Integrating the Genome into the Electronic Health Record: The Next Leap Forward

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Clinical and Research Questions

* Research
  - Why do some patients with asthma respond to steroid treatment while others do not?
  - Why does a mutation in Huntington’s gene cause a lethal defect?
  - Why do some patients with diabetes have few complications even with “poor” control whereas others with good control have severe complications?

* Clinical
  - Can I lower my cholesterol by diet alone of should I start on an anti-cholesterol drug now?
  - Will a third line anti-cancer drug be more effective as a first line drug with a patient with lung cancer?
A Vision for Personalized Medicine

- EMR with clinical decision support
- Genomic research with high capacity IT
- Integrated genomic and phenotypic data repository
- Facilitated translational research leading to
  - Diagnostic discovery
  - Drug development
- Improved individualized medicine &
  pre / post symptomatic disease management

Partners Personalized Medicine Infrastructure

Research
- I2B2 (HIVE / RelNet / Pharmacologic Surveillance)
- Computational Cluster and Storage Resources
- Enterprise Tissue Banking Infrastructure

Clinical
- Clinical Decision Support Rules Engine
- Electronic Health Record (EHR)
- Genomic Variant Interpretation Engine (GVIE)

Consent Tracking System (CTS)
Genomic Knowledge Base (GenelInsight)
Enterprise LIMS Superstructure (GIGPAD / PowerPath)
Research Patient Data Registry (RPDR)
**RPDR Allows Exploration and Delivery of Clinical Data**

1) Queries for aggregate patient numbers
- Warehouse of in & outpatient clinical data
- 4.1 million Partners Healthcare patients
- 1 billion diagnoses, medications, procedures, laboratories, & physical findings coupled to demographic & visit data
- Authorized use by faculty status
- Clinicians can construct complex queries
- Queries cannot identify individuals, internally can produce identifiers for

2) Returns identified patient data
- Start with list of specific patients, usually from (1)
- Authorized use by IRB Protocol
- Returns contact and PCP information, demographics, providers, visits, diagnoses, medications, procedures, laboratories, microbiology, reports (discharge, LMR, operative, radiology, pathology, cardiology, pulmonary, endoscopy), and images into a Microsoft Access database and text files.

**Structuring Unstructured Image Data**

- **Classifying Interpreted Findings**
  - LEXIMER (Lexicon Medical Entropy Reduction)
  - Codification - CPT, ICD-9, RADLEX, SNOMED

- **Image Feature Extraction**
  - Modern CAD Applications
    - CT Chest
    - Lung Nodule Detection
    - Digital Mammography
      - Cancer Detection
  - Future Applications
    - Brain CT, MRI
      - Sagittal asymmetry index, Tumor detection
    - Cardiac MRI, US, NM
      - Myocardial wall thickness quantification
    - Future Imaging Modalities
      - Molecular, MRI, OCT, 3D Imaging
Enterprise LIMS Superstructure
Gateway for Integrated Genomics Proteomics Applications and Data (GIGPAD)

Research/Clinical GIGPAD

- Genotyping
- BSF
- MicroArray
- Sequencing
- Proteomics
- BWH GCRC

Interfaces to Clinical Researchers and Care Providers

GeneInsight - DNA Variant Knowledgebase

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Tissue Sample Collection

Tissue Banking
Advancing Cancer Care

Informatics for Integrating Biology and the Bedside (I2B2)
i2b2 Hive

Predictive Modeling

Predicting future hospitalizations from prior use of medications in patients that have Asthma:

Predicts Hospitalization

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Clinical Trials Performed In-Silico

Clinical trial performed exclusively in computer memory finds APOE epsilon 4 allele determines risk of seizures after trauma

Key Personalized Medicine EHR Functions

- Proper Display of Genetic/Genomic Information
- Maintenance of Patient Genetic Profile
- Real Time Clinical Decision Support
- Population Based Clinical Decision Support
- Knowledge Event Propagation

- Patient genetic profile summary view
- Disease based clinical views of genotypic and phenotypic data (e.g. Breast Cancer vs. Diabetes)
  - Leverage complete genetic profile in CDSS
  - Translate genetic profile into clinical risk, diagnosis, and pharmacogenomic guidance.
- Drug – Genetic Intervention alert, when medication is contraindicated or another is more efficacious
- Genetic aware drug dosage calculators
- Identification of ‘at risk’ patient populations for prophylaxis and screening programs
- Monitoring patient profile for high-risk situations
- Maintaining clinical validity and utility of patient genetic profile over time with changing knowledge, and alert clinician to changes in recommendations
Electronic Health Record

Genetics Clinical Decision Support
Summary

- Personalized medicine will become an integral part of care delivery
- Partners has begun to implement the information technology foundation needed to support:
  - Research into the genomic bases of disease
  - Care delivery based on knowledge of a patient’s genome