Journal of Nursing Scholarship 2013
Genomic Special Issue
Relevance of Genomics to Healthcare and Nursing Practice
Overview of the Webinar

- Perspectives of the Journal of Nursing Scholarship by the Journal Editor
- Genetics/Genomics and Relevance to Nursing Practice, Education and Research
- A summary of the content provided in the Genomic Special Issue
- Genomic Education Resources
- Outline of the ongoing Webinar series
- Discussion/Questions and Answers
Susan Gennaro, RN, DSN, FAAN

Editor, Journal of Nursing Scholarship

Dean and Professor
Boston College
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Relevance of Genomics to Nursing Practice and Education
Definitions

- **Genetics** – study of individual genes and their impact on relatively rare single gene disorders

- **Genomics** – study of all the genes in the human genome together, including their interactions with each other, the environment, and other psychosocial and cultural factors
Top 10 Leading Causes of Death

- Heart Disease 24.8%
- Malignant Neoplasms 23.5%
- Chronic Respiratory Disease 5.7%
- Cerebrovascular 5.3%
- Unintentional Injury 4.8%
- Alzheimer's Disease 3.3%
- Diabetes Mellitus 2.9%
- Influenza & Pneumonia 2.2%
- Nephritis 2.0%
- Suicide
Emerging Science/Technology
Cost per Genome

Moore's Law

National Human Genome Research Institute

http://www.genome.gov/sequencingcosts

http://www.genome.gov/27541954
The Race for the $1000 Genome

1000 Genomes Project Promises Closer Look at Variation in Human Genome

A major goal of the 1000 Genomes Project is to assemble a comprehensive catalog of genetic variation in the human genome that will help increase our understanding of the genetic basis of disease. Building on the data and insights gained from the National Institutes of Health-funded Human Genome Project and the HapMap Project, the 1000 Genomes Project is aimed at sequencing genomes from more than 1000 individuals from diverse populations around the world. The project is organized through a research consortium of seven centers, including three from the National Human Genome Research Institute in Bethesda, Md., the Wellcome Trust Sanger Institute in Cambridge, England, and the Beijing Genomics Institute in Shenzhen, China. The project will be conducted in phases, with the third phase expected to be completed in 2013.

Another Stop on the Road to the $1,000 Genome

The latest stop on the road to the $1,000 genome? San Francisco, CA, where J.P. Morgan’s 23rd Annual Healthcare Conference is in full swing. There is an abundance of real-time Twitter coverage from the conference, but certain announcements warrant a more detailed discussion.

The announcement generating the biggest buzz today came from Illumina, Inc., whose CEO, Jay Flatley, unveiled a new genome sequencing machine, the HiSeq 2000. According to Matthew Harper of Forbes.com, Illumina’s new machine will “produce 100,000 worth of impelling chimes” will begin shipping in February with a cost of $690,000. [acdc.illumina.com]

MUSINGS

The $1,000 genome, the $100,000 analysis?

Elaine R Mardis*

Having recently attended the Personal Genomes meeting at Cold Spring Harbor Laboratories (I was an organizer this year), I was struck by the number of talks that described the use of whole-genome sequencing and analysis to reveal the genetic basis of disease in patients. This is the sort of application that is required for it to occur. I therefore offer the following as food for thought.

One source of difficulty in using resequencing approaches for diagnosis centers on the need to improve the quality and completeness of the human reference genome database.
Device Brings $1,000 Genome Within Reach

Ion Torrent introduced its new tabletop sequencer at CES this week.

THURSDAY, JANUARY 12, 2012 | BY ERICA WESTLY

Thanks to advances in chemistry and software, researchers can soon sequence a human genome for $1,000 in a day.

Back in July, Jonathan Rothberg, CEO of the Connecticut-based biotech company Ion Torrent, predicted that by 2013 his company would develop a chip that could sequence an entire human genome.

This week, the company surpassed that prediction with a new tabletop sequencer called the Proton. The company introduced the device at the Consumer Electronics Show in Las Vegas on Tuesday, although the sequencer is only available to researchers at this point.

At $149,000, the new machine is about three times the price of the Personal Genome Machine, the sequencer that the company debuted about a year ago. But the DNA-reading chip inside it is 1,000 times more powerful, according to Rothberg, allowing the device to sequence an entire human genome in a day for $1,000—a price the biotech industry has been working toward for years because it would bring the cost down to the level of a medical test.
Single nucleotide polymorphism (SNP)
Published Genome-Wide Associations through 06/2011, 1,449 published GWA at $p \leq 5 \times 10^{-8}$ for 237 traits

NHGRI GWA Catalog
www.genome.gov/GWAStudies
Published Genome-Wide Associations through 07/2012
Published GWA at $p \leq 5 \times 10^{-8}$ for 18 trait categories

NHGRI GWA Catalog
www.genome.gov/GWASTudies
www.ebi.ac.uk/fgpt/gwa
Direct to Consumer Marketing and Testing

- Tests are available direct to the consumer.
- Most require only a saliva sample.
- Example: 23andMe-Evaluating more than 1,000,000 SNP’s for >200 health conditions or health related traits $99.

http://www.23andme.com
Epigenetics

➤ The study of genetic variation caused by the activation and deactivation of genes without any change in the underlying DNA sequence.

➤ The epigenome involves chemical compounds that modify, or mark the genome in a way that tells it what, where, and when to do it.

  • Can be passed on from cell to cell as cells divide, and from one generation to the next.
Scope of Genome Analysis

- Has expanded to include any whole genome analysis such as
  - Whole genome sequencing
  - Whole exome sequencing
  - RNA and RNAi sequencing
  - Whole genome SNP analysis

- Consideration for incidental findings
  - Previously unknown information
    - Clinical and analytic validity of finding
    - Immediacy and seriousness of risk
    - Actionable finding
Genomic Healthcare Applications

- Preconception and prenatal assessments
- Newborn screening
- Identification of at risk individuals before disease occurs
- Screening and/or diagnosis of disease
- Characterization of disease and its aggressiveness via gene expression
- Individualization of therapies over the disease course based on molecular profiles
- Development of genetically targeted therapies
- Monitoring for disease progression
- Individualized drug selection and/or dosing based on pharmacogenomics

Genomics

- Preconception and Prenatal Testing
- Newborn Screening
- Disease Susceptibility
- Screening and Diagnosis
- Prognosis and Therapeutic Decisions
- Monitoring Disease Burden and Recurrence
The Quest for Personalized Health Care

- Use of an individual's genetic/genomic information in addition to traditional health information to guide health care decision-making

- Disease prevention, risk reduction, diagnosis, treatment, symptom management and palliative care
  - Pharmacogenomics
    - Medication selection
    - Dose selection
    - Inhibitors
    - Inducers
Genomic Competencies for Nurses
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 to be published in 2012 which includes some updates

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

http://nursingworld.org/MainMenuCategories/EthicsStandards/Genetics-1/Essential-Genetic-and-Genomic-Competencies-for-Nurses-With-Gra...
Journal of Nursing Scholarship
Genomics Special Issue 2013

Why:
• Genomics is relevant to ALL nursing practice
• Nurses practicing in any setting will increasingly apply genomics in patient care
• Genomic Special Issue is a resource for nursing education

Purpose of the Special Issue:
• Provide evidence reviews about the genomics of common health conditions with relevance to nursing practice and services
What:

- Articles provide an overview of:
  - Genomic science and technology
  - International nursing care implications
  - Genomic variation and common diseases
  - Encompass pediatrics and adults
  - Ethical, legal, and social issues
  - Nursing research priorities
Editorial: Relevance of Genomics to Healthcare and Nursing Practice

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Editorial

Genomics

Preconception and Prenatal Testing

Newborn Screening

Disease Susceptibility Screening and Diagnosis

Prognosis and Therapeutic Decisions

Monitoring Disease Burden and Recurrence
Current and Emerging Approaches in Genomics

- Yvette Conley PhD et al.
- Technologies for collecting, analyzing, & interpreting genomic information
  - Genome Sequencing
  - Genome-Wide Association Studies
  - Epigenomics
  - Gene Expression
Ethical, Legal, & Social Issues in the Translation of Genomics into Healthcare

• Laurie Badzek LLM, JD, MS, RN, FAAN et al.

• Provides a review of:
  • Ethical and legal foundations
  • Confidentiality & privacy of genomic information
  • Informed consent
  • Genetic testing
  • Biorepositories
Integration of Genomics in Cancer Care

- Erika Santos PhD, MS, RN et al.
- Provides an overview of:
  - Cancer etiology
  - Hereditary cancer syndromes
  - Epigenetics
  - Cancer management considerations
- Case studies illustrate genomic advances in oncology
Implications of Newborn Screening for Nurses

- Jane DeLuca PhD, RN, CPNP, APNG et al.
- Provides an overview of:
  - Current newborn screening activities
  - Controversies and ethical considerations
  - Roles of nurses in the newborn screening process
  - Summary of developments in newborn screening (i.e., genome sequencing)
An Update of Childhood Genetic Disorders

• Cynthia Prows, MSN, CNS, FAAN et al

• Provides an overview of:
  • Common childhood genetic disorders
  • Encompasses infants, children and adolescents
  • Important resources and services
Cardiovascular Genomics

- Shu-Fen Wung PhD, RN, ACNP-BC, FAAN et al.
- Provides an overview of cardiovascular genomics:
  - Myocardial infarction/Coronary artery disease
  - Stroke
  - Sudden cardiac death
  - Genetic testing for sudden cardiac death
An Overview of the Genomics of Metabolic Syndrome

- Jacquelyn Taylor, PhD, PNP-BC et al.
- Provides an overview of metabolic syndrome:
  - Diagnostic criteria
  - Contributions of cardiovascular, obesity, and diabetes genomic risk factors
  - Genes and polymorphisms associated with MetS
Physical, Psychological, & Ethical Issues in Caring for Individuals with Genetic Skin Disease

- Diane Seibert PhD, ARNP, FAANP & Thomas Darling MD, PhD
- Provides an overview of:
  - Five genetic skin disorders
    - Inheritance patterns
    - Genomics
    - Treatments
  - Issues and concerns important to caring for patients with genetic skin diseases
Genomics and Autism Spectrum Disorder (ASD)

• Norah Johnson PhD, RN, CPNP et al.
• Provides an overview of ASD:
  • Identification
  • Diagnosis
  • Family implications
• Genomic contributions to ASD risk
• Limitations and complexities of ASD research
The Implications of Genomics on the Nursing Care of Adults with Neuropsychiatric Conditions

- Debra Schutte PhD, RN et al.
- Provides an overview of the genomic contributions to neuropsychiatric disorders
  - Irreversible dementias
  - Alzheimer disease
  - Huntington Disease
- Implications for nursing practice and research
A Blueprint for Genomic Nursing Science

- Kathleen Calzone PhD, RN, APNG, FAAN et al.
- Summarizes recommendations of a 2012 Genomic Nursing State of Science Advisory Panel
- Blueprint provides framework to further genomic nursing science to improve health outcomes
2013 Genomics Special Issue

• World perspective of the impact of genomic knowledge on clinical nursing practice

• Accessible at:
  http://www.genome.gov/27552093

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  • Journal of Nursing Scholarship (JNS)
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Genomics Special Issue 2013

• **Lead Editors:**
  - Kathleen Calzone, PhD, RN, APNG, FAAN
    National Cancer Institute
  - Jean Jenkins, PhD, RN, FAAN
    National Human Genome Research Institute
  - Nick Nicol, PhD, FCN
    Universal College of Learning in New Zealand

• **Editorial Advisory Board:**
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    Cincinnati Children’s Hospital
  - Diane Seibert, PhD, RN
    Uniformed Services University of the Health Sciences
  - Greg Feero, MD, PhD
    National Human Genome Research Institute
Other Genomic Resources

- Genomic educational resources:
  - Genetic/Genomic Competency Center for Education
    http://www.g-2-c-2.org/
  - Global Genetics and Genomics Community
    http://g-3-c.org/

- Competency and Curricular Resources
  http://www.genome.gov/27527634
2011 Journal of Nursing Scholarship
year long series
- Genetics/Genomics and Nursing Education
Welcome to the Talking Glossary of Genetic Terms
The Human Genome Defined by Professionals at the National Human Genome Research Institute

- Designed to help learners at any level better understand genetic terms
- Guided by national science standards
- Explained by scientists at the NIH

http://www.genome.gov/glossary
Surgeon General Family History Tool

My Family Health Portrait
A tool from the Surgeon General

Using My Family Health Portrait you can:

- Enter your family health history.
- Print your family health history to share with family or your health care worker.
- Save your family health history so you can update it over time.

Talking with your health care worker about your family health history can help you stay healthy!

Learn more about My Family Health Portrait

Create a Family Health History

En Español

Use a Saved History

Em Português

In Italiano

https://familyhistory.hhs.gov/
Genomic Competency Listserv

To Join email:
calzonek@mail.nih.gov
Webinar Series

Please visit http://www.genome.gov/27552312 for log-in information

February 19, 2013
3:30PM EST-Current and Emerging Technology Approaches in Genomics

March 5, 2013
3:30PM EST-Cardiovascular Genomics
4:00PM EST-An Overview of the Genomics of Metabolic Syndrome

March 20, 2013
3:30PM EST-Implications of Newborn Screening for Nurses and Nursing Faculty
4:00PM EST-Ethical, Legal, and Social Issues in the Translation of Genomics into Healthcare
Webinar Series, continued

April 2, 2013
3:30PM EST-Integration of Genomics in Cancer Care
4:00PM EST-Physical, Psychological and Ethical issues in Caring for Individuals with Genetic Skin Disease

April 26, 2013
3:30PM EST-Genomics and Autism Spectrum Disorder
4:00PM EST-An Update of Childhood Genetic Disorders

May 7, 2013
3:30PM EST-A Blueprint for Genomic Nursing Science