Informatics Architecture for Personalized Medicine at the University of Florida

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Overview

Previous research in genetic determinants of response to clopidogrel has shown that patients may benefit from genetic testing before clopidogrel is prescribed[1]. At UF&Shands, we are implementing testing and an information pathway for both clinical care and research[2].

Description

An information pathway for personalized medicine has been developed that begins with physician/patient discussion and consent, proceeds through sample collection and processing, genotyping 256 single nucleotide polymorphisms (SNPs), data storage and transmission, and ends with treatment guidance for the physician. The pathway is implemented through iterations as an information system.

A second portion of this pathway enables research by storing the genetic data from consented patients in the UF&Shands Integrated Data Repository (IDR).

Currently, only clopidogrel metabolism responses are reported in the electronic medical record using CYP2C19 genotype. However, the system is designed with future expandability so future additions can be easily accommodated. The schematics of this process is depicted in Figure 1.

System Components

1. Epic EMR: The CYP2C19 genotyping test is ordered as a regular lab test initiated from the UF&Shands Electronic Medical Record (EMR) system (Epic).
2. This lab order is then transmitted to the Lab Information System (LIS) for sample collection and testing.
3. Microarray analysis is then performed.
4. Data output from the microarray is stored in an intermediate clinical database.
5. A locally developed software application generates the star alleles for CYP2C19, lab test code, and test results. Test results are entered into the LIS, where descriptive text is generated and transmitted to the EMR.
6. A mobile device is used at the point of care to seek consent from patients to allow their genetic data to be used in future research. This paperless system uses the Research Permissions Management System (RPMS)[3].
7. Patient consent status is queried through a web service. Genetic data from all patients consented for participation in research are then transmitted to the IDR for storage and linkage to all other clinical data.

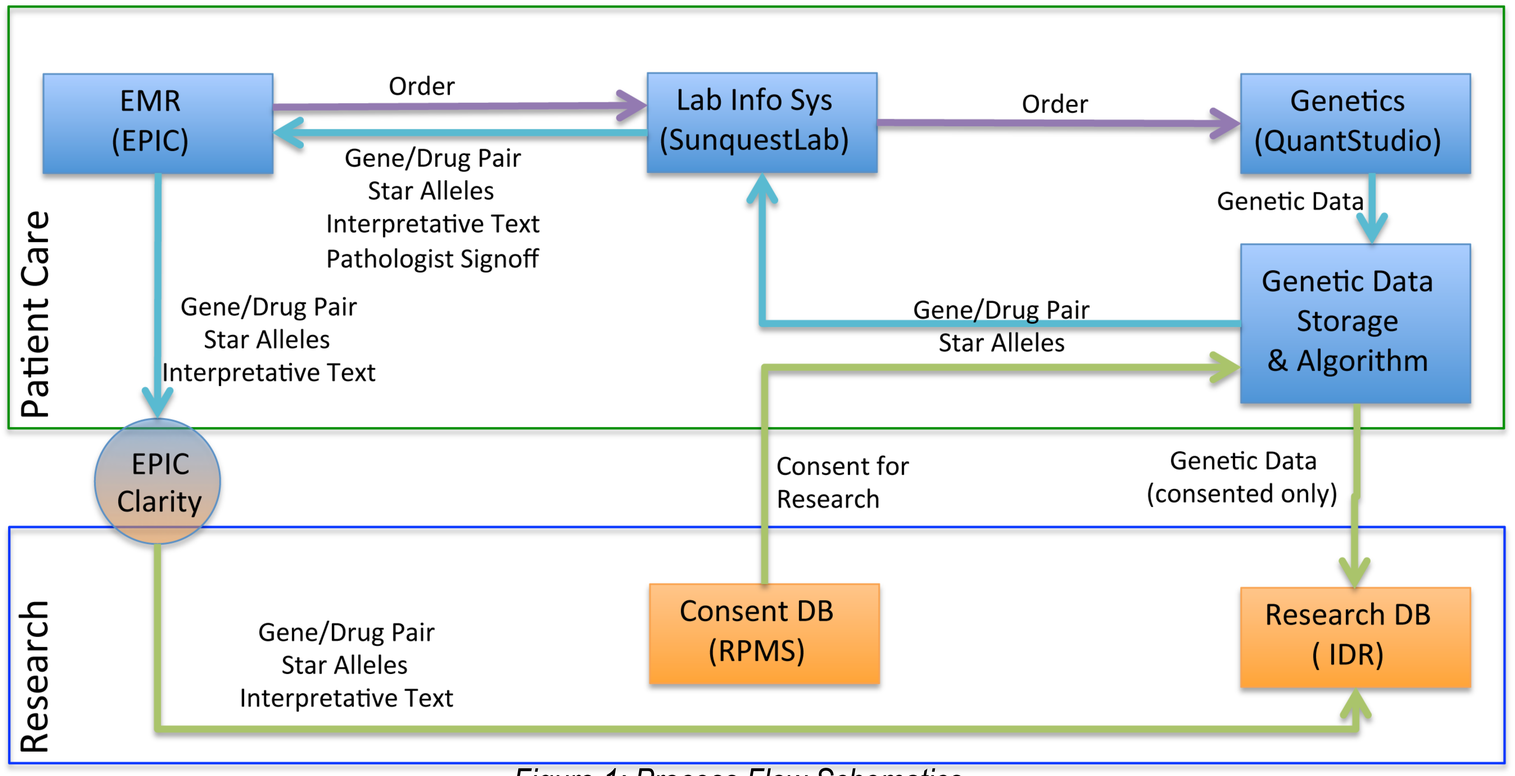

Figure 1: Process Flow Schematics

Special Considerations

1. When a patient declines to participate in future research studies, all his/her genetic data except the data used in clinical decision making are permanently deleted.
2. If a patient's consent status is unknown it is treated as declined.
3. No automated interface between the application and the LIS has been developed. This will be considered in the future.

Data Model

A simple data model has been developed for the intermediate database (Figure 2). New tests can easily be added into this schema with minor or no modification to the software application. Version numbers in various tables facilitate future changes to existing tests.

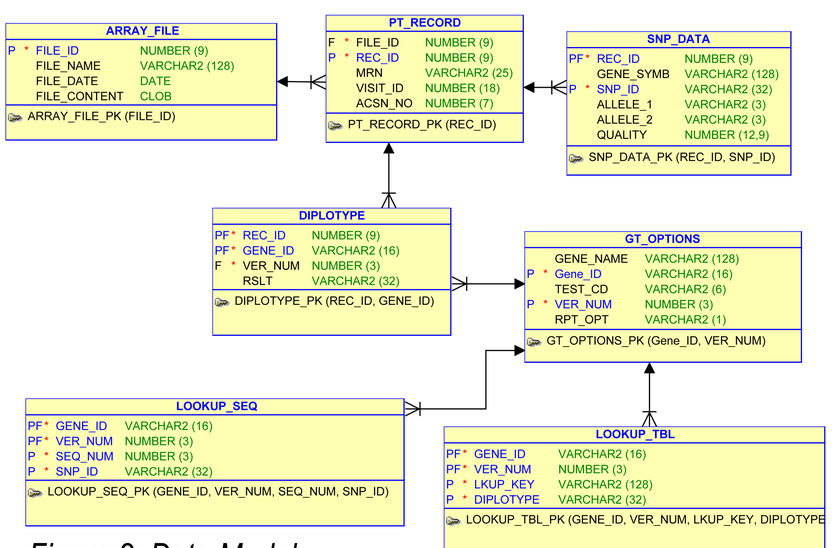

Figure 2: Data Model

Results

The pathway will initially support genetic-guided pharmacotherapy for an estimated 600 patients per year who undergo a percutaneous coronary intervention during their cardiac catheterization at UF. 900 additional cardiac catheterization w/o PCI will also have genetic information generated to be used if the relevant pharmacotherapy is prescribed in the future. Alerts in the EMR will guide physician's treatment decisions for clopidogrel vs alternative therapy based on CYP2C19 genotype. UF will store additional SNP data in its IDR for research purposes. SNP data will also be held in a pathology database in anticipation of future clinical use. The EMR contains actionable clinical findings only.

Discussion

This approach will support the first systematic use of genetic information in the treatment of patients in the University of Florida Health System, guiding choice of medication for patients, and will serve as a model for additional UF personalized medicine applications, clinical care, and research partners.

References

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