The Genomics of Big Data

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Session Objectives

• Describe the latest incentives/drivers to diagnosis and treatment using genetic and genomic data

• Identify the known Genetic and Genomic influences across the continuum of care and the implications for information technology in your EHR

• Define the most recent advances in Pharmacogenetics and Pharmacogenomics and the implications for information technology in your EHR

• Identify roadmaps and resources to integrate genetics and genomics into IT and your EHR and the implications for nursing

• Acknowledge Dr. Kathleen Calzone-Co-author Essentials of Nursing Informatics, V6, 2015. Big Data Initiatives: Genomics and Information Technology for Personalized Health
Latest Incentives
The Incentives

• $1000 Genome is here
• Regulatory reimbursement is here- lawsuits are beginning for coverage
• Direct to Consumer tests are being utilized by consumers
• Companies are now available to conduct Genetic Testing
• Monetary Incentives are HERE
• Lawsuits are beginning in pharmacogenetics/pharmacogenomics
• ANA Professional Standards in Informatics includes Genetics/Genomics
• The new Precision Medicine initiative
• Nursing has identified the role of genetics/genomics through the continuum of care
• IOM states there is sufficient evidence - 2/26/2015
$1000 Genome Test is HERE

• January 14, 2014 - Today, Illumina, the leading maker of DNA sequencers, announced a milestone in biotechnology: it is introducing a new machine that can sequence the genetic code of a human cell for $1,000.
Regulatory Reimbursement
MONETARY Incentives are HERE

• February 2014 - The McKesson Diagnostics Exchange provides an infrastructure that will support the AMA CPT efforts to advance personalized medicine, promote access to innovative diagnostic capabilities and improve patient outcomes in ICD-Z codes for genetic testing
Direct to Consumer (DTCV) Marketing and Testing

• Tests available direct to the consumer without an ordering healthcare provider
  • Varied test types
    • High penetrance diseases
    • Polygenic diseases
    • Risk Assessment
    • Enhancement tests
      • Pharmacogenomics
      • Nutrigenomics
• Most require only a saliva sample
• Costs vary based on test but can be as low as $99

http://www.cancer.gov/cancertopics/pdq/genetics/risk-assessment-and-counseling/HealthProfessional/page5#Section_362
Companies Are Now Available to Conduct Genetic Testing

- GenetWorx - Virginia tests in 50 states
- Greater Houston Healthconnect, a 20-county exchange in Texas, announced in January that it would be one of the first HIEs in the country to sign-on a pharmacogenomics lab, Companion Dx, for dissemination of genetic tests
Lawsuits Have Begun

- Lawsuit in Hawaii against Plavix Sponsors Alleges Burden is on Pharma to Market PGx Information - March 19, 2014
  - Plavix has "diminished or no effect" on people of East Asian or Pacific Islander descent because they metabolize it poorly
  - Suits also filed against Plavix Sponsors in Louisiana, Mississippi, West Virginia and California

- California Clinical Laboratory Association sues HHS over local coverage determinations-May 2, 2014
  - The complaint goes on to discuss (starting on page 11) LCDs developed by two influential MACs, Noridian and Palmetto GBA, which together control 20 states, for molecular genetic testing, and how these LCDs restricted access genetic testing for Medicare beneficiaries
Emerging areas

- All diseases and conditions have a genetic or genomic component
- Basic Genetic and Genomic competencies have been established for all nurses regardless of their academic preparation or specialty
- The informatics nurses must be able to:
  - Incorporate genetic and genomic technologies and informatics into practice
  - Demonstrate in practice the importance of tailoring genetic and genomic information and services to clients based on their culture, religion, knowledge level, literacy, and preferred language
New Money in Precision Medicine

• $215 Million budget proposed between NIH, ONC, FDA in the 2016 President’s budget

• 1 Million or more Americans proposed in cohort

President’s State of the Union Address - whitehouse.gov/sotu 28:58-29:56


www.nih.gov/precisionmedicine
Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees

• Define essential genetic and genomic competencies for ALL graduate nurses regardless of level of academic preparation, practice setting or specialty

• Established by a process of consensus
Essentials of Genetic and Genomic Nursing

- Define essential genetic and genomic competencies for **ALL** nurses regardless of level of academic preparation, practice setting or specialty
- Endorsed by 50 nursing organizations
- October 22-24 2006 Strategic Implementation Meeting
- 2nd Edition incorporated Outcome Indicators
  - Specific Areas of Knowledge
  - Clinical Performance Indicators
- 3rd Edition will be published in 2014 which includes some updates

http://www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf
New Perspective
Implementation of Pharmacogenomics: Evidence Needs

The evidentiary needs for genomic technologies are a significant barrier to the translation of genomic testing into clinical use. However, in many instances there is sufficient evidence to justify the use of genetic testing to information choice or dosage of medications. This discussion paper, the last of seven individually authored commentaries that explores the evidence needed to support the use of genome sequencing in the clinic, examines the policy issues and evidence needs for implementing pharmacogenomics testing.
Three Resources to Follow

• Genetic and Genomic Competencies for all nurses

• http://www.genome.gov/Pages/Careers/HealthProfessionalEducation/geneticscompetency.pdf

• ANA Learning the Basics of Genomics -
  http://ananursece.healthstream.com

• Journal of Nursing Scholarship - First Quarter 2013 -
  Updates to follow = http://www.genome.gov/27552312
Genetic and Genomic Influences Across the Continuum of Care
Genetic and Genomic Influences Across the Healthcare Continuum

- Preconception/Prenatal
- Newborn Screening
- Risk Identification
- Disease Characterization
- Screening/Diagnosis
- Individualized Therapy
- Management At End of Life
- After End Of Life

Preconception Prenatal Genetics

- Preconception
  - Testing for carrier status prior to pregnancy, often for autosomal recessive disorders
    - i.e. cystic fibrosis
- Prenatal testing
  - Performed during pregnancy
  - Indications include
    - Advanced maternal age, increases the risk for chromosomal abnormalities i.e. Down Syndrome
    - Family history of an inherited condition i.e. Duchenne muscular dystrophy
    - Ancestry/ethnic background of parents associated with a higher chance of an inherited disorder

Do YOU know Tracy and David?

As soon-to-be new parents, Tracy and David have a lot of questions. Do they have the right books? The right gadgets? The right name? But thanks to their primary care provider, they don’t have questions about their baby’s health.

When Tracy and David decided to try to conceive, Tracy visited a new health care provider who took a thorough family history at the first preconception visit. That history revealed that both she and David were of French-Canadian ancestry, putting them at an elevated risk of having a baby with Tay-Sachs disease, a lethal inherited disorder affecting the nervous system.

The provider explained the risk to Tracy and David, who chose to undergo genetic counseling and carrier testing. Having learned that they were both carriers of gene alterations that could cause Tay-Sachs, Tracy and David chose to have prenatal genetic testing to determine if their baby would be affected. What a joy to find out that the baby had not inherited Tay-Sachs!

The next time you meet "Tracy and David," take the time to consider and discuss the possible implications of their family history.

To learn more about how genetics is relevant to your practice visit www.genome.gov
Newborn Screening

- Newborn screening consists of a public health approach to the identification and management of health conditions identifiable in the newborn
  - Approximately 4 million newborns screened annually
  - About 12,500 new diagnoses as a result of testing
  - Newborn screening constitutes the most extensive use of genetics for public health benefit
  - All states provide newborn screening

- US Secretary of Health and Human Services Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC) provides national guidance about which health conditions should be included
Screening

• Genetic information is being used to personalize health screening recommendations

• SNP test results are being studied as a means to increase the specificity of risk calculation models (i.e. Gail model for breast cancer risk)

• Screening tests that include DNA analysis are being developed such as the DNA stool test, a less invasive means to screen for colon polyps or cancer
Risk Assessment

• More than 55 hereditary cancer syndromes have been identified

• The most common cancer syndromes are those associated with breast, ovarian, and gastrointestinal cancers
  – Tumor features at diagnosis are now being used as an indication for genetic assessment

• Risk assessment also performed in other healthcare arenas such as cardiovascular diseases

• Germline susceptibility gene testing is available
  – Relevant to individuals whose disease management may be altered
  – At-risk family members
<table>
<thead>
<tr>
<th>Gene</th>
<th>Genetic Alteration</th>
<th>Tumor Type</th>
<th>Therapeutic Agent</th>
</tr>
</thead>
<tbody>
<tr>
<td>EGFR</td>
<td>Mutation, amplification</td>
<td>Lung cancer, glioblastoma</td>
<td>Gefitinib, erlotinib</td>
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<tr>
<td>ERBB2</td>
<td>Amplification</td>
<td>Breast cancer</td>
<td>Lapatinib</td>
</tr>
<tr>
<td>FGFR1</td>
<td>Translocation</td>
<td>Chronic myeloid leukemia</td>
<td>PKC412, BIBF-1120</td>
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<tr>
<td>FGFR2</td>
<td>Amplification, mutation</td>
<td>Gastric, breast, endometrial cancer</td>
<td>PKC412, BIBF-1120</td>
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<tr>
<td>FGFR3</td>
<td>Translocation, mutation</td>
<td>Multiple myeloma</td>
<td>PKC412, BIBF-1120</td>
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<tr>
<td>PDGFRα</td>
<td>Mutation</td>
<td>Glioblastoma, gastrointestinal stromal tumor</td>
<td>Sunitinib, sorafenib, imatinib</td>
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<tr>
<td>PDGFRB</td>
<td>Translocation</td>
<td>Chronic myelomonocytic leukemia</td>
<td>Sunitinib, sorafenib, imatinib</td>
</tr>
<tr>
<td>ALK</td>
<td>Mutation or amplification</td>
<td>Lung cancer, neuroblastoma, anaplastic large-cell lymphoma</td>
<td>Crizotinib</td>
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<td>c-MET</td>
<td>Amplification</td>
<td>Gefitinib-resistant non–small-cell lung cancer, gastric cancer</td>
<td>Crizotinib, XL184, SU11274</td>
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<td>IGF1R</td>
<td>Activation by insulin-like growth factor II ligand</td>
<td>Colorectal, pancreatic cancer</td>
<td>CP-751,871, AMG479</td>
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<tr>
<td>c-KIT</td>
<td>Mutation</td>
<td>Gastrointestinal stromal tumor</td>
<td>Sunitinib, imatinib</td>
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<td>FLT3</td>
<td>Internal tandem duplication</td>
<td>Acute myeloid leukemia</td>
<td>Lestaurnitin, XL999</td>
</tr>
<tr>
<td>RET</td>
<td>Mutation, translocation</td>
<td>Thyroid medullary carcinoma</td>
<td>XL184</td>
</tr>
<tr>
<td>Non-receptor tyrosine kinase</td>
<td></td>
<td></td>
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<tr>
<td>ABL</td>
<td>Translocation (BCR-ABL)</td>
<td>Chronic myeloid leukemia</td>
<td>Imatinib</td>
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<td>JAK2</td>
<td>Mutation (V617F), translocation</td>
<td>Chronic myeloid leukemia, myelo-proliferative disorders</td>
<td>Lestaurnitin, INC018424</td>
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<td>SRC</td>
<td>Overexpression</td>
<td>Non–small-cell lung cancer; ovarian, breast cancer; sarcoma</td>
<td>XX2–391, dasatinib, AZD0530</td>
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<td>Serine–threonine–lipid kinase</td>
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<td>BRAF</td>
<td>Mutation (V600E)</td>
<td>Melanoma; colon, thyroid cancer</td>
<td>SB-590885, PLX-4032, RAF265, XL281</td>
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<tr>
<td>Aurora A and B kinases</td>
<td>Overexpression</td>
<td>Breast, colon cancer; leukemia</td>
<td>MK-5108 (VX-689)</td>
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<td>Polo-like kinases</td>
<td>Overexpression</td>
<td>Breast, lung, colon cancer, lymphoma</td>
<td>BI2536, GSK461364</td>
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<tr>
<td>MTOR</td>
<td>Increased activation</td>
<td>Renal-cell carcinoma</td>
<td>Temsirolimus (CCI-779), BEZ235</td>
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<tr>
<td>PI3K</td>
<td>PIK3CA mutations</td>
<td>Colorectal, breast, gastric cancer; glioblastoma</td>
<td>BEZ235</td>
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<tr>
<td>DNA damage or repair</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>BRCA1 and BRCA2</td>
<td>Mutation (synthetic lethal effect)</td>
<td>Breast, ovarian cancer</td>
<td>Olaparib, MK-4827 (PARP inhibitors)</td>
</tr>
</tbody>
</table>

* PARP denotes poly(adenosine diphosphate–ribose) polymerase.
Interface of Genomics and Informatics with the Healthcare Continuum
Interface of Genomics and Informatics with the Healthcare Continuum

- Availability of clinical practice guidelines
  - necessitate data derived from large studies
- Limited healthcare workforce genomic knowledgebase
  - Novel strategies for education given the current fiscal climate
  - And the infrastructure needed to integrate genomics into healthcare delivery systems such as the electronic health record (EHR), and point of care decision support (McCarthy, 2013)
  - At the cusp of the $1000 genome, managing the breadth of genomic health information that will continue to evolve over time is a remarkable undertaking yet is has the opportunity to truly improve and personalize healthcare
  - Informatics plays a vital role in every phase of the application of genomics to the healthcare continuum and as such emerges as the keystone of effective genomic clinical translation
Interface of Genomics and Informatics with the Healthcare Continuum

• Infrastructure is needed to integrate genomics into healthcare delivery systems
  – Electronic health record (EHR)
  – Point of care decision support

• Manage the breadth of genomic information and the rapidity in which it is changing
  – Infrastructure to track and communicate clinically relevant information
Transition to Personalized Healthcare

• Interprofessional Teams
  – Education
    – Patient/public and Provider

• Data
  • Clinical, behavioral, economic
  • Lifestyle and medical intervention
  • Genomic/molecular
  • Health/risk
  • Family health history

• Infrastructure
  – Healthcare delivery models
  – Biobanks
  – Evidence, outcomes, quality metrics

Define the most recent advances in Pharmacogenetics and Pharmacogenomics
Pharmacogenomic Influences

Efficacy

Toxicity
- inducers
- inhibitors

Pharmacodynamics

Pharmacokinetics

Target

PK = absorption, distribution, metabolism and excretion
PD = mechanism of action, drug concentration and effect
MASTERING PHARMACOGENOMICS
A Nurse’s Handbook for Success

DALE HALSEY LEA | DENNIS CHEEK
DANIEL BRAZEAU | GAYLE BRAZEAU
Genomics Contributions to Individual Drug Response

• Genomics is estimated to contribute to 20-50 % of individual drug response

• Even MEDICARE approves genetic testing of 9 common P450 (CYP) enzymes involved in drug metabolism
Pharmacogenetics Defined

- Pharmacogenetics is the Individual Inheritance of DNA sequences that contributes to individual responses to drugs.

- Pharmacogenetics is the variations in DNA sequences as related to drug response (FDA, 2008, p.3)

From: Lea, DH, and Winkelman, C.
Basics of Genetics and Genomics, pp. 1-22 in Mastering Pharmacogenomics: A Nurse’s Handbook for Success, Lea, DH, Cheek, DJ, Brazeau, D, and Brazeau, G.
Pharmacogenomics Defined Again Related to Nursing

• The study of how an individual’s genetic inheritance affects the body’s response to drugs

• Matching the individual’s genome profile to deliver the best drug for the person at the best dosage that is the most effective and least likely to cause side effects (Personalized Treatment - In Precision Medicine)
Nursing Implications - Symptom Management

• Priority area of nursing research is the study of the genetic influences of symptom clusters

• Pharmacogenomics
  – Inhibitors and/or Inducers
    • Implications for:
      – Medications used for other health conditions
      – Selecting medications to control
      – Use of over the counter medications like St. John's Wort
      – Consumption of certain foods or supplements like grapefruit/grapefruit juice
What is PharmGKB?

• A knowledge base that collects, curates, and disseminates knowledge about the impact of human genetic variation on drug response

• This knowledge base represents Primary pharmacogenomic literature, Knowledge extraction, Knowledge annotation, aggregation and integration, Clinical interpretation, and Clinical Implementation (where available)

• PharmGKB contains clinical information including dosing guidelines and drug labels, potentially clinically actionable gene-drug associations and genotype-phenotype relationships- links to CPIC

• It is NIH Funded and include consortium for Warfarin, Tamoxifen, SSRI, and Clopidogrel
What is the Clinical Pharmacogenetics Implementation Consortium (CPIC)

- CPIC produces guidelines or known clinical implementation based upon 4 levels of evidence:
  - Level 1 a and b - High level of evidence becomes a CPIC guideline
  - Level 2 a and b - Moderate level of evidence is represented as a variant in PharmGKB Very Important PGx gene summaries (VIP)
  - Level 3 - Low level of evidence
  - Level 4 - Preliminary data only
CPIC guidelines in EHR and using Clinical Decision Support (CDS)

• Today preemptive test results should be placed in the EHR months or years before relevant drug is used

• It is necessary to have the capability to deliver drug specific information based on genetic results at the Point of Care

• CDS facilitates use of pharmacogenetic results over a patient’s longitudinal healthcare
Sample Warning

“Based on the genotype result, this patient is predicted to have intermediate TPMT activity. The patient is at risk for myelo-suppression with normal doses of 6-mercaptopurine. Consider starting 6-mercaptopurine doses at 30-70% of normal dose. Please consult a clinical pharmacist or review the pharmacogenetics tab for more information.”

Action statement:

- Cancel Entry
- Dose Altered Accordingly
- Modify

Source: James Hoffman, St. Judes
Roadmap to CDS from CPIC into EHR

• Provide access and education on the use of the PharmGKB and CPIC guidelines
• Describe workflow
• Develop algorithm pathway
• Develop comprehensive translation tables from genotypes to phenotypes for specific drugs
• Define structure and process to efficiently develop and maintain user friendly formats of CDS text
• Publish results and enter results into PharmGKB and CPIC guidelines
HLA-B*57:01 Pharmacogenetic Test Result: Clinical Implementation Workflow for EHR

1. Enter test result in EHR
2. Add consultation/interpretation to EHR
3. Priority result?
   - Yes: Pt on high-risk drug now?
     - Yes: Medication evaluation or reassessment with the clinicians on service
     - No: Add coded genotype phenotype summary to EHR
   - No: No additional gene-based CDS
4. Result is available for Post-Test CDS
5. Blue shading indicates interaction with provider
<table>
<thead>
<tr>
<th>Test Name</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cytochrome P450 2C19</td>
<td>This test detects variants in genes which may affect an individual’s response to approximately 5% - 10% of all medications.</td>
</tr>
<tr>
<td>Cytochrome P450 2C9 &amp; VKORC1</td>
<td>This test detects variants in genes which may affect an individual’s response to approximately 15% of all medications (including Warfarin Sensitivity via 2C9 and VKORC1).</td>
</tr>
<tr>
<td>Cytochrome P450 2D6</td>
<td>This test detects variants in genes which may affect an individual’s response to approximately 25% of all medications.</td>
</tr>
<tr>
<td>Cytochrome P450 3A4 &amp; 3A5</td>
<td>This test detects variants in genes which may affect an individual’s response to approximately 40% - 45% of all medications.</td>
</tr>
<tr>
<td>Factor II Prothrombin Genetic Profiling</td>
<td>This test detects a genetic change in the Factor II gene called Factor II Prothrombin. Patients with this Prothrombin variant are at an increased risk of blood clot formation (thrombosis) when exposed to other risk factors such as smoking, pregnancy, obesity, oral contraceptive use, and immobility. The risk is approximately 3-10 times higher in individuals who have one copy of the genetic variant. The risk in people who carry two copies of the genetic variant is unknown. Individuals who do not have a Factor II Prothrombin mutation may still be at increased risk. Other changes in the Factor II gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis.</td>
</tr>
</tbody>
</table>
## Pharmacogenetic Testing and Drug Response (Part II) 9/23/2014

<table>
<thead>
<tr>
<th>Test</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Factor V Leiden Mutation Test</td>
<td>This test detects a genetic change in the Factor V gene called Factor V Leiden. Individuals who have this variant are at an increased risk of blood clot formation. This risk is approximately 2-10 times higher in individuals who have one copy of the genetic variant, and greater than 10 times higher for individuals who carry two copies of the genetic variant. Individuals who do not have the Factor V Leiden mutation may still be at increased risk. Other changes in the Factor V gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis.</td>
</tr>
<tr>
<td>MTHFR Mutation Testing</td>
<td>This test detects two genetic changes in the MTHFR gene. Individuals who are found to have two mutations are at an increased risk for serious blood clot formation. Individuals who have only one or no copies of either genetic change in the MTHFR gene may still be at increased risk. Other changes in the MTHFR gene that were not tested for, changes in other genes, and non-genetic factors may still increase your risk for thrombosis.</td>
</tr>
</tbody>
</table>
What is it going to take?

- A robust, **interoperable**, health IT environment with expert human interface that brings together:
  - Electronic Health Records
  - Personal Health Records
  - Personalized Health Information
  - Public Health Information
  - Standards (data, technical and security)
  - Interoperable Health Information Exchanges
Implemented vs. Potential Scope of Content

PREDICT Drug-Genome Interactions | 5

CPIC Guidelines (published or planned) | 83

FDA Pharmacogenomic Biomarker on Drug Labels | 155

PharmGKB Annotations | 560

Number of Drug-Pgx Interactions
Questions still raised

• Structure of the data
• Standardization of the data
• National and international data sharing
• Structure of guidelines
• Structure of the decision supports
Roadmap to the Future
Call for a new taxonomy from research knowledge networks:

- Exposomes
- Signs and Symptoms
- Genome
- Epigenome
- Microbiome
- Other Type of Patient Data
- Individual Patients
- INTEGRATED WITH OBSERVATIONS COMING FROM THE EHR AND BIOMEDICAL RESEARCH (where the report puts nursing)
A Case for Integrated Data in EHR

- Resources are available on current pharmacogenetics and pharmacogenomics from:

- Guidelines produced by the Clinical Pharmacogenomics Implementation Consortium (CPIC) - free, peer-reviewed, updated and detailed gene-drug clinical practice guidelines
Nursing Informatics Examples

- Assure the family history section in an EHR elicits a minimum of three generation family history and the physical assessment section includes information regarding genetic and environmental information and risk factors.
- Assist in identification of current genetic and genomic information resources that should be included in clinical practice guidelines orchestrated in the EHR.
- Works on policies regarding access to genomic information stored within the EHR.
- Understands the issues around genomic privacy and identifies appropriate state legislation, legal and social issues related to use and potential misuse of genomic information.
Types of IT Support

• Finding the data
• Accessing the data
• Sharing the data
• Storing, organizing, managing, and processing the data on enterprise architectures versus Big Data platforms
• Analyzing biomedical Big Data - genomic, phenotypic, environmental, and clinical
• Training faculty, clinicians, patients
Institute of Medicine 2012 Report
Recommendations

- Standardized, open source data bases with professional annotation, analytics, and curations
- Integrating research and clinical data
- Supporting open source platforms for the development of software;
- Consider secondary uses of IT infrastructure as a way to reduce overall costs

Further IOM 2012 Recommendations

- Standardize clinical data similar to high quality research data
- Develop new statistical methods and study designs for use with clinical trials
- Develop better data mining and filtering approaches to sort through massive datasets
- Connect genomic and molecular data with clinical data
- Structure clinical data to support the research data
- Integrate data that are already in the public domain to generate new hypotheses for testing
- Ensure processes are guided with a research framework
- Use a systems view of disease, which postulates that disease is a result of perturbations of one or more biological networks that lead to altered expressions of information
Sharing the Data

- Best practices of diagnosis and treatment can be leveraged to many patients and to many providers
- Policies and procedures are required for sharing data
- Governance policies and structures need to be apart of architecture designs
- Data have to be accessible for one patient over a lifetime - consider this when migrating to new systems or data repository storage or clouds
- Assure privacy and security of patient information - same principles as the EHR- authentication, access, encryption
- Note: On September 20, 2013, NIH released a draft Genomic Data Sharing Policy (GDS Policy) for a 60-day public comment period that closed November 20, 2013
Storing, Organizing, Managing, and Processing the Data

• Moving from siloed data to platforms
• Moving from enterprise to Clouds
• Moving to Surveillance Methods
• Integrating Research with Clinical with Patient
• Moving to the Penultimate Evidence
Resources to Assure Security and Privacy of Clouds

  http://www.nist.gov/manuscript-publication-search.cfm?pub_id=909494

In Summary Preparing IT for the Future

- The volume requirement to match phenotype (who the patient is and their current conditions), with genotype (what genes they possess and what the genome structure is of their clinical condition) is beyond any big data types that we have described for measuring quality, costs, and effectiveness.
The Volume is estimated at 1 Terabyte of data

• The genetic analysis of one patient can produce about 1 terabyte of data in a single encounter (Savage, 2014)
The Volume is Exacerbated by Integrating with Additional Data

- If the patient is in a care pathway requiring diagnosis and treatment based upon their genomes, then the data needs are multiplied by the number of times the genomes have to be measured to determine if the patient is improved, stabilizing, deteriorating, or on the path to mortality. Included in this pathway analysis is the need to have imaging data (X-ray, MRI, CAT Scan, which is already massive data), tissue biopsy data, laboratory data, and clinical observations of signs and symptoms of improvement, toxicities to treatments, or rejections of transplant tissue.
The Volume is Exacerbated by Determining Genomic Changes over Time and Number of Persons Tested

• These data are also utilized to determine the stage of the disease during the diagnosis and treatment. Multiply this by the number of patients with a condition (in the case of cancer, 1.7 million people diagnosed in 2013, then the data growth is massive
A Peek Into the Future

- Complete a mutational atlas for all cancer tumors, all types of heart, immunological, neurological diseases
- Expand beyond the atlas to: metastases, recurrence to full recovery
- Systematic functional annotation
- Systematic clinical utilization
- Data sharing nationally and internationally
- Attempts to genotype 1 million Americans and share data
Interested in More Depth, Case Studies, and More Resources?

ANIA Preconference Workshop
April 23, 2015 in Philadelphia, 1-5 PM Session 040

“Getting ANIA Members Involved in Genomics and IT to Support Personalized (Precision) Health”
Questions/Discussion

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