



Journal of Nursing
Scholarship 2013
Genomic Special Issue

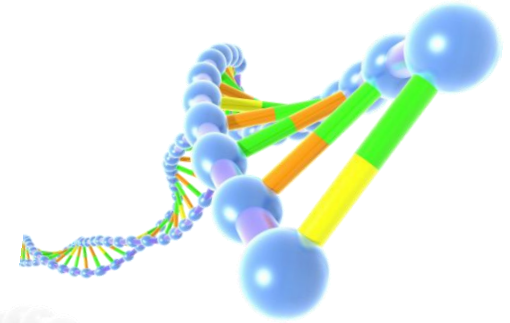
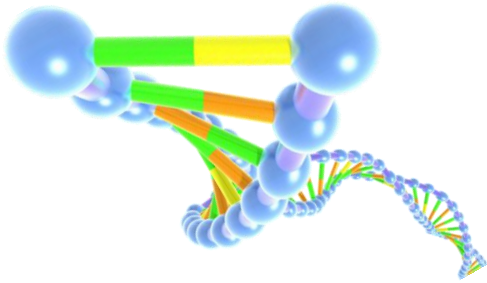
**Relevance of Genomics
to Healthcare and
Nursing Practice**

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**

William F. Connell School of Nursing



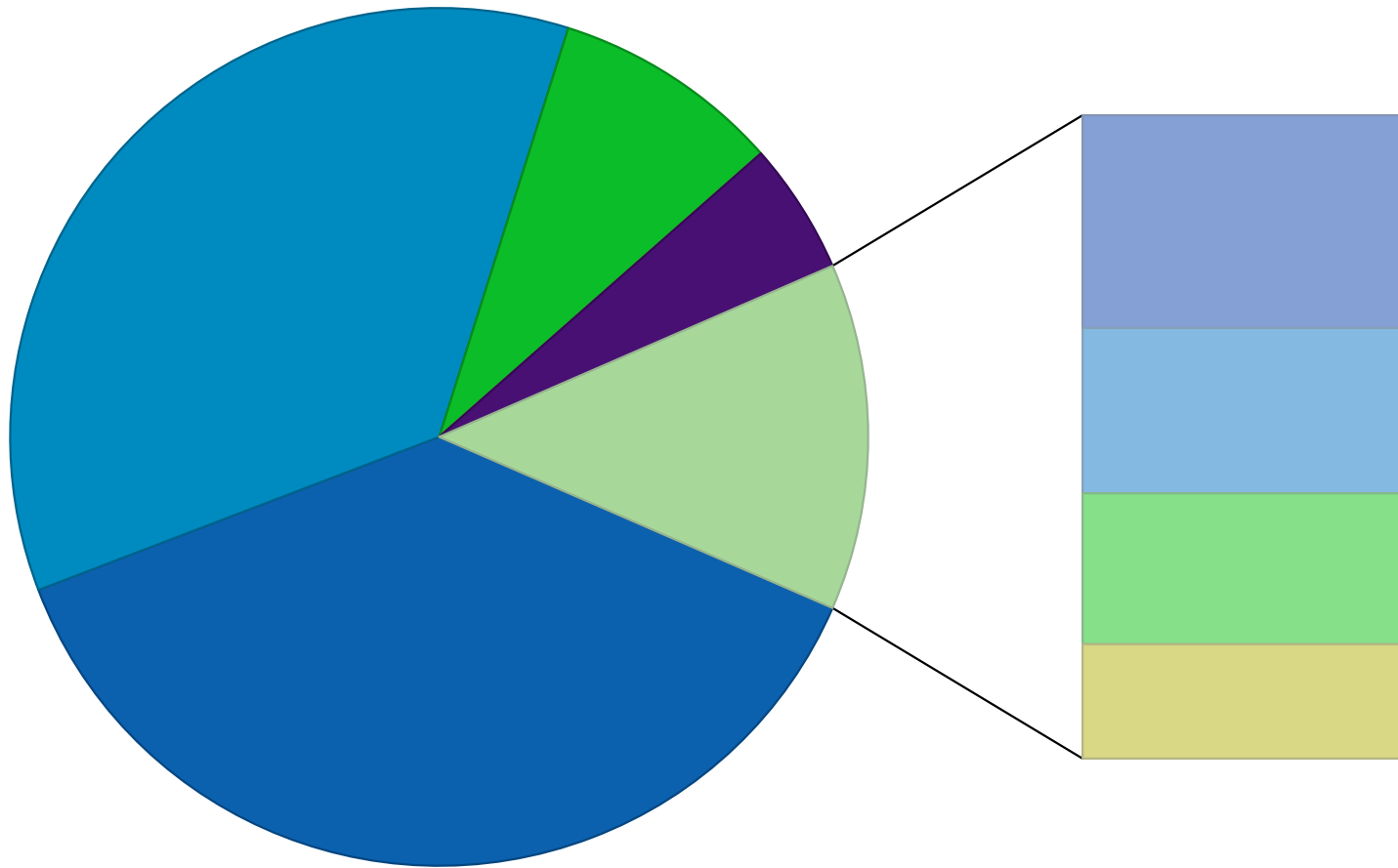
Relevance of Genomics to Nursing Practice and Education

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%

■ Chronic Respiratory Disease 5.7%

■ Unintentional Injury 4.8%

■ Diabetes Mellitus 2.9%

■ Nephritis 2.0%

■ Malignant Neoplasms 23.5%

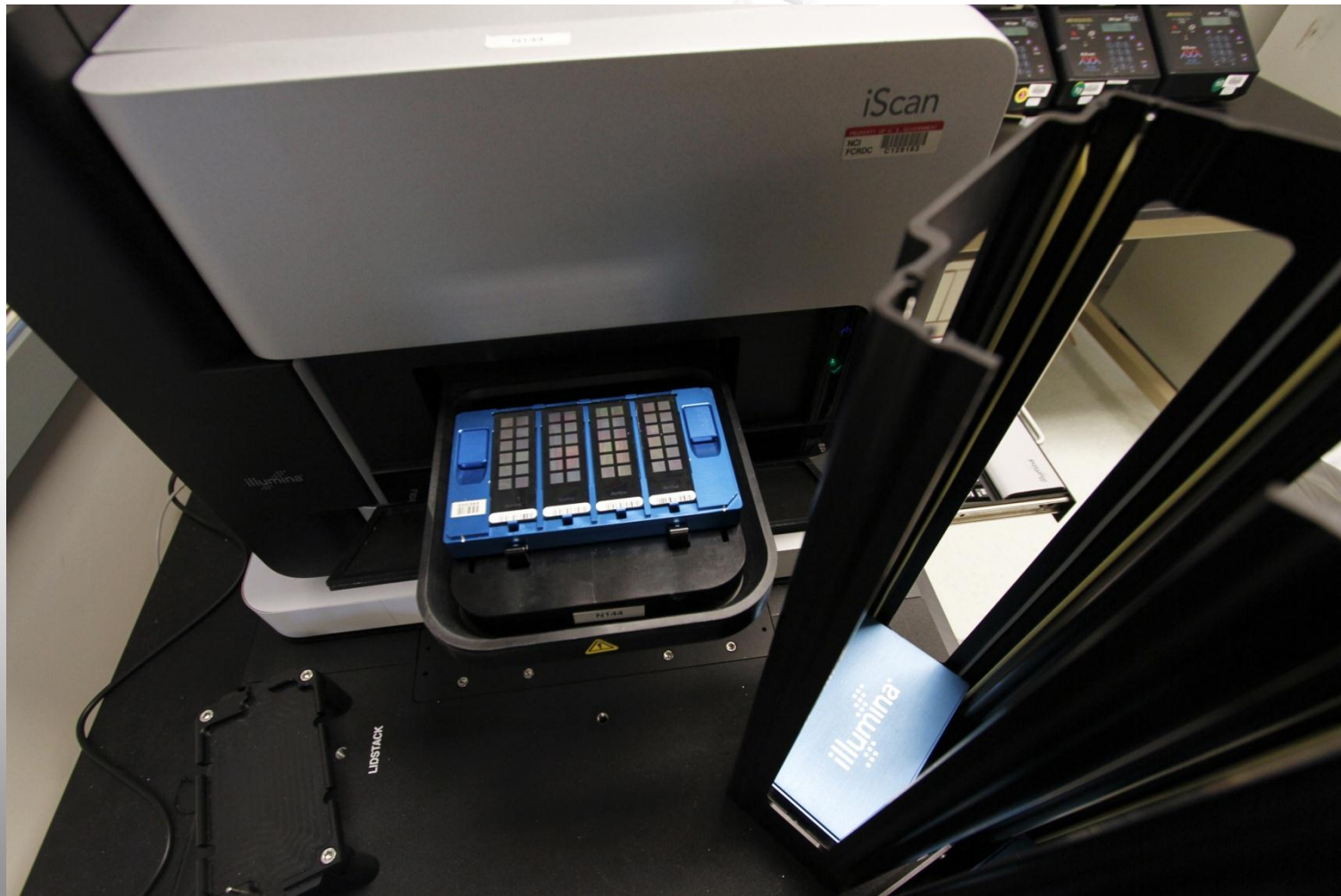
■ Cerebrovascular 5.3%

■ Alzheimer's Disease 3.3%

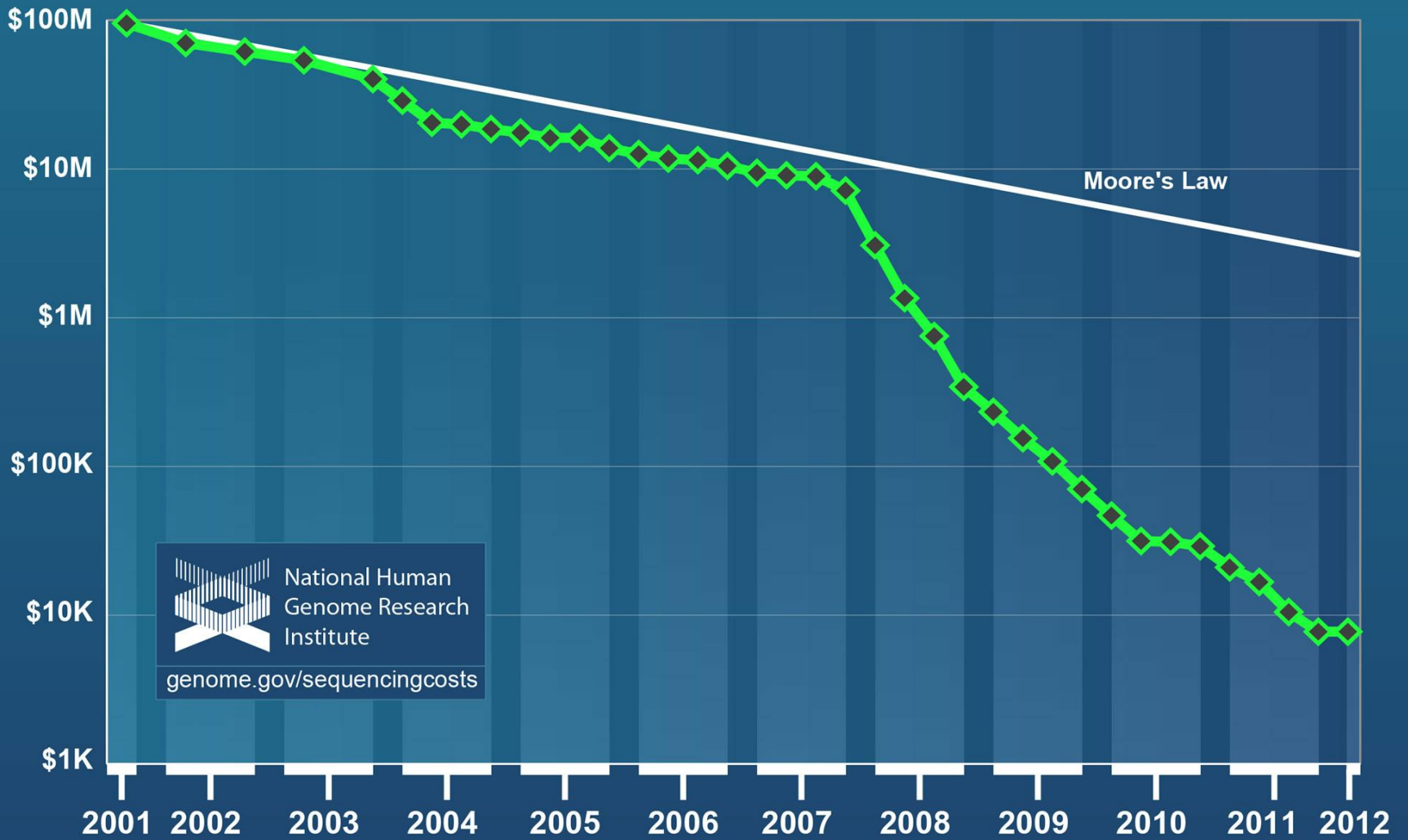
■ Influenza & Pneumonia 2.2%

■ Suicide

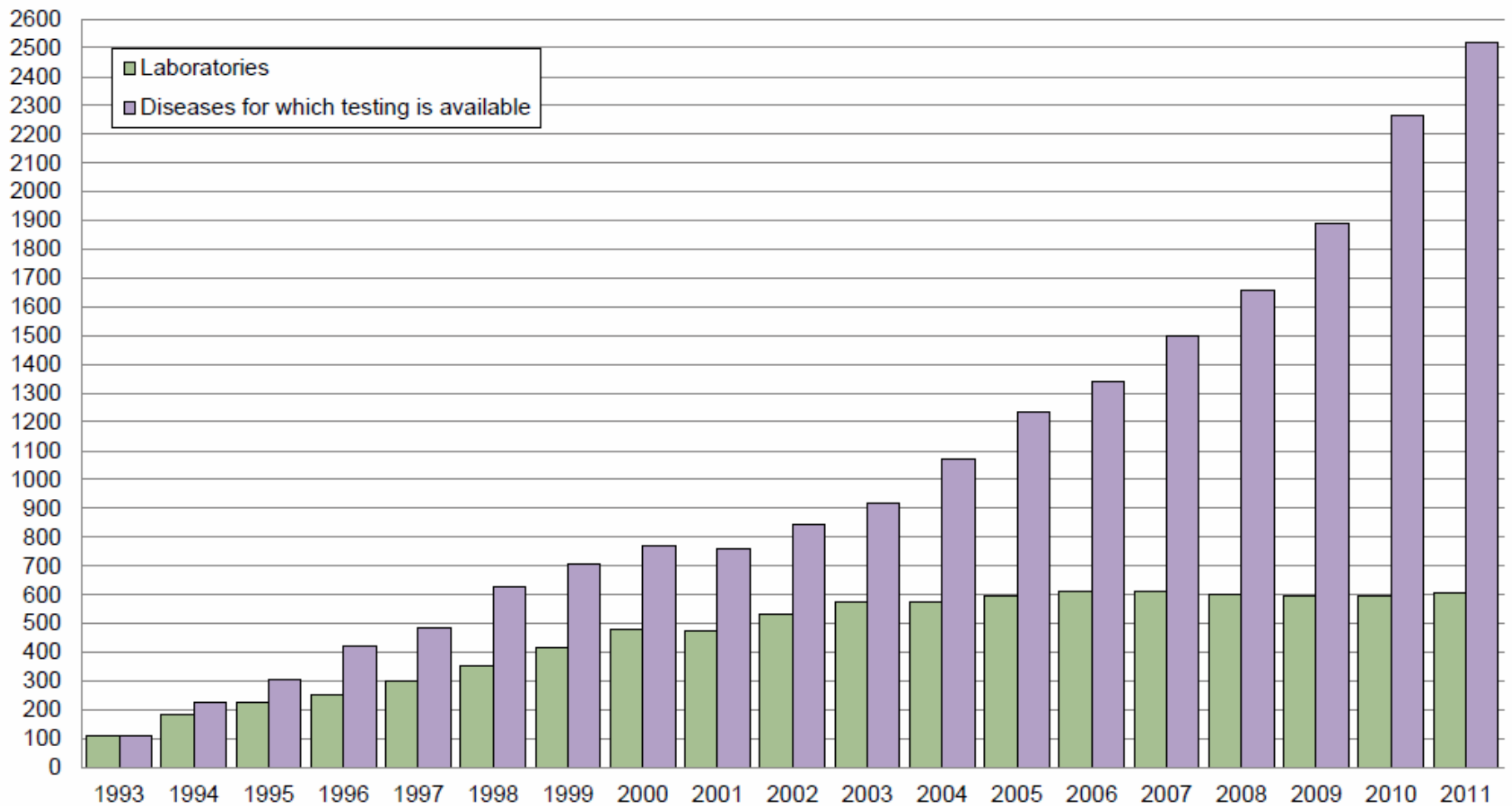
Emerging Science/Technology



Cost per Genome



GENE Tests: Growth of Laboratory Directory



Data source: GeneTests database (2011)/ www.genetests.org

The Race for the \$1000 Genome

1000 Genomes Project Promises Closer Look at Variation in Human Genome

Bridget M. Kowlin

A NEW, LARGE-SCALE PUBLIC SCIENCE project is developing a more detailed picture of variations in the human genome that may one day aid scientists' understanding of the genetic basis of disease.

Building on the data and technology generated in previous "big science" projects, such as the Human Genome Project and the HapMap (an effort aimed at describing the common patterns of genetic variation in humans), investigators for the 1000 Genomes Project plan to develop an extensive catalog of variation in the human genome by sequencing the genomes of at least 1000 individuals from around the world. The project is being carried out by an international consortium of researchers, including scientists from the National Human Genome Research Institute in Bethesda, Md., the Wellcome Trust Sanger Insti-

tute in Hinxton, England, and the Beijing Genomics Institute in Shenzhen, China. The first official data from the project will be released in January 2009, said David Altshuler, MD, PhD, co-chair of the consortium and professor of genetics and medicine at Harvard Medical School in Boston, Mass.

While it took years to sequence a single human genome during the Human Genome Project, new techniques and technologies are allowing the researchers to sequence DNA with much greater speed and at much lower cost. In fact, the project will be able to sequence approximately 8.2 billion bases per day—the equivalent of about 2 human genomes every 24 hours. Although the cost of the project has been estimated to be between \$30 and \$50 million, Altshuler noted that a precise estimate of cost is not currently available because the technological advances continue to reduce the costs of sequencing.

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Reprinted from JAMA, December 17, 2008—Vol 300, No. 23 2715



Genome Medicine

Genomics Law Report

News and analysis from the intersection of genomics, personalized medicine and the law

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

Posted by [Dan Vorhaus](#) on January 12, 2010

The latest stop on the road to the \$1,000 genome? San Francisco, CA, where [J.P. Morgan's 28th 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 Herper of Forbes.com](#), Illumina's new machine "will

000 worth of mpeting chines will begin shipping in February with a cost of \$690,000 (del). Illumina's [own product page for the HiSeq 2000](#) age (~30x) and read length (2x100 bp). There have also been equipped with an [iPhone user interface](#), a concept that Flatley [cs Show](#).



NHGRI Funding Opportunities: Research

- [NHGRI Funding Opportunities](#)
- [NIH-Wide Parent Funding Opportunities](#)
- [NIH-Wide Topic-Specific Funding Opportunities](#)

The National Institutes of Health (NIH) is replacing p transition to requiring electronic submission of grant the electronic application and transition timelines an [era.nih.gov].

Inquiries about NHGRI's program interests should be addressed to the [Division of E Program Staff](#)

Mardis *Genome Medicine* 2010, 2:84
<http://genomemedicine.com/content/2/11/84>

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.

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



technology review

Published by MIT

arpa·e energy innovation summit
2012 feb 27-29 | washington
Gathering the best minds in academia, business, and government

English | en Español | auf Deutsch | in Italiano | In Hindi

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The \$1,000 Human Genome: Are We There Yet?

The race for genomes-in-a-day heats up as two companies promise this service by the end of 2012

By Erika Check Hayden and Nature News Blog | January 10, 2012 | 3



Sequence machine: The Ion Proton Sequencer (left) from Ion Torrent Technologies

BIOMEDICINE

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

Audio

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 Ion 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)



Individual 1

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy1 ... GCTATAAGG**A**TAGCTTACAG...

Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy2 ... GCTATAAGG**G**TAGCTTACAG...

Individual 4

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy1 ... GCTATAAGG**A**TAGCTTACAG...

Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy2 ... GCTATAAGG**G**TAGCTTACAG...

Individual 2

Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy1 ... GCTATAAGG**G**TAGCTTACAG...

Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy2 ... GCTATAAGG**G**TAGCTTACAG...

Individual 5

Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy1 ... GCTATAAGG**G**TAGCTTACAG...

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy2 ... GCTATAAGG**A**TAGCTTACAG...

Individual 3

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy1 ... GCTATAAGG**A**TAGCTTACAG...

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy2 ... GCTATAAGG**A**TAGCTTACAG...

Individual 6

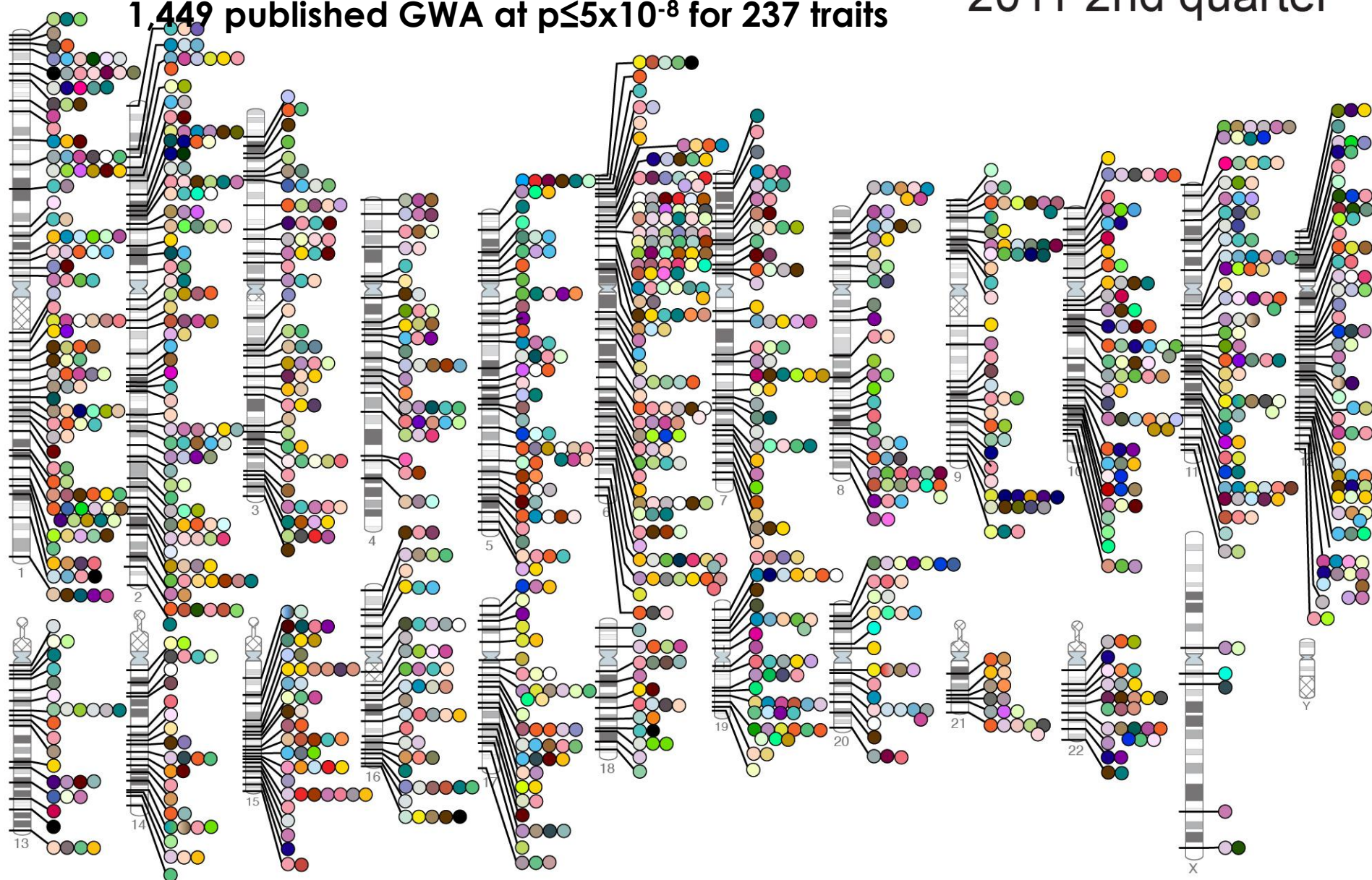
Chr 2 ... CGATATTCC**C**ATCGAATGTC...
copy1 ... GCTATAAGG**G**TAGCTTACAG...

Chr 2 ... CGATATTCC**T**ATCGAATGTC...
copy2 ... GCTATAAGG**A**TAGCTTACAG...

Published Genome-Wide Associations through 06/2011

1,449 published GWA at $p \leq 5 \times 10^{-8}$ for 237 traits

2011 2nd quarter



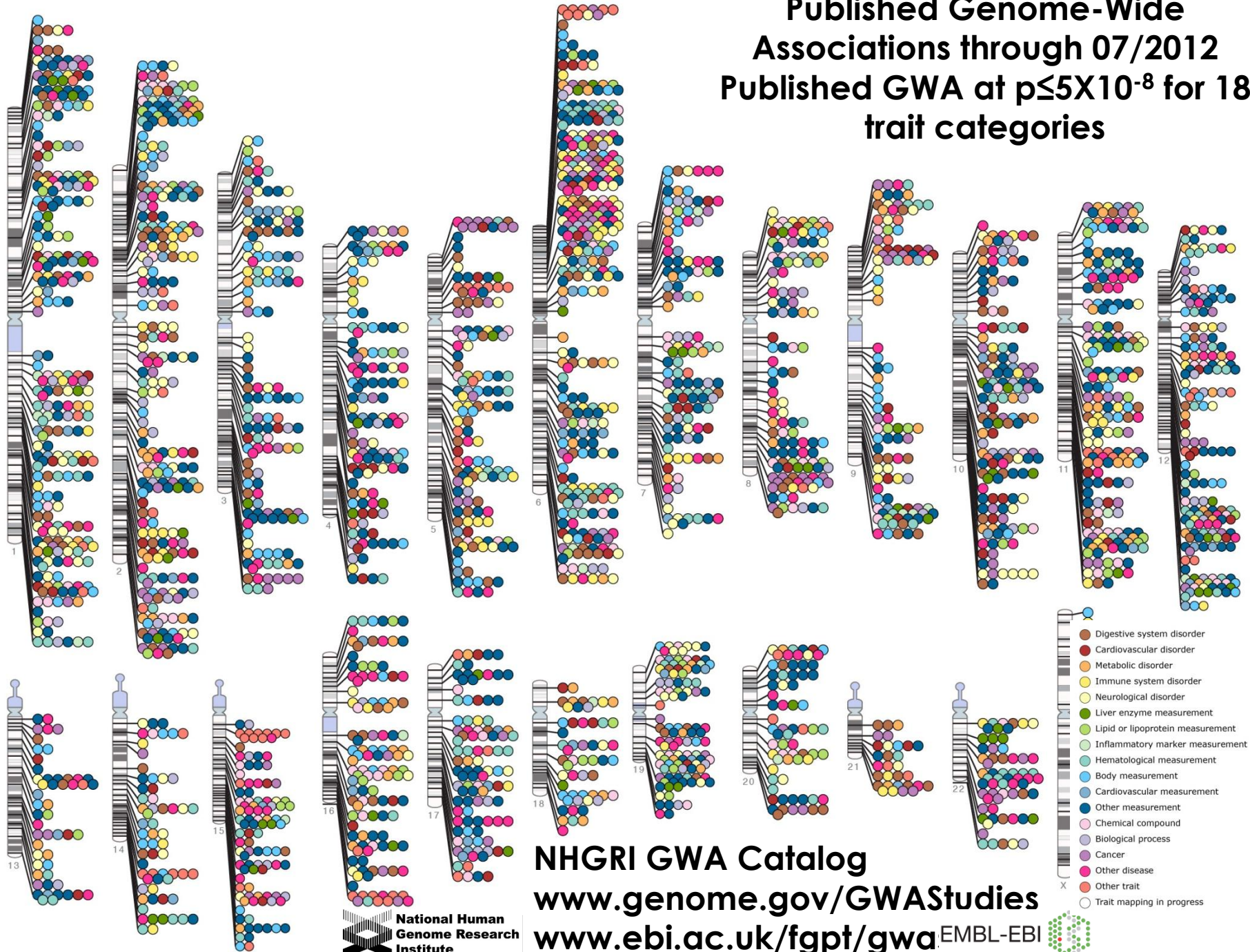
NHGRI GWA Catalog

www.genome.gov/GWAStudies

- Abdominal aortic aneurysm
- Acute lymphoblastic leukemia
- Adhesion molecules
- Adiponectin levels
- Age-related macular degeneration
- AIDS progression
- Alcohol dependence
- Alopecia areata
- Alzheimer disease
- Amyloid A levels
- Amyotrophic lateral sclerosis
- Angiotensin-converting enzyme activity
- Ankylosing spondylitis
- Arterial stiffness
- Asparagus anosmia
- Asthma
- Atherosclerosis in HIV
- Atrial fibrillation
- Attention deficit hyperactivity disorder
- Autism
- Basal cell cancer
- Behcet's disease
- Bipolar disorder
- Biliary atresia
- Bilirubin
- Bitter taste response
- Birth weight
- Bladder cancer
- Bleomycin sensitivity
- Blond or brown hair
- Blood pressure
- Blue or green eyes
- BMI, waist circumference
- Bone density
- Breast cancer
- C-reactive protein
- Calcium levels
- Cardiac structure/function
- Cardiovascular risk factors
- Carnitine levels
- Carotenoid/tocopherol levels
- Celiac disease
- Celiac disease and rheumatoid arthritis
- Cerebral atrophy measures
- Chronic lymphocytic leukemia
- Chronic myeloid leukemia
- Cleft lip/palate
- Coffee consumption
- Cognitive function
- Conduct disorder
- Colorectal cancer
- Corneal thickness
- Coronary disease
- Creutzfeldt-Jakob disease
- Crohn's disease
- Crohn's disease and celiac disease
- Cutaneous nevi
- Cystic fibrosis severity
- Dermatitis
- DHEA-s levels
- Diabetic retinopathy
- Dilated cardiomyopathy
- Drug-induced liver injury
- Drug-induced liver injury (amoxicillin-clavulanate)
- Endometrial cancer
- Endometriosis
- Eosinophil count
- Eosinophilic esophagitis
- Erectile dysfunction and prostate cancer treatment
- Erythrocyte parameters
- Esophageal cancer
- Essential tremor
- Exfoliation glaucoma
- Eye color traits
- F cell distribution
- Fibrinogen levels
- Folate pathway vitamins
- Follicular lymphoma
- Fuch's corneal dystrophy
- Freckles and burning
- Gallstones
- Gastric cancer
- Glioma
- Glycemic traits
- Hair color
- Hair morphology
- Handedness in dyslexia
- HDL cholesterol
- Heart failure
- Heart rate
- Height
- Hemostasis parameters
- Hepatic steatosis
- Hepatitis
- Hepatocellular carcinoma
- Hirschsprung's disease
- HIV-1 control
- Hodgkin's lymphoma
- Homocysteine levels
- Hypospadias
- Idiopathic pulmonary fibrosis
- IFN-related cytopeni
- IgA levels
- IgE levels
- Inflammatory bowel disease
- Insulin-like growth factors
- Intracranial aneurysm
- Iris color
- Iron status markers
- Ischemic stroke
- Juvenile idiopathic arthritis
- Keloid
- Kidney stones
- LDL cholesterol
- Leprosy
- Leptin receptor levels
- Liver enzymes
- Longevity
- LP (a) levels
- LpPLA(2) activity and mass
- Lung cancer
- Magnesium levels
- Major mood disorders
- Malaria
- Male pattern baldness
- Mammographic density
- Matrix metalloproteinase levels
- MCP-1
- Melanoma
- Menarche & menopause
- Meningococcal disease
- Metabolic syndrome
- Migraine
- Moyamoya disease
- Multiple sclerosis
- Myeloproliferative neoplasms
- Myopia (pathological)
- N-glycan levels
- Narcolepsy
- Nasopharyngeal cancer
- Natriuretic peptide levels
- Neuroblastoma
- Nicotine dependence
- Obesity
- Open angle glaucoma
- Open personality
- Optic disc parameters
- Osteoarthritis
- Osteoporosis
- Otosclerosis
- Other metabolic traits
- Ovarian cancer
- Pancreatic cancer
- Pain
- Paget's disease
- Panic disorder
- Parkinson's disease
- Periodontitis
- Peripheral arterial disease
- Personality dimensions
- Phosphatidylcholine levels
- Phosphorus levels
- Photic sneeze
- Phytosterol levels
- Platelet count
- Polycystic ovary syndrome
- Primary biliary cirrhosis
- Primary sclerosing cholangitis
- PR interval
- Progranulin levels
- Progressive supranuclear palsy
- Prostate cancer
- Protein levels
- PSA levels
- Psoriasis
- Psoriatic arthritis
- Pulmonary funct. COPD
- QRS interval
- QT interval
- Quantitative traits
- Recombination rate
- Red vs.non-red hair
- Refractive error
- Renal cell carcinoma
- Renal function
- Response to antidepressants
- Response to antipsychotic therapy
- Response to clopidogrel therapy
- Response to hepatitis C treat
- Response to interferon beta therapy
- Response to metformin
- Response to statin therapy
- Restless legs syndrome
- Retinal vascular caliber
- Rheumatoid arthritis
- Ribavirin-induced anemia
- Schizophrenia
- Serum metabolites
- Skin pigmentation
- Smoking behavior
- Speech perception
- Sphingolipid levels
- Statin-induced myopathy
- Stroke
- Sudden cardiac arrest
- Suicide attempts
- Systemic lupus erythematosus
- Systemic sclerosis
- T-tau levels
- Tau AB1-42 levels
- Telomere length
- Testicular germ cell tumor
- Thyroid cancer
- Thyroid volume
- Tooth development
- Total cholesterol
- Triglycerides
- Tuberculosis
- Type 1 diabetes
- Type 2 diabetes
- Ulcerative colitis
- Urate
- Urinary albumin excretion
- Urinary metabolites
- Uterine fibroids
- Venous thromboembolism
- Ventricular conduction
- Vertical cup-disc ratio
- Vitamin B12 levels
- Vitamin D insufficiency
- Vitiligo
- Warfarin dose
- Weight
- White cell count
- White matter hyperintensity
- YKL-40 levels

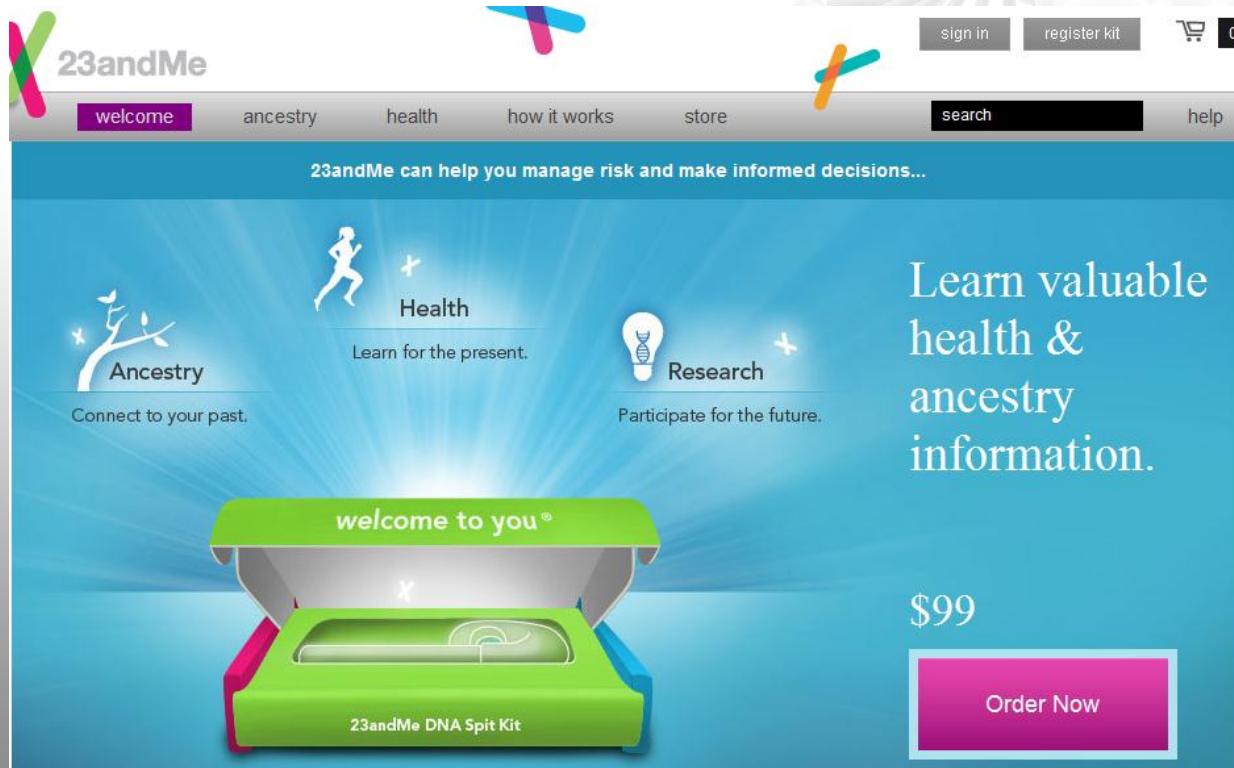
Published Genome-Wide Associations through 07/2012

Published GWA at $p \leq 5 \times 10^{-8}$ for 18 trait categories



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**.



The image shows a screenshot of the 23andMe website. At the top left is the 23andMe logo. To the right are links for 'sign in', 'register kit', and a shopping cart icon with '0' items. Below the logo is a navigation bar with 'welcome' (highlighted), 'ancestry', 'health', 'how it works', and 'store'. A search bar with 'search' and a 'help' link are also present. The main content area features a blue background with a central banner that reads '23andMe can help you manage risk and make informed decisions...'. Below this are three icons: 'Ancestry' (a tree), 'Health' (a person running), and 'Research' (a lightbulb). Each icon has a sub-headline: 'Connect to your past.', 'Learn for the present.', and 'Participate for the future.' respectively. On the right side of the main content area, there is a large text block that says 'Learn valuable health & ancestry information.' Below this is a price tag '\$99' and a large pink 'Order Now' button. At the bottom center, there is an image of an open green '23andMe DNA Spit Kit' with the text 'welcome to you' on the lid.

<http://www.23andme.com>

Epigenetics



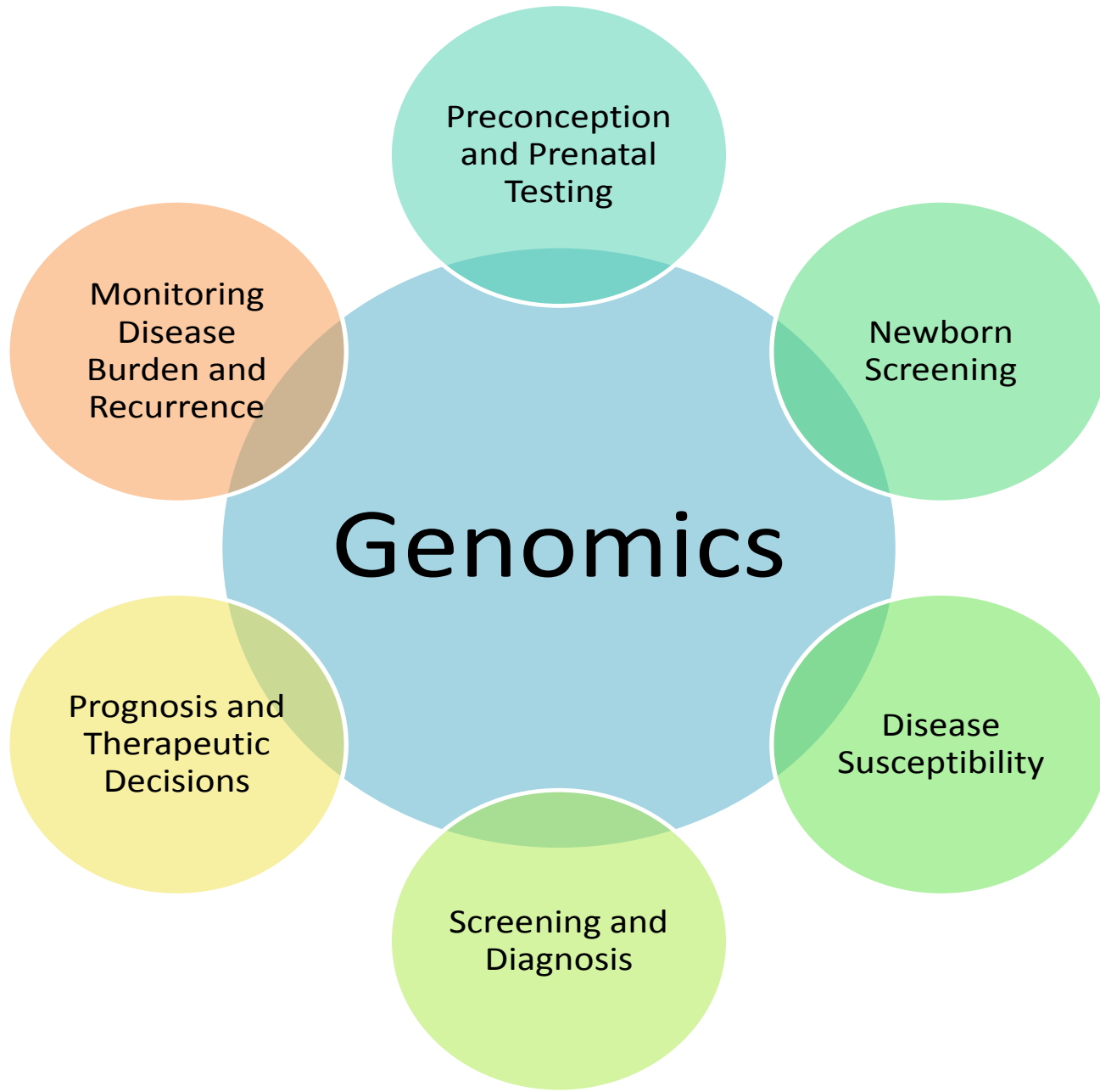
- The study of genetic variation caused by the activation and deactivation of genes without any change in the underlying DNA sequence.
- The epigenome involves chemical compounds that modify, or mark the genome in a way that tells it what, where, and when to do it.
 - Can be passed on from cell to cell 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



The Quest for Personalized Health Care



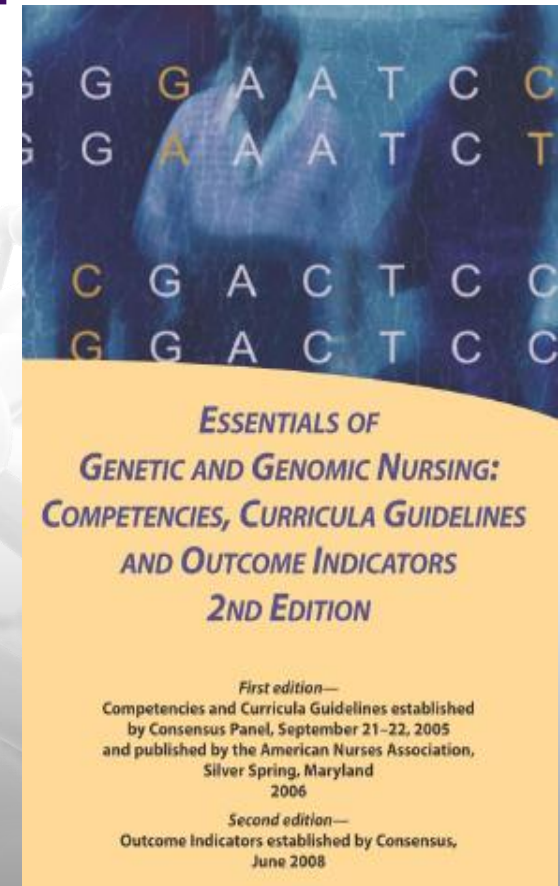
- Use of an individual's genetic/genomic information in addition to traditional health information to guide health care decision-making
- Disease prevention, risk reduction, diagnosis, treatment, symptom management and palliative care
 - Pharmacogenomics
 - Medication selection
 - Dose selection
 - Inhibitors
 - Inducers



Genomic Competencies for Nurses

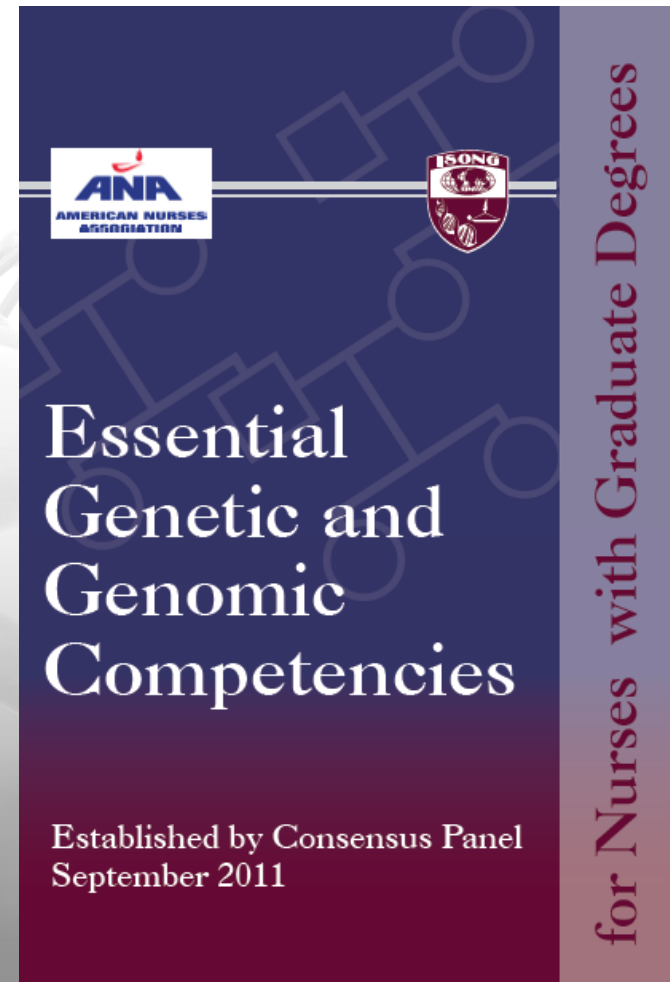
Essentials of Genetic and Genomic Nursing

- 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



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



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

Genomics Special Issue 2013

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



Authors:

Kathleen Calzone PhD, RN, APNG, FAAN

Jean Jenkins PhD, RN, FAAN

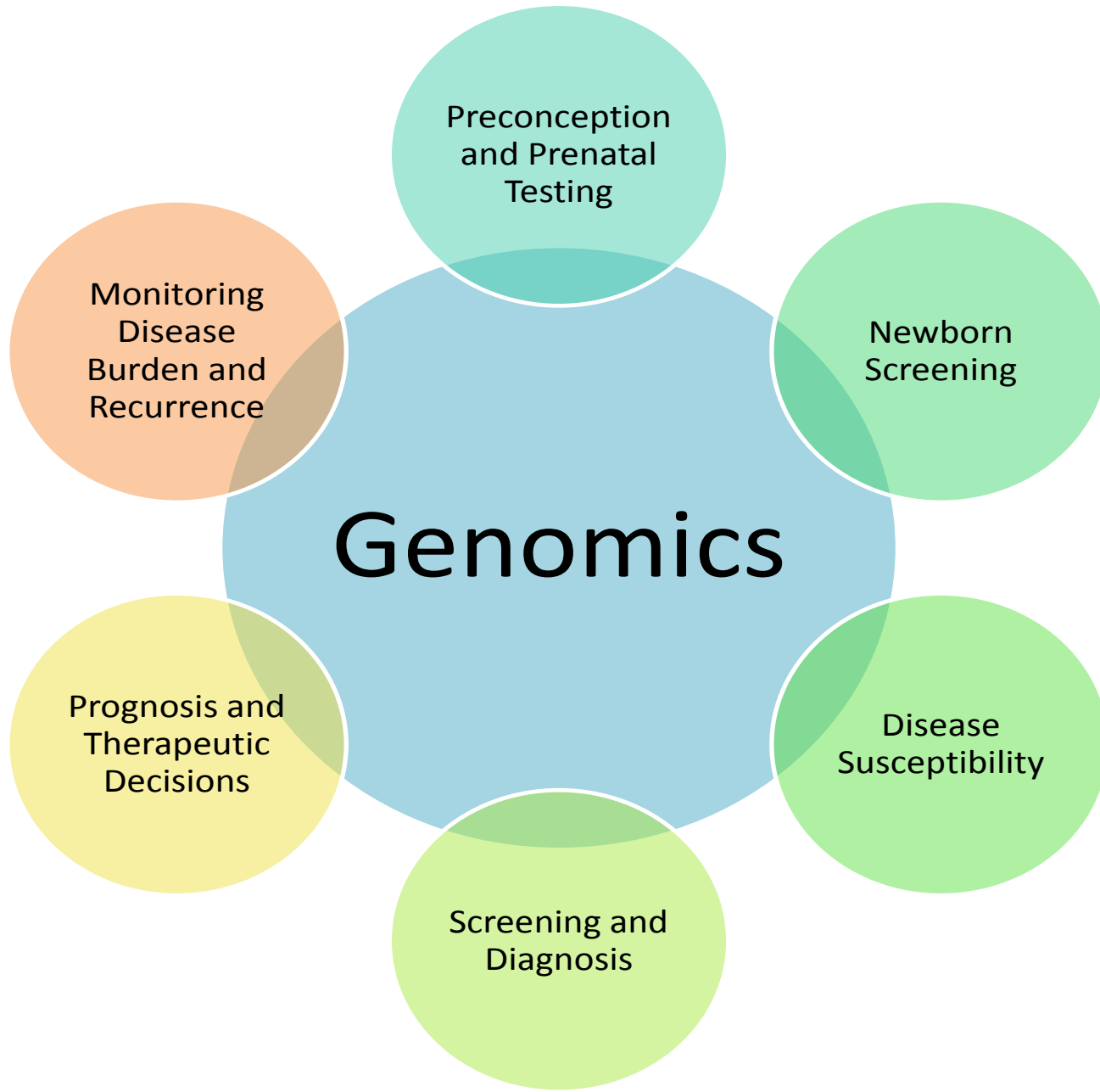
Nick Nicol PhD, FCN

Heather Skirton PhD, RGN, RGC, QMW

W. Gregory Feero MD, PhD

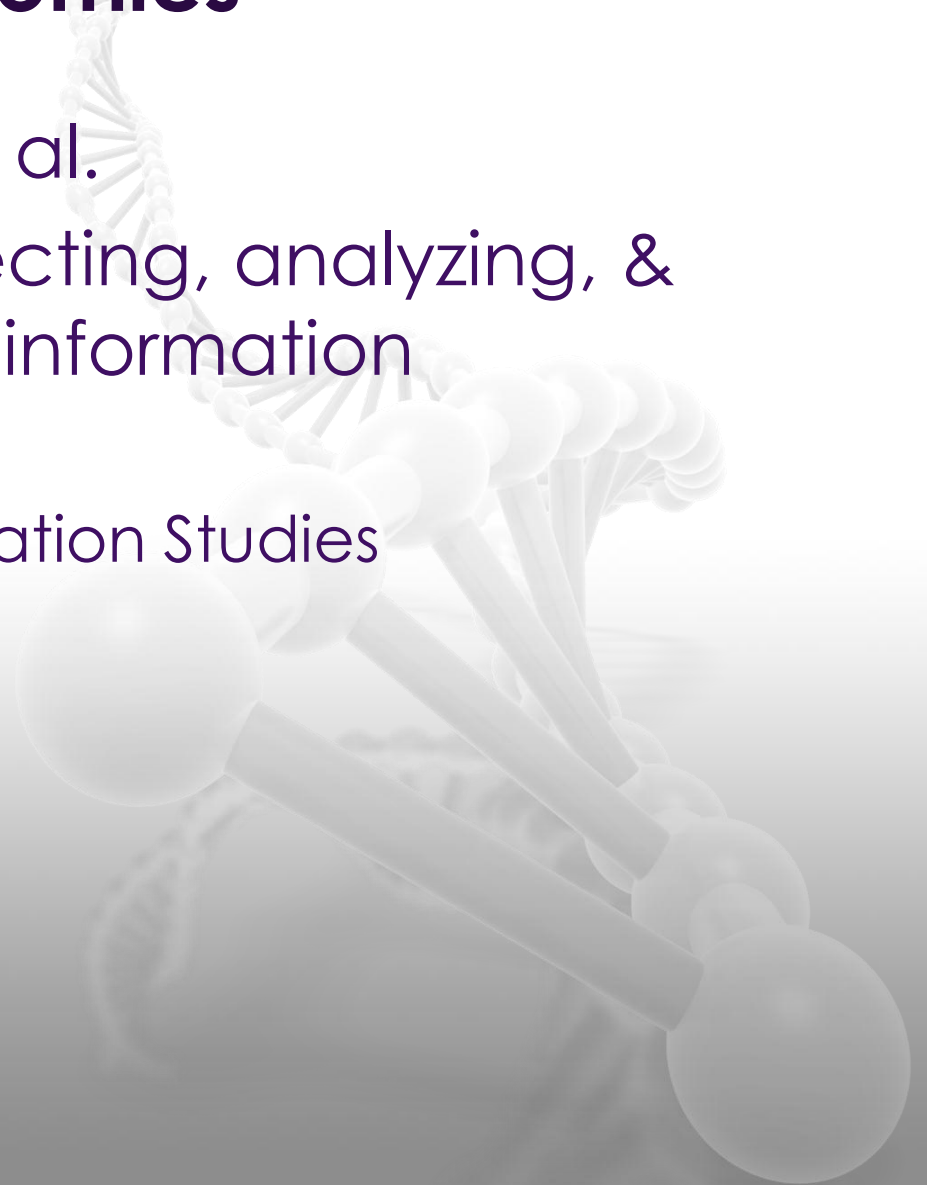
Eric D. Green MD, PhD

Editorial



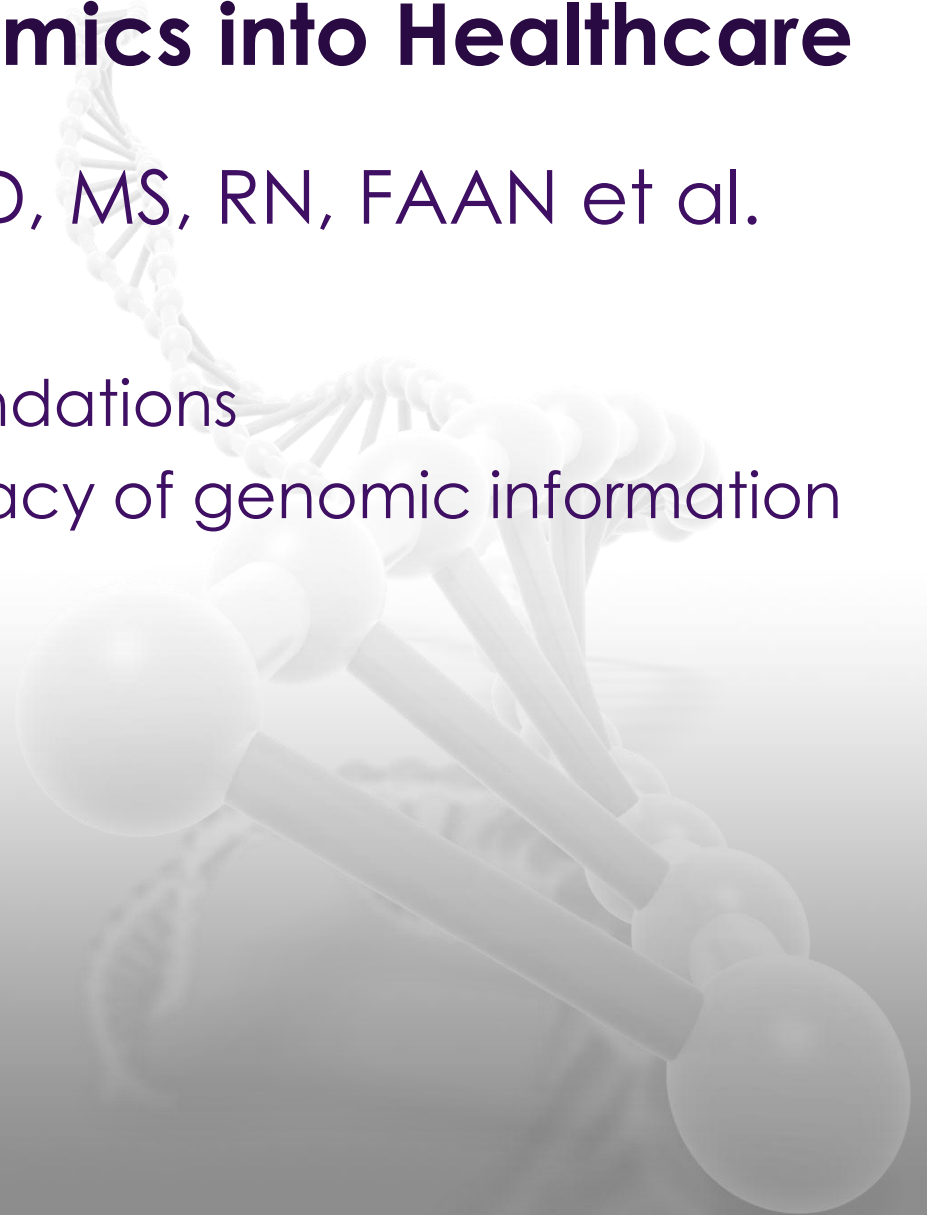
Current and Emerging Approaches in Genomics

- 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

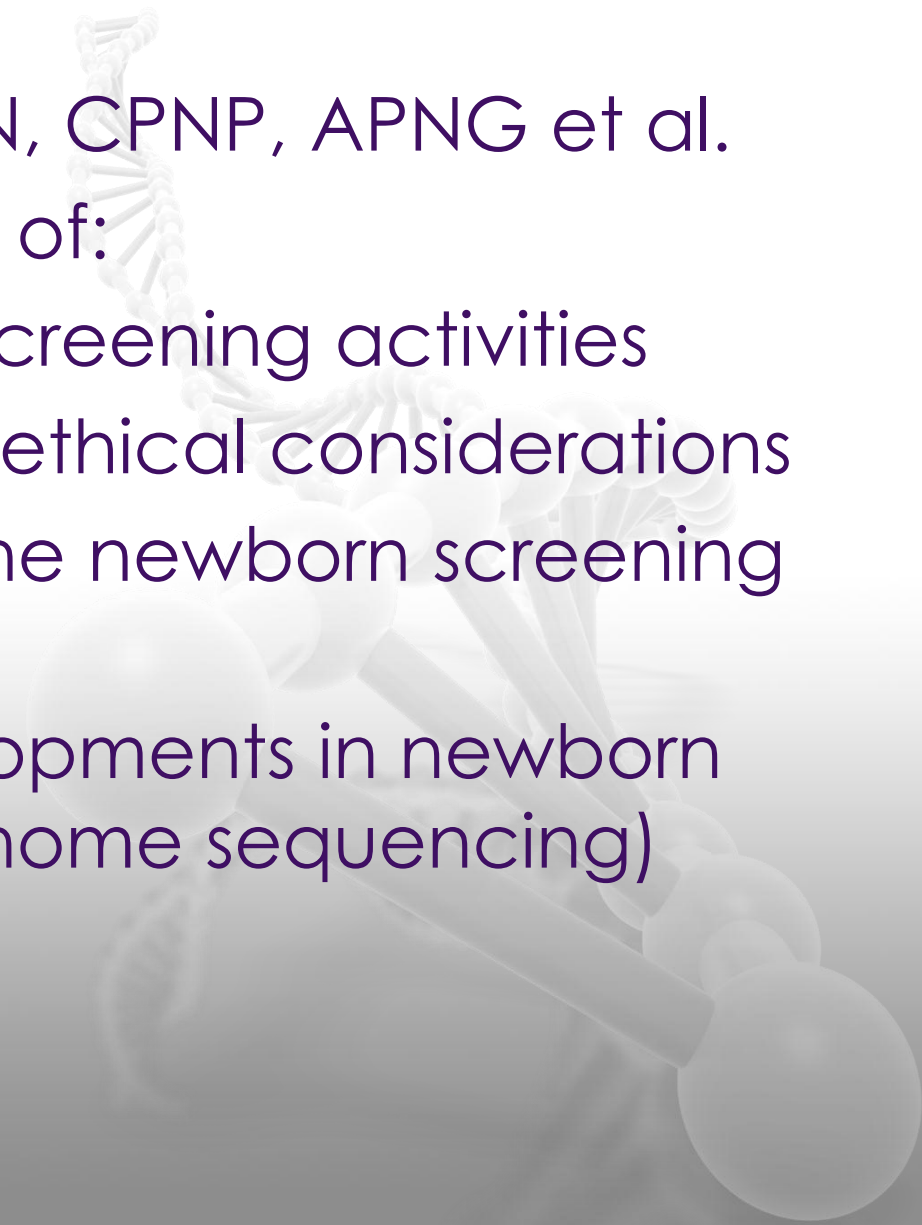


Integration of Genomics in Cancer Care

- 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)
- 

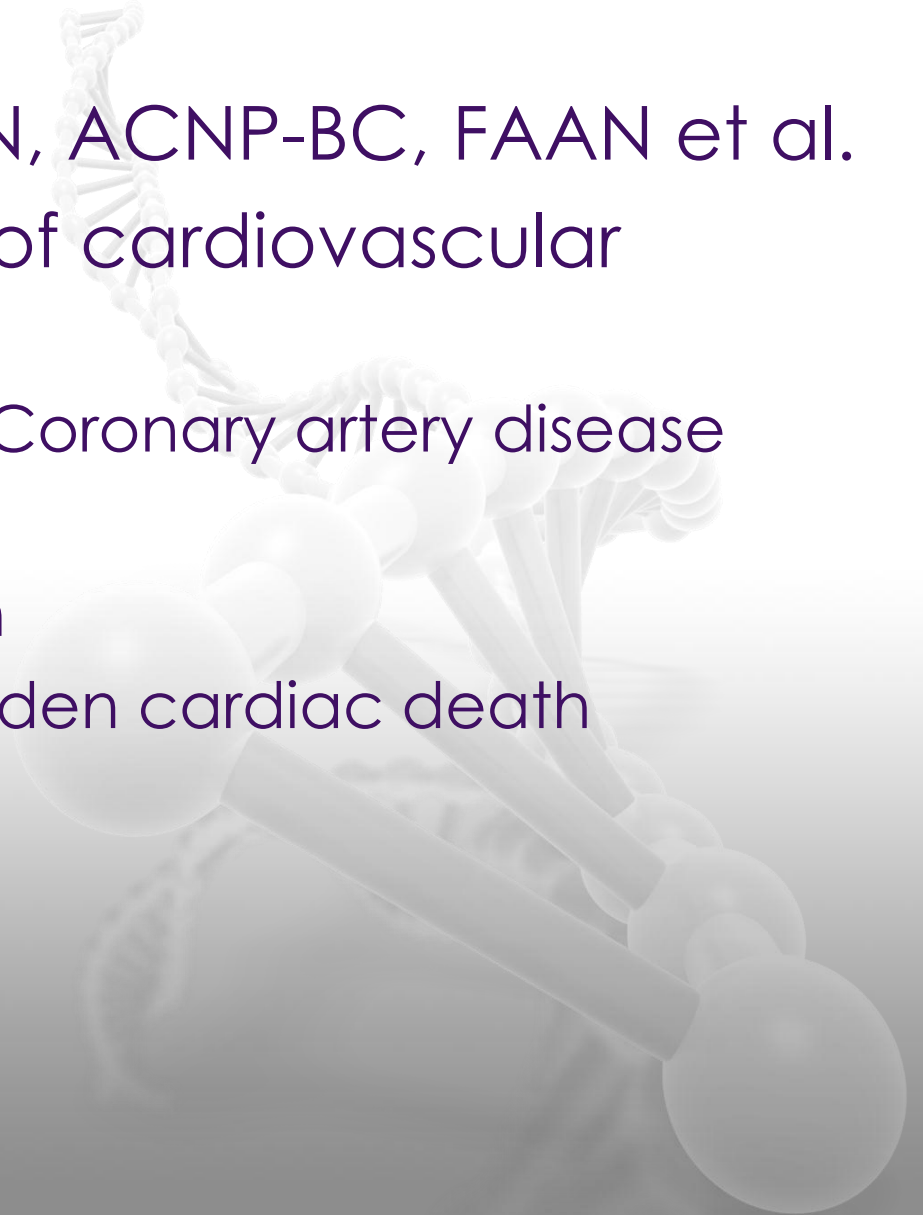
An Update of Childhood Genetic Disorders

- Cynthia Prows, MSN, CNS, FAAN et al
- Provides an overview of:
 - Common childhood genetic disorders
 - Encompasses infants, children and adolescents
 - Important resources and services

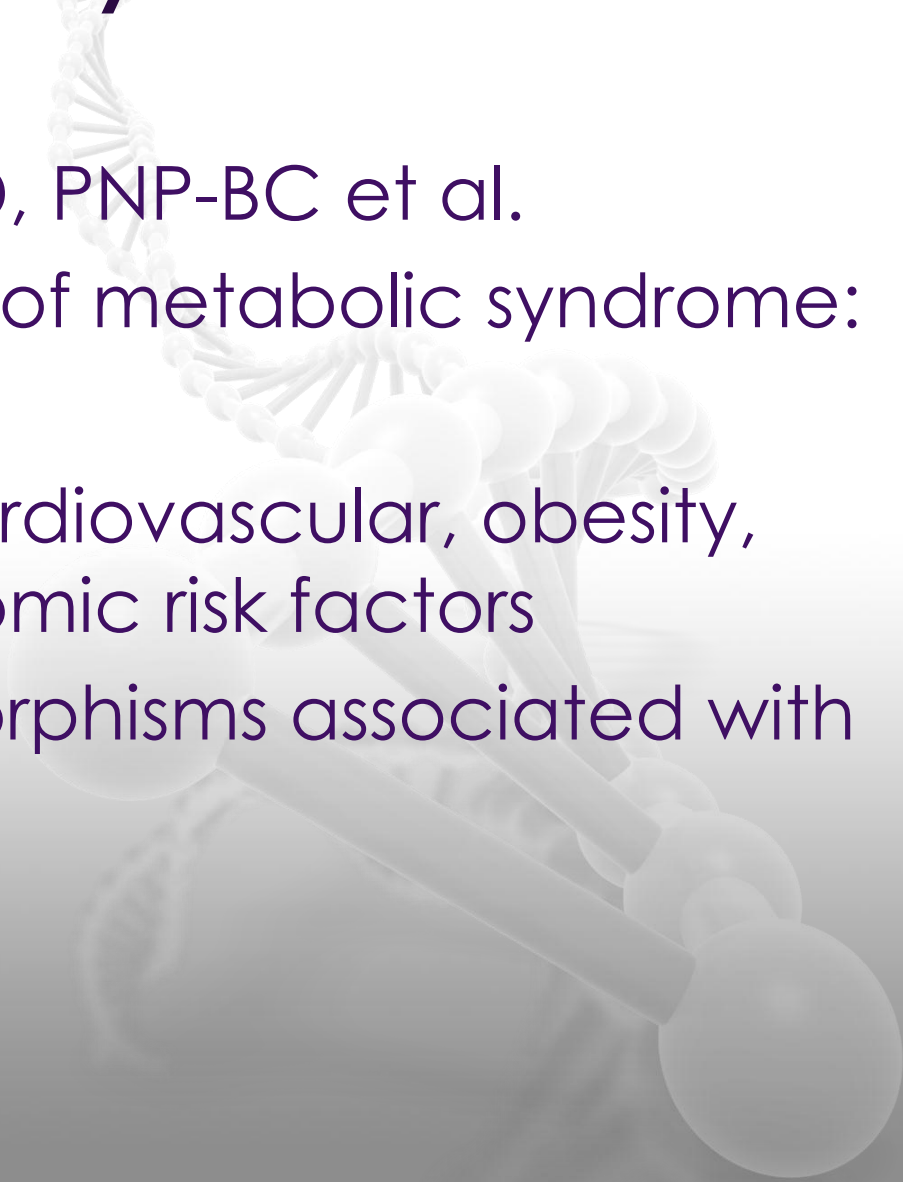


Cardiovascular Genomics

- 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

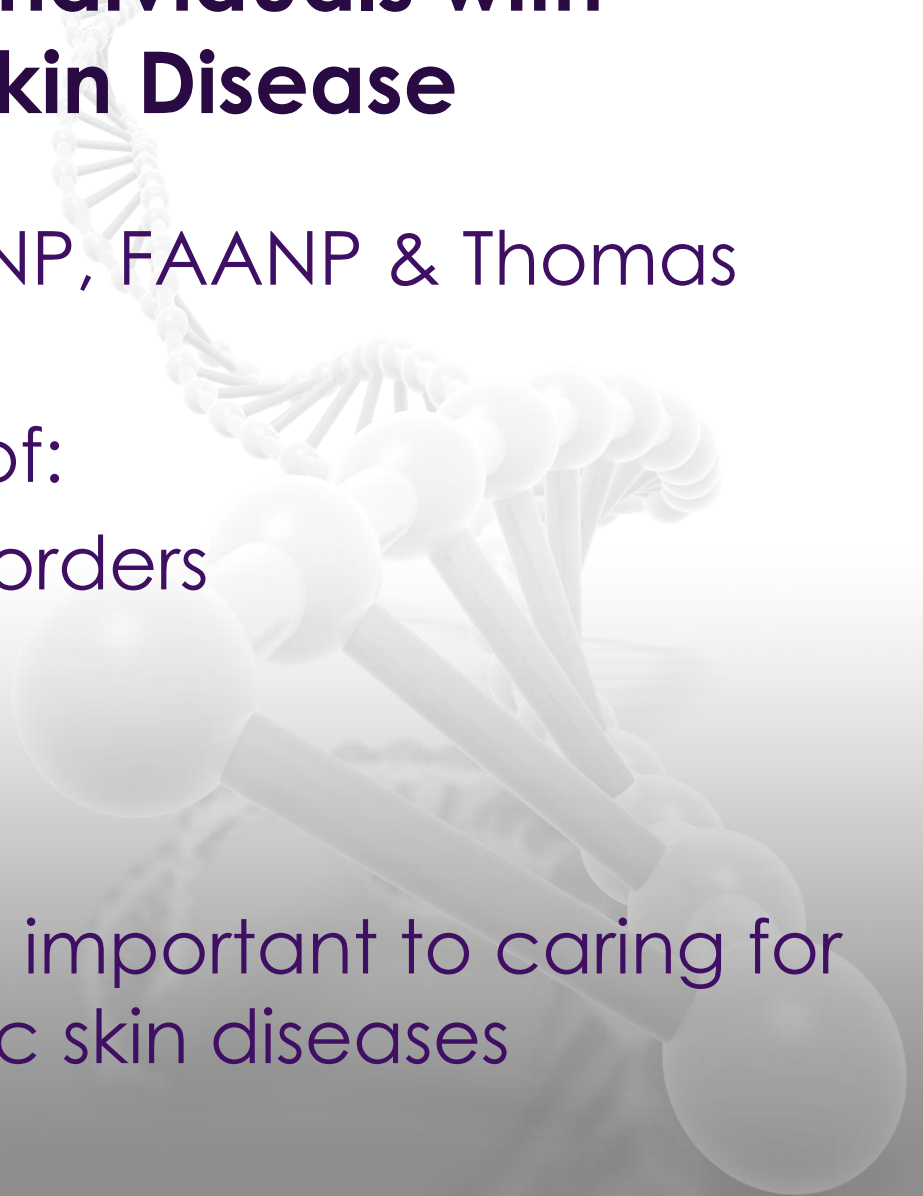


An Overview of the Genomics of Metabolic Syndrome

- 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

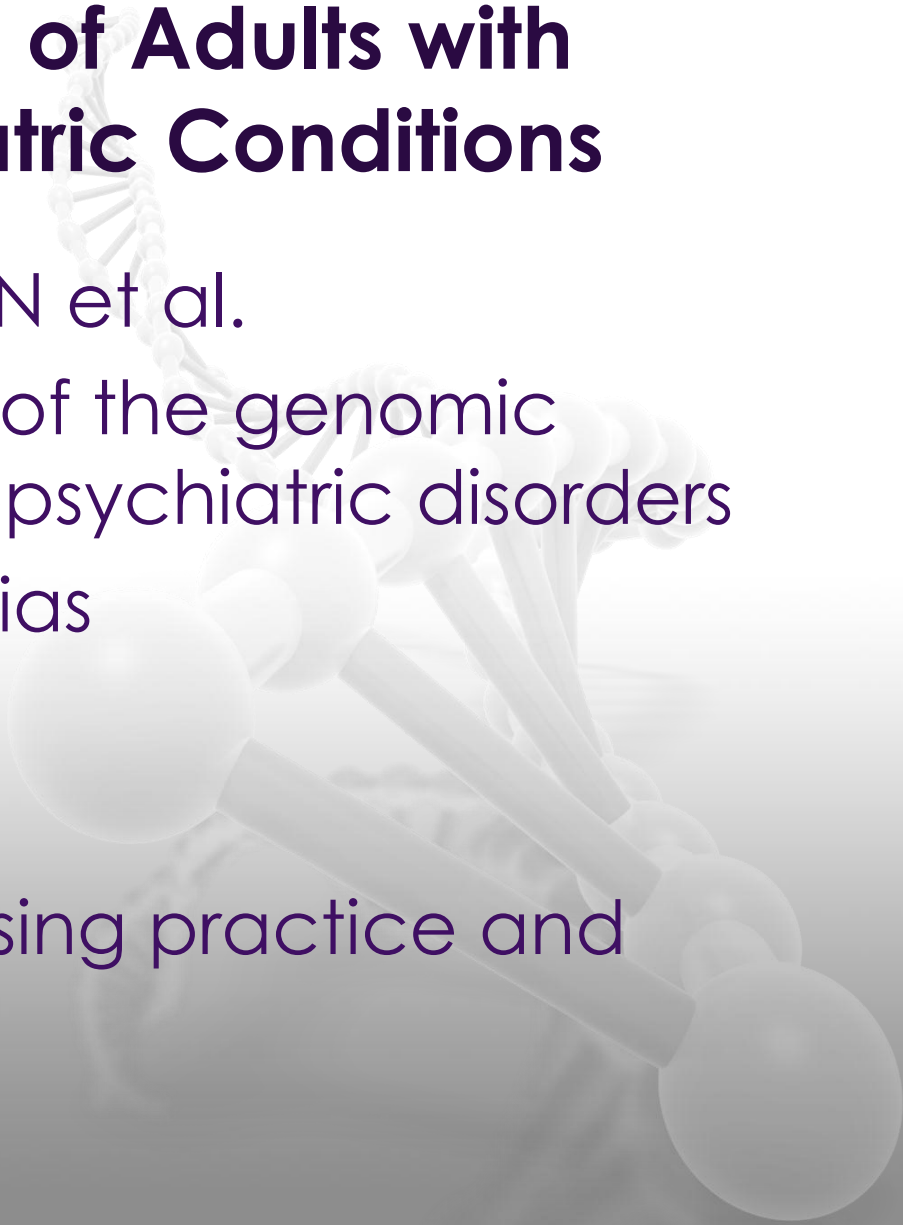
- Diane Seibert PhD, ARNP, FAANP & Thomas Darling MD, PhD
- Provides an overview of:
 - Five genetic skin disorders
 - Inheritance patterns
 - Genomics
 - Treatments
 - Issues and concerns important to caring for patients with genetic skin diseases



Genomics and Autism Spectrum Disorder (ASD)

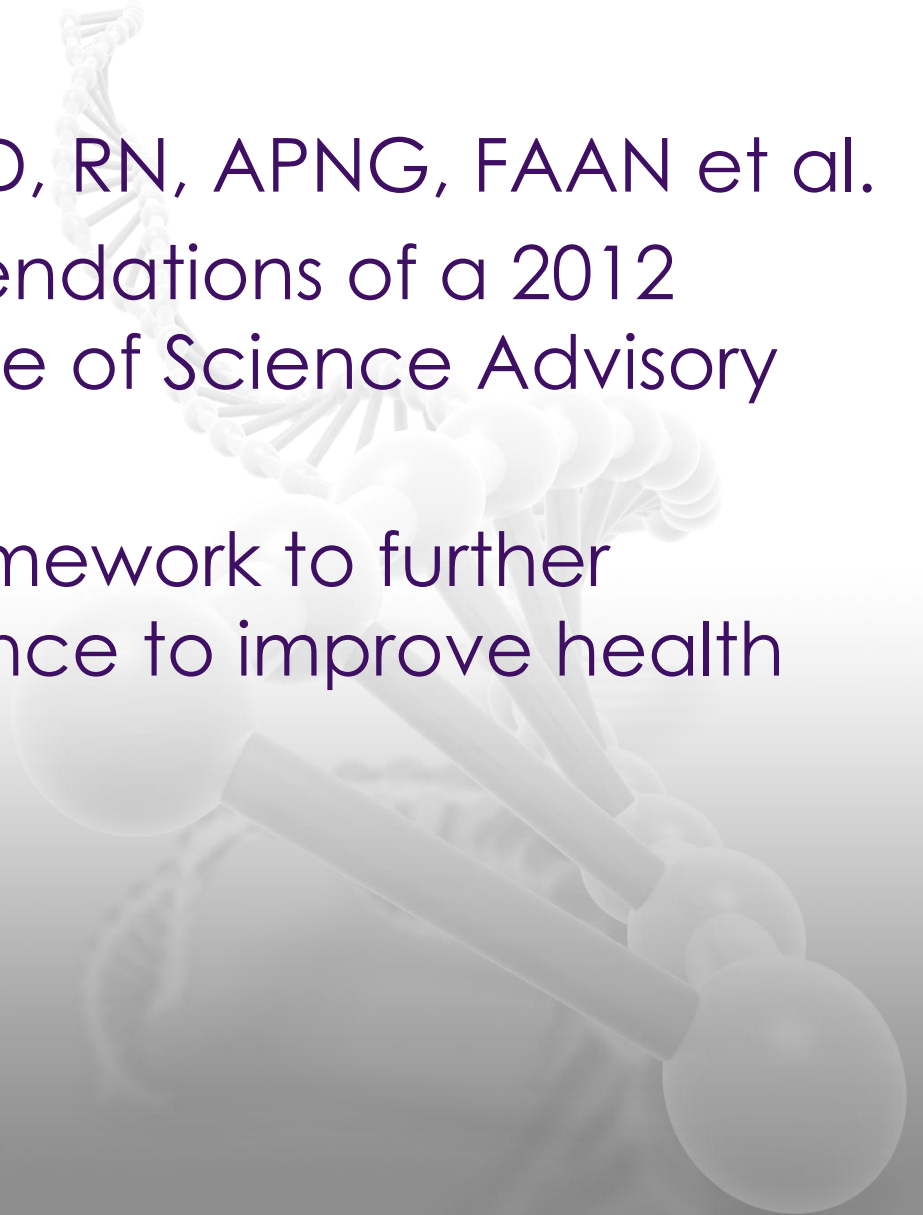
- Norah Johnson PhD, RN, CPNP et al.
 - Provides an overview of ASD:
 - Identification
 - Diagnosis
 - Family implications
 - Genomic contributions to ASD risk
 - Limitations and complexities of ASD research
- 

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>
- **Special Thanks to:**
 - Journal of Nursing Scholarship (JNS)
 - Susan Gennaro RN, DSN, FAAN Editor
 - JNS Editorial Board

Genomics Special Issue 2013

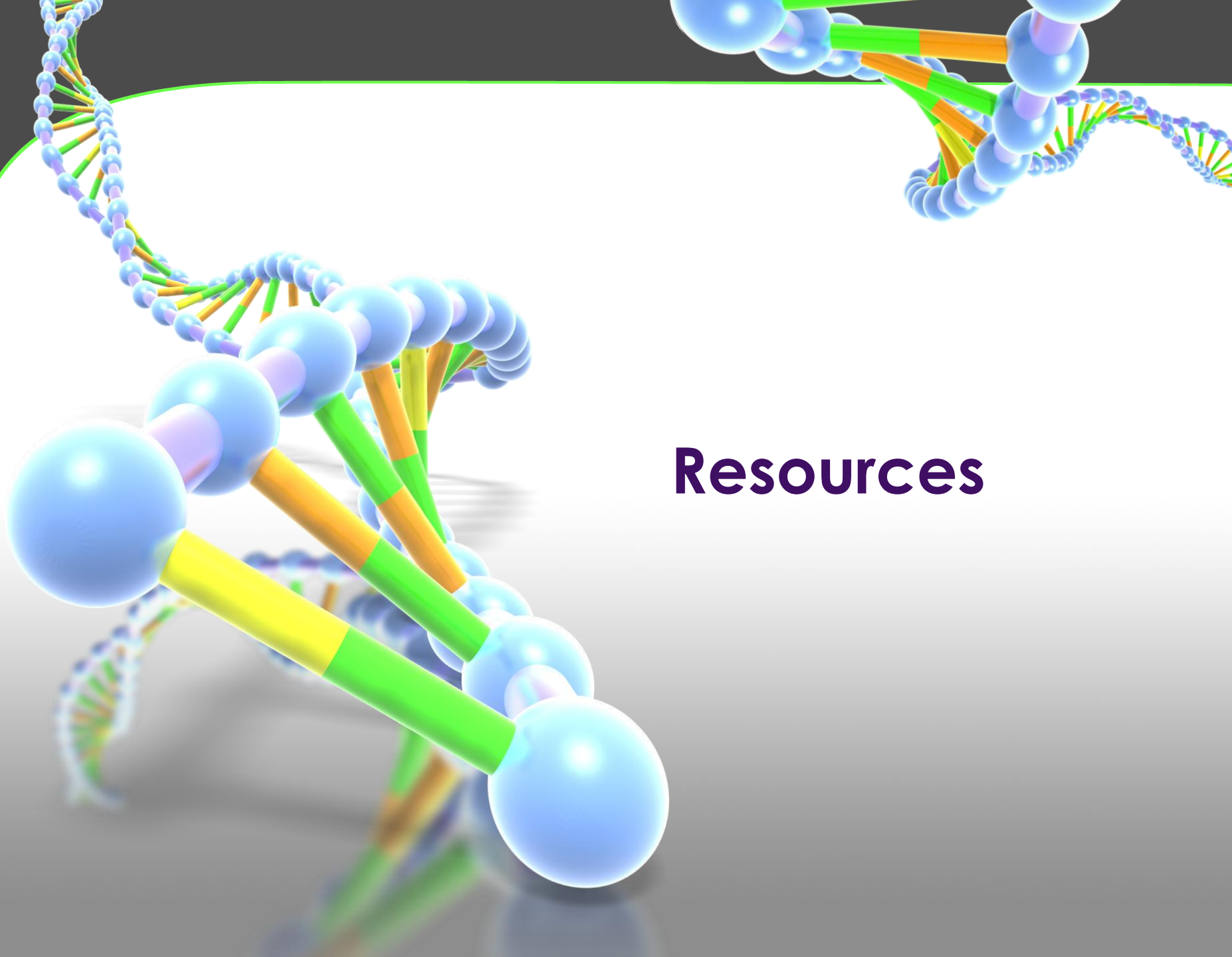


• **Lead Editors:**

- 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:**

- Cynthia Prows, MSN, RN, APNG, FAAN
Cincinnati Children's Hospital
- Diane Seibert, PhD, RN
Uniformed Services University of the Health Sciences
- Greg Feero, MD, PhD
National Human Genome Research Institute



Resources

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/>

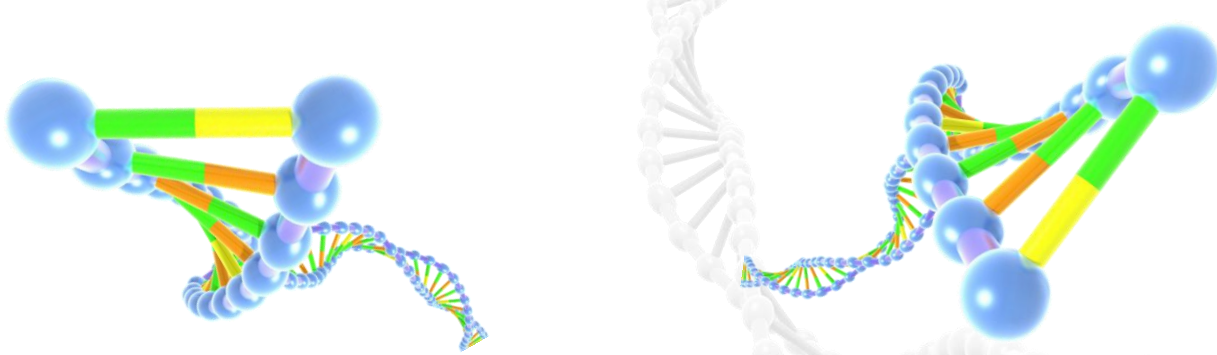
- Competency and Curricular Resources

<http://www.genome.gov/27527634>

The screenshot shows the homepage of the Genetic/Genomic Competency Center for Education. At the top, there is a navigation bar with the GGC logo and links for Home, About the Project, Curriculum Map / Guidelines, Feedback, and Help. Below the navigation bar, there are three main sections: Search, Genetic Counselor, Nurse, and Physician Assistant. The Search section includes a search box and a dropdown menu for selecting a discipline. The Genetic Counselor, Nurse, and Physician Assistant sections each feature a representative image and a magnifying glass icon. Below these sections, there is a section titled 'What you can do on this site' with four sub-sections: Search by Discipline for Genetics/Genomics Resources to Use in Your Classroom, View My Resources Found on This Site, and Submit Resources to be Included on This Site. A small video player is visible on the right side of the page.

The screenshot shows the homepage of the Global Genetics and Genomics Community. At the top, there is a navigation bar with the GGC logo and links for Home, Resources, About GGC, and Faculty. Below the navigation bar, there is a section titled 'To Get Started' with three numbered steps: 1. Click on a Case icon, 2. Read the patient's record, and 3. Start asking questions. Below this section, there is a section titled 'An Unfolding Case Study' with a brief description of the interactive learning experience. The main content area is titled 'Case Studies:' and features six case study cards, each with a patient's name and a 'View >' link. The cases are: Jeff and Maria, Tom, Lisa, Lena, Maria, and Gabi.

Publications



- 2011 Journal of Nursing Scholarship
year long series
 - Genetics/Genomics and
Nursing Education

Search the Glossary

A B C D E F G H I J
N O P Q R S T U V W

- ACGT
- Acquired Immunodeficiency
- Adenine
- Allele
- Amino Acids
- Ancestry-informative Mark
- Animal Model
- Antibody
- Anticodon
- Antisense
- Apoptosis
- Autism
- Autosomal Dominant
- Autosome

Click here to



Talking Glossary of Genetic Terms



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

Listen to a detailed explanation by the experts at the National Human Genome Research Institute's Genetic Information Research

Audio Definitions

Illustrations & 3D Animations

Test your Gene Knowledge

 A collection of small thumbnail images representing the content of the glossary: an audio player interface, a 3D molecular model, and a multiple-choice test question.

Surgeon General Family History Tool

Get Help

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

Use a Saved History

En Español

Em Português

In Italiano



OUTCOME INDICATORS PUBLISHED WITH COMPETENCY 2nd EDITION
K. CALZONE, M.Ed., Ph.D., APRN, FAAN
1 AND 3

ISSUE 4 | VOLUME 3 | YEAR 2009

SPOTLIGHT: ONLINE GENETICS COURSES 2

COMPETENCY WATCH
GENETICS
PUBLICATION UPDATE
CONFERENCE UPDATE
RESEARCH HIGHLIGHTS
JOURNALS
ADDITIONAL PAPER
ADDITIONAL WEBSITES
WEBSITE HISTORY TOOL



Competency focus

NURSING COMMUNITY ON THE IMPLEMENTATION OF NURSING COMPETENCIES AND CURRICULA: GENETICS AND GENOMICS.

The Genetics/Genomics Nursing Competency initiative finalized each of the individual competencies. They are being published in the 2nd Edition of the Essential Nursing Competencies for Genetics and Genomics.

Indicators Published with Second Edition

Domain: Professional Responsibilities		
Recognize when one's own attitude and values related to genetic and genomic science may affect care provided to clients		Clinical Performance Indicators Engage in reflective practice about one's own beliefs and values related to client care that integrate genetic and genomics.
Specific Areas of Knowledge Values, attitudes and beliefs that influence genetic and genomic science e.g. views on pregnancy termination.		Clinical Performance Indicators Articulate one's attitudes, values and beliefs that influence one's perspective about difficult genetic or genomic health care decisions.
Impact of personal values, attitudes and beliefs on genetic and genomic health care.		
Advocate for clients' access to desired genetic/genomic services and/or resources including support groups.		Clinical Performance Indicators Demonstrate appropriate care and concern for all clients throughout their referral, provision of direct care and follow-up to genetic services.
Specific Areas of Knowledge Access for healthcare professionals and lay clients about disease susceptibility, diagnostic conditions, treatment, and uses e.g. testing literature, evidence-based practice such as the National Human Genome Research Institute (www.nhgri.nih.gov) and the Centers for Control National Office of Public Health (www.cdc.gov/genetics/default.htm).		Clinical Performance Indicators Demonstrate knowledge about accessing local/regional genetic and/or genomic resources. Include genetic health care professionals in team building and collaborative strategies to optimize client outcomes. Help client navigate system barriers that limit access to genetic/genomic services including access to clinical trials.
Advocate for genetic and/or genomic referrals to community.		Clinical Performance Indicators Identify strategies that could be used to facilitate reimbursement for access to genetic/genomic services and/or tests.

(continued on page 3)

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ESSENTIAL GENETIC AND GENOMIC COMPETENCIES FOR NURSES WITH GRADUATE DEGREES
WRITTEN BY KAREN GRECO 1

SPOTLIGHT: Pharmacoe Education in the Era of Genomic Medicine
WRITTEN BY GRACE FLO 2

NEW OPPORTUNITY: PUBLICATION UPDATE 3

PARADIGM SHIFT: JNS ISSUES 4

TALKING GLASSARY LAUNCHES MOBILE APP 4

CDC OFFICE OF PUBLIC HEALTH GENETICS 4

DNA DAY 2012 4

NOVEMBER: NURSE GENOMICS 4

GRADUATE COMPETENCIES GO 4

Competency focus

UPDATING THE NURSING COMMUNITY ON THE IMPLEMENTATION OF THE ESSENTIAL NURSING COMPETENCIES AND CURRICULA: GENETICS AND GENOMICS.

Essential Genetic and Genomic Competencies for Nurses With Graduate Degrees

The advancement of genomic information and technology continues to merge into the fabric of healthcare, transforming how healthcare services are defined and delivered. Consequently all nurses must have the fundamental genomic competencies necessary to provide competent genomic health care. In addition, nurses educated at the graduate level need to be prepared to assume clinical and leadership roles in healthcare systems into effective healthcare and genomic advances in translating these into the Essential Nursing Genetics and Genomics Guidelines for Nurses with Graduate Degrees for all nurses were established by AACN in 2005, published in 2006 and a second edition published in 2009 with the addition of outcome indicators. In 2009, these competencies served as a foundation for the Association of Colleges of Nursing (ACN) facilitating the integration of genomics into the Association of Baccalaureate Education for Essential of Baccalaureate Education for Advanced Practice and the revision of the Essential of Masters Education for Advanced Practice and the Genetics/Genomic Nursing Competency Initiative leaders recognized the need to facilitate the integration of genomics into the revised Essential of Genomics (Competency Focus, <http://www.genome.gov/Pages/Health/Health/Genetics/Info/Competencies/Competency2010.pdf>). It became evident that the essential genomic competencies that build on the essential genetic/genomic competencies and inform the education and practice of nurses with graduate degrees were needed. This led to the development of the "Essential Genetic and Genomic

Competencies for Nurses With Graduate Degrees," finalized by consensus panel in September 2011. These competencies are in the process of being published on the American Nurses Association's Primary American/World website under the Ethics, Nursing/World website under the Ethics, Genetics & Genomics section. The primary purpose of these graduate level Genetics & Genomics competencies is to establish essential genetic and genomic competencies for individuals prepared at the graduate level in nursing. The overarching goal is to improve the genetic/genomic competence, academic and research leadership roles. These competencies apply to all individuals functioning at the graduate level in nursing, including but not limited to advanced practice registered nurses (APRNs), clinical nurse leaders, nurse educators, nurse administrators, and nurse scientists. These practice registered nurses (APRNs), clinical nurse leaders, and nurse scientists. These graduate level competencies build upon and are complementary to the 2009 Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators at the graduate level that nurses functioning at those competencies. that nurses functioning at those competencies. have already acquired in the Spring of 2009 Steering Committee, comprised of Karen Greco, Sue Tinley, and Diane Seibert was formed to develop these graduate level competencies. Development of the initial draft competencies included review and analysis of the following documents:

- published literature concerning what nurses prepared at the graduate level need to know about genetics/genomics

- genetic/genomic content on eleven APRN credentialing exam materials
- other key nursing documents
- Based on the above review and analysis, the Steering Committee identified common themes among key documents and created an initial draft of 51 competencies. An advisory board of nursing leaders and genetics experts was created to review and revise the draft document. Representatives from diverse number of nursing and advanced practice nursing organizations were later added to create a Consensus Panel, consisting of 31 genetics experts and other nursing communities and APRN organizations. The draft competencies document was posted on the American Nurses Association website for public comment in the Fall of 2010. The competencies were revised and the revised competencies were sent to the feedback panel for validation. A Consensus Panel for validation was formed in December of 2010 using Survey Monkey. Based on the survey feedback the competencies that did not achieve consensus were revised, consolidated or deleted due to duplication. A second survey was sent out in 2011 for

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THE ESSENTIAL NURSING COMPETENCIES FOR NURSES WITH GRADUATE DEGREES
K. CALZONE, M.Ed., Ph.D., APRN, FAAN
1

NEW FACILITY IN UNDERREQUIREMENTS
FOR REVISION
IN GENETICS, Ph.D., RN, FAAN 2

KEY MESSAGES 2

HEALTHCARE 2

Competency focus

NURSING COMMUNITY ON THE IMPLEMENTATION OF NURSING COMPETENCIES AND CURRICULA: GENETICS AND GENOMICS.

Implementation Are We Now?

Strategic Implementation Plan has been in place, but progress has been made. Please contact one of the Competency Initiative leaders if you would like to be involved!

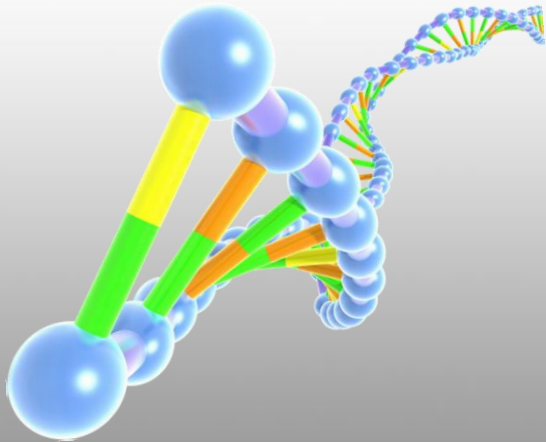
- Association of Colleges of Nursing (ACN) practice change survey meeting
- Signa Three Ten, National Nursing Staff Development Organization, American Association of Colleges of Nursing, American Association of Genomics (AACG), International Society for Nurses in Genetics, National League for Nursing
- Develop a genetic/genomic chapter for the Core Curriculum for Population/Genetics/Genomics (Genetics) for their competency
- Signa Three Ten, American Nurses Association
- Launched for inclusion of the Competency in the newly developed Breast Care certification to be offered by the Oncology Nursing Certification Corporation
- Completed a series of genetic/genomic in-person meetings in the Journal of Pediatric Subspecialty
- Topic: new genetics.gov/7751979
- All names will have a descriptive of knowledge in basic to nursing practice
- Developed and received funding for a meeting with AACN to establish implementation plan. Endpoints: organization to update to the highlighted text tomorrow. If you would like to be involved, please contact us at calzone@acn.org or Joan.Hall@acn.org
- Participated and presented in the Competency at the American Association of Colleges of Nursing Emerging Emerging
- Developed the Competency in all 1,000 Schools of Nursing and received approval for a session at the American Association of Colleges of Nursing Baccalaureate Education Conference November 2007
- Partnered with the Human Resources and Service Advancement on issues of Nursing to identify opportunities for finding practice aspects who would be willing to present
- Developed
- GOAL: Replicate Quality Control Genetics and genomics is included practice content on membership of quality health care outcomes resulting in evidence research practices that will serve as the underpinning for the development of research that produces the strongest evidence base needed to incorporate genetic and genomics into NCCLEX and
- Director of the Competency in all 50 US and territories in 10 years to complete the competency implementation plan. Endpoints: organization to update to the highlighted text tomorrow. If you would like to be involved, please contact us at calzone@acn.org or Joan.Hall@acn.org



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

Questions/Discussion

calzonek@mail.nih.gov
301-435-0538

jean.jenkins@nih.gov
301-496-4601

