empowerment so well exemplified by John, and a few other Alpha leaders. My hypotheses about stages of empowerment, and the qualitative differences in organizational leaders, thus grew out of my recognition of the important, but unique, role John has played in the community.

Anthropological Research in the Community

The current Alpha-1 research network consists of over 50 academic and medical centers representing various geographic regions in North America, and the local patient support groups that grew up in association with many of these centers. Membership in the Alpha-1 community has, thus, been formalized, first through the NIH program, then through the Alpha-1 network, as enrollment in the Registry, and participation in research. This emphasis on research, and enrollment in a Registry, differs significantly from the looser, less formal membership in patient organizations such as the American Heart Association, the American Lung Association, and the March of Dimes, or among disease specific communities like Cystic Fibrosis, or Muscular Dystrophy. Membership in these other organizations involves signing up to receive mailed or emailed information, sometimes involves paying annual dues, and frequently includes access to either medical professionals, health educators, or other patients for information or patient

support. However, membership in these other organizations may not involve participation in any type of research, membership in a formalized network of centers of excellence, or an increased sense of identity as a particular type of patient. Membership in the Alpha-1 community, in contrast to other disease community networks, connotes a greater role in research, or at least a familiarity with the parameters of research.

There have been numerous opportunities for diagnosed Alphas, and their family members, to participate in research studies and clinical trials, and many avail themselves of the invitations to participate that are periodically sent to enrollees in the Alpha-1 Research Registry. However, this very familiarity with biomedical research led to an initial uncertainty among participants in the patient empowerment study as to their role, and my expectations as a researcher. They were not being asked to give blood, undergo a lung lavage², have liver cells removed with a “scary looking 8 inch probe” (Zern, Pers. Comm., 2001), or even answer fixed questions on a survey. Instead, they were given a freedom of expression about their own experience that some found intimidating, and others found liberating.

The format of the focus group sessions was designed to elicit narratives, rather than short, or yes-no answers to questions. Two of the sessions were held in an Alpha’s home and two were held in hotel meeting rooms. In the two home-based sessions, my Alpha host and I prepared a buffet meal, and everyone who attended helped themselves to lunch, and sat around a dining table while we held our discussion. The sessions held in hotels also involved luncheon, but despite the more formal aspect of a public venue, I strove to create as casual and inviting an atmosphere as possible. In each session, for example, considerable time was spent on introductions, catching up with former acquaintances, and sharing illness stories and, likewise,

² a washing out of lung tissue for microscopic investigation of inflammatory cells and other biomarkers of disease progression

concluded with long, drawn-out farewells, exchange of telephone and email information, and more socializing. I allowed time for this phatic communication as it was very revealing of the attitudes, psychological and physical states, and level of involvement of the participants in previous research studies.

I chose not to tape the sessions, as passing around a microphone would have created a more structured discussion, with formal turn-taking, and a beginning and end to each person's comments. Instead, I encouraged an open ended discussion, without formal turn-taking, which resulted in conversations that flowed between participants, and that included many overlapping comments. Because I did not insist on staying central to the discussion, many of the participants' remarks were directed to each other, rather than at me. This interaction allowed me to observe variation in the levels of awareness between participants, and insight into how support is sought, and provided, between Alphas or between caregivers. It also allowed the participants to set the tone of the conversation, and introduce and expound on topics of interest, and relevance, to their experience as Alphas and caregivers. Although I had IRB-approved sample questions, and topics, to cover in the session, once the conversation had begun, I rarely had to refer back to my original questions. I did, however, ask the group to help me summarize our discussion, and overtly took notes of the final list of topics that the group felt important to include. I had also taken notes throughout the session of individual remarks, and demographic information on the participants.

Many participants in the first focus group session initially hesitated about giving more than a one sentence answer. They were clearly used to the fixed question and answer format of survey questionnaires, and unaccustomed to elaborating on their experience. My research seemed to be one of the first times that Alphas were asked about their emotional responses, the details of how

Alpha-1 impacted their work and home life, or about the creation of a new selfhood through their diagnosis and illness experience. By inviting elaboration on topics that were novel, or had only been explored as yes/no questions on previous questionnaires in the community, participants in the study were able to explore individually, and as a group, how their condition impacted family members (who were often present during the focus group sessions, and who provided their own point of view); the ways in which disability is defined; and the “manner in which self of the impaired person is preserved by family members” (Rubinstein, 1995).

Based on the experience at the first focus group session, subsequent sessions, and interviews, were prefaced with an overview about anthropological research, and explanation of how it differed from the quantitative approaches used in biomedical, and epidemiological, studies. I opined, during these explanatory remarks, that biomedical research was not necessarily attuned to the lives participants lived as Alphas, nor did biomedical research explore social issues, or people’s own concerns. Participants were encouraged to share experiences in their own voices, and from their own unique perspectives (Rubinstein, 1995; Colby, 1966). Although the value of qualitative research was not overtly discussed, there was an emphasis on the lived experience of participants, rather than on a statistical analysis of their yes-no answers to questions, or measures of their biological samples. The qualitative approach in this study was also used as an overt attempt to re-introduce the importance of the social components of illness experience in Alpha-1, which have been increasingly overshadowed by the dominance of a biomedical focus (Rubinstein, 1995).

Before each focus group session, or interview, an appeal was made to participants to assist in defining the parameters of the study itself, and to help define empowerment, and its components, emically, which was defined as ‘from your unique perspective’. I had discussed my

motives, and introductory remarks, with each Alpha host, and welcomed their reiteration of my study purpose in lay terms. This approach was entirely new to participants, who were used to the scientists' total control of the conduct of interviews, the definitions used in their studies, and the framing of experience in biomedical terms. The idea of developing their own understanding of the purpose of the research, and helping to create a definition of empowerment through community participation was novel, but not unwelcome. Many of the Alphas in the sessions had opinions about what was important about their condition that had not been addressed in scientific, or epidemiologic, studies.

Access: Hiring-In and Participant Observation

This study is based on more than nine years of involvement with, and close observation of, the Alpha-1 community. This involvement, or hiring in, included six years as Director, Research and Grants Programs, and three years as a Research Consultant for the Alpha-1 Foundation. Hiring in is a recognized position for an anthropologist to assume, and can provide a level of familiarity that supports a more accurate assessment of the particular domain (or community) under study (Downey and Dumit, 1997). In my position at the Alpha-1 Foundation, I was responsible for managing, and expanding, the research network, and regularly interacting with Alpha-1 researchers, industry partners, government officials, lobbyists, and organizational leaders from other disease communities. I had some contact with the patients in the community, but, the focus of my job was to provide research resources, and maintain contact among scientists, and clinicians, working in the field of Alpha-1.

This hiring in experience gave me insight, and access, that would not have been possible through participant observation alone, and I was able, through my position, to evolve from being an outsider in the Alpha-1 community to being an insider (Keith, 1980). My employment also enabled me to reach a stage where data collection was refined to an investigation of specific

areas of interest, and I was able to give an increased personal attention to key informants. Through these activities, I was able to gain insight into informants' subjectivities and "the inner workings and meanings" of Alpha-1 culture (Keith, 1980). It was also my position in the community, and contact with my acquaintances, that allowed me to recruit a sufficiently diverse cohort for the study (Penrod et al. 2003). The normal channels of recruitment are highly controlled in the Alpha-1 community, both by the researchers and IRB approvals, and by the Alphas' awareness of allowable protocols in research. But, I was able to contact people I knew directly, and initiate recruitment without having to go through the time-consuming process of obtaining approval from organizational administrators. I was also able to easily arrange interviews, and observations, of researchers, physicians, and nurses from among a wide field of acquaintances in the research community. I had served the research community for many years, had many friends among the researchers, and, from feedback I had received, knew I had gained a reputation as someone fully invested in the Alpha-1 community – this proved very useful during recruitment of medical professionals for this study.

The motivation to observe the Alpha-1 community, while working among them, grew out of my background in both anthropology and research administration, and prior work among marine scientists. I had been exposed to the cultural domain of scientists for over 28 years, and this led to research questions about the differences between types of scientists, and the reasons why individuals chose a particular branch of science as their focus. Noticeable differences, for example, were immediately apparent between marine scientists and physician/researchers, and these differences seemed significant, particularly in terms of the level of commitment to patients, the consumers of scientific investigation, as opposed to the detachment of research scientists from the general public. The opportunity to observe the variations in the conduct of medical vs.

marine research, and a growing curiosity about why the patients in the Alpha-1 community seemed so much more empowered than any other type of patient I had ever encountered, became the basis for this study.

In addition, as a person with relatively minor lung problems, I was also motivated to understand how to develop a positive image about a physical disability, and how to achieve a level of empowerment, and self-management, related to my own condition. I became aware, during the course of the study, that my medical condition, and having a mother with advanced Chronic Obstructive Lung Disease (COPD), gave me much greater access to the patients in this community, although this had little effect on my interaction with physicians and nurses. I became, in study participants' eyes, both a researcher and a patient myself, with many of the same concerns, and issues, as they had. This was a crucial transition to make, as many patients noted that they first experienced me as one of the scientists, using terminology they did not understand, and even more problematically, as a representative of the Alpha-1 Foundation rather than an objective, disinterested researcher. The Alpha-1 Foundation does not provide support for patient activities, and is, therefore, considered as operating fully within the medical, not lay, domain. As a representative of that organization, I could have been experienced as less than interested in the patients' concerns, and efforts were made during interviews, and focus group sessions, to clarify my role as an anthropology student at the University of Florida, rather than an employee of the Alpha-1 Foundation. There was also the need to make this distinction to the physicians and nurses interviewed, and to ensure them that their responses to my questions and their opinions about the Alpha-1 community would not be shared, or provided, to the Alpha-1 Foundation, but, would be considered academic research data conducted under the auspices of the University of Florida, Institutional Review Board's approval.

Without my previous years of employment in the community, this study might have taken years to accomplish. I would have had to learn the medical terminology, familiarize myself with a complex community infrastructure, become acquainted with key members of the scientific and patient communities, and spend time traveling to Alpha-1 centers around the United States, and Europe. In addition, without the hiring in experience, the study might have remained firmly rooted in an etic perspective. Instead, my study has been an evolving examination of empowerment created by me with the active involvement of members of the community. Through this coordination and interaction, the study results will more likely address the issues of importance to the community, reflect empowerment as it is experienced in the community, and provide policy recommendations that are based on existing resources, and potential for implementation.

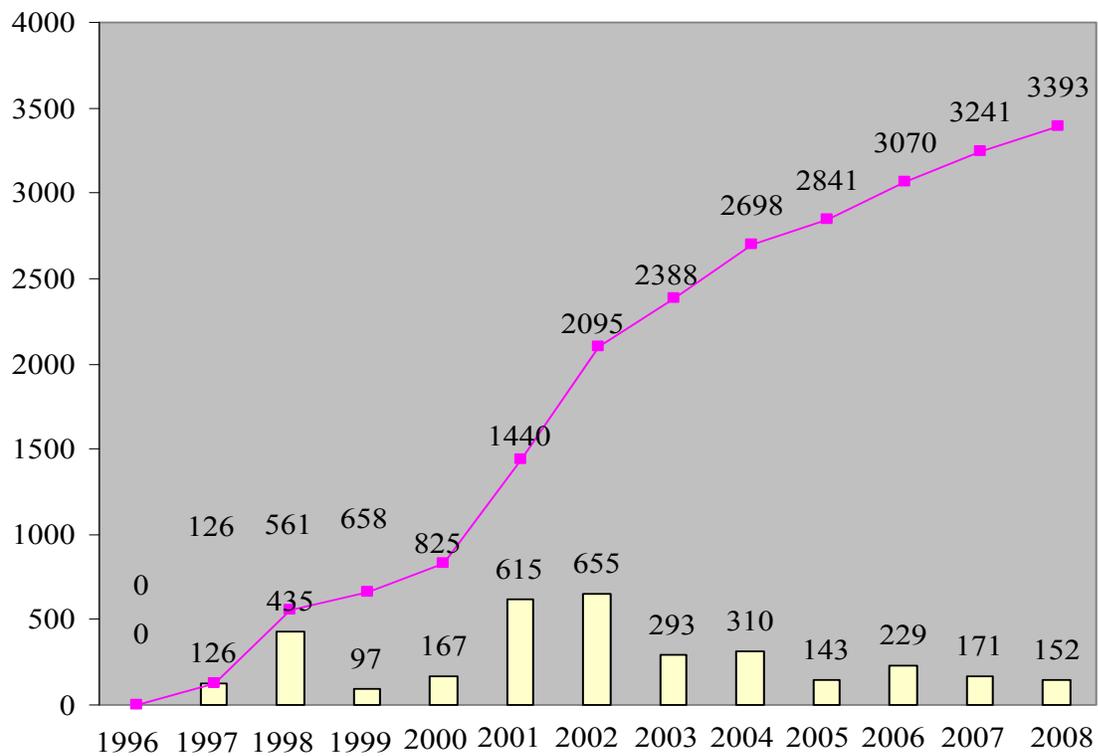


Figure 2-1. Alpha-1 Research Registry enrollment by year

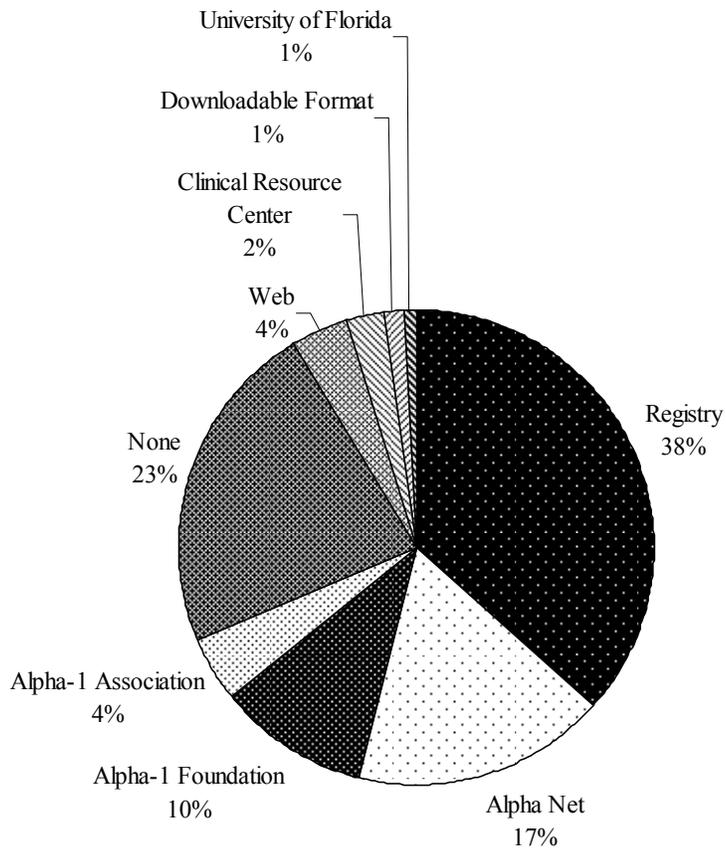


Figure 2-2. Source of Research Registry applications since 2000

CHAPTER 3 THEORETICAL FRAMEWORK

Introduction

In Chapter 2, Alpha-1 Antitrypsin Deficiency (Alpha-1) is described as a variety of modalities. It is first described as an individual's experience of increasing illness, then as a genetically caused, but rare, condition affecting the health of infants, children, and adults, as a potentially stigmatizing social identity as a patient, as an economic burden, and, as an interesting and challenging biomedical research topic. Examining Alpha-1 through such multiple locations and enactments may help to realign the importance, even centrality, of the patient's experience. However, a more socially oriented approach is not meant to diminish the importance of the biomedical understanding of Alpha-1, but to fully incorporate all aspects of the Alpha-1 experience into our anthropological assessment of the condition, and the community. For, much of what is known about Alpha-1 is biological. Since the discovery of Alpha-1 in 1963, a great deal of progress has been made in the biomedical domain to identify those affected (Anonymous, 1989, 1997 and 2003; Needham and Stockley 2004, Sandhaus, 2004), and to elucidate the underlying factors contributing to the pathogenesis of disease associated with this genetic condition (Blank and Brantly 1994, Sveger et al. 1997, Stoller 1998, Marcus et al. 1998, Soy et al. 2006). Although, there are some commonalities with other rare genetic lung diseases, such as Cystic Fibrosis (CF), Alpha-1 was discovered more recently, and during a time when societal attitudes about the inviolability of science, and the practice of medicine, were undergoing a major shift.

Understanding Alpha-1, and empowerment within the Alpha-1 community, must, therefore, be considered in the context of the existing societal attitudes at the time of its discovery. For, unlike CF, which was discovered in the late 19th century, Alpha-1 was first

diagnosed during an era when individuals and groups were embracing the concept of the power of the patient, and patients' right to information, and privacy, during the course of their medical diagnosis and treatment (Botkin, 2001, Merz et al. 2002, Sharp and Foster, 2002). There were concurrent changes occurring in United States society in the late 20th century relating primarily to loss of trust in authority (Teff, 1994; Hall et al. 2001). I believe the loss of trust in authority promoted a change in public attitudes relating to the role patients play, and, may have been a central factor in the increased power of the patient, particularly in the rare genetic disease communities. But, these same social factors (loss of trust, rise of patient empowerment), may also signify a shift in the traditional hegemonic relationship between doctors and patients in the United States (Teff, 1994; Hall et al. 2001). This study explores both aspects – the factors leading to empowerment in a particular rare, genetic disease community, and the larger implications of patient empowerment relative to the role medical professionals play in identifying, and managing, disease. And, the purpose of this chapter is to delineate the underlying anthropological and linguistic theories that frame the study design, and the relation of this research to a larger body of medical anthropological studies on disease communities, and the illness experience, in the United States.

Contextualizing Empowerment

Social Conditions

Patient empowerment has been variously defined in the literature as patient autonomy, knowledge, and as representing the potential for social transformations (Teff, 1994; Leps, 1995). In studies of empowerment, the patient is characterized as a consumer rather than recipient of medical care, and as a “counter-availing authority” to medical professionals (Clarke et al., 2003, Schlesinger 2002). Although, ‘consumer’ instead of ‘recipient’ may seem a subtle distinction, a

consumer connotes someone who makes active choices, whereas a recipient, or patient, connotes a more passive role (Clarke et al., 2003).

Preliminary research explored several key social and historical factors that may have contributed to a shift in power between healthcare professionals and patients, including the loss of faith in the American medical profession that Schlesinger (2002) characterizes as a “deprofessionalization”. This deprofessionalism refers to the loss of doctors’ autonomy and authority. At the heart of this authority is the idea that physicians possess a special knowledge that helps them make appropriate decisions about treatment of illnesses. A secondary source of medical authority is the belief that professionals will act as “reliable agents”, protecting the patient interests and well-being (Schlesinger, 2002). However, this authority, and the legitimacy of the medical profession, has been increasingly challenged by the growing influence of other stakeholders in healthcare and illness, whose agendas conflict with those of the medical community. This countervailing authority includes, the empowerment of patients who are the consumers of medical care, and the increasingly active role of patient organizations, and patient advocates (Schlesinger, 2002).

The growing knowledge about Alpha-1, and its potential impact on those affected, was also embedded in a social framework that had become increasingly biomedicalized. The growing reach of medical jurisdiction over health itself, in addition to illness, disease, and injury, and the commodification of health, led to the production of new “technoscientific identities” (Clarke et al., 2003). These identities are made possible by biomedical technologies that identify high-risk statuses, such as the electrophoretic and gene array technologies that allowed for the identification of deficient states of Alpha-1 Antitrypsin Deficiency.

In the biomedicalization era, the focus is no longer centered around a deterministic view of illness, disability, and disease, but on “health as an ongoing moral self-transformation” (Clarke et al. 2003). This new focus on health, risk, and surveillance is transforming the production, distribution, and consumption of biomedical knowledge, and transforming our image of bodies and identities in illness. The focus on health and surveillance may paradoxically lead to a less medicalized view of health, as the primary site of responsibility shifts from the medical profession to responsibilities shared in collaboration with the individual patient-consumer (Clarke, et al. 2003, Gunderman 2000; Loughlin 2003; Anie et al. 1996). Historically, medical decision-making was paternalistic, dominated by medical professionals maintaining their primary role through non-disclosure, and an insistence on deference to their authority. In this traditional medical paradigm, good communication, and effective interaction with the patient, was viewed as marginal to medical practice (Teff, 1994). A new model for decision-making is emerging, however, that places greater emphasis on patient autonomy, and therapeutic alliances that move beyond the view of medical decisions as the application of scientific knowledge to a passive patient (Hanna, 1998; Warren et al. 1998).

The decline in social attitudes about medical responsibility for health may also be due to an erosion of trust in the medical profession, and doctors. Mechanic (1996) notes confidence in medical leaders has declined from 73% in 1965, to 22% in 1993. Although, this trend reflected a general decline in trust in social institutions, the changes in medical practice to managed care, and the availability of information about medical errors, have also contributed to loss of trust in medicine (Mechanic 1996). In addition, as longevity in America increases, and medical practice increasingly serves chronic care patients; several studies have demonstrated a widespread discontent with the medical approach to chronic care. These studies credit the growth of patient

self-help groups as a new social movement that will address chronic care concerns more effectively than traditional biomedical approaches (Hanna, 1998; Mechanic, 2001). Patients are urged to be more thoughtful and skeptical as consumers of medical care, and to question their medical treatment (Mechanic, 1996). As noted above, the change from patient to consumer may seem simply a change in terminology, but, it is suggestive of a significant change in how we think of physicians, and the previously unquestioned dominance of their role in healthcare and disease management (Mechanic, 2001).

Another factor in a shifting hegemony between patients and healthcare professionals can be attributed to the wide availability of Internet technologies, since the mid-1990s, that promoted a more rapid dissemination of information on medical conditions (Armstrong, 2003, Finkelstein and Friedman, 2000; Goodman, 2005). Those with access to the Internet, and affected by chronic or genetic illnesses, now had the means to educate themselves about their condition, and be in contact with others with the same condition. Current developments in telematics may represent a new social movement centered on the “need to know”, and where information becomes a consumer good (Leps, 1995; Finkelstein and Friedman, 2000). This need to know, in turn, activates social, economic, and political relations, and is a major factor in what Pierre Bourdieu calls the *habitus* of Western industrialized societies (In: Leps, 1995). Through the increasing dominance of computer resources, the discursive affirmation that knowledge is power will increase the possibility for large-scale empowerment through information (Leps, 1995). Wagner et al. (1996) note increases in self-efficacy, and the confidence that one can manage illness, will actually improve disease self-management, change key behaviors and affect illness outcomes for many chronic conditions. Wagner, and Leps also cite the use of lay leaders in developing education programs, and patient registries, as particularly empowering approaches.

Anthropological Considerations

As an applied anthropologist, my primary concerns are what I can do about a situation I observe. However, my research is based on theoretical frames, and research methods, that have been successfully utilized by anthropologists and linguists working in the medical domain. Since little research on patient empowerment, or other social issues relating to genetics, has been conducted in rare disease communities, I had to find appropriate models from a variety of disciplines to use as the basis of my research design. Using such established frameworks to guide my research design situates my research within a larger body of knowledge. And, by incorporating a number of different, well tested approaches, my research results may be more accessible, and understandable, to both social scientists and medical scientists. I use this Chinese menu approach, (one from Column A and two from Column B), because it is useful without being constraining.

I began with elements of a grounded theory approach (Addison, 2002; Agar, 1980, Irurita, 1996; Kearney et al. 1995; Strauss, 1992). I consider it an appropriate approach in that I seek to elucidate the social, cultural, historical, and linguistic background features that are the ground, or frame, of the practice of medicine, and the role of patients. And, I chose this approach because my work is ‘grounded’ in the everyday practices of members of the Alpha-1 community, and their physicians. Addison (2002) had proposed central principles of grounded research that I found useful as a way to approach the issue of empowerment. These central principles include, immersing oneself in the participants’ world, maintaining a constantly questioning attitude, and analyzing, in a circular progression, between parts and whole, foreground and background. Having immersed myself in Alpha-1 culture for nine years, I was able to make assessments from a concerned, involved perspective, and to analyze what I observed as situated within a cultural, and historical, context. Grounded theory principles also provided me with an understanding of

the possibilities in the role I play as an anthropologist in the Alpha-1 community - I could offer my own account of what I had observed, describe how the issue developed, and recommend future actions, or positive changes (Addison, 2002)

A grounded approach requires a certain degree of flexibility, and the need for a heightened sensitivity to the actual responses of participants, as the primary means of generating hypotheses about empowerment (Charmaz, 1994; Abrahamsson, 2002). Using this approach, I focused on examination of the social processes and existing problems in the Alpha-1 community, and looked (and listened) closely at how Alphas handled, or resolved, their problems. There was no need for me to fit data into preconceived or standardized categories, and I was able to interpret the data freely, and modify initial hypotheses or understandings, after analysis of interview and focus group data. By using this method in the Alpha-1 community, crucial components of empowerment, (or lack of empowerment), represented by the diagnosis event, the cost of illness, and the often stressful experience of caregivers, were identified emically, and incorporated into subsequent interviews and focus group sessions.

Elements of participatory research also figured in the research design. This included, the emphasis on an emically derived definition of empowerment, and the involvement of community gatekeepers (AlphaNet coordinators), in recruitment of an appropriately diverse participation in the project (Penrod et al. 2003; Johnson, 1990). As a proponent of participatory action research, I believe that the people most affected should have the most say in the way their experience is characterized, and analyzed. The affected individuals should also have considerable input into the research questions being asked, and, ultimately, have ownership of the information derived from a study about them. Through their involvement in this participatory research project, Alphas had the opportunity to look into their own lives, and explore the meanings of their

experiences with Alpha-1, as recommended by Ervin (2005). But, study participants also had the opportunity to influence the construction of the theoretical models of empowerment, set priorities among the potential components of empowerment I identified, and, even alter the basic questions being asked in this study. Because the patient focus groups centered on the lived experience of Alpha-1, as articulated in lay terms (rather than the more traditional approach to Alpha-1 as a medical condition that is described in medical terminology), the sessions served as a powerful, and empowering, means for participants to help construct knowledge about Alpha-1, and feel fully invested in the process. By allowing this active community participation in the research process, the sessions, and interviews, appeared to provide an energizing, and educational, experience for the majority of participants. Subsequent feedback from participants confirmed this impression of participation as a positive experience.

Linguistic Theories

I also utilized key concepts from anthropological linguistics as a theoretical framework for the study. Linguistic relativity, or the domain-centered approach described by Ochs and Capp (1996), was employed as a means of understanding the significance of differences in the languages used by stakeholders in the community. Domains, as defined by Hymes (1962), represent specific types of speaking, rather than speech communities as unified wholes. In more recent sociolinguistics research, domain is used to refer to a sphere of life in which verbal, and non-verbal, interactions occur, such as the institutional discourse heard in medical contexts (Boxer, 2002; Borgatti, 1998; Maynard, 1991). Conceiving of the interactions between patients and physicians as the intersection of distinct domains, led me to consider empowerment as a transitional process bridging the personal and medical domains (Maynard, 1991; Zwitter, 1997). Using a domain-centered approach might explain the reason for the differences between how doctors, nurses, Alphas, and family members experience Alpha-1, think of how to treat it, or

explain it to others. I operationalized this by noting how each study participant selected from versions of medical, or lay, terminology to organize the overarching domain of Alpha-1. The domain-centered approach also provided a means for understanding the commonalities in belief between Alphas and their doctors, even when expressed using markedly different terminologies.

Linguistic relativity, apart from identification of domains, influenced my thinking about how to analyze empowerment, and led to consideration of the nature, and extent, of the distinctions between the language of medicine, and the patient's narrations of diagnosis and illness (West, 1984). The doctrine of linguistic relativity had evolved as a Boasian reaction to the denigrating attitude toward unrecorded languages prevalent in the late 19th and early 20th centuries. Boas influence on his student Sapir, and Sapir's student Whorf, led to the formulation of their oft-cited hypothesis about linguistic relativity, namely, that languages have great structural diversity, and, because of their differences in structure, influence the world-views of their speakers (Whorf, 1956, Sapir, 1951). .

The Sapir-Whorf hypothesis has spawned radical adherents, and vociferous opponents who propose what they claim are more rationalist assumptions (Kay and Kempton, 1984; Whorf, 1956; Brown and Levinson, 1993; Lucy, 1997, Coupland and Jaworski, 1997; Wassman and Dasen, 2000). Several studies, seeking empirical proof of linguistic relativity, have focused on color perception (Davies, 1998; Davies and Corbett, 1997; Berlin and Kay, 1979; Brown, 1976; Heider, 1972; Kay and McDaniel, 1978). The color studies prior to 1969 tended to reinforce the validity of linguistic relativity, however, several of the studies after 1969 contested linguistic relativity, at least as it relates to color perception (Kay and Kempton, 1984). There are other studies that demonstrate at least weak linguistic relativity; one study relates to differing time perceptions between English and Hopi speakers (Whorf, 1956), another study looks at language

and spatial perception. The latter studies compared spatial perception among Balinese, and other Asian societies, that differ markedly from the academically predominant, Western European spatial schema.(Wassman and Dasen, 2000; Levinson, 1996; Barnes, 1993).

Because the Sapir-Whorf hypothesis has been so variously interpreted, I focused on particular quotes, by Sapir and Whorf, which I found particularly applicable to my study of empowerment:

“The real world is to a large extent unconsciously built up on the language habits of the group (Sapir, 1951).

“The categories and types that we isolate from the world of phenomena we do not find...because they stare (us)... in the face. On the contrary the world is presented in a kaleidoscopic flux of impressions which have to be organized in our minds. This means, largely, by the linguistic system in our minds (Whorf, 1956).

Too many of the Alphas that I interviewed had noted the overwhelming feelings they had when first diagnosed, and the difficulties they had assimilating masses of new information into an understandable scheme they could cope with. A linguistic approach would help me elucidate the organization, or structure, of their knowledge about Alpha-1. For, if Whorf’s hypothesis is true, the structure of the patient’s ‘native tongue’, or lay language about Alpha-1, would strongly influence, or fully determine, the world-view the patient acquires. In addition, if linguistic relativity exists, then the language of the illness experience, that the patients use, is equal in validity to the medical terminology that the physicians, and nurses, use to describe, and cope with, Alpha-1.

The theory of linguistic relativity also suggested there might be viable links between the lexical categories expressed so differently by doctors and patients. To a patient, being ‘very sick’ is a meaningful expression, and situates a current experience in relation to previous, or anticipated, states of illness. To a nurse or physician, however, ‘very sick’ expresses little that is useful in terms of diagnosis and treatment, until the phrase is translated into quantitative

measurements of body temperature, expiratory volume, heart beat, blood pressure, and audible cues from the lungs. A radical linguistic determinist would propose that, instead of reconfiguring the patients narrative into the predominant language of the medical domain, the divergent narrative could be ‘calibrated’, or incorporated, through “linguistically unbiased description of the same situation or experience” (Lucy, 1997). Although, I remained open-minded about the utility of linguistic relativity, I also considered other linguistic theories and approaches, such as Speech Act Theory, critical discourse analysis (CDA), narrative analysis, the ethnography of speaking approach, and interactional sociolinguistics (IS) (Boxer, 2002; Jaworski and Coupland, 2000).

Critical Discourse Analysis seemed an appropriate theoretical frame for a study on empowerment, as it could help me to expose important social problems stemming from discriminatory, or manipulative, language use by medical professionals. For, if linguistic relativity was not a valid assumption, than the predominant dynamic in medical care would be the maintenance of power through language, rather than the concurrent development of different, but equally valid, ways of expressing and experiencing disease (Maynard, 1991; West, 1984). Likewise, IS, with its specific focus on miscommunication between different groups, would allow me to study miscommunication in order to discover how it evolves, and how it can be avoided (Gumperz, 1977; Boxer, 2002). And, the ethnography of speaking (EOS) approach would provide what I consider, as an anthropologist, the essential framing for the speech acts under study – the larger social context. The EOS and Speech Act Theory methods, used in the study, are described in detail in Chapter 5: Looking and Listening: A Linguistic Analysis of Medical Encounters.

The theories underlying narrative analysis helped me to understand the potential of this study to empower participants, and, gave me specific tools for understanding language choices within the Alpha-1 community. Using a linguistic framework, Alphas' narrations were seen as empowering for their ability to provide patients, and caregivers, with an opportunity to impose order on otherwise disconnected events, and to create continuity between past, present, and imagined worlds (Ochs and Capps, 1996; Labov and Waletzky 1967). Narrating their stories, also allowed Alphas to socialize their emotions, and identities, as well as consolidate their membership in the Alpha community (Hansen and Liu, 2001; Giles and Johnson, 1987; Tajfel, 1981). Since, their narrations are versions of reality, I saw how Alphas were able to re-construct their condition in the most positive terms, frame their past experiences in terms of present knowledge, and empower their individual experience through identity, and membership, in the supportive network of the Alpha-1 community (Hansen and Liu, 2001; Waitzkin and Britt, 1993; Labov and Waletzky 1967; Lucy, 1997; Colby, 1966).

Using linguistic analysis, I was able to perceive qualitative differences between the disjointed narrations of newly diagnosed individuals, and the more positively expressed, and cohesive, narrations of those diagnosed year's earlier (Waitzkin and Britt, 1993). Those diagnosed earlier had the chance to refine their narrations over time, re-order sequences of events to make them more understandable, and fit various topics into an overall schema, or structure, called Alpha-1. Newly diagnosed individuals are still trying to digest information, and relate their newly defined experience as a diagnosed Alpha to entire fields of medicine and genetics. According to those I interviewed, diagnosis, and introduction to the world of Alpha-1, was a universally overwhelming experience, and early narratives of the experience reflect a degree of confusion about the meaning of Alpha-1 that is missing from later accounts.

In addition to the theoretical influences from anthropological linguistics, I also employed specific linguistic analytic tools. These are described below.

A review of the linguistic literature led to consideration of medical encounters and diagnosis events as locutionary speech acts (Austin, 1962; Jaworski and Coupland, 2000). To a linguist like Austin, a diagnosis is an illocutionary act because it is asking or answering a question, giving some information or warning, making identification, or giving a description (Jaworski and Coupland, 2000). Diagnostic utterances can also be considered as performative speech acts, as they lead directly to actions and responses (Jaworski and Coupland, 2000). Performative speech acts in medical care can include neutral utterances such as announcing an intention to conduct a procedure ('now I will listen to your lungs'), provide identification, or make an appointment. Performative acts can also include utterances that are more apt to produce an emotional, psychological, or physical response, such as criticizing the patient, warning about risky behaviors, pronouncing a specific diagnosis, advising, suggesting, ordering, or demanding. However, there is a continuum between a physician's advising, or merely suggesting, or actually ordering, and the subtleties inherent in lexical choices can have drastic consequences when the intent of a diagnosis, and recommendations for disease management, are misunderstood. A speech analysis of medical encounters was seen as a potentially useful approach for discerning effective performative choices.

I also evaluated the three medical encounters I observed for their Gricean cooperative principles (Grice, 1991; Jaworski and Coupland, 2000), and as distinct speech genres (Bakhtin, 1986; Jaworski and Coupland, 2000). Using the Gricean approach, the diagnostic event is seen as violating several features that signify cooperation, namely that the participants have some common immediate aim, and that the transaction continues until both parties agree it should end

(Jaworski and Coupland, 2000). Although, both participants in a diagnostic event are concerned about the condition Alpha-1 Antitrypsin Deficiency, doctors and patients have distinct, and not necessarily mutual, aims regarding a diagnosis, and the doctor, in the majority of cases, controls the length of the encounter. The doctor's aim, in a diagnosis, is to demonstrate expertise and knowledge, to represent the final authority for decision making, and to provide sufficient information for the patient to comply with recommended treatment plans. The doctor may also be motivated by the goal of publishing new information in unexplored fields, and in carving out their professional competence as distinct from other practitioners. Unfortunately, the doctor is also under the constraint of time and case management, in this era of managed care, and brevity may be institutionally mandated, and an overriding aim in a diagnostic event.

The Alpha-1 patient, on the other hand, is in the process of creating a new self identity based on designation as a rare, genetic disease patient (Ochs and Capps, 1996; Clarke et al., 2003). This new identity includes emotional, and psychological, responses that can veer between feeling victimized or feeling empowered; what becomes evident in the course of this study, is that an empowered identity may give the individual a better chance of coping with their diagnosis.

In the process of creating a new identity, a diagnosed individual must construct lay meanings about complex biological processes, and relate often newly acquired understanding of human biology to their own symptoms. The diagnosed individual's concern is not necessarily what Alpha-1 is, or how it behaves on the molecular level, but how their condition affects their day to day work and family life, their ability to breathe, and the length of their life. In this regard, the diagnostic speech event may also violate the Gricean principles of quantity, manner, and relevance, for the physician seems to have a different focus than the patient on what is important

about Alpha-1, may tell them more than they need to know about certain topics, and, unfortunately, make lexical choices that are obscure, ambiguous, or lengthy.

The reason for poor lexical choices was discerned using Bakhtin's theory of speech genres, or subsets of language developed by culturally specific communities. Given the emphasis on mastery of medical terminology as indication of professional competence, the often obscure language used in medical encounters may be because doctor's talk is so firmly rooted within the medical speech genre. Physicians specific utterances in a diagnostic speech event are links in a "very complexly organized chain of other utterances" (Bakhtin, 1986), and the doctor's diagnosis is based on a well developed, prior knowledge of biology, genetics, and more narrowly, disease pathogenesis and clinical outcomes. The doctor's description of the significance of a diagnosis may, therefore, utilize descriptive terms that for them, as medical professionals, sum up complex processes, but, for their patients, are words they can barely pronounce, much less understand. Patients have often expressed feeling overwhelmed during their diagnosis, and subsequent office visits, with the amount of new terminology they are asked to understand, and respond to. These patients may find themselves unable to fully comply with treatment options, because they lack understanding of their condition, and it's progressive, and degenerative, nature.

An analysis of medical visit conversations was also evaluated according to Grice's theory on implicature, or the kinds of implication that speech exemplifies in ordinary conversation. Together with Bakhtin's definition of culturally specific speech genres, the conversations in a medical office are seen as existing within, and dependent upon, larger linguistic and social contexts. These contexts, including the use of a particular culturally-specific speech genre, may not be consciously recognized by the physician or nurse, or alluded to directly. When a nurse communicates information to the doctor, for example, there are unspoken references and

implications in the phrases used. A comment on heightened white cell count, or higher number of eosinophils, communicates, through implication, the presence of infection regardless of any observed external symptoms, comments by the patient, or explicit statement regarding infection.

Grice's principles, and Bakhtin's approach, both seem to confirm the ongoing existence of a traditional hegemony between medical professionals and patients, however, analysis of the speech event of office visits is necessary to evaluate this in practice. The study, therefore, examined speech gaps, and miscommunications, through micro-sociolinguistic phenomena such as code switching, strategies of neutrality, and choices between formality and informality (Scotton 1976; Irvine 1979). Code switching research is of particular interest, as I had observed code switching as one means of maintaining the boundary between physician/nurse and patient/caregiver (Hansen and Liu, 2001). Code switching has been proposed as a means of contesting hegemonic relations (Gumperz, 1982; Giles and Johnson, 1987; Heller, 1988; Nelson, 2001) since, in the medical domain, code switching is common, and frequently used to mark utterances with authority and accuracy. I observed nurses frequently code-switching from the lay language they use with patients, to the medical terminology that they use with physicians. Once they switch to medical terminology, nurses are able to summarize long, sometimes rambling narratives into discrete symptoms, and precisely define physical measurements indicating the patient's status on the disease/health spectrum.

In the Alpha-1 community, I observed evolutionary stages of empowerment relating to the patient's ability to code-switch into medical terminology. Rather than using the opportunity of the office visit to challenge the predominance of medically-centered definitions of Alpha-1, Alphas tend to recast their experience in medical terms, even though such terminology may "lack the semantic power to communicate (their) intended meaning" (Nelson, 2001). Although,

embracing the value of code-switching may denote patient's taking responsibility to learn the language of their illness, it also denotes the centrality of the medical domain in defining their illness.

Formality, and informality, was also considered as linguistic markers of existing hegemonic relations in medical care. Formality is the prevailing affective tone of medical encounters, and my observations of patient and physician interactions noted the increased structuring of their discourse, the ritualized expressions, and the respect for the normative social order so characteristic of situational formality (Irvine, 1979; Bloch, 1975). The degree of formality, and level of politeness, expressed during medical office visits indicates seriousness, and a heightened respect for a traditional social order. Formality, since it expresses normality and tradition, may therefore also indicate a "coercive political establishment" that constrains the individual to conformity (Irvine, 1979). The maintenance of formality in physician-patient relations may serve to counteract any gains patients have made in learning about their condition – they still must express their experience formally, and in medical terms, to be heard by the physician.

Linguistic analysis was ultimately chosen as the best means of highlighting the often enormous gap between medical speech genres and patient speech genres in a way that promotes more effective bridging of that gap. Nurse-patient interactions were also examined, since I had observed how miscommunications that arise in the doctor-patient encounters were often resolved, or overcome, by the nurse or physician assistant.

Language use in the community

The choice to conduct a study of the Alpha-1 community using linguistic methods, also grew out of the observation that, in the Alpha-1 community, the use of the words 'patient' and 'disease' is discouraged in public discourse, and in publications. Since, Alpha-1 does not

necessarily lead to disease (Sandhaus, 2004), individuals, who are diagnosed, refer to themselves as Alphas, not patients, and, even some researchers prefer to call Alpha-1 a genetic condition, not a disease (Carrell, 2004). Capturing the full flavor of the Alpha-1 community would, therefore, be more appropriate as an examination of how well individuals in this community cope with their condition and maintain their quality of life, rather than a study of the extent of disability or disease.

Similar to the subtleties associated with the use of the word consumer vs. patient, referring to themselves as Alphas, rather than patients, may denote a conscious attempt to influence perception through vocabulary. Whether a conscious choice or not, the common connotations for the word Alpha in United States culture evoke images of Alpha male primates, or Alphas as first, best, or primary; referring to themselves as Alphas is to embrace these common, positive connotations. A further linguistic advantage in this community was the switch from Orphan Disease status in the 1990s to Rare Disease status. Rare connotes a more positive image than orphan, giving members of the Alpha-1 community a further linguistic advantage, and self-perception that they are somehow special, rather than bereft.

In addition, the community has chosen to express their goal as a search for a cure. Initially, scientists involved in Alpha-1 research objected to the use of the word cure, preferring to think of their efforts as developing therapies, or searching for a potential cure. However, the patients insisted on using the word cure and, eventually, the scientists also began to think of their work as a search for a cure, and to use the word cure to refer to their efforts.

Recognizing the community standard for the use of positive expressions, empowerment was defined in pre-study recruitment of Alphas as ‘those things that make you feel in control, help you cope better with Alpha-1, understand your condition, or control your illness better’. I

also noted during recruitment of both Alphas, and medical professionals, that empowerment is defined in the literature as including components of ‘better communication with your doctors and nurses, access to resources, education, and services, good family support, and interaction, and support, from fellow Alphas’.

Communicative competence

I am well known in the community in my former position as Director, Research and Grants for the Alpha-1 Foundation. I have been told by colleagues in the community that I have a reputation as a science geek, which I consider a compliment. For, I am unendingly interested in the science of Alpha-1, and willing to spend considerable time to understand the biology of Alpha-1, and the research being conducted on this genetic condition. I spent the past nine years asking questions, reading scientific articles, visiting genetic testing labs and basic science labs, observing medical and diagnostic procedures, and organizing scientific meetings about Alpha-1. I want to be competent in the language of Alpha-1, and have some degree of understanding of this genetic condition.

One of my jobs at the Foundation was to maintain, and regularly update, a very large bibliography on Alpha-1 research. This bibliography had been originally compiled by Dr. Mark Brantly, and contains over 12,000 titles relating to Alpha-1 research and clinical care. I was trained by Dr. Brantly and Dr. Sandhaus on the use of EndNote software to maintain the bibliography, and how to search for relevant articles. Dr. Sandhaus gave me additional instruction in how to review abstracts, and how to compile topic bibliographies. In 2000, I visited the NIH, National Institute of Environmental Health and Safety facility, in Research Triangle Park, where a computer consultant gave Dr. de Serres and me a lesson on how to search PubMed for recent publications, and in the use of EndNote to manage our bibliographies. From 2000 until 2006, Dr. deSerres conducted a twice-yearly review of recent publications, and sent me the

listing, which I then incorporated into the larger bibliography. Throughout my tenure at the Alpha-1 Foundation, I was contacted, on average, once a month, and asked to search the bibliography, and provide a scientist, a student, or a patient with the resulting list of relevant articles on a specific topic. Typical search topics included, co-morbidities of Alpha-1, environmental impacts and Alpha-1, psychosocial impacts, articles about screening and detection, or a list of articles describing diagnostic technologies.

Another responsibility I had at the Foundation was to organize scientific meetings and workshops. This included participating in planning sessions with the scientists, who would designate the topic, and begin to outline the agenda. Over time I was able to understand enough about who did what in Alpha-1 research, to suggest appropriate speakers and moderators, draft the conference book, and assist with conference grant proposals. During the many months of planning with conference chairs, I had numerous opportunities to learn about specific areas of Alpha-1 research. Dr. Sifers, who chaired the 2000 Conformational Disease Conference, was particularly patient, and spent many hours explaining his molecular research, and its implications for Alpha-1 patients.

A third key responsibility, during my years in the community, was to help create and manage a peer-reviewed grants program. Although, I was not expected to understand the science on the level of the reviewers, I did assist in summarizing the proposals in lay language for the Foundation's President and Board members, and reviewing the proposals myself as a way to learn more about Alpha-1. In addition, as noted in Chapter 2, I made a number of visits to laboratories and research centers, always with the purpose of understanding the science of Alpha-1.

My immersion in Alpha culture provided a useful context for understanding the oft-encountered Alpha-centric point of view shown by both Alphas, and the medical professionals considered expert in this field. At times, I felt like I was attending a ten-year pep rally, as there is a pervasive sense of rooting for the Alpha cause in this community, an almost obsessive concentration on finding a cure, an ongoing commitment to community building, and a competitive drive to be the first, the best, and the most effective among the rare genetic disease communities. But after nine years, these attitudes have become more understandable, and I fully embrace the positive identity of the Alpha-1 community. More importantly, knowing the Alpha-1 culture firsthand, and experiencing my own identity as a member of this community, have proven invaluable as a framework for understanding empowerment in this community.

Discussion

In light of socio-historical factors, the increase in patient awareness, and access to resources, linguistic indicators, and over nine years' participant observation of the Alpha-1 community, I hypothesized that members of the Alpha-1 community are noticeably empowered. However, questions remained - did empowerment in this community also signify a shift in the traditional power relations between doctors, nurses, and patients? Are patients challenging the logic for a biomedically-determined knowledge of this rare disease, or trying to gain mastery within the medical domain? Was empowerment unidimensional, with one form following another in linear fashion, or, was it multidimensional, and highly variable among individuals with the same condition? Were there core components of empowerment for all types of patients, or, was the form of empowerment in the Alpha-1 community unique to that community? And, finally, could components of empowerment be quantified in ways that allowed application of principles to other disease communities?

Anthropological and linguistic analysis might confirm whether Alpha-1 patients are empowered enough to be thoughtful consumers, forge therapeutic alliances with their caregivers, and question their medical treatment, or, if historical paternalistic modes of interaction between doctors and patients still prevail in practice.

Examining the issue of empowerment in the Alpha-1 community is not intended as an assault on the ultimate value of biomedical knowledge. Rather, it is an exploration of the role of the patient's experience in co-constructing knowledge about disease. Data on nurse-patient interactions was included to help elucidate the key role nursing staff, and physician assistants, play in effective communication with patients. In addition, a discussion of the role caregivers, and organizations, play in disease management and empowerment is included to provide the appropriate context for increasing our understanding of how patient empowerment is achieved.

CHAPTER 4 ANALYSIS OF COMMUNITY INPUT

Introduction

This chapter presents the methods used to elicit and analyze data, provides results of the narrative analysis of interview data and focus group sessions, and provides a preliminary definition, and model, of empowerment. An important premise of the narrative analysis is the value of patient-derived data, and emic definitions of empowerment. My commitment to the participatory nature of the study made it especially important that I fully understand the core communications of the narratives, and derive appropriate core themes, and sub-themes, on the nature of empowerment from the community's perspective. Through a careful analysis, and interpretation of the data, I could ensure that the study results reflect the nature of empowerment, as Alphas themselves experience it.

The analytic techniques I chose, and that are described below (in Narrative Analysis), led to development of a codebook of themes, and overarching meta-themes, relating to the experience of Alpha-1. These thematic components, in turn, became the basis for the design of a statistical analysis, and the means of testing the preliminary model of empowerment. .

Sample

The empowerment study sample consists of 49 members of the Alpha-1 Antitrypsin Deficiency (Alpha-1) community (see Appendix C). Of the 49 participants, 34 are Alphas, and 15 are caregivers or family members. Data for the study were gathered from June, 2006 to June, 2008 during interviews, and five patient/caregiver focus group sessions, at geographically dispersed regions in the United States. The focus group sessions involved an average of 8 participants in each locale; the largest group session involved 11 people, the smallest session involved 4 individuals. Data was also obtained during 10 semi-structured interviews with key

Alpha informants from 2006 to 2008. The study also included interviews, conducted in 2006, with 8 physicians and 3 nurses, and interviews conducted in 2007 and 2008 with 1 physician, 1 physician assistant, 1 nurse, and 4 laboratory personnel. The interviews with other medical professionals and laboratory personnel did not provide demographic data; and, are therefore not included in the narrative analysis described below, nor in the statistical analysis (Chapter 6: Quantifying Empowerment).

The relatively small sample size for this study was predicated on the requirements of ethnographic and linguistic research, the need for in-depth narrative data, and, the rarity of this condition among the general population. However, the small sample size, and preponderance of individuals diagnosed for many years, and more aware of their condition, ultimately influenced the information derived. This is, therefore, a study that does not fully delineate the comparison between empowered and un-empowered individuals, but one that describes, and defines, components of empowered individuals. Given the availability of a much larger cohort in the Alpha-1 Research Registry who can participate in the next phase of study, the limitations of the purposive sample is expected to be addressed by a larger, more random sampling of the community.

Methods

A purposive recruitment was conducted to maximize the quality of data through choice of articulate, and expert, key informants. Informants were chosen from among my acquaintances in the community, or after a recommendation from an organizational leader, or patient support group leader. Once key informants were identified, chain referral recruitment was conducted, primarily using AlphaNet coordinators, the official gatekeepers of the Alpha-1 community (Penrod et al. 2003; Juengst, 1998; Johnson, 1990). Four of the five gatekeepers who helped recruit for the study hold employed positions as AlphaNet coordinators. Each AlphaNet

coordinator is responsible for a large number of diagnosed Alphas in their geographic region, and, through their extensive network of contacts, was able to help me recruit a diverse set of participants for interviews, or a focus group session, in their area. The fifth recruiter, who was recommended by a nurse involved in Alpha-1 care, is an individual with a large network of Alpha acquaintances, and who is known as the “go-to” Alpha in her region when events need to be organized.

The diversity of participants was necessary, so that the data would adequately represent variance in the Alpha-1 community population. However, despite every effort to include the full range of experience of Alpha-1, there was limited numbers of newly diagnosed individuals willing to participate, and only one parent of a liver affected Alpha-1 child participated³. Although, the range of ages of participants was also limited – there was only one participant younger than 40 (a parent of a liver affected child), and only one participant over the age of 68 (the mother of an adult Alpha) – this did not seem a limitation, as the average age of participants (56.9) falls within the range of the average age of enrollees in the Alpha-1 Research Registry (50.2 years old at enrollment).

Interviews were held in the homes of Alphas and care givers, however, physician and some nurse interviews were conducted in hospital and university settings. I stayed at the home of one of the nurses, while visiting that region for a focus group session and interviews; she was to become a key informant, and provide information and feedback in a total of three interviews. Our first interview was conducted over breakfast, and ranged over her entire history with the Alpha-1 community, her relation with a recognized Alpha-1 physician, and changes in the community

³ Children were excluded from participating in the study; the average Alpha child is too young to articulate their experience, and despite the minimal risk of this study, concerns remain high in the community regarding involving Alphas younger than 15 years old in research.

that she had witnessed over 20 years. I also stayed in the home of one of my Alpha recruiters, and key informant, and in a guest room at the assisted living facility of one of the Alphas interviewed. Five of the ten interviews with Alphas were held in their homes; the three individuals I knew well before the study participated by phone; and two interviews were held in a restaurant and hotel, respectively. On these two latter interviews, I was accompanied by the Alpha recruiter, who in each case knew the interviewee well, and had recommended these individuals as particularly interesting subjects. During home based interviews, I was able to observe and assess the extra-linguistic context, such as the socio-economic level, availability and quality of family support, and the depth of the emotional impact of Alpha-1 on that individual. The opportunity to interview the individual in the comfort, and security, of their home was a particular advantage for discerning the emotional reactions, which in my experience in this community were more often hidden during medical the encounters, or down-played during focus group sessions.

Focus group sessions began with a description of the primary goal of my study (to define empowerment from the community's point of view), and a short explanation of what medical anthropology is, and how anthropologists approach a research question. I would then explain how the anthropological approach differs from the biomedical and epidemiological research and, that many of my study subjects were familiar with. I had a written sheet with my IRB approved sample questions and topics (see Appendix A), and after referring to it suggested that the group might want to explore some topics that academic literature had defined as components of empowerment. I noted specific topics, such as information and awareness, networking, family support, communication with physicians and nurses, and disease self-management. I did not, however, note the diagnostic event in the first focus group session. Based on the number of

narratives about diagnosis during the first session, and the obvious impact this pivotal event, and the way it is communicated, has on an Alpha and their family, this did become the introductory topic in subsequent sessions, and during interviews. It proved a useful introductory topic for, once the individual had explored their diagnostic experience, they seemed to naturally progress to discussion of initial periods of denial and depression, the compounding nature of their family's reactions, and their attempts to become more aware, connect with others with the same condition, or begin to manage their illness episodes. I had little need of prompting, and once an individual had begun to narrate their experience, they would touch upon additional, and common, impacts of their condition. All those interviewed, for example, mentioned the costs of illness and disability, the costs of therapies and devices; and the difficulties with insurance coverage. The majority (8 of 10) also noted the initially challenging interaction with family members frightened by the diagnosis, and by the implications of a genetic condition for their own health.

Research participants in the study responded to inquiries about their activities with assessments about their level of involvement in the Alpha-1 community, the amount of contact they have with individual patients, or patient support groups, and their interaction with medical professionals, all which are of direct concern in this study. Analysis of the patient focus group sessions yielded information on a variety of activities associated with patient empowerment, such as understanding of Alpha-1 Antitrypsin Deficiency and its pathogenesis as lung, or liver, disease, active involvement in healthcare management, advocacy on the local or national level, and active participation in research studies, and clinical trials.

There were, however, factors that were consistently noted in the sessions not anticipated by the literature review, or identified by the thematic sample questions, such as the oft-noted lack of

empowerment regarding the cost of illness. And, based on the responses, one of the most crucial areas of impact for Alphas appears to be associated with the event of diagnosis; an area not specifically identified etically, but, that proved useful in eliciting narratives relating to identity, and coping, among Alphas. There was a notable lack of discussion, or mention, of end of life or advanced planning, and little discussion about transplantation in the focus group sessions. In addition, during 3 of the 5 focus group sessions, and 5 of the 10 interviews, participants asked questions about the science of Alpha-1. In these 8 instances, I was asked to provide a lay description of my understanding of Alpha-1, or to answer a specific question that the Alpha's physician or nurse had not sufficiently explained. Although not a goal of the study, these types of questions, and requests for information, indicate the need for, and value of, providing information for every level of understanding. It may also indicate the commitment Alphas, and family members/care givers, have to understanding Alpha-1, and their ability to utilize the resources of the community to gain the information they need.

Interviews and focus group sessions were transcribed within 2 days of each session. Follow-up telephone calls were made to each focus group session organizer, (the local gatekeeper and recruiter), and to several of those interviewed, at least once during the transcribing of notes to verify information, and to ensure accuracy. Two of the gatekeepers, and 4 of my informants, were contacted on an average of once a month throughout the entire project, to discuss preliminary results, ask additional questions, or clarify ambiguous notes. I also re-contacted 3 of the nurses interviewed, and 4 of the physicians, to review their interview responses, and to discuss preliminary results.

Narrative Analysis

The analysis utilized techniques developed by Jehn and Doucet (1996), MacQueen et al. (1998), McLellan et al (2003), Ryan and Bernard (2003), and Carey et al. (1995). The techniques

involve identifying themes, and, when the patterns in the data become repetitive, or thematically saturated, metathemes are identified, and a codebook developed (see Table 4-1). Principles for thematic coding included attention to repetition, indigenous typologies, linguistic connectors, and, importantly, missing data (Jehn and Doucet 1996; Ryan and Bernard 2003; Bernard, 2003).

To code my data, I prepared a two-column document. In the left column was the transcript of the interview, or focus group session data. The right hand column had blank lines. As I read through the transcript, I made notes in the right hand column of the theme, or topics, in each line. I then ran word searches on each theme, and, when appropriate, combined phrases, or single words, under one thematic label. After reviewing the list of themes, I determined that several naturally grouped into metathemes, as they represented various aspects of a single domain, such as the descriptions of disease progression, or comments about relationships and networking. By the fourth focus group session, patterns in the interviews and focus group sessions became repetitive, and I considered the data as thematically saturated (Rubinstein, 1995). It is not always obvious ahead of time, how many informants are needed to learn about a particular subject, however, attention to thematic saturation helped me to recognize when sufficient data was collected (Rubinstein, 1995).

Interpretive strategies I used also includes, an overview analysis to develop theoretical sensitivity, and, a second line by line coding, using the thematic codebook I had developed, for identifying concepts, and confirming the conceptual relationships between concepts (Abrahamsson, 2002; Gottschalk, 1997). Metathemes were derived through a constant comparative method (Boeije, 2002). The thematic coding not only allowed for the systematic investigation of the qualitative data, it provided a means for identifying, and modifying, hypotheses (Willms, 1992).

Codebook Development: Identifying Core Elements of Empowerment

In the preliminary analysis, three higher-order categories were developed and labeled *Disease Etiology*, *Forms of Patient Empowerment* and *Networks and Relations* (see Table 4-1). All are considered core categories, central to the meaning Alphas, family members, caregivers, and the Alpha-1 community, as a whole, experience in relation to the condition Alpha-1. Each of these core categories includes several descriptive categories that were grounded in the interview data, and, that together, “comprise a model framing the process” of empowerment in a rare, genetic disease community (Abrahamsson, 2002).

Disease Etiology

The frequent descriptions of the medical condition Alpha-1 include, narratives detailing the pathogenesis of disease in the individual (symptoms evolving over time; identification of possible environmental causes), and narratives about how Alphas are currently coping with their chronic condition (disease management regimes, therapeutic options, and transplantation). A noticeable omission in the data were references to end of life, mortality (except in general epidemiological terms), and advanced planning. Although, it was not an area explored in the sample questions, it remains a significant omission, given the chronic and progressive nature of Alpha-1 Antitrypsin Deficiency. Many Alphas did note comments their physicians made about predicted mortality, but, none specifically commented about their own expected mortality other than to reject the often devastating “life sentence” their physician had given them. This selective memory is best summed up by one Alpha’s comments:

AB (male, 54 years old): “I may be in denial but until I went for transplantation evaluation, no one ever said ‘end stage or terminal’, except the psychologist”.

In light of the physician interviews, this statement may not be true for many patients, but it remains the perception, and memory, of many of those interviewed. The patient and caregiver

focus remained, throughout the interviews and focus sessions, on available and future therapies, transplantation, and mental attitude.

Diagnosis

Remarks coded under Disease Etiology include a consistent, and marked, emphasis on the event of diagnosis, and importance of the individual's age at diagnosis. It became clear during the first patient focus group session, and in subsequent sessions, that the diagnostic event is central to an Alphas' identity. Alphas' narratives characterize (and divide) their life experience into the pre-diagnosis era and the post-diagnosis era. in terms of their identity as, first, a chronically ill person, and, then, as an "Alpha", and participant in a well-defined community (Waitzkin and Britt, 1993). Depending on the quality and content of the diagnosis event, Alphas are either empowered shortly after diagnosis, or have an uphill battle to overcome the negative impression given them by physicians regarding their condition. It is a testament to Alpha support networks, that the negative experiences were overcome by the majority of Alphas interviewed.

Some of those interviewed experienced a positive diagnosis of Alpha-1 Antitrypsin Deficiency as a relief. However, a number of those interviewed had primary care physicians who were either uninformed, or resisted testing

IJ (male, 62 years old): "Rather than fear, the diagnosis made me feel relieved. It justified the feeling it was a genetic condition".

LL (female, 52 years old): "My primary care doc pooh-poohed the idea it might be Alpha-1".

EF (male, 60 years old): "There was an aversion to testing in the 80s because there was no treatment".

NN (male, 59 years old): "My pulmonologist tested me for Alpha-1 but was reluctant since it was an expensive test".

For many, the diagnosis explained family health dynamics, or highlighted the probable cause of family deaths. At least half the respondents had known about, or chose, testing because

of the previous diagnosis of a family member (usually a sibling), or at the urging of a family member who was a doctor, or nurse, and aware of the condition. In general, however, even when actively sought by the individual, a positive diagnosis was frequently a negative, scary experience. Many were told by primary care physicians, relatively uneducated in this condition, that they “had 2 years” or “a few years to live”, or that they would have to be on therapy, or go on supplemental oxygen within 10 years.

UV (female, no age given): “I felt instant isolation upon diagnosis.

YZ (male, no age given): “I felt terror associated with turning 47” [literature proposes the average mortality for Alpha-1 is 55 years old].

XY (male, no age given): “I went on a grieving trip”.

CC (female, no age given): “It was cruel, the way my diagnosis was given. I was told ‘you’ll be dead in three years’.

A few individuals however, expressed, at least in retrospect, a more positive reaction to their diagnosis. This may be, as Ochs and Capp note (1996), an unconscious attempt to re-order past events into a meaningful experience through narration.

NO (female, 67 years old): “My husband left me when I got sick...but I was happy to be diagnosed, although it led to poverty, because I was able to stop smoking and get some appropriate treatment”.

VW (male, no age given): “I feel a sense of pride about being an Alpha....it is a unique fraternal group of people with a common genetic bond.

EE (female, no age given): “There is a true sense of family”.

Therapies

Other components of disease etiology, noted by all participants, were the therapeutic options available to those with this condition. There was constant reference to Prolastin®, the first drug developed by Bayer Corporation (now known as Talecris Biologics, Inc.) as an augmentation therapy for those with lung disease related to their Alpha-1, and frequent mention of the more recent pulmonary drugs available. This emphasis on lung therapies is because,

unfortunately, there are no pharmaceutical means of managing the liver conditions associated with Alpha-1, transplantation being the only currently available approach for those with severe Alpha-1 liver disease. The emphasis on lung therapy is also because the majority of Alphas are lung patients (over 90% of those diagnosed in the United States).

Throughout the focus sessions, the Alphas engaged in extensive sharing, and advice-giving, related to specific therapies, the best transplant centers for lung or liver, devices, such as portable oxygen containers, and insurance coverage. I observed that their physical condition is often understood as symptoms requiring specific drugs, rather than a clear understanding of the internal biological processes associated with Alpha-1. The sessions were a clear demonstration of how those more recently diagnosed are able to learn from Alphas on the same drugs, or using the same therapies as themselves, and how those further along in disease progression reach out to the newer Alphas to educate and inform. This was despite the fact that Alpha-1 includes lung, liver, and, even more rarely, skin disease, and that Alphas are frequently on completely different drugs. At least half of those interviewed, displayed a sophisticated knowledge of therapies for their condition, and had accumulated information regarding many alternative, or complementary, therapies that might ease their condition, or improve their quality of life.

BC (female, 57 years old): “I met an acupuncturist who probably helped me as much or more than any person. Have you ever felt weight on your chest? That feeling went away and never returned!

Transplantation

In addition to the distinction between the experiences of lung and liver Alphas, the focus sessions brought out the distinction between those on therapy, those believing a cure is possible, and those considering the more drastic, surgical option of transplantation. Many of the Alphas seemed aware of the current inequities in the allocation system, and the potential impact of

transplantation on insurance, family income, and longevity (Lynch et al. 2006; Zamberlan 1992, Sharp, 2006).

WX (male, no age given): “CF [Cystic Fibrosis] patients get transplants but Alpha-1 patients are lower on the allocation list.”

TU (male, no age given): “Yeah, and the unfairness of the allocation scheme. Alphas shouldn’t have to wait 4 to 4 ½ years for a transplant.”

PO (male, 57 years old): “The East Coast of the United States has the least availability of transplantable organs, especially lungs”.

PO seemed very knowledgeable about the United Network for Organs Sharing (UNOS) system of allocation, which prioritizes needs on a regional basis. He suggested working with a nationwide (rather than regional) system that is not constrained by insurance. In his opinion, a nationwide system would avoid, “territorial pissing matches” over organ availability.

One Alpha drew a correlation between gender and choice of transplantation that may be of interest to pursue in further research:

CD (female, 57 years old): “The sex of the person (considering transplantation) makes a difference. I met another Alpha who takes infusion where I do mine. He was already on disability and he was younger than I was. I suggested going to rehab and he was totally resistant and thought of transplantation as an end all and be all.”

CD implied that, as a male, the other Alpha preferred the easy way out, which may simply indicate her exist attitude; however, it is an interesting suggestion that gender be evaluated as a dependent variable, particularly as it relates to therapeutic choices..

Forms of Patient Empowerment

The thematic topics broached during the focus group sessions included components of patient empowerment identified in the medical literature. The primary components, information and awareness, advocacy and activism, and identity, were explored fully by each focus group. Of these three, information and awareness was the form most often chosen by Alphas and their family members, and medical professionals, as the key to empowerment. This suggests a clear

correlation may be found between the length of time since diagnosis, and the level of empowerment, presuming an increase in the amount an individual learns about their condition in the intervening time. This supposition was tested statistically, and a moderate correlation confirmed ($P=0.0367$) (Figure 6-1). However, years since diagnosis and level of awareness proved to have a strong correlation ($P=0.0003$) (Figure 6-2), as did the total empowerment score and awareness ($P=0.0001$) (Figure 6-3). The total empowerment score was also highly correlated with organizational leadership ($P=0.0001$) (Figure 6-4) and with a positive diagnosis ($P=0.0002$) (Figure 6-5). Chapter 6, Quantifying Empowerment, describes in greater detail how these variables were derived from the narrative analysis, and the methods used to characterize and measure them statistically.

Information and awareness

The process of becoming informed was openly demonstrated by the interaction among the Alphas who attended the focus group sessions. I observed how the participants maintained ongoing dialogues, throughout each session, to inform each other, and raise awareness of the condition for those more newly diagnosed. As described in more detail in Chapter 6: Quantifying Empowerment, the Alphas coded as Level 3 or 4 awareness displayed familiarity with web sites, and educational resources about Alpha-1, and, in general, seemed very well informed about laws governing genetic research, privacy regulations (such as the Health Insurance Portability and Accountability Act of 1996, or HIPAA), the rights of patients, transplantation allocation, who was expert in treatment of Alpha-1, available therapies, and, importantly, how to connect with other Alphas. One Alpha brought a notebook with his entire health history in chronological order, with each record neatly categorized, and color coded. He offered very useful tips to the others about how to manage their relations with a variety, or procession, of doctors and nurses, and still maintain control of their own disease management.

Many of those participating cited the regional Education Days as helpful in their growing understanding of Alpha-1, and, in particular, the scientific talks given by experts in the field. These Education Days are organized by the Alpha-1 Foundation, Alpha-1 Association, COPD Foundation, and AlphaNet, and involve a speaker's bureau of experts in Alpha-1 liver and lung care, bioethics, and transplantation. Alphas, and family members, also cited the positive experience of participating in breakout sessions at the Education Days, as being particularly educational. There was wide acknowledgement among the participants that the Alpha-1 organizations in the United States (Alpha-1 Foundation, Alpha-1 Association and AlphaNet), are pivotal in creating reliable, professionally validated resources for the community. There was also frequent mention of the National Institutes of Health (NIH), Centers for Disease Control (CDC), and other federal websites, and easy access to university and hospital websites containing accurate information about Alpha-1. If empowerment were measured solely on the basis of the level of awareness community members possessed, one would have to rate the Alpha-1 community on the high end of empowerment.

Networks and relations

As the interviews and focus sessions made clear, information and awareness is empowering, but information and connection (networking) are more empowering. The participants noted, over and over, the importance of meeting other Alphas, and the support, help, and empathy they experience as part of the Alpha community. Other key interactions were frequently cited, such as the importance of family relations, good communication with doctors, nurses, and other caregivers, and membership in a well-defined community; these factors may prove as important as information and awareness in defining empowerment. It is for this reason that the statistical analysis is designed to test the strength, and relation, of these variables in the construction of empowerment in the Alpha-1 community.

Attendance at local support group meetings was frequently cited as the first step in becoming more aware. BC expressed most poignantly the importance of meeting other Alphas:

BC (female, 57 years old): “Oh, to walk in and there are people like you, where you don’t stand out. I got to be myself. For someone to see me, not the O2!

GG (female, no age given): “At the first support group that I attended, I was asked what is my FEV-1 (Forced Expiratory Volume in 1 second), and I didn’t know. After I had a transplant evaluation I began to pay attention to what my FEV was.

BB (male, no age given): “After attending support group meetings I as able to ask more intelligent questions (of the doctor).

Comments regarding the difference between Alpha-1 and COPD support groups were interesting, and confirmed the initial hypothesis that there may be fundamental differences in how genetic, as opposed to chronic, disease communities perceive, and experience, empowerment.

HH (male, no age given): “COPD patients are generally older and have more conditions, like cardiac problems.

PQ (male, 57 years old): “COPD patients talk about their condition in a whiney manner.

Many of the study participants, including the majority of the physicians interviewed, felt that Alphas are more positive in their outlook than COPD patients. These statements are not borne out by medical fact, since many Alphas have co-morbidities including heart conditions, but, the assessment regarding age is true, and may be an important factor in the sense of community, and purpose, experienced among Alphas. They have more of their lives ahead of them than many older COPD patients, and, perhaps, are more willing to fight for better health, or a cure, than someone in their seventies or eighties.

Six of the eight physicians interviewed noted their impression that Alphas were more positive and courageous than the other types of lung or liver patients they treat. It is not an opinion these physicians keep to themselves, but is one that is often shared with their patients:

BC (female, 57 years old): “I said something optimistic the first time I met Dr. XX and he responded, ‘that’s one quality of Alphas is that they’re optimistic. [He told me], ‘You have more courage and heart [than my COPD patients]’.

Cost of illness

The patient focus groups, and individual interviews, identified a broad range of activities, attitudes, and events that Alphas experienced as empowering. However, the cost of illness was mentioned in 4 of 5 focus group sessions, and during 5 of 10 interviews, as an area where Alphas and their families feel the most un-empowered. Although the impact varied, according the socio-economic situation of each participant, all those who mentioned costs described their situation as a burden, or as having a negative impact on their family, and long term financial plans:

AA (male, no age given): “Patients are at the mercy of the economics. Prices for augmentation therapy are higher even though there are more products available. Companies don’t listen. It is important for us to raise concerns. Alphas are more effective as a group.

Several participants pointed to the only outlet they felt is effective regarding the cost of illness – activism, advocacy as a group and the combined clout of more than one patient-centered organization.

Individually, Alphas expressed that they are not empowered relative to the cost of their illness. Some are denied the benefit of available therapy because of the cost.

LM (male, 52 years old): “I didn’t want to be tested, because I was afraid of the cost of medicine

CD (female, 57 years old): “I had insurance problems since I had pneumonia – I had a rider on my policy. I was lucky later to get insurance. Work paid for my illness for a long time. But when I went on disability I had a problem. I was supposed to get on augmentation therapy but I didn’t for awhile...when I started on Medicare supplements were cost prohibitive for me...Prolastin was so expensive and I couldn’t pay the 20% co-pay. I was diagnosed in 2000 but [was] not on Prolastin until 2002”.

Although, the organizational leaders interviewed noted that they feel empowered in relation to industry, the data from the interviews, and focus group sessions, suggests that this is not a feeling shared by the majority of Alphas in the community.

Discussion

Analysis of the interview data, and focus group sessions, resulted in identification of 3 core categories, and 19 descriptive subcategories, related to an emic definition of patient empowerment. The Alpha-1 community's self-assessment confirmed many components previously identified as comprising patient empowerment, but, also revealed critical components that may be unique to this community, or more prevalent among well-defined communities, such as another rare genetic disease community.

Identity, awareness, advocacy, communication, and control of one's own medical condition all show up in the data as central to empowerment as an Alpha. Several Alphas were emphatic about their own empowerment. When asked if they felt empowered, they responded quickly, and confidently:

“Yes, I can make decisions, I can ask good questions.”

“I have a personality type that drives me on a quest for knowledge; especially when it's about my own health”.

“Yes, I've received a lot of help from people in the community on line. I am not afraid to discuss issues.”

“Yes, it stems from knowledge about this disease. Just sitting here I realize how helpless [patients with] other conditions are”.

This final remark confirms one of the original hypotheses, that a rare, genetic disease community may provide a unique strength to an individual patient. The visibility of the Alpha-1 community structure, and the strong identification individuals have as members of this community, may, therefore, hold the key to empowerment as an Alpha. These attributes are wonderfully summed up by the written comments sent to me by a particularly articulate Alpha, when she notes:

JK (female, 64 years old): “I am a lung patient and somewhat disabled in that I cannot do certain things easily, if at all, any more. I try not to dwell on either of these, however.

Rather, I try to focus on the community of which I am a part (both rare genetic disease and chronic disease) and try to be a patient activist and advocate for that community. For each person, patient empowerment means different things as we are all different people with different backgrounds, strengths, and weaknesses and attitudes towards life in general. I believe that all of these carry over and affect how we deal with our disease. For myself, I have always been a very active and positive person. So it was natural, after I got over the initial shock of my diagnosis, that I would seek out other Alphas and try to find a way to turn that diagnosis into something positive. Additionally, it was and continues to be natural for me to find out everything I can about Alpha-1, my individual medical condition and how to best treat it through my own research and regular conversations with my local physicians and my Alpha doc. I then try to act on that information, including using supplemental O2 for exercise and sleep, implementing an exercise program, making certain diet modifications, limiting stress where possible, getting sufficient sleep, etc.

I am empowered and other Alphas I know are. I participate in patient advocacy activities. My physician involves me in decision making about my treatment. I am better informed than some doctors about my condition and believe Alphas are more empowered when they know more about their condition. I believe Alphas play a role, along with medical professionals in defining what Alpha-1 is.”

The narrative analysis confirmed that many of the attributes cited in the literature as empowering exist in the Alpha-1 community, and, that those interviewed were either empowered, in the process of empowerment, or at least in a situation that was potentially empowering.

Table 4-1. Codebook development: identifying narrative themes

CORE CATEGORIES

Disease Etiology
Forms of Patient Empowerment
Networks and Relations

DESCRIPTIVE CATEGORIES

Disease Etiology (Pathogenesis and Treatment of Alpha-1 Antitrypsin Deficiency)

Disease Symptoms

Disease Causes

Diagnosis

 Age at Diagnosis

Disease Management

Therapies

Transplantation

Patient Empowerment

Forms of Patient Empowerment

Information and Awareness

Identity

Advocacy and Activism

Networks and Relations

Networking

Relation to Doctors

Relation to Nurses, PAs, RTs

Relation to Family

Relation to Organizations

Relation to Industry

Relation to, Role in Research

Cost of Illness, Cost of Research, Cost of Networking

CHAPTER 5
LINGUISTIC ANALYSIS OF MEDICAL ENCOUNTERS

Language in the Medical Domain

Language is at the heart of healthcare and disease management, and each aspect of the encounter between a patient and healthcare provider - from the initial visit to a doctor's office and description of symptoms, to the provision of a diagnosis, and discussion of health maintenance and disease management options - depends on communication. Although nursing journals have traditionally focused on the communicative aspects of patient care, research since the 1960s indicate a growing awareness on the part of physicians, and medical educators, of the importance of effective dialog with, and education of, patients (Deveugele et al. 2001; Francis et al., 1969; Korsch et al., 1968; Levinson et al., 1997; Stewart et al., 1995, Stewart, 1999; Makoul, 2003; Haas et al., 2006; Makoul and Curry, 2007; McDaniel et al. 2007). For these authors, effective communication has been linked to patient and physician satisfaction, increased adherence to treatment plans, more appropriate medical decisions, and quantifiably better health outcomes (Makoul and Curry, 2007). Based on these obvious benefits, acquiring interpersonal and communication skills are now considered a core area of competency for physicians in the United States, and Canada (Makoul and Curry, 2007). Linguistics, therefore, seemed an appropriate theoretical framework for examining medical encounters within the context of empowerment in the Alpha-1 community, and I conducted a literature review to determine a specific approach that would best capture elements of empowerment in the speech patterns of study participants. As this phase of the study differed from the interview and focus group session phase, I obtained additional IRB permission to observe medical encounters, and to conduct follow-up interviews with the physician, and Physician Assistant, who agreed to participate.

I conducted a linguistic analysis of these physician/patient interactions to discern the language components involved in empowering patients in the Alpha-1 community. The analysis was based on four distinct research components: 1) Participant observation of the interaction between a doctor and patients during office visits; 2) observation of the interactions between nurses and patients; 3) ethnographic interviews with the doctor and Physician Assistant; and 4) ethnography of speaking analysis of the office visit speech events, and interviews.

Permission was obtained from a pulmonary physician, with expertise in Alpha-1, to observe his interaction with lung patients during pulmonary clinic hours in a large teaching hospital. I know this physician well, and have worked with him over the past 9 years on numerous Alpha-1 projects and activities.

Permission was also obtained from the Physician Assistant (PA), and Clinical Programs Coordinator for this physician, to observe her interactions with patients on three occasions in October, 2007. In addition, this PA agreed to pre-screen patients, to assist in identifying those willing to be observed during their office visits.

Follow-up ethnographic interviews were conducted with the physician (ML, male, 55 years old), the PA (JB, female, 38 years old), and another pulmonary clinical nurse associated with the Alpha-1 clinic (PA, female, 57 years old). The interviews were designed to explore their perceptions of the medical encounter, and the observed interactions, and to invite feedback on the preliminary linguistic analysis. Phone interviews were also conducted with key Alpha-1 informants, for additional feedback on the preliminary linguistic analysis, and for their independent observations of office visit dynamics.

The data collected was analyzed primarily from an ethnography of speaking approach, but utilized additional linguistic approaches, including consideration of Gricean principles of

cooperation, to determine appropriateness of quantity, manner, and relation aspects of a diagnosis; interactional sociolinguistics, to identify micro level linguistic choices, the uses of code switching, and Speech Act Theory, to identify the felicity conditions of the communications, and the specific reasons for misfires, and abuses, of the speech acts.

Fieldwork in Medical Offices

Observations were conducted of three separate office visits during the month of October, 2007. This included medical appointments with one Chronic Obstructive Pulmonary Disease (COPD) patient, and two Alpha-1 patients. Comparative observations were conducted to confirm whether a qualitative difference exists between how COPD patients and Alpha-1 patients relate to their condition, communicate with their physician and nurse, and react to diagnostic and treatment options. Although, this represents a very small sampling of patients, follow-up interview questions, and contact with five key informants, confirmed that the dynamics observed were typical of these two types of lung patients.

The observations, and data, from the three encounters I observed did confirm the anecdotal evidence from both patients, and physicians, that a marked difference exists. Additional research is planned for observations of a larger, and more statistically significant, number of medical encounters. However, the linguistic analysis for this research study was intended as an in depth examination of the dynamic between an Alpha-1 patient and doctor, and a COPD patient and her doctor. Three examples were sufficient for me to understand why such anecdotal evidence exists, and to spot the key differences that are likely typical, and that distinguish Alpha-1 from chronic lung patients.

The office visits were conducted in a pulmonary clinic associated with a large regional teaching hospital. One's initial experience at the clinic is the often frustrating job of trying to find a parking place in one of three large parking garages. Once parked, a long walk led to the

newly constructed outpatient clinic facility. This walk can be significant for lung patients, and represents one of many physical challenges related to their office visit. As I approached the clinic, I noticed several uniformed security guards stationed next to the reception desk, in the entrance way, and a large number of staff, patients, and caregivers snacking in the cafeteria, asking questions at the reception desk, standing in line at the pharmacy, or waiting for elevators. I joined those at the elevators, and noted the often anxious tone of the conversations around me – for, this is a specialty care clinic where patients are often referred when their primary physician cannot help them. It struck me that the entrance to the clinic, although not imposing in an architectural sense, still represents a departure from the patient’s normal life, and a visible reminder of their status as patient. It is a symbolic, as well as physical, entrance into the domain of medical care.

At the pulmonary clinic, at least 30 patients waited in the reception area. I approached the front desk, and identified myself as a researcher with a pre-arranged appointment with pulmonary staff. I was then shown into an interior waiting area, separate from the patient’s waiting room. Once inside, I observed how busy the clinic was - the constant flow of traffic, the five staff members attending patients, taking physical measurements, making appointments, or serving as cashiers, and the inter-professional dynamics of staff members. Each physician, intern, nurse, or PA who entered the inner offices was greeted by other staff; and there was a considerable amount of phatic communication in the inner reception area, such as chit-chat and gossip, but little medical discussion. However, once JB arrived, I was taken into her work area, where I observed a great deal more medical talk, and professional interaction between staff.

The physician/nurse work area is a long narrow room, with four computers set up next to each other, shelving holding informational brochures for patients, and bulletin boards with

information on medical coding, scheduling, and hospital regulations. There are posters on the wall with medical diagrams, information about HIPAA rights, and reminders to check patients for certain conditions. During my visits, a physician or nurse or PA sat at each computer, pulled up electronic records, checked their schedules, and entered data. In this room, the physicians also make phone calls, such as calls to primary physicians to pass on their specialists' diagnosis, and treatment plan. The specialists included both pulmonologists and hepatologists, i.e., lung and liver specialists, based on the terminology I heard, and the types of diagnoses discussed. None of the medical providers in the room seemed distracted by the flow of people, the conversations around them, or the phone calls; rather they all seemed used to conducting their business in a group setting. This seemed counter-productive to HIPAA regulations, which are intended to protect the privacy and confidentiality of individual patients, as I overheard very detailed discussions of patient conditions, judgmental remarks by physicians about their patients, and personal remarks about each other, despite the fact that I was present, and was not wearing clothing indicating medical status. I could only conclude that my invitation to sit in the work area was sufficient, tacit authorization to permit me to overhear conversations, and that the other medical providers assumed I was 'one of the team' and, therefore, privy to confidential remarks. It may also indicate how inured they are to other staff, or that their undivided focus is on getting through their case load.

Although, the majority of conversations were related to medical cases, I did note additional examples of phatic communication in the work area, as when a physician arrived who obviously knew JB and ML, and who spent approximately ten minutes describing her husband's planned research in Europe, her search for a house-sitter, and personal remarks on health and upcoming holiday plans. It occurred to me that these conversations were taking place

during the time scheduled for patients, and explained some of the (unnecessary?) delay patients often experience with their physicians.

ML and JB conferred for approximately 10 minutes regarding the afternoon's schedule, and the patients they would be seeing, and consulted the computer for relevant medical records, and previous diagnosis and treatment of these patients. JB also spent some time explaining to me who I would be seeing, how long they had been a patient, and the general purpose of their visit on that day. For two of the three patients I observed, JB asked the patient if they would participate in a research study, and when they agreed, introduced me. On the third occasion ML saw the patient first, and introduced me.

On each of my three clinic visits, the patient readily agreed to be observed, and seemed to understand that I would not otherwise participate in the discussion. However, it was obvious that they consented without thinking too much about it, and did not question the fact that a stranger would be observing their office visit. Both ML and JB opined, in their follow-up interviews, that patients expected to be observed because it was a teaching hospital, and that this is a common occurrence (at least once a month interns, and medical students, attend office visits as part of their training). In fact, during the second and third office visits, an intern also observed the proceedings. When the intern attended, the physician spent additional time in the work area explaining the cases to the intern, and talking about Alpha-1 Antitrypsin Deficiency. ML also described to the intern why he had become a pulmonologist, with a focus on Alpha-1, and, in his interview with me, explained that this was a way to stimulate interest in pulmonology among young doctors, and a way for him to mentor the next generation of physicians.

After consenting to my presence, JB brought me into the office, introduced me, and began her interaction with the patients. She asked about their symptoms, referred to previous office visit notes to follow-up about their treatment plan, and took notes of the responses throughout the conversation. JB was openly sympathetic, like other nurses and PAs I have observed prior to this study, and responded to the patient's complaints with both verbal and facial expressions of caring, concern, and commiseration. She spoke in a warm conversational tone of voice, and used a minimum of medical terminology. She expressed her questions in terms of the patient's home activities (i.e., "did you have trouble breathing going up the stairs?"), rather than asking for a measurement of FEV (Forced Expiratory Volume), or if the patient had dyspnea, the medical term for breathlessness. JB then asked each patient to step onto the examining table, and took measurements of their breathing, lung sounds, condition of throat, ears and nose, blood pressure, and heart rate. She next made diagnostic statements based on reference to previous illnesses or treatments, in addition to the current observations, remarking "sounds like you have an infection", or "seems like you may need to go on antibiotics again". These diagnostic statements were provided however, with the tacit understanding that the doctor would have to confirm the findings for a formal diagnosis to be made.

At this point JB left the room, noting she would confer with the doctor, and he would be in shortly. ML and JB discussed the case outside the room for approximately three minutes. ML then entered the room, and in each separate case, warmly greeted each of the three (female) patients, and then turned toward me and remarked, "THIS is my FAVORITE patient". He spent a few minutes joking with each patient, and their accompanying spouses and commenting on

previous encounters. He took this joking tone with each patient, and, noted during our follow-up interview, that it was the same manner he adopted with his male patients.

Although, three informants, and several patients in focus group sessions, had commented about their frustration with the repetition of questions by both the PA and the physician, ML referred to JB's notes extensively, and asked additional, and different, questions of each patient. He delved, for example, into the family dynamic, questioning how many people the patient lived with, their ages, and then remarked, when one patient admitted to babysitting for her grandchildren,

“You can't be doing that – just say no! I understand they expect you to, but you remember how you got ill before....PLEASE don't do that. You're more susceptible...you'll get sick if you do that.”

He was emphatic, and, although he said “just say no” in a joking tone, obviously referring to Nancy Reagan's infamous exhortation against drugs, he was clearly giving the patient a strongly worded professional admonition about her exposures, and their connection to illness episodes. He made other suggestions, like wearing a mask around sick family members, and washing hands at least eight times a day. He asked all of his patients about their exposures, what medications they were currently on, and whether they had had the flu shot that year. He also asked about the state of their breathing, the color of their sputum, and, interestingly, asked about what type of furnace the patients owned, and whether they cleaned the filters. One patient said she did, and he responded “good for you, GOOD for you!” He also asked about weight gain (obesity being a contributing co-morbidity with both heart and lung disease), and about any weight loss (a visible sign of progression toward end-stage lung disease).

ML gave each of the patients a physical exam, noting throughout what he observed, and what it meant. He used more medical terminology than JB had, sometimes explaining his words, but, often not. At one point, he remarked that he knew the patient was still sick because

he saw an elevated level of eosinophils in her blood, noting it was an indication of the inflammatory process. At that point ML had to step out of the office for a moment, and, although I had committed to not interacting with the patients, this patient kept trying to make eye contact with me, and seemed very interested in the notes I was taking, so I broke down and remarked, “gosh I can’t even spell eosinophils . . .do you know what it means?” She replied that it was “something in her blood”, which, although for me would not be a sufficient explanation, apparently was enough information for her, and she did not subsequently ask the doctor to explain it further. I, however, questioned him about it in the follow-up interview, noting that the patient clearly did not understand his terminology, and asked his purpose in using it. He replied that his interactions with both chronic and genetic lung patients were long term, ongoing relationships, and that patients learned what the words meant over time. He repeatedly noted that he encouraged his patients to “become more empowered” and, when I asked if that meant they had to learn medical terminology, he said yes.

After ML had given his diagnosis, he prescribed a treatment plan (which JB wrote up, and entered into the computer). He noted, laughingly, how JB “doesn’t let me keep a prescription pad. She thinks then I’d write prescriptions (without her consent)”. ML frequently made joking remarks that indicated his reliance and trust in JB, and, it was a common theme in all three office visits. He often included her, as well as the intern, and I, in the joking banter, and, through his frequent mention of JB’s key role in the patient’s care, made the two way interview with the patient seem like it involved three voices. In the follow-up interviews, both ML and JB noted the many advantages their good communication, and close coordination, had for effective medical care. They both mentioned that they had, over time, arrived at an optimal procedure for dealing with disagreements – they never did so in front of patients, but strove to

maintain a single coordinated voice in the office visit. This suggested they had had a number of disagreements, and, when asked if the physician always had the final say, ML said yes, but JB said many times ML came to agree with her when they disagreed. Given these conflicting statements, I conclude that the traditional hegemony within the ranks of medical caste system is not disrupted – JB’s authority still lies in the physician’s trust in her, but, ultimately, the physician has the final say.

The office visit concluded with an elaborate coda: first, ML quizzed each patient about things he had said during the visit,

Okay young lady (addressed jokingly to a woman in her sixties) how many times a day do you have to wash your hands? Eight times a day—you pass! You were listening. You’re the world’s best patient!

Next, both ML, and then the patient, repeated the treatment plan, and medical doses. ML then asked if the patient, or spouse, had any questions. Although, the two Alpha-1 patients had asked questions, and offered comments, throughout their visits, it was during this phase of the visit that they showed a noticeably more active approach than the COPD patient. Each Alpha had several questions to ask at this point in the office visit - each asked about new drugs, about drug interactions with their other medications, and if they were eligible for clinical trials of investigational drugs. One also asked how she might improve her eligibility for research studies. In each case, my hypothesis that Alpha-1 patients are more empowered, and more willing to take control of their medical condition than chronic lung patients, was borne out by the individuals I observed. In the follow-up interviews, both ML and JB confirmed that this was a typical difference between COPD and Alpha-1 patients, and that less than 20% of COPD patients asked any questions, much less were knowledgeable enough to request participation in research.

Medical and Social Context of the Office Visit

The office visits ended on a cordial note, but, for both the patient and the medical team, the visit represents only a small portion of their entire experience. Once ML and JB had finished seeing the patients, JB completed the notes of the visits, and ML telephoned the primary physician, who had referred the patient, and spent at least 5 minutes on the phone explaining the diagnosis, results of tests, and treatment recommendations. Prior to the visit, the physician had read the results of laboratory blood work, sputum analysis, x-rays, CT scans, and/or results of breathing tests. He also consulted medical articles, and websites, for current findings and recommendations (during my first visit, he spent several minutes surfing medical websites to find out how often to give a pneumonia shot), spoke with the primary physician and other pulmonary specialists about the case, and discussed the treatment plan with JB, and with another nurse in attendance in the clinic.

The patient, on the other hand, had to complete additional paperwork, get a flu shot, make another appointment, pay for her visit, take the long walk back to the parking garage, and find her car. For many patients, seeing this particular Alpha-1 expert involves traveling up to 1,000 miles from home by car or airplane, staying in a motel near to the hospital, and disrupting the normal work and home life. Both of Alphas I observed had traveled out of state for their yearly exam, and both were accompanied by their spouses. This meant that their spouse's work and home life was also disrupted for up to 4 days by a ½ hour office visit.

The physician aptly described the office visit as “the tip of an iceberg”. For, like an iceberg, the majority of the activities associated with the office visit occur beneath the surface, or behind the scenes. For the physician, this meant extensive readings, and research, to stay abreast in his field of expertise, and the time it took for him to learn how to communicate medical information effectively. I had first hand experience of the evolution of this physician's

communication style, and observed that he was most effective when he communicated medical conditions as metaphors, or analogies, that the patient could understand, rather than when he used medical terminology.

Given that both the physician and the patient's office encounter is the tip of an iceberg, I began to visualize the medical encounter as the point where two cones of activity, two very different domains, come together for a brief period of time. ML, JB, and the key informants I contacted all agreed with this view of the office encounter as the point where the medical domain and personal domain meet.

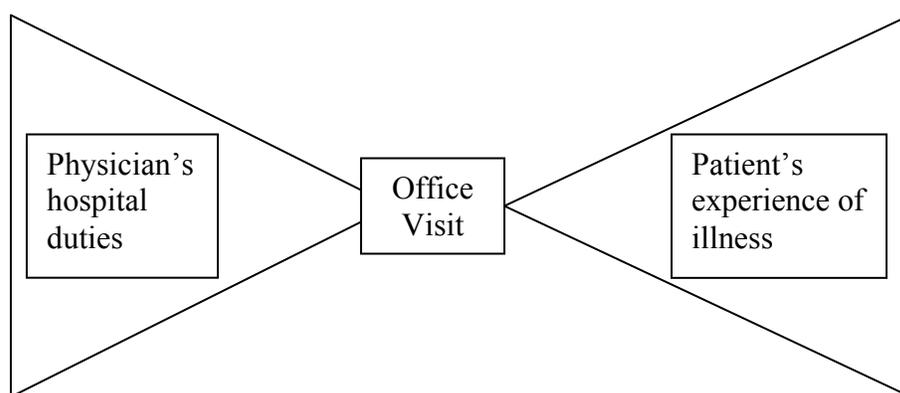


Figure 5-1. Where worlds collide: the office visit as tip of the iceberg

Physician Interview

I held follow-up interviews with ML, JB, PA, and five key informants. The questions in ML's and JB's interviews centered around five topics – the differences between COPD and Alpha-1 patients, questions about medical procedures, questions about how much patients understand of their condition, and questions about language use, and my linguistic analysis of their medical encounters (see Appendix B).

All those interviewed agreed that there are noticeable differences in how much Alpha-1 patients and COPD patients know about their illness, and how willing they are to take control of their condition. This may be explained by the fact that Alpha-1 patients tend to be younger than

COPD patients, are less prone to other conditions associated with aging (i.e. dementia), or they feel obliged, because of the genetic nature of their condition, to “do something for their children’s sake”. Based on the results of the initial research in this community, one may also cite the strong identity these patients have as a community, and the wealth of resources available to them about their condition as components of their heightened empowerment. JB noted that less than 20% of COPD patients asked any questions, as opposed to the 90% of Alpha-1 patients who come armed with a list of questions for their physician. JB made an interesting distinction regarding gender differences in coping, and the relation of socio-economic status and empowerment. She noted that socio-economics was a greater factor for COPD patients, with those less advantaged tending to be less empowered, but it was less of a factor with Alpha-1 patients, all of whom had access to information and resources.

Questions in the interviews about procedures were very enlightening, and provided insight into the reason why a nurse or PA saw a patient first, followed by the doctor, and the historical roots of this practice. ML explained that the role of PA had arisen during the Vietnamese War, when orderlies received extensive training to assist physicians in field hospital emergencies. Upon their return to the United States, these former orderlies were recognized as being more highly trained than nurses, and the medical societies pushed to utilize this valuable corps of medical trainees. The role of PA was created, and certification offered to returning veterans with this type of experience. Although, in the 1970s and 1980s the majority of PAs were male, in more recent years the majority are female, and they are rapidly replacing nurses in medical encounters as the primary gatekeeper to the physician.

When asked, why is it necessary to see the PA or nurse first, ML and JB both explained it served, ideally, as a “tremendous” time saver for the physician. When conducted properly, the

PA's often lengthy interview with the patient is transcribed into abbreviated medical terminology that the physician understands as a kind of shorthand, embedded with a precise biotechnical meaning. Although, the physician and nurse agreed in their assessment of the utility of these preliminary discussions with a nurse or PA, patients did not share this view. Instead, several of those interviewed had expressed frustration with this aspect of a medical encounter unnecessarily time consuming and redundant. One male patient (62 years old), in particular, dwelt on this topic in his interview, and despite feedback from me regarding the biomedical justification, continued to experience the imbalance in waiting times as a sign of the physician's dominance.

When asked if the symptoms the patient describes are a list or a narrative, JB said she wished it were a list, but it is often a long, rambling, disjointed narrative that she has to pick through for the symptoms, and physical conditions, that are meaningful as indicators of illness. Her view of her job is to summarize, condense, and "standardize" the information provided by the patient so that ML can respond with the appropriate diagnosis and treatment. ML noted that a PA could, when properly trained, communicate what is "important" for him to know about the patient, and that when he relied on the PA he could avoid repeating her questions to the patient, and, instead, focus on a treatment plan and recommendations.

Another area of agreement among those interviewed was the level of understanding a patient should have. Both ML, JB, and the Alpha-1 patients felt it imperative for the patient to learn as much as they "were capable of understanding", although they did not agree with how much medical terminology was necessary. They did all agree, however, that patients should consult with other patients when they needed help to understand technical topics, and that a nurse, or PA, often played the role of translator for the patients. Although, this seemed to

confirm the traditional dominance and centrality of the medical domain in illness experiences, I realized that by becoming a patient, Alpha-1 and COPD patients become players in the medical domain. As research has demonstrated, patients negotiate their own care better if they can communicate in the ‘native’ language (Makoul and Curry, 2007; Leps, 1995; Teff, 1994). It is telling, therefore, that the COPD patient observed did not seem to share the Alpha’s drive for knowledge, as evidenced by her lack of understanding about the medical terminology used in her office visit. This COPD patient seemed to lack any curiosity about the things she did not understand about her own condition, and the necessary assertiveness to resolve her understanding through questions.

My final set of questions explored the linguistic aspects of the medical encounter, and asked for feedback of my linguistic analysis. When it was explained, all those interviewed, in this phase of the study, acknowledged that they code-switched during an office visit (switched between types of vernaculars particular to a domain). JB noted her approach was to use lay language almost exclusively; her code switching was primarily when she related information to the physician. ML, although I observed how often he code switched, over estimated the amount of lay language that he employed. ML said it is important for the patient to understand the medical procedures they experience, and the terminology associated with it. Still, he showed little self-awareness of how much background had gone into his own understanding of biological processes, and how terms he used to condense medical information were embedded with years of study and practice. The three patients interviewed in this phase of study, and several others in previous focus group sessions, also thought it important to learn medical terminology. One Alpha noted he was aware that he code-switched much like a second language learner speaking in their newly acquired tongue – he used medical terminology to the

extent of his knowledge, and then code-switched back to lay language when he did not know the medical term to use.

Results

Ethnography of Speaking Analysis

The interviews concluded with a presentation of the preliminary linguistic analysis, and an invitation to augment, refute, or otherwise provide feedback on the analysis. The presentation included a brief overview of the ethnography of speaking approach, an explanation of the SPEAKING acronym developed by Dell Hymes in the 1960s (Hymes, 1962), and then, the following analysis of the medical encounter was read, and a copy given ML and JB.

S – The **setting** is institutional (hospital) and the constraints on the speech event are institutional and legal, not personal constraints. The medical setting focuses the speech event around exploration of the physical condition of the patient, although related issues (familial, social, and economic) are also explored in the visit, they are considered only in relation to the physical.

P – The **participants** in the speech event are limited to the patient, their spouse, parent or caregiver, the physician, PA or nurse and any permitted observers; HIPAA regulations, regarding the privacy and confidentiality of patients, has tightly controlled who may overhear or observe these proceedings and the formal institutional permissions needed for such overhearing (i.e, IRB approval).

E – The **ends** or goals are different for the patient and the physician. The physician needs to communicate vital biomedical information to affect an amelioration of physical symptoms, or eliminate infection or other forms of disease. The physician may also be providing training for medical students, gathering data for research or fulfilling institutional requirements for service. The speech event is completed when the information is provided. The patient's goals are to end breathlessness or relieve an exacerbation, i.e. obtain care for their physical symptoms. They may also want to address fears or uncertainties or understand why and how the symptoms arise or other information related to their condition. Another end for patients is to translate the biomedical information into understandable explanations for their physical symptoms, and through understanding the biomedical information cope better with their condition, be more compliant with treatment regimes or help other family members cope with related illnesses. The speech event may end before they have achieved understanding.

A – Although the purpose of the office visit was initiated by the patient's condition, the speech **act sequence** is fully controlled by the medical staff. The visit is generally limited in duration and preceded by often long waiting times on the part of the patient; it is

conducted primarily as a close-ended question and answer session with fixed turn-taking and the absence of overlap in speaking. The **speech act sequence** of the 3 office visits observed included:

1. JB greets patient and spouse as they enter the examining room.
2. Greetings - JB and patient continue to exchange greetings in a conversational, friendly manner
3. Questions from JB to patient regarding purpose of visit, symptoms since last visit
4. Narration by patient of their symptoms, deterioration or improvement since last visit, other medical problems affecting their lung condition,
5. Physical exam by JB – use of instruments, some medical terminology
6. JB prepares notes of visit for permanent medical record
7. JB consults with physician out of hearing of patient⁴
8. ML enters examining room
9. Greeting – joking manner, friendly, personal and interactive
10. Reading notes, questioning JB – code switch medical terminology
11. Questions from ML to patient (and occasionally to the caregiver) – code switch to mix lay language/medical terminology, interview style
12. Answers from patient – lay description of symptoms, activities, limited narration
13. Responses from ML – mix empathetic, stern, joking, use of metaphors
14. ML writing notes, and/or referring to test readings, computer screen - non verbal
15. Additional questions from ML to patient
16. Additional answers from patient
17. Additional responses from ML
18. Writing prescription(s) – formal medical style, legally bound documents non-verbal
19. Diagnosis – serious formal pronouncement by ML primarily medical terminology
20. Treatment recommendations – serious formal directives primarily in lay language
21. Questions/comments from patient – repeating and restating recommendations, questions about proposed treatment
22. Closing remarks by ML indicating end of interview and patient’s turn-taking for closing questions, joking manner
23. Questions from patient – issues not discussed in diagnostic interview or of interest to patient and not directly related to their symptoms, additional questions
24. Closing remarks – personal, friendly

K – The **keys** employed included a broad range of serious talk, empathetic responses, joking, sarcasm, and compliments. The physician used joking as a phatic communication (to achieve rapport and relax the patient), and, also, to signify a level playing field with the patient (patients could also make jokes or respond to a joke in the same manner).

I – **Instrumentalities** included spoken exchanges, writing of medical records, writing of prescriptions, use of computer to check information and enter information, use of

⁴ Although this was not observed, the assumption is that it is conducted in a formal professional manner utilizing medical terms exclusively

instruments to determine heart rate, sound of breathing, level of congestion and inflammation in ears, nose and throat

N – Norms of interaction were asymmetrically medical. The inferences were primarily medical and turn-taking was determined by either the PA or the physician. The balance of power in office visits is determined by the centrality of knowledge in medical encounters.

G – The genres included medical terminology and lay description of illness; the event although at times friendly and joking, was not casual speech but a provision of information based on a need to improve a negative state. The event was structured around resolving a problem and was therefore more serious than the outer manner of speaking might imply.

I expected some reaction to Norms of Interaction, but, surprisingly, neither ML, JB, nor the two Alpha-1 patients initially objected to any of the points above. However, the physician did comment, after a few moments thought, that, although knowledge was central to the encounter, his goal is to teach the patient how to be empowered, and, that over time most became empowered by learning more about Alpha-1. He also repeated a comment he had made earlier, that although some patients did not understand the medical terminology during the first visit, that he knew them over a long period of time, and that allowed them to assimilate the information, and learn over time. He further asserted that he had refined, and perfected, his manner of speaking to patients, and, was confident that it was effective. This assertion might have sounded arrogant on face value, but had been previously corroborated by at least six patients of his, who I had interviewed during my research, and who gratefully attested to his ability to explain complicated medical information that other physicians had not been able to explain to them.

Discussion

Although the medical encounter seemed at times inappropriately technical, and the medical staff firmly controlled the proceedings, ML and JB both communicate in ways they believe will empower their patients, and enable the patients to take greater control over the

course of their treatment. Their use of medical terminology is not done to maintain their control over knowledge, but to draw the patient further into the medical domain, and to a more precise understanding of what is happening to their body in biological terms. And, in fact, the two Alphas observed in their care, and many more who were previously interviewed, consistently demonstrate a greater understanding of the biological processes affecting them than chronic lung patients, and come across as effectively assertive with their concerns and questions.

There is a risk, however, in establishing the biomedical explanations for this particular condition, or any illness, as normative, as it may silence alternative ways of talking about disease, and over standardize the individual experience. The centrality of biomedical explanations may also valorize the physical symptoms over the emotional or psychological experience. However, in the Alpha-1 community the personal, emotional, psychological, and economic impacts of Alpha-1 are freely shared with other patients, and family members, in an atmosphere of strong community and institutional support, and the possibility for empowerment in both medical and personal terms is strengthened. Alphas, in general, seem more willing to achieve communicative competence in the medical domain in addition to exploring their common experiences of illness, and its impact on their daily lives, with other Alphas, in their own terms.

This phase of the research confirmed that variation may exist in how genetic lung patients and chronic lung patients communicate about, and understand, their condition, and how medical providers communicate about it to them. However, the question whether such variation disrupts, or endangers, the traditional hegemony may be moot after observing how empowerment exists in more than one realm of experience, and has more than one form of expression. Empowerment of patients may, therefore, not mean less power for medical

providers, as much as it signifies a more open, and central, role for the patient in defining the condition, and coping with it.

The differences I observed, between the physician's dialogues with the two Alpha-1 patients compared to his dialogue with the COPD patient, was so marked that I conclude, even from this limited data, that the ideal conditions exist in the Alpha-1 community to promote patient empowerment in both the personal and medical domains. However, further investigation of a larger cohort is necessary to confirm these findings, and delineate the specific forms of interaction, and communication, that will promote such empowerment for other types of patients.

According to the Centers for Disease Control, COPD affects millions worldwide, and lung disease is quickly becoming a leading cause of death worldwide. It would be useful to confirm the communicative components of empowerment, as exemplified by Alpha-1 patients, and the Alpha-1 community organizations, towards improving the possibility for empowerment, and the related benefits of increased awareness, preventive action, and disease self-management, for this larger body of sufferers.

CHAPTER 6 STATISTICAL ANALYSIS

Unpacking Empowerment into Definable Components

Quantifying patient empowerment is the goal of this study, and the theme of this chapter. In Chapters 4 and 5, I describe how the data from focus group sessions, interviews, and medical office visits was gathered and analyzed qualitatively. This chapter focuses on how a complex concept like empowerment was unpacked into its components, how the qualitative data was translated into quantitative data, and how the data were analyzed. The final results of the statistical analysis are assessed against the qualitative results, and demonstrate the validity of the hypothesized relations, and successful identification of appropriately defined variables. The final results are also considered in relation to future research questions and goals, specifically, whether the load scores, or total empowerment scores developed in this study are representative of empowerment, and can serve as accurate data for correlation analysis with quality of life (QOL) scores.

An important premise underlying the statistical analysis is the value of identifying the key, or intervening, variable(s) from among the components of empowerment identified in the narrative analysis. From the narrative data, it was clear to me that “time since diagnosis” may be more effective at confirming the qualitative impressions of the cohort than other components, or attributes of empowerment. By focusing on, and testing, the relation of the variable “time since diagnosis” to the other components, I was able to identify what may be the intervening variable, and eliminate several sub-domains, that essentially capture the same information, thereby refining my definition of empowerment.

Factorial analysis, described below, had demonstrated that the components of empowerment grouped into two sub domains, as hypothesized. In one of these domains, the

components were so tightly correlated that having two separate questions, or components, was redundant – two components, such as organizational membership and participation in research both characterized the same feature. In the second domain, at least one component, advocacy, had no association with a total empowerment score, and it did not track with the other components. Similar to Clark et al. (2004), who proposed use of a single item measure for disability, I sought, through the statistical analysis to confirm which, if any, of the components of empowerment might represent the core meaning of empowerment. However, like Fries et al. (2006), I ultimately refined the number of sub domains to several attributes, or markers of empowerment, that together contain the maximal information for defining empowerment.

In addition to the identification of an intervening variable, and the factorial analysis of domains, the statistical analysis also includes derivation of a “total empowerment score”. As the nine potential domains of empowerment appeared equally important, equal scores were assigned to each domain (see Table 6-1), and totaled. The total empowerment score was then analyzed against individual components of empowerment, to demonstrate the relation of the various sub domains in the extent of empowerment. An additional value of the Total Empowerment Score is that it will permit comparison with QOL scores, a future research goal.

All the final statistical results suggest that the qualitative data is a very successful definition of empowerment, and that the components, or sub domains, identified were sufficiently refined so as to provide an accurate picture of empowerment. With such results, future surveys can include only those questions necessary to accurately measure empowerment.

Identifying Appropriate Measurement Tools

Patient empowerment is a complex social construct similar to Socioeconomic Status (SES). SES uses combined measures of income, education, and occupational prestige, since none of these individual measures captures the complexity of the idea of SES. Patient

empowerment, likewise, should be measured with a complex instrument (or scale) made up of several indicators together forming a composite measure of one underlying concept. I chose a Guttman scale, since the components of empowerment seem to line up in a particular pattern indicating a unidimensional variable (Guttman, 1950; Peregrine et al. 2004, Handwerker, 1996; Johnson, 1995; and Beddington, 1977). Johnson (1995) suggested using a Guttman scaling for scoring disability, and its applicability in the context of health surveys. In addition, I chose a Guttman scale because it is inherently hierarchal, and could capture the evolutionary order of components of empowerment (Peregrine et al. 2004). To populate a scale with components of empowerment, the demographic and investigational variables identified in the narrative analysis were assigned quantitative values (McLellan et al. 2003).

However, prior to populating the Guttman scale, decisions had to be made regarding how many, and which, variables to use among the several themes identified in the community, and how to group them into sub-domains. Readings in the medical and epidemiological literature proved helpful as a large body of literature exists describing the development, adaptation and use of QOL instruments to measure complex parameters relating to the health/disease spectrum (Dowson et al. 1999; Cheng et al. 2000; Bloom, 2001; Gunzareth et al. 2001; Hajiro et al. 1998, 2000; Handwerker and Wozniak, 1997; Jones et al. 1991, 1992; Ketelaars et al. 1996; Knebel et al. 2000; Okubadejo et al. 1996; Osman et al. 1997; Renwick and Connolly, 1996; Seemungal et al. 1998, Tu, 2006; Wijksta and Jones, 1998; Wilson et al. 1997; Zamberlan, 1992). QOL research is relatively unique among biomedical research in that it recognizes the value of non-medically derived data, and the importance of standardizing the translation of this type of data into numeric values. In addition, this type of literature is more accessible and understandable to me, as an anthropologist, and complements my anthropological research of a disease

community. For these reasons, QOL studies were chosen as the model for my research, and represented a well worn bridge spanning the qualitative/quantitative divide that I feel is safe to cross.

The research on QOL instruments explored how to identify appropriate, and representative, items to be used to measure the qualitative experience of disease. Clark et al. (2004), for example, proposed use of a single item measure for disability, however I thought it more likely that I would identify a somewhat larger item pool that would provide clearly defined, reliable, flexible, quantifiable, sub domains, and had “maximal information content” (Fries et al., 2006). To achieve this maximal content, several of the QOL studies stressed the value of utilizing patient reported outcomes (PROs), or triangulating patient, physician, and nurse data. PROs have now become standard study outcomes in medicine; this previous research validated my use of both patient and caregiver data in this study, and situates my anthropological work within acceptable biomedical parameters (Fries et al. 2006; Goodacre, et al. 2007).

Fries, Goodacre, and subsequent researchers have validated the use of QOL instruments to measure the full spectrum of a patients’ experience. But, importantly, they have also demonstrated the usefulness of adapting QOL instruments for specific medical conditions (Rat et al., 2007, Goodacre et al. 2007, Fries et al, 2006, Swigris et al, 2005, Fries, 1983, Grunfeld et al, 2008, Graham et al. 2008, Wyrwich et al. 2007, 2006; Tsukino et al, 2002, Reardon et al 2006, Alvarez-Gutierrez et al, 2007; Hajiro et al. 1998; Katsura et al, 2007). The research described by the majority of these studies was conducted among lung patients, primarily COPD patients. The approaches used in these studies served as a useful model for how I might incorporate my qualitative data into a quantitative assessment of the Alpha-1 community, and,

in them, I found well-tested techniques for adapting my measurement(s) of empowerment specifically for the Alpha-1 community (Fries et al, 2006, Goodacre et al, 2007, Handwerker, 2003; Swigris et al, 2005, and Grunfeld et al. 2008.) Utilizing the adaptive techniques, I focused on issues unique to the Alpha-1 community to ensure item generation for future surveys that would actually measure empowerment.

Modeling my research on QOL studies, and incorporating techniques for measurement widely accepted and used in the medical community, has already made my work more accessible and meaningful to Alpha-1 researchers. Based on the grant review comments I received for phase 2 of my empowerment studies, my work is, and will be, accepted as useful information derived from a new perspective, but firmly situated within the body of research on patient-physician dynamics in medical care. In addition, as noted previously, using QOL as a model will ultimately allow comparison between the data from this study and QOL measures.

Quantifying Empowerment

Quantification implies measurement of numeric values. The research design, therefore, included the gathering of basic demographic data and narrative responses that could be translated into numeric values, and measured quantitatively. The qualitative data also included observation of study participants, and, by a reiterative process, new data was compared against the original scale of empowerment (Figure 1-1). Over time, and in different venues, a focus developed on observing participants' stage of awareness, understanding of their condition, level of involvement/networking in the community, and attitude about their diagnosis. The attributes comprising personal empowerment were clarified, and revised, or expanded, to more accurately capture essential variation in the process of empowerment.

The components I had hypothesized as indicative of empowerment (Figure 1-1), were refined through preliminary statistical analysis, and the components "Access to resources" and

“Disease self-management” eliminated. Since, it was determined that all those who participated in the study had access to some resources, this ceased to indicate any variance in the cohort. I also began to understand disease management, not as an investigational variable, or stage of empowerment, but as an outcome of empowerment. Although, there was variance among the study participants in the level of their disease self-management, it seemed to be the result, not cause of their responses to the other domains. However, the hypothesized domain, personal empowerment, was not only comprised of “awareness”, “research participation”, and “organizational membership”, it also seemed dependent on levels of networking. It seemed appropriate to quantify levels of networking as a variable, and include yes/no responses to three questions aimed at clarifying the level of family vs. level of community support/networking. I had data for 17 of the 34 Alphas on the number of Alpha-1 community members they knew, (their “Alpha network”), and responses from 49 out of 50 participants on whether or not “family support” was available. From the narrative data, I noted a clear distinction between these types of support, and determined it would be valuable to measure, rather than combine, a score for each of these types of support.

Attitude about diagnosis, or “type diagnosis”, was also added to the final list of empowerment sub-domains, as an indicator of the potential for empowerment at an early stage in the patient’s experience. I assigned a yes/no value to each participant based on their comments, or longer narratives, describing the diagnostic experience as either positive or negative. Diagnosis emerged as a central experience for Alphas, and their families, and all participants, except one, provided comments on their experience. From these comments, I discerned a difference in how quickly individuals moved beyond their early fears, and denial, and got involved with the community, or their own education about Alpha-1. This difference

often seemed to be directly related to the quality of their diagnosis, and how positive, or knowledgeable, (or negative, and unhelpful), their medical provider was in the initial dialog about the diagnosis.

This aspect of temporality, and the progressive nature of empowerment, suggested the final investigational, and possibly interventional, variable, “Years since diagnosis”. I was particularly interested in whether the statistical analysis would demonstrate a strong correlation between the time from diagnosis to level of awareness, (rather than a correlation with empowerment as a total score), and felt the qualitative assessments were validated when the correlation between “years since diagnosis” and “awareness” proved highly significant ($P = .0001$) (Figure 6-2). However, the total empowerment score and level of awareness were also highly correlated ($P = .0001$) (Figure 6-3), and, in addition, explained 70% of the variance, as opposed to the 18% variance in level of awareness and years since diagnosis. The statistical analysis demonstrated two other relations that explained 70% and 72% of the variance - Alpha Network and Total Empowerment Score, and Participation in Research and Alpha Network.

Although, “level of awareness” was the most well developed measurement in the study, and seemed to capture the essence of personal empowerment, I eventually chose not to use it as the only marker of personal empowerment based on the statistical results described above, and a reassessment of the interview data. DE (male, 68 years old) had been diagnosed with Alpha-1 36 years previously, making him one of the first people diagnosed in the United States. However, his level of awareness, despite his advanced educational level (PhD), was only a 3. Despite his familiarity with biology, and his understanding of how Alpha-1 affects his body, he had not synthesized his knowledge, had little knowledge of the types of research being conducted, did not seem aware of specific legislation, or the economic and legal issues

associated with Alpha-1, and, although he could articulate about Alpha-1, he clearly had not considered, prior to our interview, the impact of Alpha-1 on him and his family. He was not that involved with the community, knew very few Alphas, and had not availed himself of the support or interaction available in the community. He also did not score high on structural forms of empowerment. Although, this is only one case among 34, it helped me distinguish between levels of awareness, and an overall high score of empowerment. Qualitatively, levels of awareness could not serve as the sole attribute of personal empowerment, and the statistical analysis confirmed this.

Through this refinement of the list of components, a clearer picture of the nature of each sub domain, personal and structural forms of empowerment, was obtained. The goal, for each sub domain, is to refine the questions I use to the most central, or essential markers, of that form of empowerment, and then test for the relation between these components. A factor analysis can test the utility of grouping components into these particular specific sub-domains, but, additional, and useful, information could be gained by weighting each domain equally, and using the load score from each domain to arrive at a Total Empowerment Score. Since, data from this study will be used to compare with QOL scores, it is necessary to obtain these comparable measurements. The Total Empowerment Score should also provide a test of the validity of the factor analysis, by determining whether those characterized as structurally empowered, actually score higher than those personally empowered.

The observational data were primarily assigned yes/no values, except for “levels of awareness”, or understanding of Alpha-1. This variable was assigned values according to a formalized classification scheme, developed in the field of education, to measure stages of learning and comprehension (Bloom, 1952; Krathwohl, 2002). By quantifying the qualitative

components of empowerment, the results of this study will ultimately be measured against the physical parameters used to quantify various stages of illness and disability, as is done with QOL scores.

The final analysis of community defined components of empowerment, and a re-analysis of hypothesized components, resulted in nine key components of empowerment. This includes 1) type diagnosis, 2) family support, 3) awareness, 4) organizational membership, 5) participation in research, 6) advocacy, 7) organizational leadership, 8) membership in a supra-organization, and 9) involvement in policy-making. Given the rarity of Alpha-1, further development of an empowerment survey tool is a difficult task. Like a well developed QOL tool, an empowerment tool would require independent testing of each domain to assure the scales are linear, assessment to assure the domains are independent, and evaluation with a validation cohort to assess the appropriate weight to be given to each domain. One way to abbreviate this process is with factor analysis. A factor analysis of the potential components of empowerment of the items was conducted, and each of the potential item domains given a score, or factor loading (see Table 6-5).

If the components of empowerment form a unidimensional scale there will be a “single overwhelming factor that underlies all the items” and all items will load high on that factor (Bernard, 2003). If the scale is multidimensional, there will be a series of factors that underlie the set of variables (Handwerker, 1997). To test how closely this data reproduces a perfect scale, Guttman’s co-efficient of reproducibility (CR) was applied. According to convention, a CR of .90 or greater will be accepted as significant (Guttman, 1950).

Sample

The empowerment study sample consisted of 49 members of the Alpha-1 Antitrypsin Deficiency (Alpha-1) community (see Appendix C). Of the 49 participants, 34 were Alphas,

and 15 were caregivers or family members. Although, there was missing data for several participants on age, and numbers in close circle and Alpha network, only one individual, QR, (male, 66 years old), was missing data in categories necessary for the analysis (organizational membership, research participation, and advocacy). These missing data points were imputed as the mean from the other individuals with the same awareness level as QR.

Data for the study were gathered from June, 2006 to June, 2008 during interviews, and patient focus group sessions, at 5 geographically dispersed regions in the United States. The focus group sessions involved an average of 8 participants in each locale; the largest group session involved 11 people, the smallest was 4 individuals. There were also 10 semi-structured interviews with key Alpha informants. Not included in the statistical analysis are data from 8 physician interviews, 3 nurse interviews, and data from observations, and interviews, with 1 physician assistant, and 4 laboratory personnel. These interviews focused on the biomedical point of view relative to empowerment in the Alpha-1 community, and were referenced as a counterpoint, or confirmation, of the patient narratives.

A purposive recruitment was conducted to maximize the quality of data through choice of articulate, and expert, key informants, and chain referral recruitment was conducted using Alpha-1 gatekeepers (Penrod et al. 2003; Juengst, 1998; Johnson, 1990). These gatekeepers all hold employed positions as AlphaNet coordinators. Each coordinator is responsible for a large number of diagnosed Alphas in their geographic region, and, through their extensive network of contacts, was able to recruit a sufficiently diverse set of participants to adequately represent the Alpha-1 community population. The relatively small sample size for this study was predicated on the requirements of ethnographic research, the need for in-depth narrative data, and, the rarity of this condition among the general population.

Methods

Data for the quantitative analyses were extracted from the semi-structured interviews, the focus group sessions and over eight years participant observation of the Alpha-1 community. The goals of the semi-structured interviews and focus group sessions was twofold – to define the components of empowerment in Alpha-1 through input from stakeholders, and to identify areas of relevance not covered in the literature on empowerment. The stakeholders, those people involved in or effected by Alpha-1, include diagnosed individuals, family members, caregivers, physicians, nurses, researchers, and organizational leaders.

The interviews lasted about 2 ½ hours and the focus group sessions lasted approximately 4 hours. The interviews and focus group sessions were conducted utilizing anthropological, and linguistic, interview methodology (Spradley, 1979, Boxer, 2003). Information derived from the interviews was both demographic and investigational in nature. Investigational data from the interviews and focus group sessions were transcribed and analyzed using narrative analysis techniques developed, and utilized, by Jehn and Doucet (1996), MacQueen et al. (1998), McLellan et al. (2003), Ryan and Bernard (2003), Boeije (2002), Abrahamsson, (2002) and Willms (1992). The empowerment questions and discussions were structured to evaluate the patients' understanding of their condition, their level of involvement in local support groups, and/or national patient organizations, their participation in research and advocacy activities, the quality of their care giving and family involvement, and the number of Alphas in their close circle and wider social network. Specific items, and their description, can be seen below.

Demographic Variables

Limited demographic data was assembled for the patient participants in the study (see Appendix C). This data includes patient's gender, age, years since diagnosis, family support, the number of Alphas in their close circle, and the number representing wider acquaintances.

Participants were also classified by their geographic region as a simplified means of representing different sub-populations, and sub-cultures, within the closed population of diagnosed Alphas in the United States. Gender was included as baseline data as it is an area of increasing interest in lung related research (DeMeo, 2007; Hersh et al. 2004a, b) and, in future studies, may prove a relevant variable in terms of the patient's or caregiver's extent of empowerment. Age was provided by only 33 of 49 participants, but was included, along with years since diagnosis, to track variation in experiences that may be based on age, and to test for a correlation between level of awareness and years since diagnosis. The number of Alphas a participant knows in their close circle, and among a wider set of acquaintances, was included to provide a quantitative means for tracking increases in involvement with the community, level of exposure to knowledge and support, and for determining if level of awareness, memberships, leadership, and involvement in community are significantly correlated.

Racial and ethnic characteristics were not considered in this study for several reasons – the majority of diagnosed Alphas in both the United States and Europe are Caucasian, the known penetrance of Alpha-1 is primarily among Northern Europeans, with a secondary strain among Mediterranean rim countries of Italy, Spain, Morocco, Israel, Egypt; and Alpha-1 is rarely diagnosed among Asians and those of Afro-American descent (de Serres, 2002; 2003; de Serres et al. 2007, 2006a, 2006b, 2003; Silverman et al. 1989). Furthermore, it was not the aim of this study to investigate the validity of the known epidemiology of Alpha-1, or to question the ethics of using a social construct, such as race, as a means of classifying genetic inheritance. However, these remain problematic issues requiring future study, and if properly highlighted, could lead to reconsideration of how the prevalence of Alpha-1 is classified among ethnic sub-populations, and how specific criteria are chosen to use in targeted detection programs. It is

interesting to note, relative to the ‘known’ penetrance of Alpha-1, that there are only 12 African-American Alphas (0.36%) in the Registry, and 14 Asians (0.43%), but 213 Native Americans (6.48%), a race rarely mentioned in the epidemiological literature on Alpha-1. Due to the snowball recruitment methods used, and the small proportion of African-American, Asian, and Native American Alphas diagnosed in the United States, all those who participated were apparently Caucasian. A consideration of race may help elucidate why certain sub-populations are more likely to participate in research studies (Coultas, 2007; Pressel, 2003; Corbi-Smith et al. 2003; Condit and Bates, 2005; Cox and McGarry, 2003; Schneider et al. 2003; Schutta and Burnett, 2003). When reasons for non-participation are known, it is possible that concerted efforts can be made to include more racial, or ethnic, variety in study recruitment.

Investigational Variables

A number of variables representing content specific areas were examined in this analysis. Variables and their descriptions follow:

Awareness

Levels of awareness were scaled to represent stages in knowledge about Alpha-1. The stages of awareness are based on a Bloom-like taxonomy of education ranking awareness according to specifically defined levels of knowing (Bloom, 1956). Bloom’s original taxonomy formed a cumulative six-stage hierarchy from simple to complex, and from concrete to abstract, with mastery of the simpler categories prerequisite to mastery of the more complex ones. The simplest category denotes a stage of knowledge involving recognition or recall of information, leading to higher levels of comprehension, application, and synthesis, to a state of knowledge incorporating evaluative strategies. This hierarchal approach is consistent with a model of empowerment as a Guttman scale, and the categories described by Bloom, and later revised by

Marzano and Krathwohl, were adapted for this study. In the revised taxonomy, there are four stages of knowledge (Table 6-1).

Although, Bloom's taxonomy was based on six stages of knowledge, only four distinct stages of awareness of Alpha-1 were observed. Like Marzano and Krathwohl, I therefore found it more useful to combine some of the original stages of knowledge delineated by Bloom, and to equate Awareness Level 1 with Factual Knowledge, Level 2 with Comprehension or Conceptual Knowledge, Level 3 with Procedural Knowledge and Level 4 with Synthesis and Evaluation or Metacognitive Knowledge. Using this four-tiered scheme, I classified participants as follows:

- **Awareness level 1** (Factual Knowledge) = little awareness other than knowing the name of the condition, its genetic inheritance, and the possibility of illness and death associated with Alpha-1
- **Awareness level 2** (Comprehension) = awareness of the variants of Alpha-1 Antitrypsin Deficiency (i.e., ZZ phenotypes are homozygote and MZ, MS, SZ phenotypes are heterozygote); some understanding of the possible pathogenesis of the lung or liver disease associated with Alpha-1; some understanding of the options for treatment and the necessity for respiratory therapy, supplemental oxygen and avoidance of certain exposures
- **Awareness level 3** (Procedural Knowledge) = expanded understanding of the variants of Alpha-1 and their variable risk of disease, basic understanding of the genetic and biomedical terminology used by health professionals about Alpha-1, basic understanding of the different types of research being conducted, awareness of specific legislative, economic and legal restraints associated with a genetic condition, understanding about disease self-management
- **Awareness level 4** (Synthesis and Evaluation) = mid-level to expert awareness of the science of Alpha-1, facility with and understanding of the genetic and biomedical terminologies used by health professionals, fuller understanding of the types of research being conducted, full awareness and participation in the specific legislative, economic and legal issues associated with Alpha-1, good to very good understanding of their own condition and how to manage it. Another factor denoting level 4 awareness is the ability to articulate about Alpha-1 and its impact on their personal lives.

Although, the dynamic of the focus group sessions might have obscured the opinion of shy or quieter individuals, efforts were made to solicit responses from all participants. This

included, at times, politely constraining more assertive members during focus group sessions, or introducing periods of more formal turn-taking in response to specific topics, to ensure all voices were heard. In one focus group session, the host (my Alpha recruiter) warned me that he tended to dominate group conversations, and knew he was the most assertive person of the group. In his case, we made an agreement that I would ask him to be quiet if he spoke too often, or obstructed others from participating. I did, in fact, have to take this action twice during his focus group session, but the dynamic remained friendly and constraining him did open up the floor for others to participate.

Organizational membership

Involvement in Alpha-1 organizations includes participation in local support groups, and/or membership in national Alpha-1 organizations. (0=No and 1=Yes). Of the 49 study participants, 39 were members of an Alpha-1 organization. However, the data did not distinguish between local and national memberships. Participation in a local group indicates some knowledge about resources available to Alphas, and provides exposure to those more experienced in living with Alpha-1, as well as providing a forum for connecting caregivers with each other. Membership in national organizations, however, may indicate a greater level of commitment to a potentially more demanding form of involvement. Those who join the Alpha-1 Association, Better Breather Clubs, American Lung Association, or the Alpha-1 Foundation's advisory committees frequently travel more, attend both patient and scientific meetings, or serve as representatives, or advisors, on behalf of other patients. This increases an Alphas' exposures to disease vectors during travel, and in large meetings, is disruptive to the work and home lives of the patient, their family and their caregiver, and creates extra challenges in terms of complying with daily treatment regimes. Air travel is particularly challenging for Alphas in

an advanced disease stage, and for many Alphas, travelling requires a companion/caregiver which increases the costs of participation.

Although, membership in an organization is a useful indicator of a growing commitment to networking, and increased involvement with other Alphas, caregivers, and experts in Alpha-1, the variable, as defined, did not distinguish between local involvement, and the potentially more demanding economic and physical commitment of membership in a national organization. Future studies will be designed to unpack this variable into its components, and test whether local vs. national involvement correlates with years since diagnosis, or whether national organizational membership is more indicative of, or leads to, the structural components of empowerment.

Participation in research

The Alpha-1 research network was originally created to increase the participation of diagnosed Alphas in research and clinical trials, and, is, according to many medical professionals in a variety of disciplines, a primary indication of empowerment (Anonymous 2004; Dickersin and Rennie 2003; Mannheimer and Anderson 2002; Meropol et al. 2003; Morgan 1992; Schneider et al. 2003; Weinfurt et al. 2003; Stoller et al., 2000; Sveger et al. 1999). This seemed an important variable to track and, as used in this study, includes participation in all forms of research, such as clinical trials; biomedical, bioethical or social research studies; and/or responding to a survey or questionnaire. The variable was included as it was unclear from the qualitative analysis whether individuals always reach of certain level of awareness before they participate in research, or whether knowing about Alpha-1 does not necessarily precede a commitment to research participation. Analysis of the variable 'research participation' would also elucidate whether it is an intervening variable to membership in organizations. Values used for this variable are: 0=No and 1=Yes.

Advocacy

This component of empowerment is similarly multivariate. For, advocacy can include participating in letter writing campaigns, visiting state or federal officials to advocate for Alpha-1, testifying before Congress, or assisting in developing advocacy programs. Future survey questions that separate these activities may result in a more accurate view of the progression of advocacy, and empowerment. However, in this study advocacy only indicates activities in this area, not the quality or amount of activity or the level of commitment such activities implies. Values used for this variable are (0=No and 1=Yes)

Organizational leadership

Only a few individuals in the study are classified as an organizational leader, however, it seems a key variable to include as it is presumed organizational leaders display, or demonstrate, a higher level of awareness, commitment, and empowerment than those newly diagnosed, or compared to those who were involved as members of local support groups. Membership in support groups, and policy makers, were further permutations intended to capture involvement and commitment in the community. The values for all three of these variables were assigned as 0=No and 1=Yes.

Membership in a supra-organization

Similar to organizational leaders, only a few participants are members of a supra-organization. These types of organizations represent larger constituencies than a single rare disease community, and are created to manage common goals and widespread issues. Examples include organizations such as the National Health Council that is comprised of over 150 Voluntary Health Organization members, the American Thoracic Society, with over 10,000 members worldwide, the National Institutes of Health, Public Advisory Panels, or the International Lung Association. During fieldwork, I had the opportunity to attend a National

Health Council meeting where I observed individuals at the highest, most functional end of the scale of empowerment. Membership in one of these organizations, therefore, represents for me the upper end of the Guttman scale of empowerment, and highest manifestation of empowerment in the Alpha-1 community.

Scoring Rubric

Long term observations, and analysis of interviews and focus group data, were used as the basis for scoring participants for each of the variables described above. Although, some of the variables were straightforward (membership, leadership, policymaking), one variable, Level of Awareness, proved more challenging as a category for classifying the participants experiences. As described above, a widely used educational classification scheme, based on Bloom's Taxonomy, was used to help distinguish between forms of awareness along a hierarchal scale. Level 0 was marked when an individual admitted to "knowing nothing" about Alpha-1, and, in each case, was an individual diagnosed within a few weeks of our meeting. Factual knowledge (level 1) was noted when a participant asked more questions than they answered, demonstrated a lack of understanding of the medical terminology or devices used to describe and treat Alpha-1, had little concept about the Alpha-1 research or patient support networks, and who did not assume a leadership or advocacy role, even among their local support group members. Level 2 assignees demonstrated greater understanding of their condition, and knowledge of the ways and means of dealing with the specifics of Alpha-1; this was often demonstrated by their detailed descriptions of their diagnosis, and a retrospective understanding of the implications of their condition, and their comments on different treatment methods and research initiatives. Those assigned to Level 3, on the other hand, vocalized their awareness of specific legislative, and the economic and legal restraints associated with their genetic condition. They seemed able to contextualize their experience, and to be able to

extrapolate more about the outcomes of different research initiatives. Like Level 2 assignees, these individuals were more often answering questions about Alpha-1 than asking them. An additional means of distinguishing between Levels 2 and 3 was the level of confidence the individual displayed when explaining Alpha-1 to newly diagnosed individuals; Level 3 individuals did not second guess themselves, or add caveats to their statements, as often as Level 2 individuals did.

The majority of individuals in the study were assigned Levels 2 or 3, however, 3 of the participants were assigned Level 4 awareness. Each of the individuals assigned as Level 4 assumed a leadership role in their focus group session, discussed strategic alliances, described their vision of a cure, talked about their plans for the community, or how they had participated in establishing mission statements, or advocacy initiatives, on behalf of the community. These individuals also demonstrated a heightened familiarity with the medical terminology associated with their condition, and a more synthesized understanding of the biological processes affecting them, and being studied by researchers in the field.

Statistical Analysis

Descriptive statistics were performed in JMP (SAS Institute, Cary, NC), and expressed as mean +/- standard deviation and range (Tables 6-2 and 6-6). Box plots were generated encompassing the middle 50% of data. Continuous variables were characterized, and compared to other continuous variables by simple linear regression. Comparison between subgroups was performed by analysis of variance, or chi-square analysis depending on data characteristics. P values <0.05 were considered significant.

Additional analyses were performed in R, version 2.7.1 (R Development Core Team, 2008). Phi was calculated between dichotomous investigational variables to assess correlation. Kendall's tau (1938) was calculated among dichotomous variables and awareness, ordinal in

scale. Principal components analysis (Armitage, Berry, & Matthews 2002) was used to extract factors from the nine content-specific areas (Table 6-3). Principal components were determined by using both the Kaiser Criterion (Kaiser 1960) and the scree plot (Cattell 1966).

Results

Study participants were from Midwest (n=10; 20%), Northwest (n=9; 18%), Southeast (n=19; 38%), Northeast (n=7; 14%), and Southwest (n=4; 8%). No racial or ethnic data was collected, however this sample can be characterized as being predominantly white, non-Hispanic.

Participants were categorized as being aware on a scale from zero to four, with a plurality of participants judged to have a “2” level of awareness (n=19/49, 38%). There were almost an equal number of participants having a “1” and “3” level of awareness, n=13 (27%) and n=10 (20%), respectively. Two participants were judged to have a “0” level of awareness (4%) and five participants had a “4” level of awareness. Table 6-4 shows the distribution of the levels of awareness.

Correlation analyses between individual domains of the total empowerment score showed strong correlation among; research participation and advocacy ($R^2=0.27$, $p<.0001$); organizational leadership ($R^2=0.22$, $P=0.002$); a good diagnosis experience ($R^2=0.49$, $p<.0001$) and higher level of awareness ($R^2=0.50$, $p<.0001$). Policy makers had more organizational leadership ($\phi=0.45$); and memberships in a supra-organization ($\phi=0.64$). Lesser degrees of correlation were demonstrated between level of awareness and “family care” ($\phi=0.01$) and the only negative correlation was between type of diagnosis and “family care” ($\phi=-0.08$).

The total empowerment scores were highly correlated with the individual domains of the score. Significant correlations were found with awareness ($R^2=0.70$, $P=.0001$), (Figure 6-3),

policy making ($R^2=0.36$, $P<.0001$), organizational leadership ($R^2=0.56$, $P<.01$) (Figure 6-4), advocacy ($R^2=.0.44$, $p<.0001$), research participation ($R^2=0.63$, $P<.0001$), and membership in a supra-organization ($R^2=0.20$, $P<.001$). The total empowerment score also proved to be associated with the numbers of networked individuals ($R^2=0.70$, $P=.0001$), which, similar to the relation between total score and level of awareness, explained 70% of the variance. The numbers in an individual's network and their level of awareness, therefore, seemed to be the most significant of all the correlations.

In an effort to define if empowerment was simply a function of time since diagnosis, further correlative analyses were performed. A larger network of individuals was seen in individuals diagnosed longer ($p=0.052$) (Figure 6-6). Longer years since diagnosis were seen for individuals with research experience, ($p=.002$) (Figure 6-7), but not for any of the other individual domains of the total empowerment score. And, although the total empowerment score was associated with years since diagnosis ($R^2 =0.10$, $p=0.04$) (Figure 6-1), the variance suggests that the composite score may not be as sufficiently robust a tool as individual domains.

The factor analysis was performed to see if individual domains tracked together, however, multi-factorial analysis was not performed to validate the Guttman scale hypothesis. This final analysis requires a larger cohort, for statistically significant results, and the assistance of a statistical consultant to perform these more complex calculations. However, the principal components analysis did confirm the basic groupings hypothesized as a Guttman scale, specifically, two principal components (Eigen values 3.2 and 1.2) accounting for 52% of the variance (Figure 6-8). The first principal component accounted for 30% of the variance; level of awareness, organizational membership, research participation, advocacy, and organizational leadership loaded on this component. This component was labeled *individual (personal)*

empowerment. Membership in a supra-organization and participating in policy making loaded mainly on the second component. This component was labeled *structural (community) empowerment*.

Discussion

The results confirmed the validity of modeling components of empowerment as a Guttman scale, despite the fact the distinction between personal and structural forms of empowerment fell along a different set of components than hypothesized. In the model of empowerment, organizational leadership fell within structural empowerment, not personal empowerment, but the statistical analysis did not confirm this grouping of components. However, the statistical analysis did confirm significant correlations between components similar to those hypothesized, and validated the concept that some forms of empowerment precede others (Figure 6-8).

One hypothetical correlation was identified in the analysis as a moderate correlation between membership in a supra-organization and policymaking – only those at the most empowered end of the scale participate as a representative of their community on a national and international level. However, other correlations demonstrated strong significance between personal forms of empowerment such as awareness and years since diagnosis, research participation and numbers in Alpha network, and research participation and years since diagnosis. More moderate associations were found between research participation and organizational leadership, and research participation and awareness. However, the most significant relations, total score and research participation, total score and level of awareness, and participation in research and numbers in Alpha network, explained, respectively, 63%, 70% and 72% of the variance.

A future study is planned, that will involve verifying the statistical results through re-contact with original study participants, and community feedback regarding the validity of correlations described as significant. This future study will commence in October, 2008, and has also been designed to gather demographic and investigational data on a larger cohort towards more statistically significant results. Surveying a larger cohort may result in a more precise view of the statistical relationships, particularly the Guttman scale, or may require refiguring the hypothetical model. A further possibility is the definition of the variables chosen does not adequately represent the variation within that component, and additional review of the narrative data is needed to tease out the relevant factors. A redefinition, or expansion, of the component 'advocacy', for example, may yield more accurate data by distinguishing between types of advocacy activities. Another possibility is that the purposive recruitment used in this study skewed the results, and a random sampling would be more representative of community attitudes and attributes. This may be particularly true in relation to the variables organizational leadership and participation in research. The study participants, by participating in my study, were all coded as participating in research, however, for several of them it was the first, and may be the only, time they participate, whereas others regularly participate in research and in clinical trials. This variable needs further refinement. In addition, the purposive sampling may have resulted in an over-representation of individuals already well on their way to being empowered. As noted above, a random sampling of a larger cohort may resolve this issue, and provide a clearer picture of the full range of experience in the Alpha-1 community.

Despite the obvious limitations of statistical analysis of such a small cohort, and the need to redefine or expand at least two of the components, it did prove useful to combine qualitative and quantitative methods. The statistical analysis did confirm significant correlations derived

from the qualitative data, and the validity of the relevance, and utility, of the nine identified components of empowerment. The resulting picture of stages of empowerment, based as it is on many years observation, analysis of ethnographic material, and a statistical analysis, can now be tested empirically and, as appropriate using a reiterative process, be revised according to the perceptions of community members. This approach does not denigrate the value of my own observations and preliminary conclusions, rather ensures that the final model of empowerment, and components of empowerment used in survey instruments, is firmly grounded in validated qualitative and quantitative results.

An additional advantage of combining qualitative and quantitative results is to bridge the divide between social and medical research, and to make the results accessible to the medical experts, and policy makers, involved in the Alpha-1 community. Since, empowering patients includes interaction, communication, and understanding between medical professionals and their Alpha-1 patients, it is important to make the results of this study relevant to medical professionals, and presented in forms they can understand (i.e., statistical results). Strong statistical correlations based on qualitative data, as identified in this study, may help strengthen the role qualitative data plays in understanding medical issues, and allow combined qualitative/quantitative study outcomes to be incorporated into medical practice and standards of care.

Table 6-1. Structure of knowledge dimensions

A. Factual Knowledge

1. The basic elements that must be known
2. Knowledge of terminology and specific facts

B. Conceptual Knowledge (Comprehension)

1. Knowledge of conventions, trends, sequences, categories, criteria and methodology
2. Knowledge of principles, theories, and structures
3. Translation, interpretation, and extrapolation of information

C. Procedural Knowledge (Application)

1. Knowledge of subject-specific skills, techniques and methods
2. Knowledge of criteria for determining when to use appropriate procedures
3. Analysis of elements, relationships, and organizational principles

D. Metacognitive Knowledge (Synthesis and Evaluation)

1. Strategic knowledge
 2. Production of a unique communication, plan or proposed set of operations
 3. Derivation of a set of abstract relations
 4. Evaluation in terms of internal evidence
 5. Judgments in terms of external criteria
-

Table 6-2. Descriptive statistics of demographic characteristics

	Min	1 st Qu	Median	Mean	3 rd Qu	Max	Missing	StdDev
Age	34.0	52.0	58.0	56.9	61.3	81.0	17.0	8.80
Years Since Diagnosis	1.0	6.0	10.0	10.7	14.0	36.0	4.0	6.45
Close Circle	1.0	6.0	50.0	50.4	70.0	150.	33.0	51.21
Alpha Network	24.0	100.0	400.0	378.8	585.0	1000	32.0	291.20

Table 6-3. Frequency distribution of six of seven content specific scores

	Frequency (%)	
Male	44.9%	22/49
Positive Diagnosis	54.5%	18/33
Family Care	93.8%	45/48
Organizational Membership	79.6%	39/49
Participate in Research	38.8%	19/49
Advocacy	30.6%	15/49
Organizational Leadership	24.5%	12/49
Membership Supra-Organization.	6.1%	3/49
Policymaker	6.1%	3/49

Table 6-4. Levels of awareness frequency distributions

Level of Awareness/Description	Frequency (%)	
0 Newly diagnosed	4.1%	2/49
1 Factual knowledge	26.5%	13/49
2 Comprehension	38.3%	19/49
3 Procedural knowledge	20.4%	10/49
4 Metacognition, Synthesis	10.2%	5/49

Table 6-5. Total empowerment scores for nine domains of empowerment

Domain	Score	Weight	Maximum Pts
Type Diagnosis	No=0, Yes=1	1	4
Family Support	No=0, Yes=1	1	4
Awareness Level	0= no awareness	1	4
	1=factual awareness		
	2=comprehension		
	3=procedural awareness		
	4=synthesis, evaluation		
Organization Membership	No=0, Yes=1	1	4
Participation in Research	No=0, Yes=1	1	4
Advocacy Activities	No=0, Yes=1	1	4
Organizational Leadership	No=0, Yes=1	1	4
Membership in a Supra-organization	No=0, Yes=1	1	4
Participation in Policy	No=0, Yes=1	1	4
Total Empowerment Score			36

Table 6-6. Descriptive statistics of total empowerment scores

	Min	1 st Qu	Median	Mean	3 rd Qu	Max	Missing	StdDev
Total scores	2	9.5	13.0	14.73	19	36	0	7.91

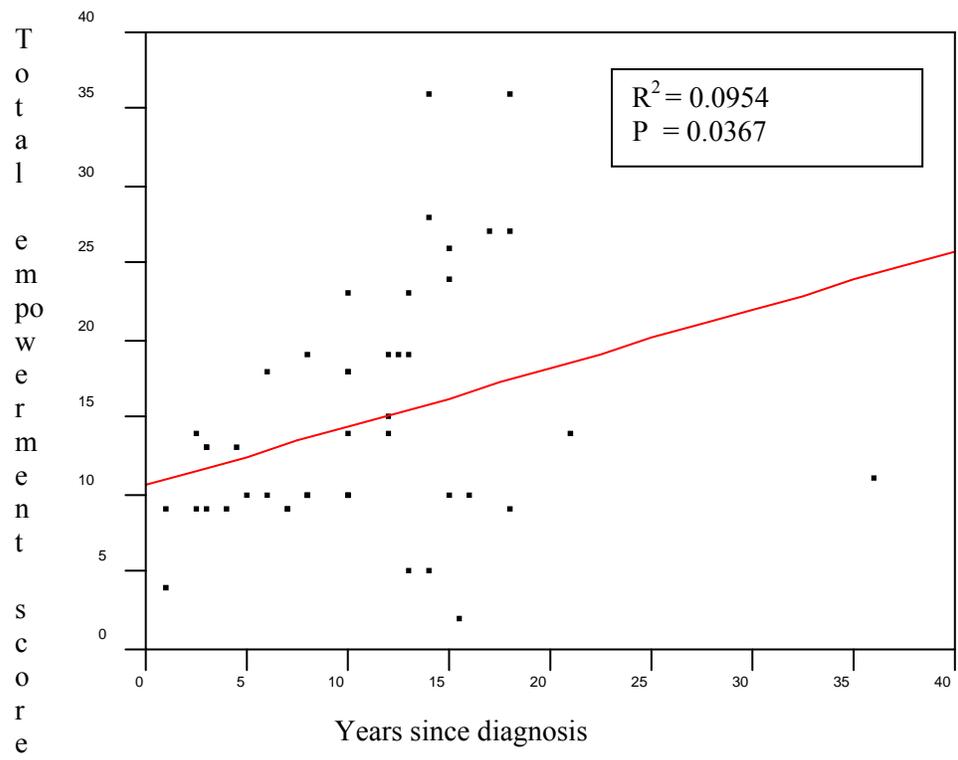


Figure 6-1. Bivariate fit of total empowerment score by years since diagnosis

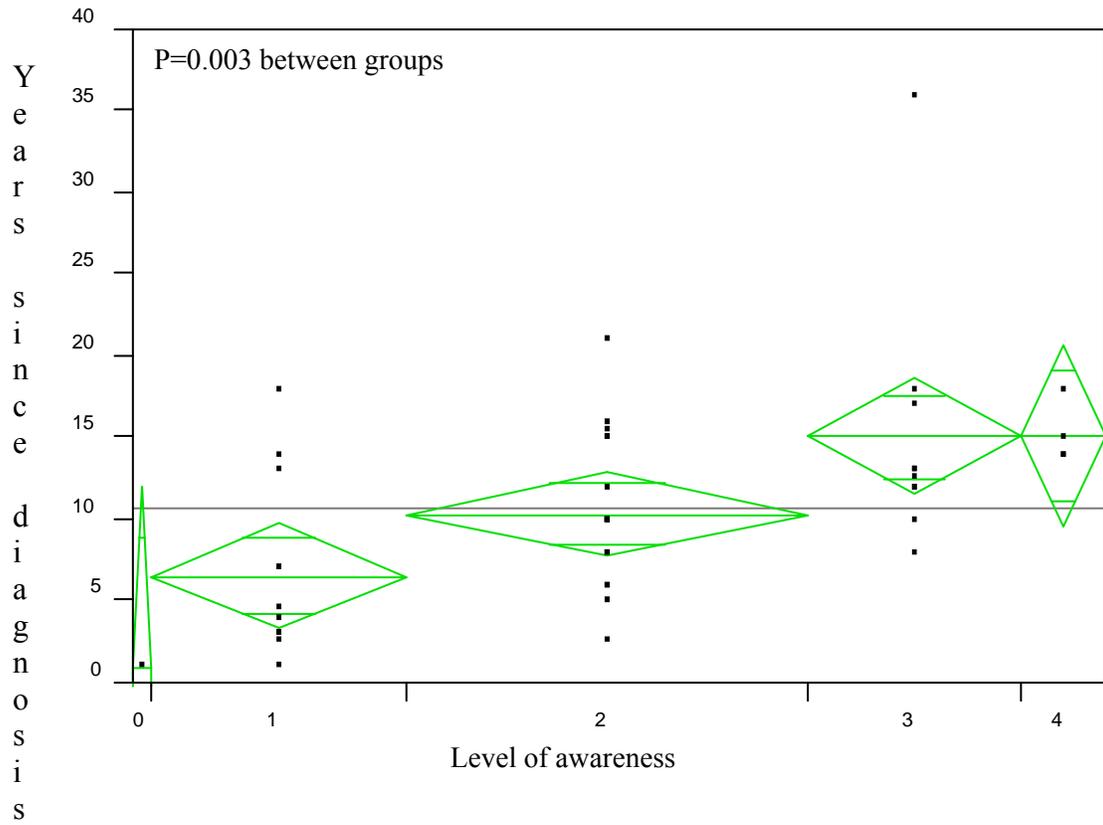


Figure 6-2. Years since diagnosis by levels of awareness

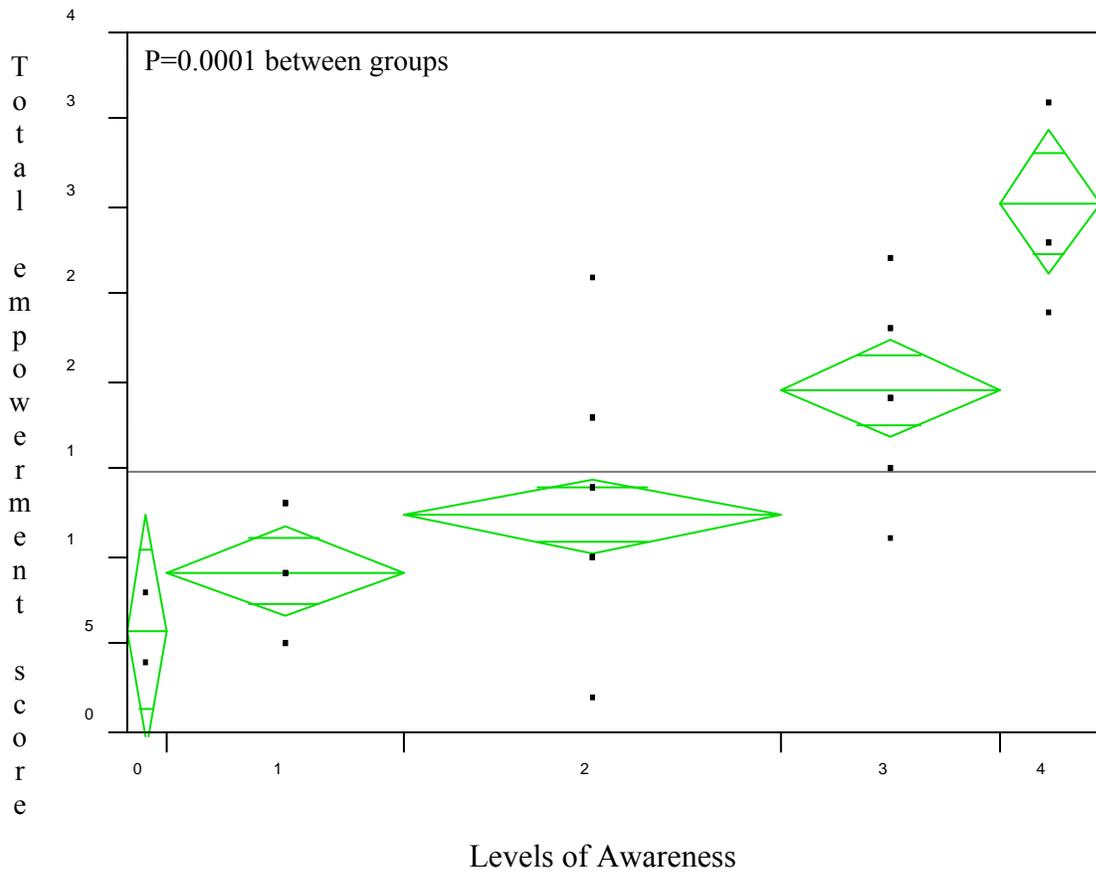


Figure 6-3. Total empowerment score by awareness

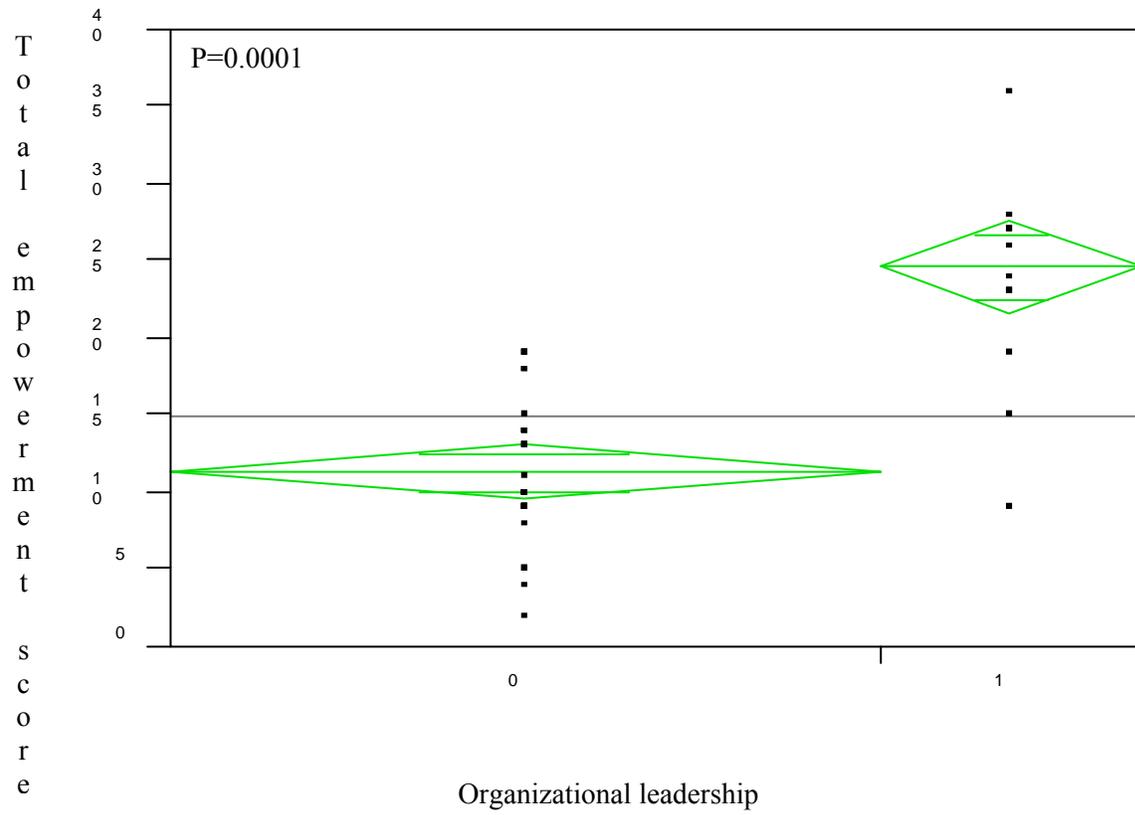


Figure 6-4. Total empowerment score by organizational leadership

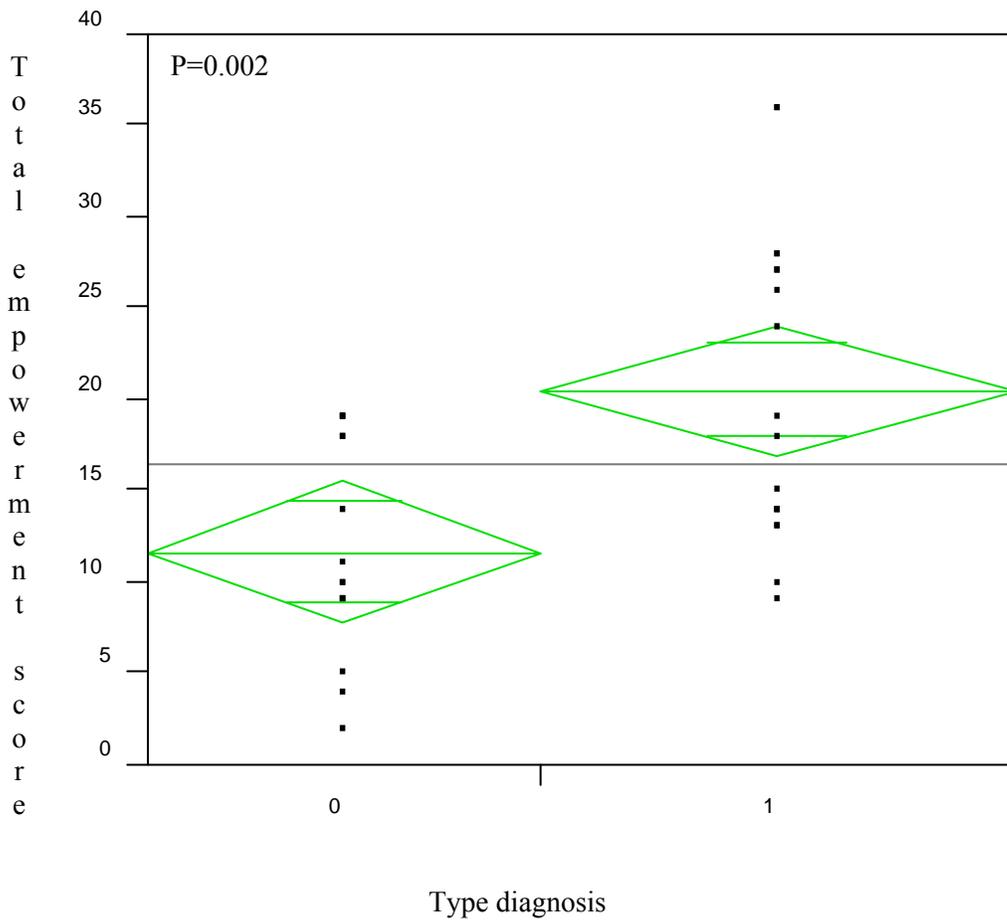


Figure 6-5. Total empowerment score by type diagnosis

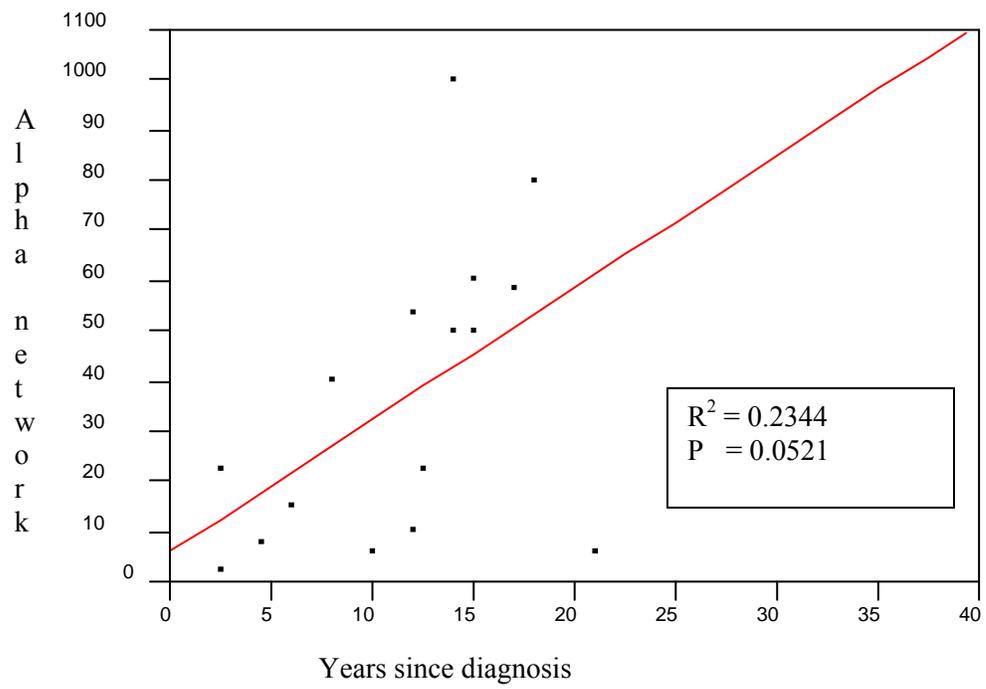


Figure 6-6. Alpha network by years since diagnosis

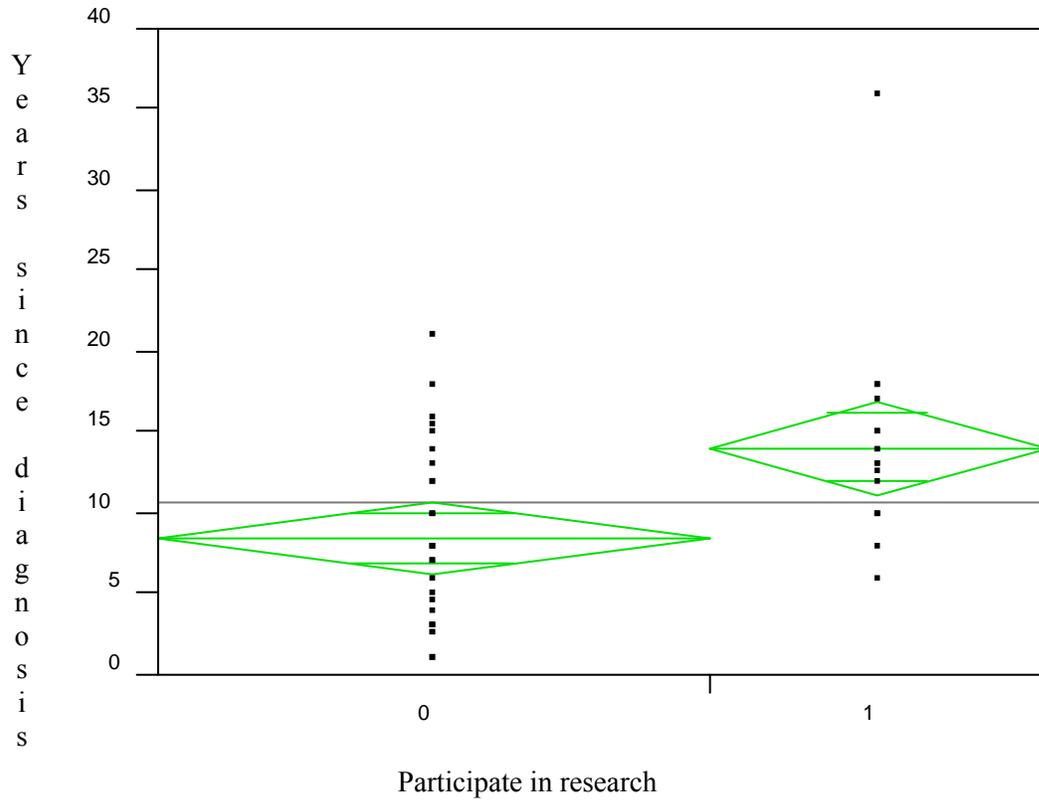


Figure 6-7. Years since diagnosis by participation in research

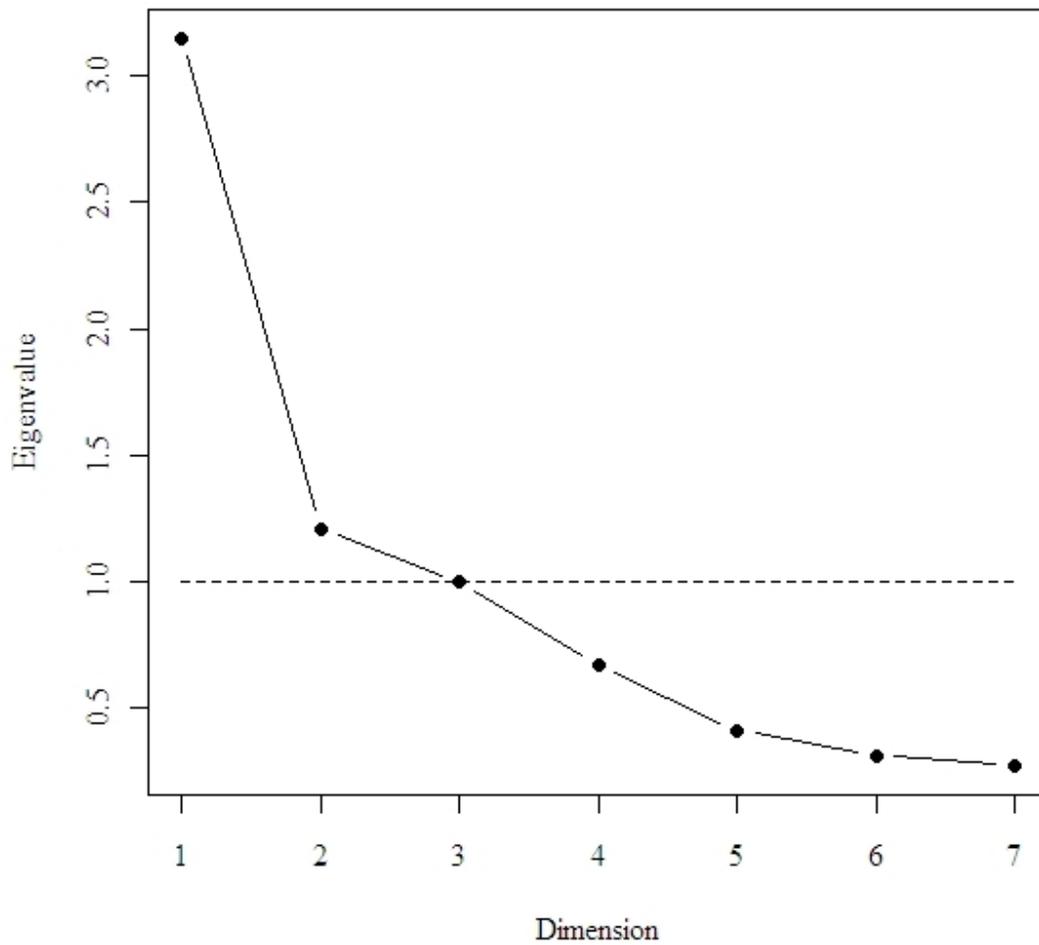


Figure 6-8. Scree plot of principal components analysis. 1 = Membership in a supra-organization; 2 = Participating in policy making; 3= Level of awareness; 4 = Organizational membership; 5 = Research participation; 6 = Advocacy; 7 = Organizational leadership. Results of principal component analysis (Eigen values 3.2 and 1.2) accounting for 52% of the variance. The first principal component accounted for 30% of the variance.

CHAPTER 7
EMPOWERMENT IS SIMPLE, EMPOWERMENT IS COMPLEX

Key Elements in Empowerment

Rarity and Genetics

Studying empowerment in a cohesive, rare disease community, as with the Alpha-1 community, may be a particularly effective way to identify, and quantify, components of empowerment. There are several reasons why this may be true, and why the Alpha-1 community, in particular, is an appropriate model of empowerment among rare and genetic disease communities. The Alpha-1 community has developed resources, and an identity, that serve all stakeholders interested in Alpha-1: there is a visible and formalized infrastructure; there is scientifically validated information available to both Alphas and the research community in publications and on websites; there are annual educational and scientific conferences; there is a professionally managed advocacy program; and many members of the community are aware, articulate, and concerned about empowerment. The access and understanding I had developed over the years about Alpha-1, and the community, made this particular rare disease group the appropriate model for my exploration of empowerment.

However, there are additional reasons why it is fitting to choose a rare and genetic disease community to understand, and quantify, empowerment. A rare disease community, by its definition, is small. In the United States, a rare disease is defined as one with fewer than 200,000 affected individuals (<http://rarediseases.info.nih.gov/>). The development of rare disease patient networks, and the extended communities that grow up around a rare disease, like the Alpha-1 community, therefore represents small, bounded populations. The criterion for membership is a diagnosed condition, or disease, or a family member with the condition, and all members of these types of communities are connected through an explicit common cause. The

medical and research networks, that are also part of these extended disease communities, are likewise linked through common interests and goals.

Chronic disease communities, in comparison, often do not have such a clearly defined common purpose, as the cause of many chronic conditions, like cardiovascular disease or COPD, is still not known, or has been identified as arising from multiple causes. The terms cardiovascular, or heart, disease, and COPD, each cover numerous distinct conditions. COPD, for example, may mean an individual has chronic bronchitis, emphysema, refractory asthma, severe bronchiectasis, frequent bouts with pneumonia, or airway disease (www.copdfoundation.org). Heart disease, as well, connotes several different types of disease associated with the heart, the interaction between the heart and lungs, or the interaction between the circulatory system and the heart. Patients may identify as a COPD patient, but have no common symptoms, medications, or treatments as another patient diagnosed with COPD (Caress et al. 2005).

Alpha-1 Antitrypsin Deficiency has been diagnosed among approximately 5,000 individuals in North America, whereas, NIH estimates that 12 million individuals have been diagnosed with COPD (Anto et al. 2003; www.copdfoundation.org). Of the 5,000 diagnosed Alphas, 3,289⁵ have enrolled in the Alpha-1 Research Registry. They may not all know each other, but, each one of these diagnosed Alphas has registered to be part of a well established community with others with the same condition. Like an exclusive club, the Alpha-1 community has very few members, and, precisely because the condition is rare, a strong sense of uniqueness and even distinction - in short, a positive identity. The community identity also comes from shared attitudes, and in the Alpha-1 community there is a tendency to focus on

⁵ Enrollment figure from 08/01/08 Alpha-1 Research Registry Quarterly Report, Medical University of South Carolina.

what can be accomplished as a group, rather than on a shared disability. Although the Alpha-1 community serves as the model for empowerment, I have also noted a similar sense of positive identity in the Lymphangiomyomatosis (LAM) rare lung disease community, and a growing sense of identity in the newly formed Pulmonary Alveolar Proteinosis (PAP) rare lung disease community.

The linguistic aspect of the word “rare” is also an important factor in the positive identity I observe among rare disease communities. NIH officials pushed for a reclassification from “orphan” to “rare” disease in the 1990s, as a conscious effort to improve the image of the smaller disease communities (Groft, Pers. Comm. 2008). Rareness has a decidedly more positive connotation than “orphan”, and may represent a potential linguistic strengthening, and empowering, for rare disease communities that is not be equally available to the millions of people suffering from chronic diseases.

The genetic aspect is another factor that made the Alpha-1 community an appropriate model for a study of empowerment. There is no stigma attached to a lung condition that was not caused by cigarette smoking, or to a liver condition that is not caused by alcohol or drug abuse. Although, Alphas are frequently asked if they smoked, or drank, they can respond without a sense of guilt, that no, what they have is genetic. Cigarette smoking, drinking, and other environmental impacts have been identified as increasing the risk factor for development of Alpha-1, but even when an Alpha has smoked, they can still credit their genetics as the primary cause of their disease (Sharp et al. 1969; Mayer et al. 2006; Teckman, 2007). COPD patients, on the other hand, must live with a sense of guilt that they caused their own emphysema by smoking. The burden of guilt should not be dismissed lightly in an era when patients are told their lifestyle choices put them at risk of disease, and media informs us that the indoor air

environment humans create is a greater risk than outdoor exposures. There is pressure on patients to take responsibility for their health, and by extension, when their health fails, they must also be responsible (Hanna, 1998; Gunderman 2000; Loughlin 2003).

I conclude that stigma is un-empowering, and must be addressed in disease communities where there is no known genetic cause. In the Alpha-1 community, the patients can bypass a negative sense of responsibility for their illness, and focus instead on more positive, and productive, attitudes. Lack of stigma is thus a primary element in the Alpha-1 community's empowerment. Other rare disease communities have an equal potential for empowerment, but addressing stigma may help to promote empowerment for many other types of patients.

A genetic condition has another positive advantage in the 21st century. Biotechnology and genetics, instead of representing what I term 'the fear of Frankenstein' that earlier genetic research had predicted, have become defining and ubiquitous themes in United States, and global, culture (Stock, 2003). Instead of the public fear of science, and scientists, exemplified by the Frankenstein archetype, and other 'mad scientist' science-fiction films, we have become eager in our society to "say **Yes** to science and technology" (Downey and Dumit, 1997, authors emphasis). Like other social scientists, I perceive the growing appeal of biological explanations in our culture (Nelkin, 1996a), and how the younger generations are embracing the "techno-eroticism" of science and technology, that Hardaway first noted in the 1990s (Hardaway, 1993).

The appeal of biological explanations created the larger social framework for the rapid advances being made in genetic research since the early 1990s. Legislators, like many of us, were caught up in the excitement about mapping the genome, and other genetic discoveries, and designated increasing funding for genetic research in the United States. Since that time, society in the United States seems to have shifted from trepidation about the use of genetics, to the

sometimes unreasonable expectation that, in the human genome map, we will find definite answers about the causes of health and disease (Collins et al. 2003). Newspaper headlines tell us almost daily of new genes that have been discovered relating to some disease, or mental state, or how new technologies are allowing scientists to manipulate genes and cells to cure diseases (see Table 7-1). Despite the huge miscommunication gap between the scientific discoveries, and how they are reported in the media (Nelkin, 1996b), news about genetics is generally positive, and has helped to establish the value of genetics, and genetic research, in our society.

The rapid incorporation of computers into daily life, that began to occur in the early 1990s, has also had a profound impact on societal attitudes about technology (Goodman, 2005). A generation that has grown up with computers is now hooked on mechanical devices, and computer technologies, that prevail over personal interaction, physical activities, and public entertainment. It is a sad day, indeed, when someone is diagnosed with WiFi shoulder from playing a computer game, rather than dealing with a tennis elbow obtained on an actual court, but, that day is here, and it connotes a culture that valorizes all forms of technology. I see this embrace of technology as impacting the practice of medicine - it makes the use of highly technical equipment and tests more acceptable, and the role of genetics, and scientific manipulation of genes, a welcome form of modernity. Boasting among scientists is frequently based on who has the newest, most state-of-the-art equipment, or who has developed a new cutting edge biotechnology. Scientists know new is best.

Having a genetic condition like Alpha-1 is also a signifier of modernity. To be diagnosed implies having access to advanced technologies that can scan your genes, identify genotypes and phenotypes, and determine whether you have a genetic flaw. And, there are no home

remedies for Alpha-1, no history of folk medicine cures, only scientific and genetic solutions. Alpha-1, and empowerment in this community, thus becomes an examination, and example, of how we deal with disease in the modern era – a topic of general interest in our society. It does not matter that Alpha-1 is rare, it can, and does, represent how society conceives of disease in the genetic era, and attitudes about illness and its management.

Socio-Political Context

Dr. Moseley, a bioethicist I interviewed for this study, listed several key socio-politic influences that have figured into the context for empowerment. He included a general questioning of authority in the 1960s, 1970s, and 1980s in political, academic, and cultural arenas (Chernichovsky, 2002; Mechanic, 1996; Teffs, 1994; Clarke et al. 2003; Schlesinger, 2002); the proliferation of self-help and alternate medicine books in the 1980 and 1990s; the accessibility to information, and other patients, the Internet provided by the mid 1990s; and the rise of bioethics as a distinct field in medicine.

Bioethics, as a distinct field, is almost as new as the discovery of Alpha-1 Antitrypsin Deficiency, and, equally dependent on discoveries in genetics. It arose as a discipline in response to specific issues relating to the genome mapping. The first formal Ethical, Legal, and Social Issues advisory committee was formed as a component of the Human Genome Project at the NIH (Juengst, Pers. Comm. 2005; Collins, 2004; Collins et al. 2003). For, it had become almost immediately apparent to the scientists involved in the genome project, that genetics research could lead to several very slippery ethical slopes. There now existed the possibility of human enhancement, for those who could afford it, cloning, inappropriate use of an individual's, or family's, genes, genetic discrimination, and the specter of eugenics (Caulfield, 1998; Caulfield et al. 2008; Coultas et al. 2007; DeRenzo et al. 2001; Juengst, 1998; Juengst and Fossel, 2000; Dugan et al. 2003; Dressler and Juengst, 2006; Driscoll, 1998; McGee and

Juengst, 1999; Foster and Sharp, 2006; Kicklighter and Sharp, 2005; Pressel, 2003; Slaughter, 1994, 1997; Collins et al. 2003; White, 1999). Would you, as an insurance executive, want to provide coverage to someone with a known genetic flaw? Or, would you, as an employer, want to hire someone with a genetic condition for a crucial job in your company? Or, should families test children if there is no known therapy, or cure, for a condition they might, or might not, develop? But, what if the flaw did not definitely lead to disease, and the risk was difficult to define for any one individual, like happened with the BRCA2 gene? These types of questions are the basis of current bioethical consultations, but as genetics advances, additional issues will arise, and the field will continue to evolve.

Bioethics has subsequently developed as a philosophical check on other types of medical activities, such as advanced planning, but remains a potent force within genetics research, and an appropriate intervention when medical or research goals fail to account for the human dynamic. It is this interventional aspect that relates to empowerment. By focusing on the individual's rights, and the ethical concerns relating to families, privacy, and confidentiality in genetic conditions, bioethics has emphasized the social dimensions of the illness experience.

Bioethics was my introduction to social research in the Alpha-1 community, and through several bioethical projects I was involved in, I began to see a potential for anthropological research to complement the work of the ethicists. Together, I believe medical anthropologists and bioethicists can more effectively intervene on behalf of the patient community, and ensure that the lived experience, and ethical implications of a genetic condition, is not subsumed by the domination of the scientific perspective.

An additional factor underlying patient empowerment is the aging of the population, and the growing proliferation of chronic disease patients. With extended life spans, many more

individuals are living with chronic and disease conditions for 10, 20, even 30 years after diagnosis. The culture of medicine has had to shift from an emphasis on critical care to chronic care (Hanna, 1998; Warren et al. 1998). In chronic care, the patient, caregivers, and family members play a greater role in disease management, and must become more aware of sometimes subtle symptoms, what keys off exacerbations, how to effectively communicate with medical staff, and how to manage illness at home. Since awareness, disease management, and communication with medical staff are hallmarks of empowerment, chronic conditions may yet provide the potential for empowerment

Maintaining a Traditional Hegemony

Socio-political factors, rare and genetic conditions, and a technophilic culture all play a role in promoting patient empowerment. Responsibility for knowledge production, however, is an area where patient empowerment is still challenged. I have conducted an ongoing survey of newspaper articles about science and medicine since 1993, to discern trends in medical, and environmental science, and to identify the social factors that impact the cultures of science and medicine. I grouped selected articles by topic (see Table 7-1) and began to see clear patterns that forebode patient empowerment. Media stories, for example, had begun, in the 1980s and 1990s, to challenge the objectivity of scientific truths (Condit and Bates, 2005, Geller et al. 2002). This exposed an ongoing tension between those within academia producing knowledge, and those interpreting its products from the outside (Nelkin, 1996b). Medical sociologists and anthropologists also began to call attention, in the period beginning in the 1970s, to the cultural bias of investigators, and the subjective nature of medical practice (Mol, 2002; Downey and Dumit 1997; Kaufert and O'Neil, 1993; Lindenbaum and Lock, 1993; Rosaldo, 1989; Geertz, 1973; Good, 1994; McGee & Warms 2003; Segal 2001).

The social literature also provides insight into the dynamics of knowledge production. Historically, knowledge production occurs in restricted-access locations, what Downey and Dumit call the fortress, or “Citadel”. In the Citadel, scientific and academic knowledge is created behind closed doors, and subsequently disseminated to those outside the Citadel. Despite the centrality of the Citadel in knowledge production, several investigators propose that the cultural significance of a discovery is only actualized outside the Citadel through the diffusion of knowledge to the public arena (Downey and Dumit, 1997). And, that this diffusion into the public arena has been the traditional place for patients, and the general public, to have any power over knowledge, for knowledge that is ignored gains no cultural significance, and remains without cultural power (Geller et al. 2002, Condit and Bates, 2005).

It would seem, that challenges to the dominance of medical experts in knowledge production, would not only create opportunities for patients to participate, but shift the exclusive hold medical professionals have as the sole creators of knowledge of disease. Like the view expressed in the social literature and media, I therefore embraced the notion that patients have become empowered, and that knowledge production is more evenly shared with medical professionals. This led to my research question about changes in the hegemonic relation between doctors and patients.

Unfortunately, in the case of rare or genetic diseases, there is widespread fear on the part of patients that ignoring the ‘truth’ language of biomedicine about their rare condition could literally be life-threatening. As one well-educated, and articulate, rare disease patient told me, “No matter how many textbooks I read, I will never have the necessary medical expertise to make truly informed decisions about my healthcare” (Chaite, Pers. Comm. 2005). This individual is not an Alpha, but the President of a different, rare disease organization. When I re-

contacted key Alpha-1 informants, they confirmed this opinion, despite the heightened awareness many of them have about Alpha-1.

Through conscious and unconscious reference to this fear, I believe science gained, and has kept, its authority to direct truth practices, and constitute power relations in illness experiences. The fieldwork I conducted in medical offices confirmed the ongoing dominance of medical authority in Alpha-1, as did the interviews with individual patients, and medical staff. It is no surprise that the physicians and nurses I interviewed would valorize the centrality of medical knowledge; but I did not anticipate how many of the Alphas, that I classified at the high end of empowerment, still believe that only their physician, and nurse, produce knowledge about Alpha-1. These patients may feel they contribute in meaningful ways to the community, and to the support of other Alphas, but they do not perceive how what they know, and what they experience, contributes to the body of knowledge about Alpha-1.

My conclusion is that the hegemonic relations have not shifted, at least in the Alpha-1 community, and medical experts remain firmly in control. Instead, empowerment for patients, family members, and caregivers is mastery of the language of the medical domain, and, ultimately, participation in a sub-ordinate role to their physician in their own disease management. However, empowerment is a process; patients evolve in their understanding, their sense of control, and their degree of self-management of their condition. It is not inconceivable, given the advances in patient empowerment to date, that the hegemonic relations in medical care may shift over time, and patients will share more in knowledge production. And, that this equalizing of relations may come sooner in rare, genetic disease groups, or through the efforts of particularly empowered disease communities, like the Alpha-1 community.

I also conclude that, Alpha-1, as a rare, genetic condition, may not be representative of the illness experience of the majority of patients, who do not have a rare, or genetic, disease, and who potentially may not feel as daunted about understanding their condition. Genetic conditions, as I saw in the laboratories, are complex, a difficult topic to understand even for many doctors, and involve technologies, and an understanding, of molecular biology that remain obscure for the majority of patients. If, after nine years, hundreds of articles, and numerous conversations with (very patient) basic scientists, I am unable to fully grasp some of the details of what goes wrong in Alpha-1, I can only imagine how intimidating achieving this knowledge must be for the average Alpha. Counteracting the challenge of understanding a genetic condition, however, is the wealth of information that the Alpha-1 community has developed. To understand which factor, difficulty of the topic vs. availability of educational materials, takes precedence is a question that I hope to answer by further research in a larger cohort of Alphas, and by conducting similar research in other rare disease communities, and in a chronic disease community, like COPD.

Future Research

This doctoral research has provided a definition, and quantification, of empowerment in one rare disease community. However, there are other questions I believe are important to answer, and that may benefit the Alpha-1, and other disease, communities. The first of these questions is, “What is the extent of empowerment among Alpha-1 patients?” Using data from the doctoral research, and postdoctoral funding from the Alpha-1 Foundation, I will be designing a survey based on the emic definitions of empowerment, to be administered to enrollees in the Alpha-1 Research Registry, a cohort of 3,289 Alphas. This second phase of my empowerment studies aims to create a validated survey instrument that will measure the extent of empowerment in the Alpha-1 community, but, that can also be used to test empowerment in

other disease communities. An empowerment survey among Alphas, compared to surveys among other types of patients, may answer additional questions such as, “Is the Alpha-1 community, and Alphas, more empowered than other rare, genetic disease patients?” Or, “Are Alphas more empowered than chronic lung disease patients, and if so, why?”

A third phase of the empowerment study is also planned to answer the question that may have the greatest clinical significance for patients, and their physicians, “Does the extent of empowerment correlate to QOL?” Through these additional studies, I hope to clarify not only the extent of empowerment in one rare genetic disease community, but elucidate key components of empowerment that are relevant to all types of patients. My ultimate goal is to draw attention to the possible correlation between empowerment and an improved quality of life.

As noted earlier, in Chapter 2, this correlation with QOL is of great interest to clinicians who treat patients, to patients, and their families, and to the organizations in the Alpha-1 community. Since, Alpha-1 is not currently curable, except through transplantation, (which is only a temporary solution), and there are no therapies that improve the lung and liver destruction once it has begun, QOL is an important goal for Alphas.

There is only one type of drug therapy available for Alpha-1, augmentation therapy. Three companies currently produce this drug which attempts to provide the missing protection that misformed Alpha-1 cannot provide, and other companies are developing new forms of the drug. However, there is no drug therapy for the liver patients, and other than respiratory therapy, exercise, vigilant preventive measures, and limited nutritional advice, there is not much to offer patients in terms of illness management.

The focus of the phase three study, therefore, will be to draw parallels between the extent of empowerment, and the possibility this leads to improvements in QOL. A demonstration of a significant correlation would be meaningful to any type of patient. For, despite the disease-specific components of empowerment that I observed in the Alpha-1 community, it would not matter which form the empowerment took; what would matter is that empowerment and QOL are related, and therefore becoming empowerment is a healthy choice for patients. These additional, planned phases of the empowerment study will also give me the opportunity to refine the methodological approaches, in particular, the design of the statistical analysis.

Reevaluating the Statistical Approach

The statistical analysis, provided in Chapter 6, confirmed many of the qualitative assessment, such as the very strong correlations between components of empowerment and the total empowerment score, for example, or between years since diagnosis and awareness. However, some of the statistical results suggest that the size of the cohort may have unduly skewed the results, and that the purposive recruitment, and sample used in the study, may not have adequately represented the full range of empowerment in the Alpha-1 community. The statistical analysis did confirm the utility of using a scale to model empowerment, although the exact order of components, and grouping into sub domains, differed slightly from the hypothesized arrangement. However, like Peregrine et al., I believe that, since a Guttman scale is intended to model an evolutionary process, testing it in a single time period is not satisfactory (Peregrine et al. 2004). A refigured Guttman scale will be tested using multi-factorial analysis in the upcoming empowerment study, and the results of that analysis used to elicit feedback from a larger cohort, to more precisely define the hierarchal order of components of empowerment in Alpha-1.

In addition, Likert-like scaled response choices will be used in survey questions, instead of yes-no responses, to ensure that the data elicited more accurately portrays distinct forms of empowerment, and to give participants more options for how to characterize their experience. Rensis Likert had developed the scale in 1932, to measure people's internal states, such as attitudes, emotions, and orientations (Likert, 1932). Realizing these states are multi-dimensional, and no single question, or dimension, could characterize a multi-dimensional, internal state, Likert developed a composite scale. The Likert-like scaling will also provide more precise data for a statistical analysis (Bernard, 2003).

Measuring Disability: Assessing Methodologies

An important conclusion of this empowerment study is the value of utilizing validated survey instruments, developed within the medical domain, as the templates for the survey topics, and measures, I use to characterize empowerment. Over many years, investigators within the field of medicine have refined the use of QOL surveys to quantify qualitative attitudes among patients, and many of the techniques I needed to translate qualitative information into quantitative data have likewise been used for years in the pulmonary community (Carone et al. 1999; Dowson et al. 1999; Cheng et al. 2000; Bloom, 2001; Gunzareth et al. 2001; Hajiro et al. 1998; Jones et al. 1991, 1992; Ketalaars et al. 1996; Knebel et al. 2000; Okubadejo et al. 1996; Osman et al. 1997; Renwick and Connolly, 1996; Seemungal et al. 1998, Tu, 2006; Wijksta and Jones, 1998; Zamberlan, 1992). In addition, researchers in medicine, and behavioral science, have identified precise ways to develop measures of disability, and to operationalize difficult to assess, and complex, social constructs (Rubinstein, 1995; Fries et al. 2006; Peregrine et al. 2004).

I also recognize the value of the narrative analytic techniques that I employed, and that were developed in the field of anthropology and linguistics (Abrahamsson, 2002; Bernard,

2002; Bernard and Ryan, 1998, 2005, Bernard et al. 1986; Boeije, 2002; Gottschalk, 1997; Jehn and Doucet, 1996; Ervin, 2005; Jehn and Doucet, 1996; MacQueen et al. 1998; Rubinstain, 1994; Ryan, 1999; Ryan and Bernard, 2000, 2003; Ryan and Weisner, 1998; Ryan et al. 2000; Ryan and Martinez 1996; Willms, 1992). Using these techniques allowed me to discern the topics of importance to members of the community among the long, sometimes rambling narratives. One of the techniques I chose, a coding system, made it easier for me to spot recurrent themes, and to manage my extensive interview and focus group session data. However, it was through my readings in research design (Gottschalk, 1987; Bernard, 2003), that I learned how to translate the qualitative factors I had identified into a numeric values, and to operationalize my theories into research questions, research activities, and, ultimately, answers.

My use of any one technique, however, does not indicate my adherence to a fixed approach for hypothesizing about, or analyzing, data. Instead, I am a firm proponent of incorporating all relevant techniques, theories, and templates, and creating my own mixed methods approach. Since, a study of empowerment encompasses both social and medical factors, economics and politics, measurement issues and quantification, and, importantly, how we speak about all of these areas, it seemed appropriate to choose techniques and methodologies from each of these fields. I could then test which ones would best compile, then handle, the dataset, and enable me to bridge the qualitative/quantitative divide.

My final assessment of methodologies relates to the purpose of this research. I have undertaken an examination of empowerment as an intervention. Although, I am conscious of the need to situate my work within the field of medical anthropology and linguistics, I am primarily concerned with promoting empowerment among disease communities. To achieve

this aim, my research results must be understandable within the medical and lay domains I hope to affect. It is for this reason, that I used both qualitative and quantitative analysis. My commitment to using both methods has already validated my work among the Alpha-1 research community, and led to additional funding to further my examination of empowerment. However, the qualitative results are what will make sense to the patients, and their families, and, by providing qualitative results, my study can be understood, and used, by individuals, and Alpha-1 organizations, to promote empowerment.

Overarching Themes in Empowerment

Historically, when uncontested knowledge flowed outward from the “Citadel”, it separated the producers from accountability for their products (Downey and Dumit, 1996). However, societal changes in recent decades began to challenge the inviolability of the “Citadel”, and, despite what I see as the maintenance of the physician-patient hegemony, there are a growing number of individuals in the public arena who insist on accountability for how knowledge is constructed about disease, and which types of knowledge will be the most useful for addressing specific disease states (J. Walsh, Pers. Comm. 2006). Recognizing the usefulness of knowledge in patient’s terms is a pragmatic approach that I believe has been instrumental in the empowerment I witnessed in the Alpha-1 community. But, putting knowledge to good use is not the only manifestation of empowerment. As I researched and wrote the ethnography of the Alpha-1 community, I recognized a variety of forms and enactments of empowerment. At times it seemed, and as the quote below expresses, that empowerment is as varied as the number of people in a disease community.

NO (female, 67 years old): “For each person, patient empowerment means different things as we are all different people with different backgrounds, strengths, and weaknesses and attitudes towards life in general. I believe that all of these carry over and affect how we deal with our disease. I have always been a very active and positive person.

So it was natural, after I got over the initial shock of my diagnosis, that I would seek out other Alphas and try to find a way to turn that diagnosis into something positive.

In this regard, empowerment is complex – it reflects individual personalities, motivations, and choices. However, there are common themes to empowerment, and, from what I observed in the Alpha-1 community, empowerment can be articulated as relatively simple, core components that can be extrapolated to other types of patients.

Core components of empowerment include, the value of networking with other patients and caregivers, the importance of increasing awareness about the disease, accessible resources, and the key role family, and community support, play in disease management. Other attributes that I identified in the Alpha-1 community as empowering, but, that would be applicable to other types of patients, are membership in patient support groups and participation in research. Analysis of interviews, and focus group sessions, confirms my view of Alpha-1 community members as active participants in research and thoroughly versed in the procedures associated with scientific studies. This research participation is empowering, as it includes increased awareness of, and familiarity with, the process of informed consent, the need to protect their privacy as individuals with a genetic diagnosis, the procedures for taking biological samples through sometimes painful or uncomfortable procedures, and the importance of the relationship between bench science (basic research) and their own clinical care. Other empowering activities noted in the Alpha-1 community, but applicable to other groups, are becoming an organizational leader, getting involved in advocacy initiatives, and having a seat at the table in policy making.

Underlying all of these specific components of empowerment is a basic theme of disease-management (Figure 7-1). If, it is empowering to become more aware of one's condition, or to effectively communicate with one's doctor, it is for the purpose of managing

one's disease, lessening negative impacts on daily activities and family dynamics, and improving quality of life. Likewise, community wide efforts, such as advocacy or organizational leadership, may grapple with issues larger than one individual's experience of illness, but are also meant to manage disease.

I have noted, in earlier chapters, the reluctance in the Alpha-1 community to use expressions that define their condition in terms of disability and disease. So, although my ultimate conclusion is that empowerment represents disease management on both the individual and structural levels, it remains important, for the sake of a positive identity in the community, to express disease management as empowerment, and to link empowerment to improvements in Quality of Life.

Lessons from the Alpha-1 Community

There are specific components of structural empowerment in the Alpha-1 community that should serve as a model for disease communities of all types. This includes the leadership of visionary, and committed, individuals, and the creation of a strong organizational infrastructure and resources. I observed that effective leadership in the Alpha-1 community included proven business skills, networking at the national and international level, good interpersonal and communication skills, an understanding of scope of the problem, strong personal motivation, and, importantly, a vision of a solution. Having leaders with a vision led to creation of medical advisory boards, the Registry (with its growing cohort of patients willing to participate in research), and resources that would serve both the patient and research networks. Effective leadership also helped make the Alpha-1 organizations grow to a size that began to have a positive impact on advocacy efforts. And, with a solid infrastructure, all members of the community gained a sense of identity, and purpose.

Having a solid and visible infrastructure increased the ability of Alpha-1 researchers, and organizations, to attract funding. An effective organizational structure also promoted creation of key research resources, like the Registry and Tissue Bank, and led to increased interaction among researchers in diverse fields of Alpha-1 research. But, it was the recognition that all stakeholders had to work together, that has been the most powerful force in the empowerment of Alphas. When contacted for a final remark on empowerment, Dr. Brantly was quick to note that networking is the core component of empowerment in the Alpha-1 community. Through involvement of Alphas, family members, caregivers, organizational staff, and Alpha-1 medical experts, the Alpha-1 community has established strategic alliances, a positive reputation among VHAs, and visibility of the needs of the Alpha-1 community to funding agencies, pharmaceutical companies, and governmental officials.

Recommendations

My final conclusions are that members of the Alpha-1 community are empowered to various degrees, using any recognized standard of empowerment. And, that empowerment is a process, not a static condition (Figure 7-2). Definitions of empowerment derived from this study, therefore, encompass an evolutionary element. For, even during the three years of this study, the level of empowerment of individuals changed, as did the level of empowerment of the Alpha-1 community. I see achieving empowerment not as a single event, or stage of awareness, but as an ongoing, and progressive, process. Other conclusions relate to the original research questions:

- **Research Question 1:** Patient empowerment in the Alpha-1 community does not signify a shift in the hegemonic relation between physicians and patients.
- **Research Question 2:** There is a qualitative difference between leaders in the community and the majority of Alphas: structural empowerment is embodied by organizational leaders and Alpha gatekeepers; personal empowerment is embodied, at the local level, by individuals and their families. However, the vision of finding a cure, held by

organizational leaders, is being incorporated, over time, into a lived reality throughout the Alpha-1 community.

- **Research Question 3:** The strength and effectiveness of organizational leadership, and community structure, adds to both the structural and personal empowerment of individuals in the community. In addition, access to resources improves the chances for patients to more quickly become empowered.
- **Research Question 4:** The Alpha-1 community exemplifies empowerment along a unidimensional scale.

In addition, I found that the involvement of all stakeholders, as partners in the search for a cure, elevates the patient's power, and the community's clout. Based on these conclusions, I recommend that the Alpha-1 organizations continue to provide their community resource templates to other disease communities, as these resources have proved particularly effective in the strengthening, and empowerment, of the Alpha-1 community. I, also, recommend that the results of this research study be made available, as lay reports, to patient networks within the Alpha-1 community, and in other disease communities. The core components of empowerment are relevant to more than just Alpha-1 community members, and may help other communities achieve the levels of empowerment I witnessed in the Alpha-1 community. In addition, the results of this study, when publicized, may serve to increase the positive identity of Alpha-1 community members, and validate the efforts they have already made that promote empowerment.

My final recommendation is that other disease communities undertake investigations on the relation between empowerment and QOL, and assist in defining what empowerment is, and how to promote it, within their communities. Through such studies, the distinctions between various community's needs, and forms of empowerment, can be elucidated, and, when necessary, appropriately disease-specific empowerment activities and resources can be developed.

Table 7-1. Thematic assessment of media reports on science and medicine

Topic	Source	Date	Title	Sub-title or [Topic]
Trust in science	Gainesville Sun	07/16/08	Study: Impending death often kept from patients	“People crave these conversations, (or).they feel abandoned and forlorn.”
	Gainesville Sun	2005	States make it easier for doctors to say “I’m sorry”	[Physician responsibility rather than liability in medical mistakes]
	Gainesville Sun	03/29/06	Scientists: NOAA hiding truth on global warming	[Tension between politics and science]
	Miami Herald	2007	Poll: Little faith in government crisis response	Americans are wary of likely disease outbreak.
	Miami Herald	2007	Is talking to your doctor a pain? Licensing board is listening	New MDs to be tested on communication skills
	Gainesville Sun	03/29/08	Fed. Survey finds mixed feelings on U.S. hospitals	The results reflect a growing uneasiness with medical care and comfort.
Patients Rights	Gainesville Sun	09/07	How does ethnicity affect your health care?	Area workshops will focus on teaching women skills to deal with health care providers
	www.salon.com	03/07/00	Who owns your DNA?	Genetic research that can save lives is often stymied by biotech cos. greedy patient claims
	Miami Herald	12/02/00	FDA raises red flag on Red Cross blood supply	The FDA has filed court papers alleging that the RC violates regulations on processing blood.
	Roll Call	06/06/02	She knows why it’s so hard to breathe.	Now explain why her lifeline is being squeezed.
	Gainesville Sun	2006	Medical illiteracy could be death of you	Almost 40% of those deemed medically illiterate died during the study.
	Gainesville Sun	2007	In Britain, patients turn to computers instead of therapists	The program is a quick, easy way to get basic therapy
	Miami Herald	2007	Microsoft launches health management site	Electronic medical records
	Gainesville Sun	2007	Options abound for Web sites on health information	The best way to sift through online health information is to use many sites.
	Gainesville Sun	02/11/08	Minnesota Web site creates online health marketplace	The Web site will allow Twin Cities users to look for medical services.

Table 7-1. Continued

Topic	Source	Date	Title	Sub-title or [Topic]	
Genetics	Miami Herald	01/13/01	Ox gestated in cow lives 2 days	Cloning	
	Miami Herald	02/12/01	Data of genome project a window to life's secrets	Mental illness, addiction cures among scientists' expectations	
	Newsweek	07/09/01	Cellular Divide	Harvested from stem embryos, stem cells may cure Alzheimer's, Parkinson's and a host of other diseases.	
	Miami Herald	2003	Humans have fewer genes than thought, new research shows	People have fewer than 25,000 genes, a new study of the genetic code found.	
	Gainesville Sun	2005	New breast cancer gene found	The damaged gene caused only a few cases.	
	Gainesville Sun	2007	Paternity tests sold in stores	DNA home testing	
	Gainesville Sun	03/31/08	Gene work unravels mysteries of diseases	A wide range of diseases may be subject to new treatments	
	Gainesville Sun	04/01/08	Your personal gene scan	People can find potential medical problems before they show up	
	Bioethics	Miami Herald	10/22/99	Study shows fear of genetic tests	Privacy concerns focus on job, health insurance discrimination
		Miami Herald	Undated	Insurers can use genetic testing	British decision draws criticism
Miami Herald		03/15/99	An idea whose time has come	Parents hesitate at DNA sample to identify kids	
Miami Herald		07/21/99	Organ Transplants	Livers should be given to the sickest	
Miami Herald		2000	'Hoarding laws' keep organs within state borders	I have a Boston liver	
Newsweek		07/09/01	Battle for Bush's Soul	The president is trapped between religion and science over stem cells. and votes – are at stake.	
Newsweek		07/09/01	A question of life or death	Untangling the knottiest of ethical dilemmas	
Gainesville Sun		11/11/07	Fears of fueling racism accompany DNA data differences	Geneticists fear that new findings could undermine principles of equal treatment	

Table 7-1. Continued

Topic	Source	Date	Title	Sub-title or [Topic]
Voluntary Health Organizations	Wall Street Journal	04/14/99	The Lung Association, its donations waning, casts about for a cause	Christmas Seals grow tired as “AsthmAttack” flops, spawning internal strife
	Science	04/16/99	Foundations and Science Policy	[Foundation support of research]
	Miami Herald	06/14/00	From courtroom to crusade	Bernard Siegel left a law career to champion scientists doing stem cell research
	Miami Herald	03/27/00	For health-care groups, issues include clinical trials, genetics	[Column discussing liability to foundation board members of targeted screening programs]
	Wall Street Journal	04/25/02	Hiring your own scientist to find a cure	Families of terminally ill set up research foundations; here’s where to get help

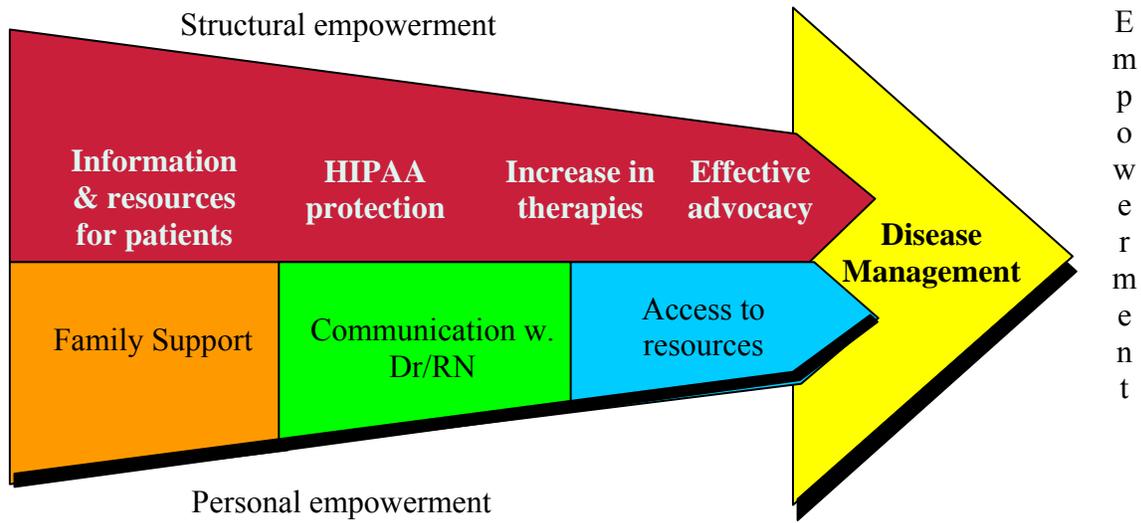


Figure 7-1. Empowerment is disease management

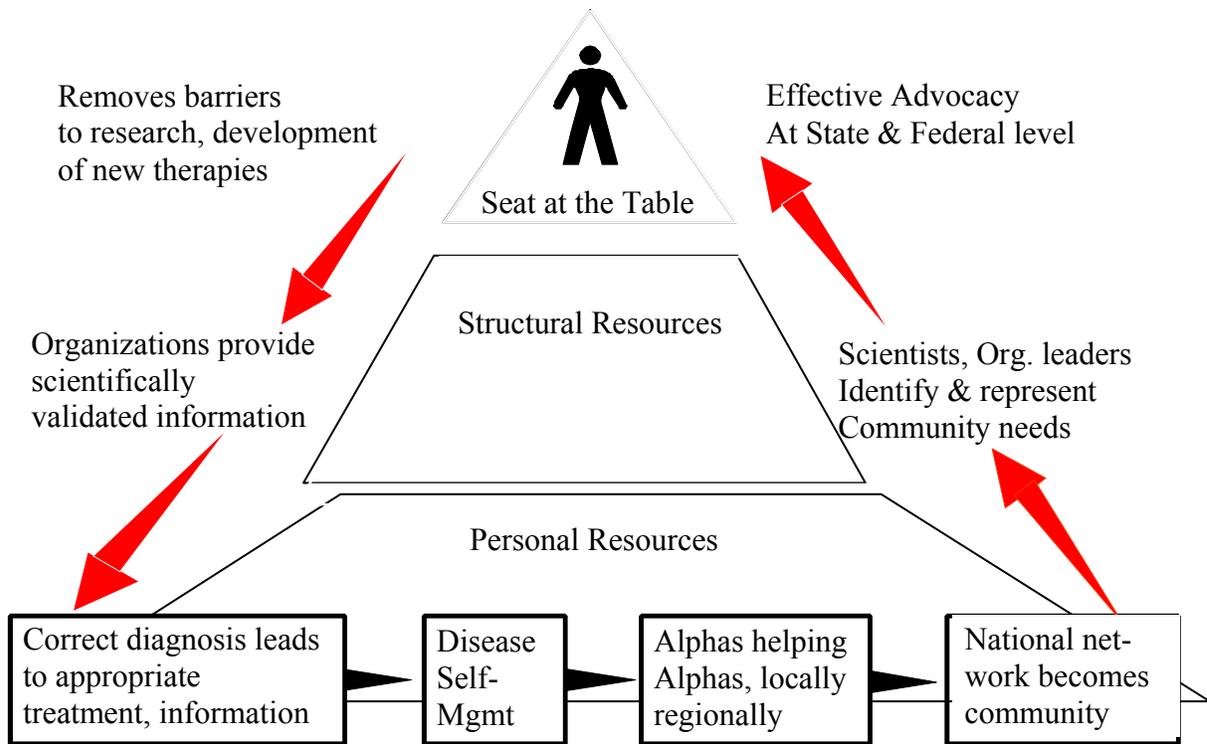


Figure 7-2. The process of empowerment

APPENDIX A
SAMPLE QUESTIONS 2006 INTERVIEWS AND FOCUS GROUP SESSIONS

Semi-Structured Interviews with Physicians and Researchers

The interviews are intended to explore physician and researcher attitudes about their role in the practice of medicine. Questions explore social context, psychological motivation, and perceived power between doctors and patients in terms of knowledge about a medical condition and control or cure of the condition. Questions also explore a physician/researchers self-assessment of the following: communication with patients, empathy with patients, expertise with the condition, and commitment to medical practice. Input is sought for defining components of patient.

It is anticipated that not all questions will be covered in any one interview, but that participants will focus on topics that are more relevant to them.

I. Social background

- a. Why did you choose a scientific career? [*Were parents, older relatives, siblings, spouse and/or children a hindrance or encouragement for the choice of a scientific career? Was your religious background or social background a hindrance or advantage for your choice of career?*]
- b. How do you identify yourself in a social context? By your birthplace, ethnic group, religion, gender, country of origin, occupation, race, age group, ideological stance (*participants are NOT being asked what their ethnicity, race or country of origin, just how they self-identify*)
- c. Does a scientific career complement, oppose or is irrelevant to your social identity?
- d. Do members of your family (*parents, older relatives, spouse, children, siblings*) support your commitment to career? The particular demands of your career as a scientist?

II. Identity as a Scientist

- a. At what age did you decide to be a scientist, study science or practice medicine? Are these the same thing or distinct goals?
- b. What factors are you aware of that may have motivated your choice (family member, mentor/teacher, religious leader, peer, event (illness or death in family), social altruism?)
- c. Is it what you expected when you first choose it?
- d. Does it suit your personality?
- e. Does it fulfill your intellectual goals, spiritual or economic goals? (or) is it a meaningful occupation

III. Identity as a Specific Type of Scientist

- a. When did you choose your specific sub-specialty?
- b. What factors may have motivated your choice? (*Mentor, supervisor, peer, intellectual curiosity, expertise in an area, economic benefit*)
- c. How long have you practiced your specialty?
- d. Do you consider yourself expert in your area?

IV. Relation to Patients

- a. Did you choose to be a clinician in order to interact with patients?
 - a2. Did you choose to focus on research to interact less with patients?

- a3. As an MD, PhD, do you have a preference for time at the bench or time with patients?
- b. What do you enjoy the most about interacting with patients? What is the worst about dealing with patients?
- c. Did medical training include specific written or verbal instruction in how to relate, interact or cope with patients or were you expected to pick it this information in residency? Did your med training include readings in psychology, linguistics, bioethics or sociology? Did you study epidemiology or public health as a component of med training? If not, do you think these types of courses are useful? If yes, did you find these courses useful?
- d. Do you know many patients? Is your relationship formal, casual, both?
- e. Of the patients you know, what is the average length of time of your acquaintance, treatment?
- f. What proportion of your patients is diagnosed with a genetic condition?
- g. What proportion of your patients is diagnosed with a fatal or chronic condition?
- h. Do you experience a difference in attitudes associated with your diagnosis, treatment or care for a genetic as opposed to chronic disease patient?
- i. Has the relation between patients and physicians changed in your lifetime? In the course of your practice? Should it change?
- j. Are the patients you know capable of giving informed consent? Do you think they understand what will occur on studies and in trials? Should they understand more?
- k. Do you think it appropriate for parents to make informed consent decisions for teenagers?
- l. Are you good at conveying technical information in lay terms? Where do you stand on how much a patient should know about their condition? Their family?

V. Relation to Patient Organizations

- a. Do you attend patient support group meetings or arrange them at your facility?
- b. Do you attend or belong to national patient organizations?
- c. Do you serve in an advisory capacity or as an officer of a patient-centered organization?
- d. *do you donate to such organizations?(Optional question)*
- e. Does your involvement with a genetic disease patient community or individual patients differ from your experience of interaction with chronic disease patients
- f. Do you believe in the efficacy of patient-driven organizations to fund research? How could they be more effective?
- g. How much should such organizations determine the specific direction research will take? Provide funding only?

VI. Relation to Modes of Patient Empowerment

- a. How would you define patient empowerment? Is it the same as patient activism? (what else might it include besides advocacy, pharMO-economic control, disease management, information and awareness, role in research development and funding)
- b. Are the patients you know empowered?
- c. Do you participate in any patient advocacy activities?
- d. Do you involve patients in decision making regarding treatment, research activities or communication about medical issues?
- e. Do you know patients who you consider well informed about their own condition?
- f. Do you know patients who know more than health care providers about their own medical condition?

- g. Are you comfortable when patients ask for a specific drug, treatment or diagnostic test?
- h. Are you comfortable with patients looking up information on websites or obtaining information from other patients? Do you think such information is accurate?
- i. Are you comfortable with how medical and scientific information is disseminated in lay language?
- j. Do you believe that patients are empowered when they know more about their condition and how to treat it? When they lobby for their rights?
- l. Are patients empowered by a seat at the table in policy making, legislation effecting health care or decision making related to research initiatives?
- m. Are patients empowered by controlling drug pricing, Medicare carve-outs, fast-tracking drug development or the sources of drug distribution?
- n.. Do patients have a role along with physicians and researchers in constructing medical knowledge? Should they have any role?

Focus Group Sessions with Alphas

I. In addition to social identities (where we were born, our religion, ethnic group, race, age group, occupation, ideology/philosophy) some people, through diagnosis, become identified with their medical condition. For those with Alpha-1 Antitrypsin Deficiency this means a set of nested, or interlocking identities, like Russian dolls, one within another. Identity becomes medical categories:

- as an Alpha
- as a Member of a Rare Genetic Disease Community
- as a lung (or liver) patient
- as a family member of an Alpha
- as a member of a chronic disease community
- as a disabled person
- as a patient activist, as a patient advocate

How Would You Define Patient Empowerment? Is it the same as patient activism? (what else might it include besides advocacy, pharmo-economic control, disease management, information and awareness, role in research development and funding). (Free listing exercise)

II. Modes of Patient Empowerment (questions to stimulate discussion)

- are you and other patients you know empowered?
- do you participate in any patient advocacy activities?
- does your physician involve you in decision making regarding treatment, research activities or communication about medical issues?
- are you, or do you know patients who you consider well informed about their own condition?
- do you or other patients you know better informed about aspects of their own medical condition than their doctors?
- should patients ask for a specific drug, treatment or diagnostic test?
- do you look up information on websites or obtaining information from other patients? Do you think such information is accurate?

- are you comfortable with how medical and scientific information is disseminated in lay language?
- do you believe that patients are empowered when they know more about their condition and how to treat it? When they lobby for their rights?
- are patients empowered by a seat at the table in policy making, legislation effecting health care or decision making related to research initiatives?
- are patients empowered by controlling drug pricing, Medicare carve-outs, fast-tracking drug development or the sources of drug distribution?
- do patients have a role along with physicians and researchers in constructing medical knowledge? Should they have any role?

III. Relation to Physician

- what is your relation to your physician? Do you trust them, rely on them, can you relate, do you understand them, do they treat you with respect, are you afraid of them?
- does your physician enjoy interaction with patients?
- Does he or she do it well?
- do you think your physician's medical training included specific written or verbal instruction in how to relate, interact or cope with patients or were you expected to pick it this information in residency? Should med training include readings in psychology, linguistics, bioethics or sociology, epidemiology and public health as a component of med training?
- do you deal with one physician or several? Is your relationship formal, casual, both?
- of the doctors you know, what is the average length of time of your acquaintance, treatment?
- has the relation between patients and physicians changed in your lifetime? In the course of your treatment? Should it change?
- are you capable of giving informed consent? Do you think you understand what will occur on studies and in trials? Do you want to understand more?
- do you think it appropriate for parents to make informed consent decisions for teenagers?
- are your caregivers good at conveying technical information in lay terms? Where do you stand on how much a patient should know about their condition? Their family?

V. Relation to Patient Organizations

- do you attend patient support group meetings?
- do you attend or belong to national patient organizations?
- do you serve in an advisory capacity or as an officer of a patient-centered organization?
- *do you donate to such organizations?(Optional question)*
- does your involvement with a genetic disease patient community help you deal with your condition? Does it give you a greater sense of identity, community or optimism? Has it harmed you to be involved?
- do you believe in the efficacy of patient-driven organizations to fund research? How could they be more effective?
- how much should such organizations determine the specific direction research will take? Provide funding only?

APPENDIX B
SAMPLE QUESTIONS 2007 INTERVIEWS

Physician and Nurse Interviews

Questions about Differences COPD And Alpha-1 Patients

- 1) I observed that both Alpha-1 patients were more knowledgeable about their condition and asked many more questions than the COPD patient who was more passive in her interaction with you during the office visit. Is that typical?
- 2) If that dynamic is typical, what percentage of Alphas and COPD patients exhibit those traits?
- 3) In this particular case, or in general, do you think socioeconomic or educational level is a factor in engaged vs. passive patients?
- 4) Do you perceive differences in coping and/or willingness to self-manage their condition between male and female patients?
- 5) Do you perceive gender differences in who seeks transplantation? Is the difference in when this treatment option is requested (i.e., early vs. late in course of disease).
- 6) How common is it for a male caregiver to accompany a female Alpha to an office visit? female caregiver to accompany a male Alpha? Is this typical of COPD patients as well?
- 7) Is the variable the distance to clinic, economics or stage of disease?
- 8) How often do COPD patients ask about clinical trials, investigational drugs or how they might participate in research?

Questions About Procedures

- 8) What is the purpose of the physician assistant or nurse to first interview the patient?
- 9) What do you think are the historical roots of this practice?
- 10) Does it serve your purpose as a physician or is it institutionally mandated?
- 11) Is the physician or PA in charge of the office visit? Who is responsible for the notes in the medical record? Can and does the physician alter the PA or nurses notes after the office visit?
- 12) How often do medical students or researchers participate or observe in office visits?
- 13) Do you think it alters the way you talk to or treat a patient?
- 14) How often do patients refuse to allow observers or students into the office visit? Do you think some patients display discomfort but do not say no?

Level of Understanding

- 15) Do you think patients understand their rights in informed consent? Is the motivation to please the physician, demonstrate trust of the physician, or altruism?
- 16) How much of the science of Alpha-1 or COPD do you think patients need to know? Do you always provide information or only when a patient wants to know about the underlying biology?
- 17) At what stage do you use medical terminology to describe a patient's condition?
- 18) Does this depend on the patient's education, time since diagnosis, indication of interest or is it a generally structured and stepwise discourse?
- 19) How often do patients ask you to define a specific medical term, i.e., eosinophils?
- 20) Do questions about medical terminology or specifics of the biology of their condition generally come early in the course of treatment, later, or is it ongoing?
- 21) How often do you have to repeat information during an office visit and translate medical terms into lay language or analogies, metaphors?
- 22) Do you think it is the PA or nurse's role to explain things in lay terms? Do you think patients rely on support staff for a lay understanding of Alpha-1 or COPD?
- 23) Is the nature of managed care constraining the amount of time you can spend with a patient? Would you want to spend more time with some patients? Do you have that latitude?

Sociolinguistic Questions

- 24) Are you aware of code-switching during an office visit in the way you address patients vs. the way you address the physician assistant, nurse or medical student? Does the PA or nurse code switch? Does the patient?
- 25) Do you think patients are more empowered when they learn some of the medical terminology and can discuss and describe their symptoms in medical terms?
- 26) Do you think relating of symptoms is a list or a narrative?
- 27) When you ask questions, based on the interaction between the PA or nurse and patient, are you interested in eliciting narratives or are they close ended questions?
- 28) What do you do when a patient wants to tell a long story related to their condition?
- 29) Do you think length of practice enables you, as a physician, to understand the context for remarks from a few verbal or physical cues?

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BIOGRAPHICAL SKETCH

Symma Finn's background includes studies in communications, environmental studies, and anthropology. She has an undergraduate degree from Adelphi University (communications), a master's degree from the University of Florida, Rosenstiel School of Marine and Atmospheric Sciences (anthropology of coastal communities), and has studied medical anthropology at the University of Florida since 2005.

Ms. Finn has worked with environmental, marine, and medical scientists since 1984, and has extensive experience in the scientific and medical domains. Her master's thesis dealt with the human component of Everglades' restoration, and she remains involved in research of the intersection between human health and the environment. Her current research focuses on the social issues relating to genetics, and empowerment of patients. Her doctoral research quantifies empowerment in the Alpha-1 Antitrypsin Deficiency community, a network of individuals diagnosed with a rare, genetic condition, and related stakeholders.