Journal of Nursing Scholarship 2013 Genomic Special Issue

Relevance of Genomics to Healthcare and Nursing Practice

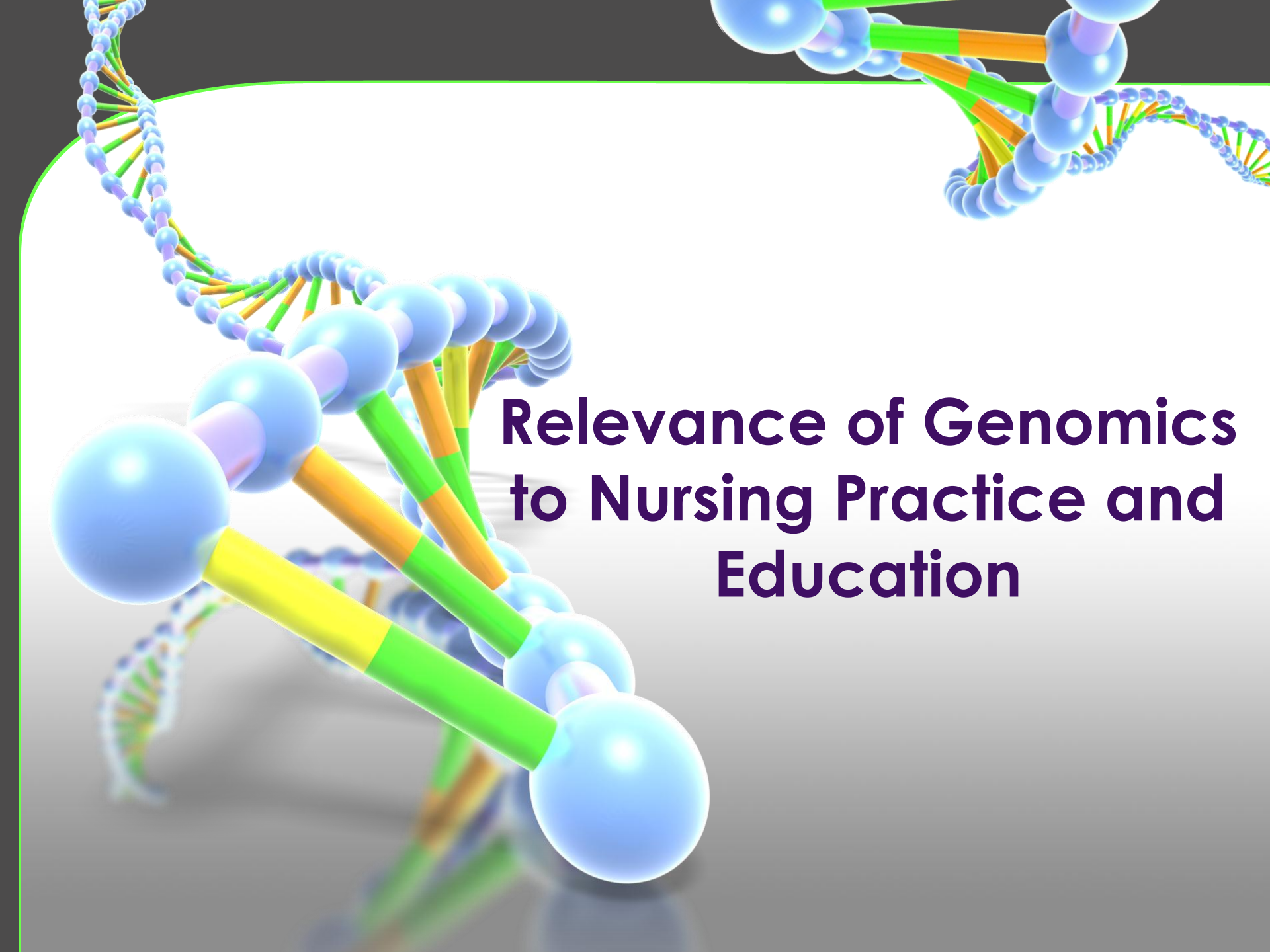
Overview of the Webinar

- 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



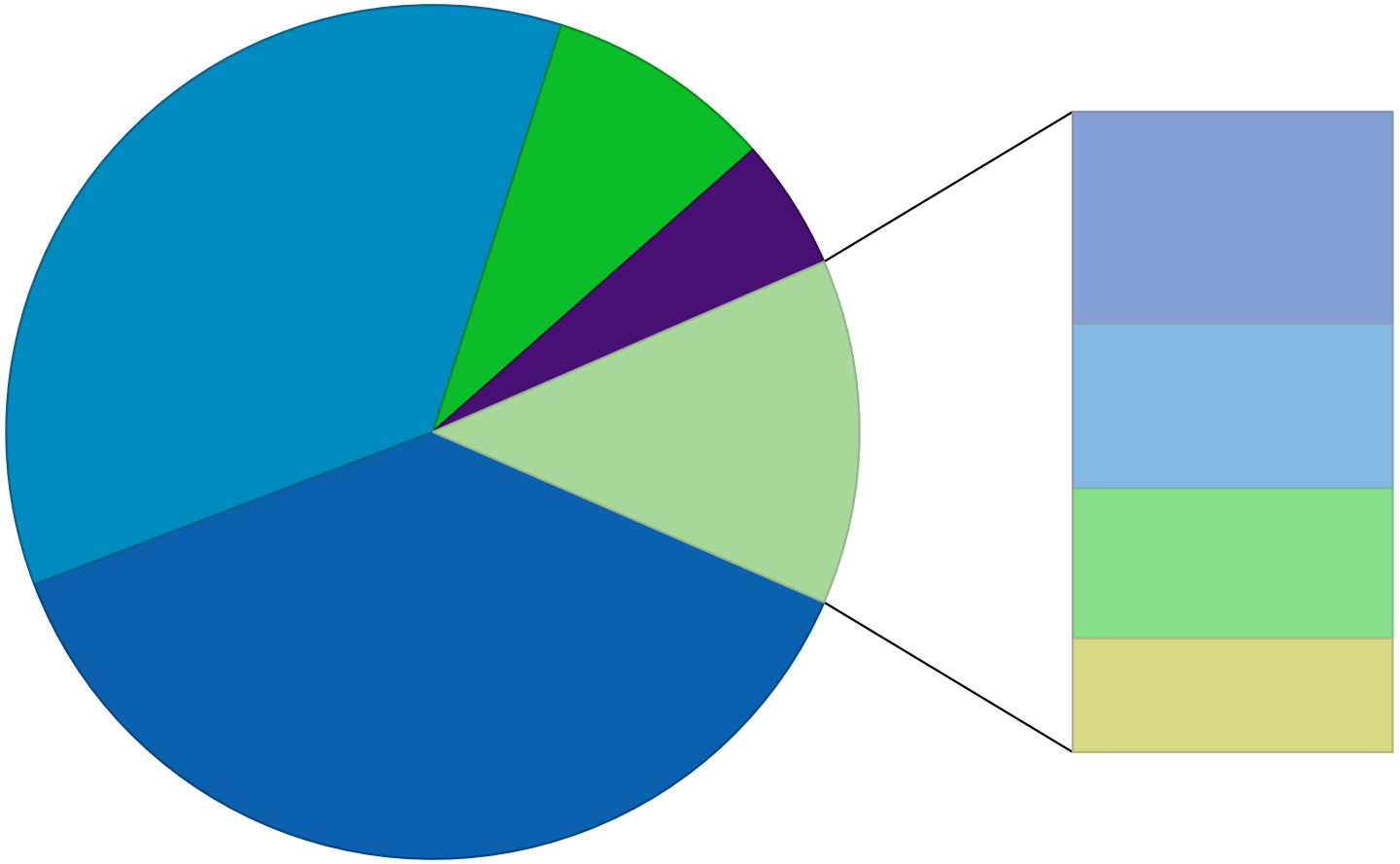
Relevance of Genomics to Nursing Practice and Education

Definitions

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

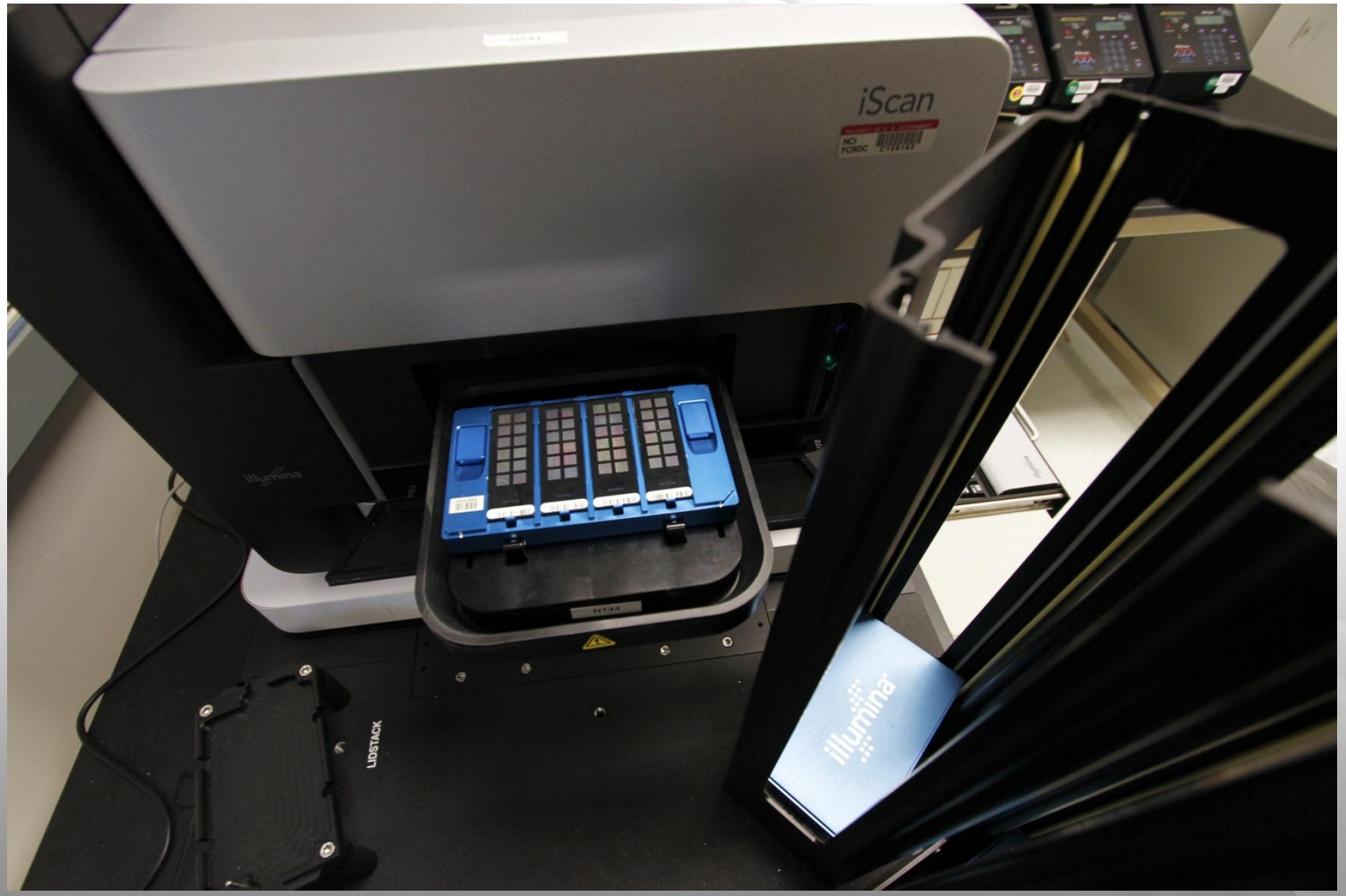
- **Genomics** – study of all the genes in the human genome together, including their interactions with each other, the environment, and other psychosocial and cultural factors

Top 10 Leading Causes of Death

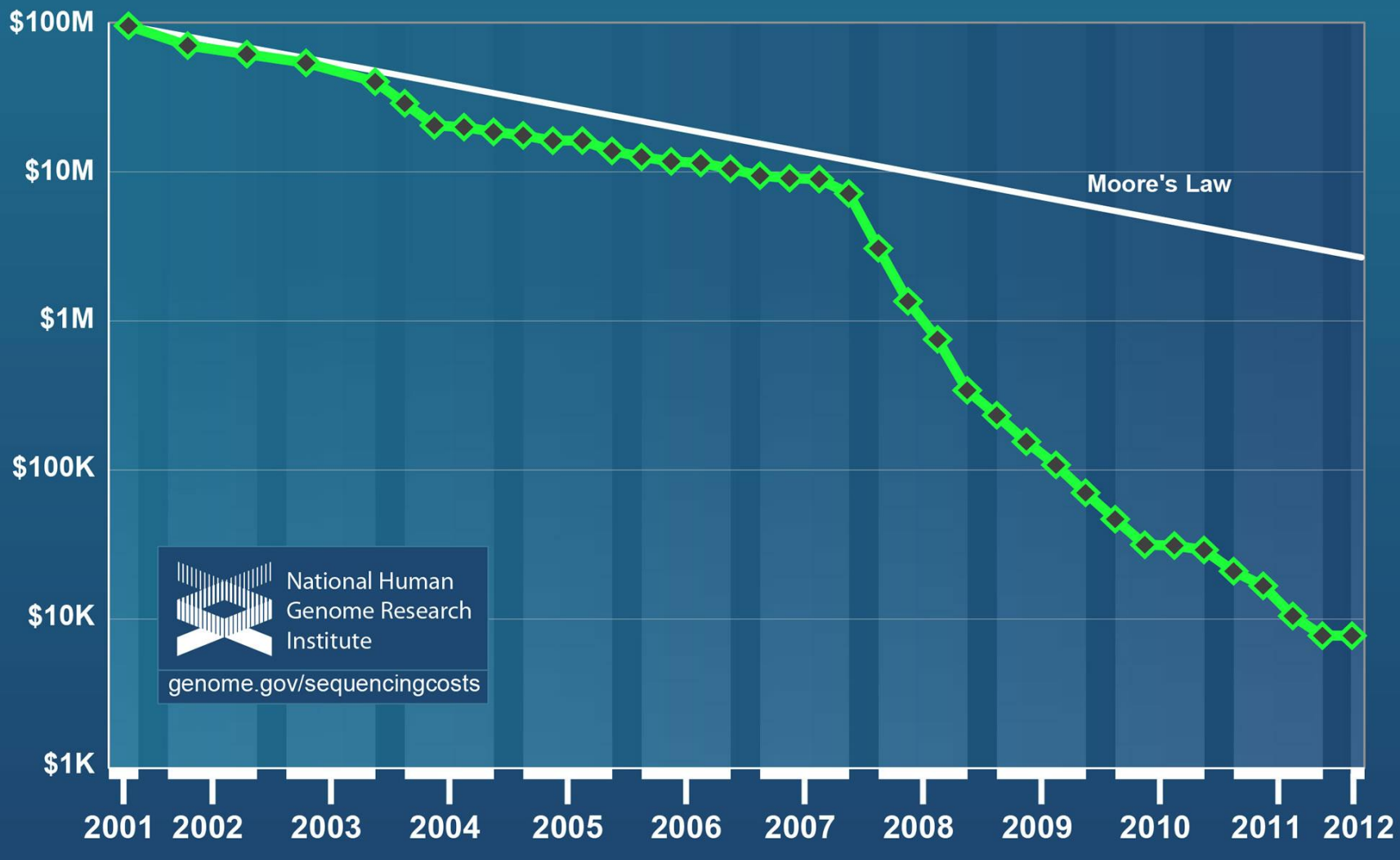


- Heart Disease 24.8%
- Malignant Neoplasms 23.5%
- Chronic Respiratory Disease 5.7%
- Cerebrovascular 5.3%
- Unintentional Injury 4.8%
- Alzheimer's Disease 3.3%
- Diabetes Mellitus 2.9%
- Influenza & Pneumonia 2.2%
- Nephritis 2.0%
- Suicide

Emerging Science/Technology

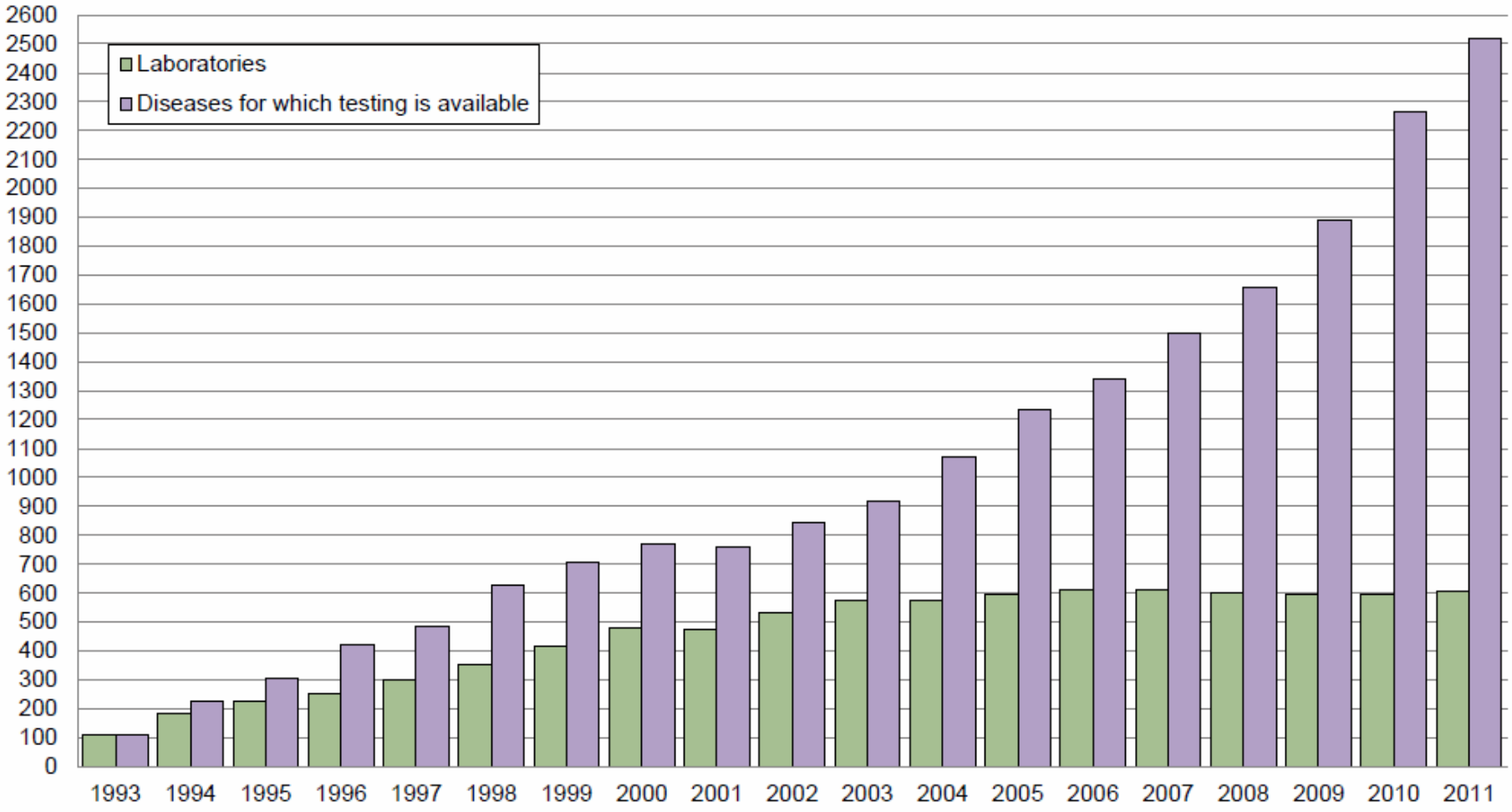


Cost per Genome



http://www.genome.gov/27541954

GeneTests: 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. Kuehn

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 Institute in Hinxton, England, and the Beijing Genomics Institute in Shenzen, 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.

The project will reveal variation in the human genome at a higher resolution than previously possible, documenting single nucleotide polymorphisms and larger genetic variations, such as copy number variations. It also will produce a database that scientists will be able to search for variations in regions of the genome that have been linked to disease. Researchers will then be able to further "interrogate" those data to identify disease-causing variations and elucidate the underlying disease pathways, explained David Valle, MD, director of the Institute of Genetic Medicine at Johns Hopkins School of Medicine in Baltimore.

For example, Valle said, retinitis pigmentosa has been linked to variations in more than 100 genes; the fact that all these genes are involved in the phototransduction cascade suggests that targeting this pathway might be a fruitful avenue to pursue for potential interventions. □

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

NHGRI Funding Opportunities: Resea

- NHGRI Funding Opportunities
- NIH-Wide Parent Funding Opportunities
- NIH-Wide Topic-Specific Funding Opportunitie

The National Institutes of Health (NIH) is replacing pa 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

Genomics Law Report

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

HOME ABOUT SUGGEST A TOPIC CONTACT US

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 odel). Illumina's own product page for the HiSeq 2000 age (~30x) and read length (2×100 bp). There have also been equipped with an iPhone user interface, a concept that Flatley cs Show.

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

Genome Medicine

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

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 »

equence machine: The Ion Proton Sequencer
lp.
e Technologies

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-ased biotech company Ion Torrent, predicted that by 2013 his company would develop a chip iat could sequence an entire human genome.

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

t $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.

SCIENTIFIC AMERICAN™

Winner of the 2011 National Magazine Award for General Excellence

News | Health

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

Single nucleotide polymorphism (SNP)

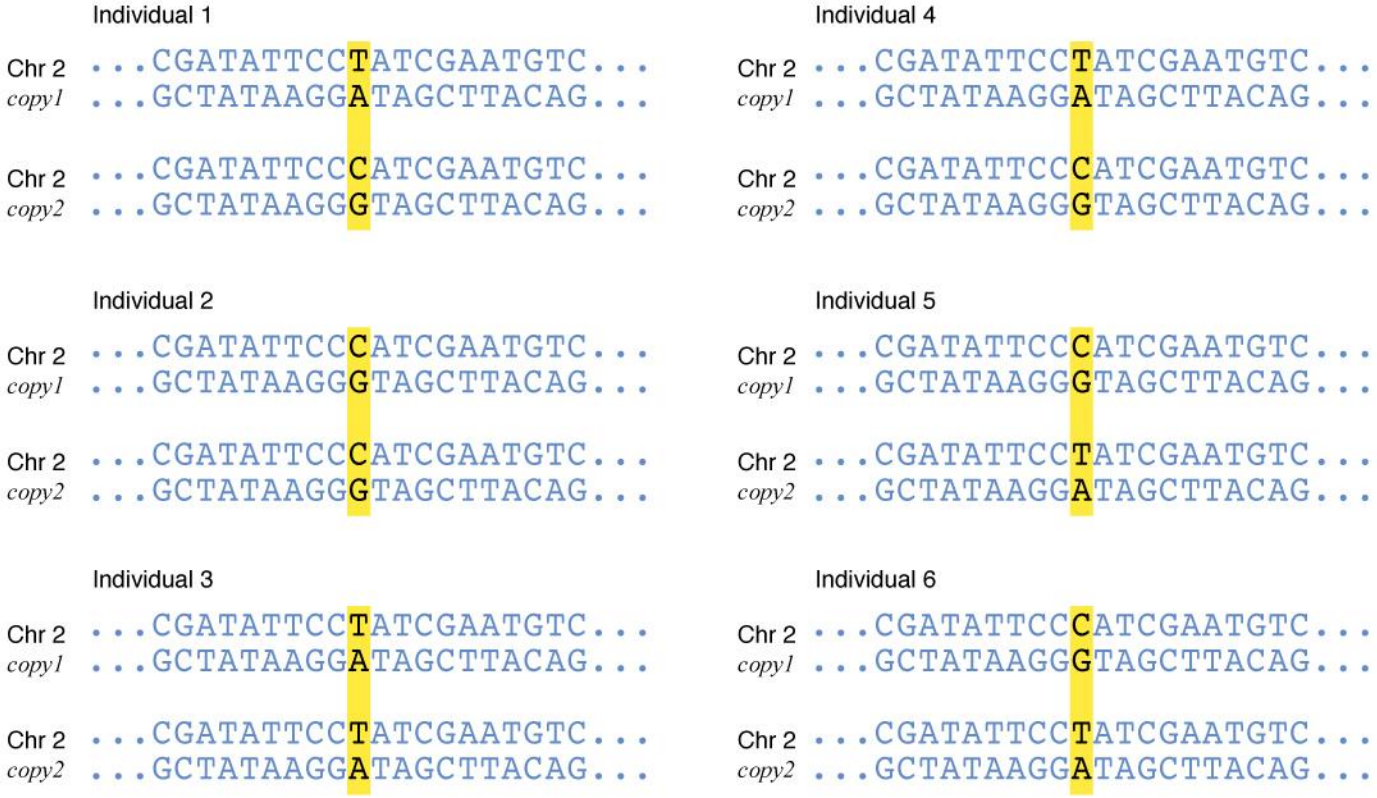
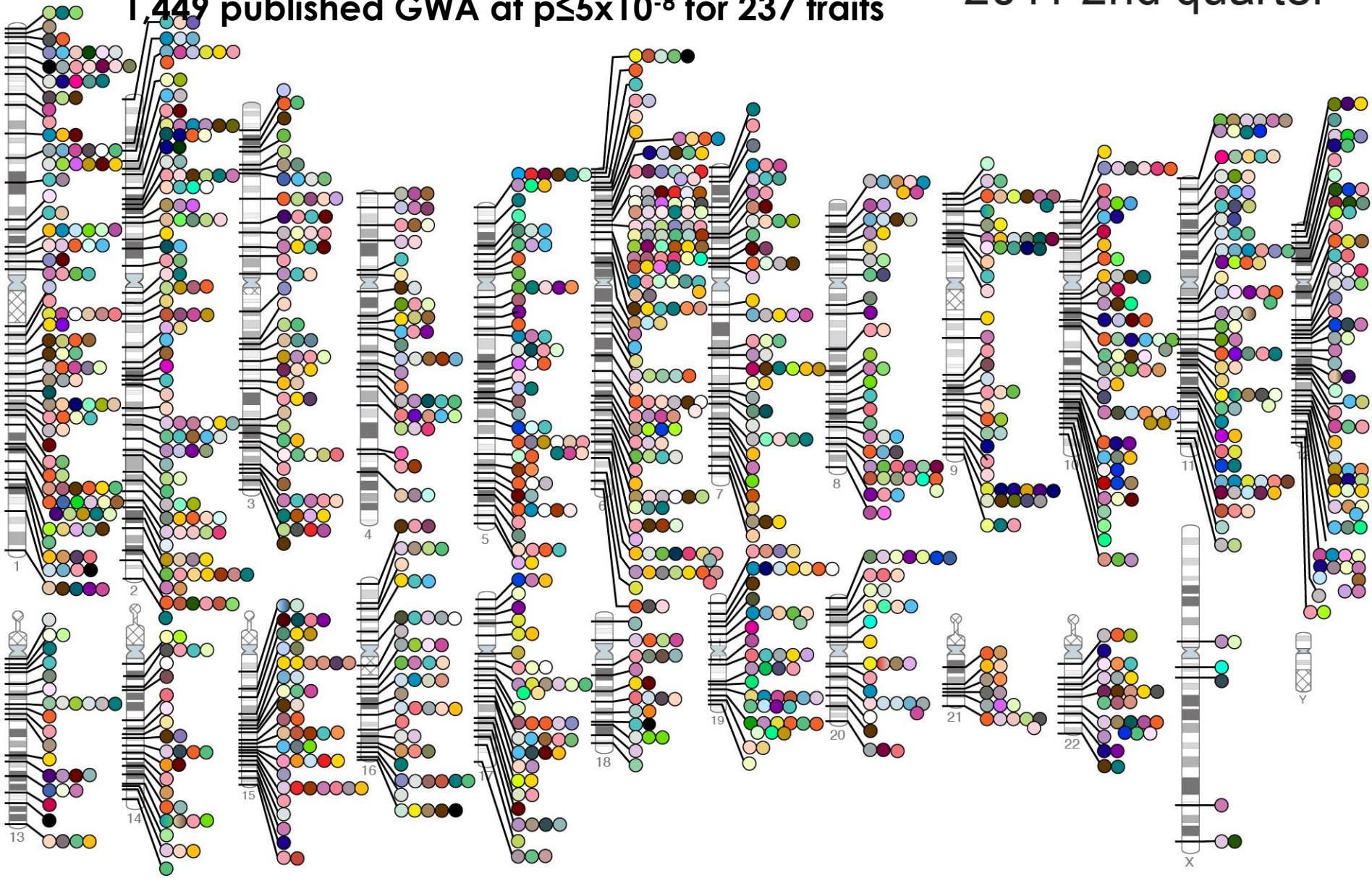


Illustration by Darryl Leja, NHGRI

Published Genome-Wide Associations through 06/2011,
1,449 published GWA at p≤5x10^{-8} for 237 traits

2011 2nd quarter

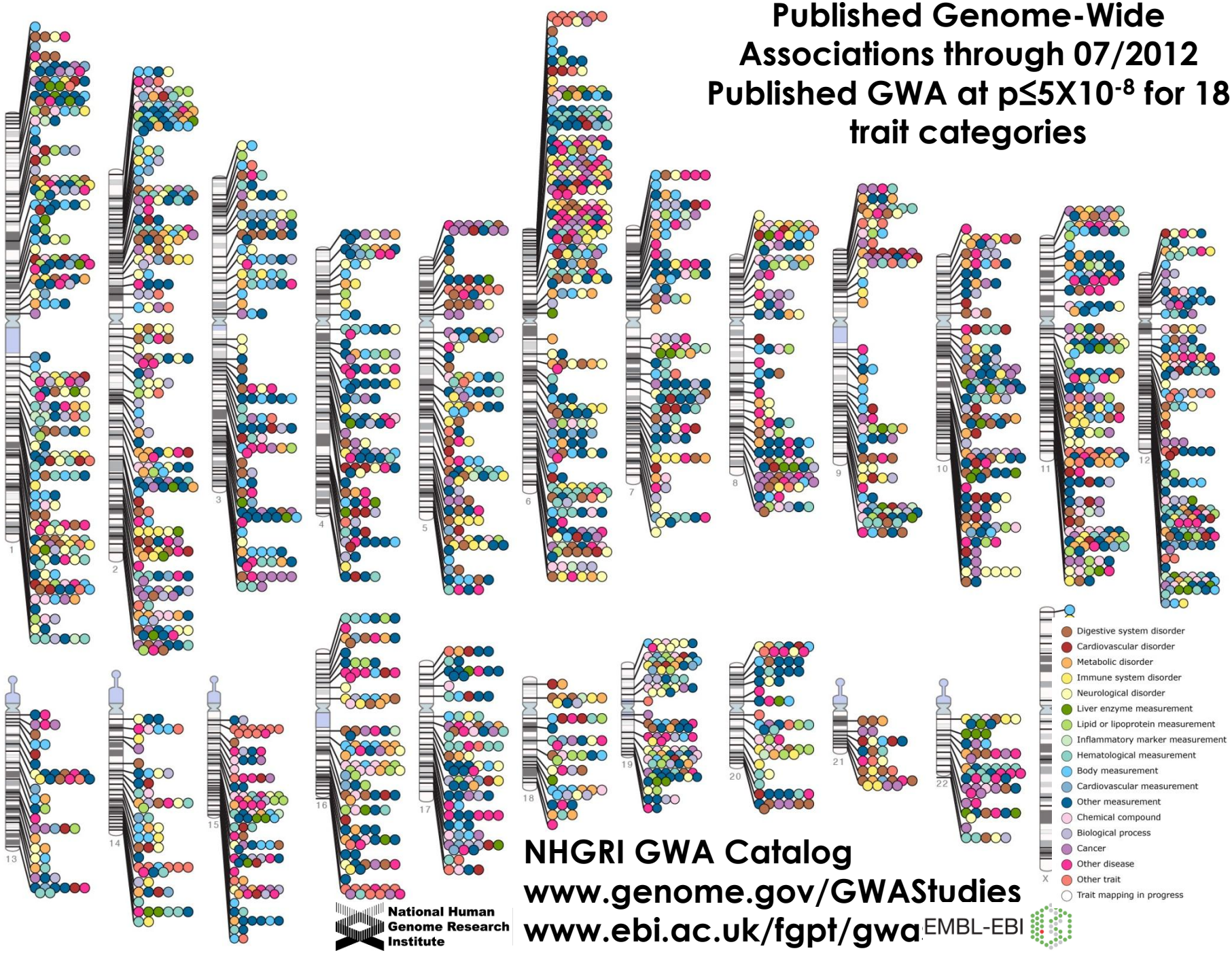


NHGRI GWA Catalog
www.genome.gov/GWAStudies


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 carbamazepine
Response to clopidogrel therapy
Response to hepatitis C treat
Response to interferon beta therapy
Response to metaformin
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 insuffiency
Vitiligo
Warfarin dose
Weight
White cell count
White matter hyperintensity
YKL-40 levels

Published Genome-Wide Associations through 07/2012

Published GWA at $p \leq 5X10^{-8}$ for 18 trait categories



NHGRI GWA Catalog

www.genome.gov/GWAStudies

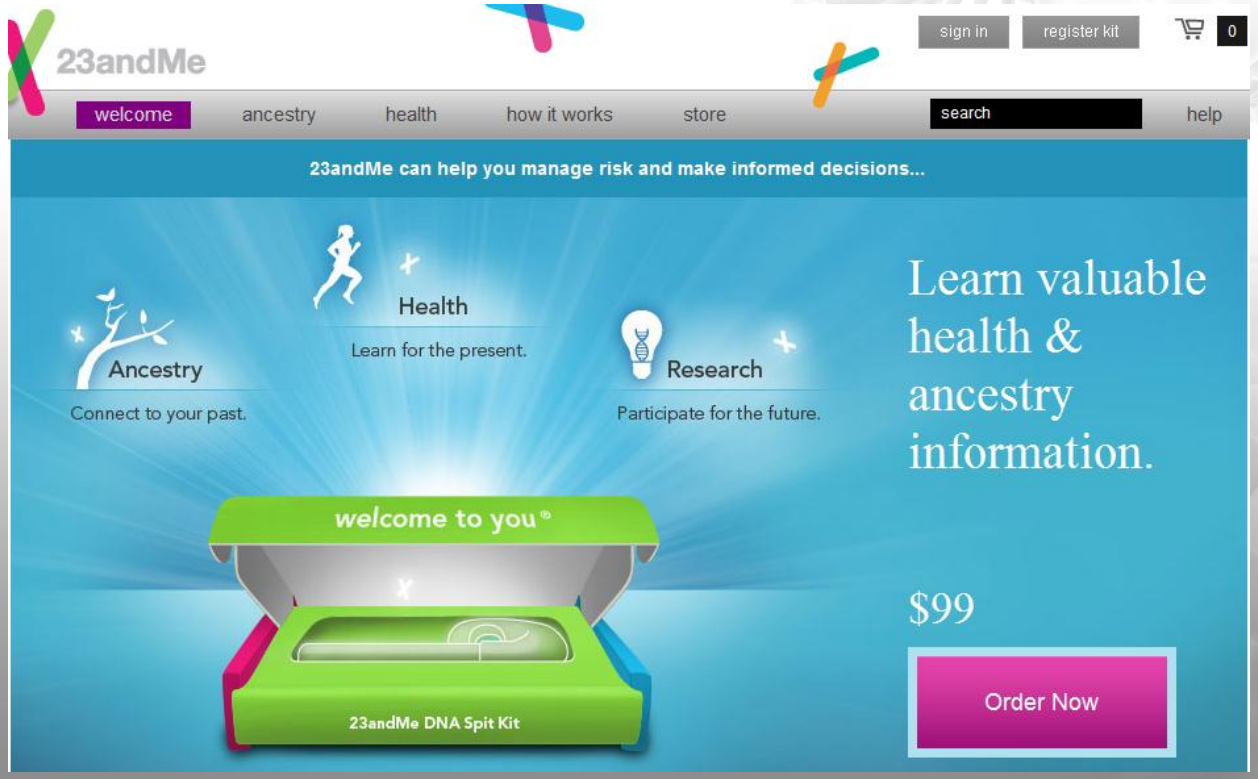
www.ebi.ac.uk/fgpt/gwa

National Human Genome Research Institute

EMBL-EBI

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

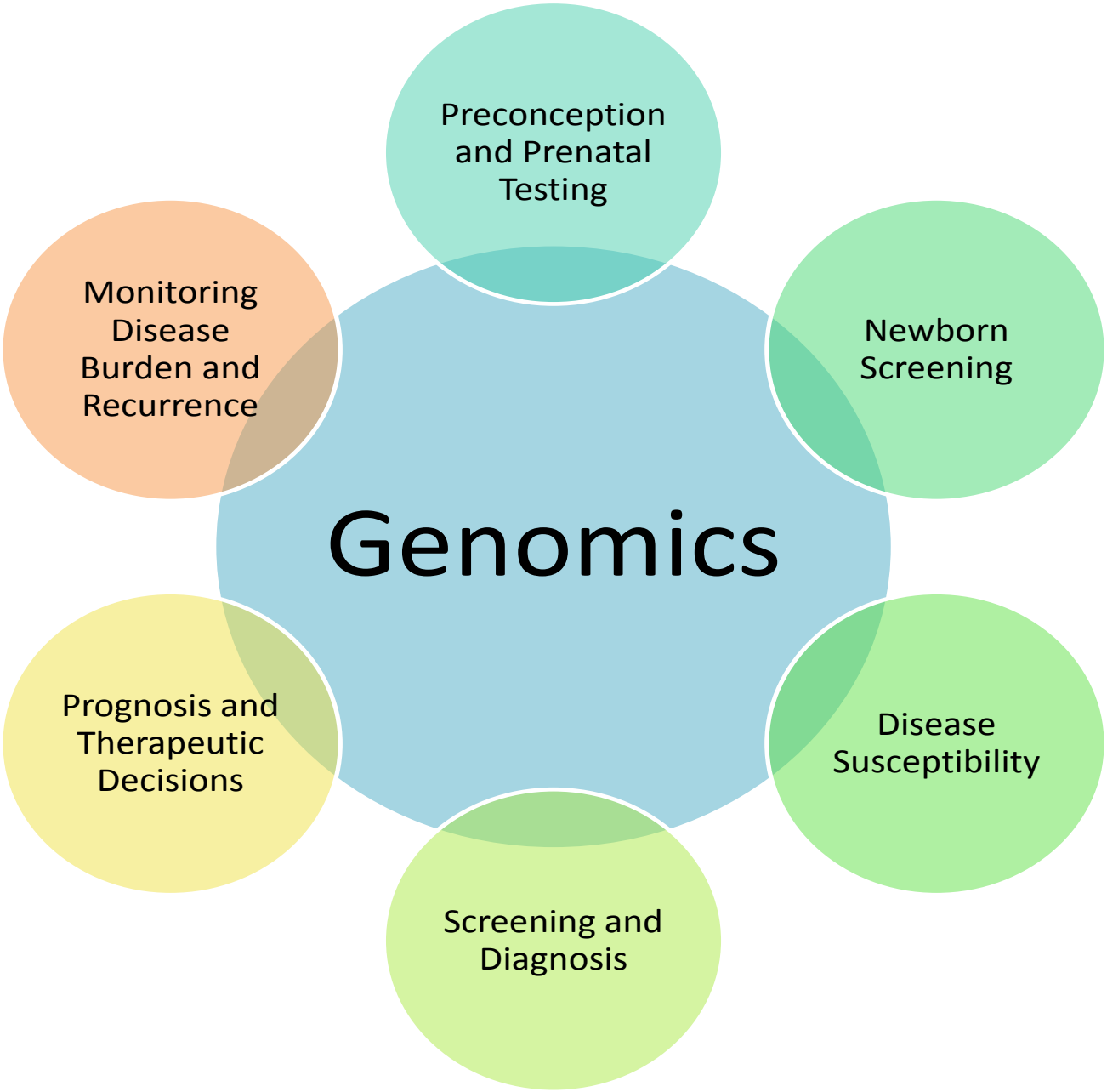
Epigenetics

- 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

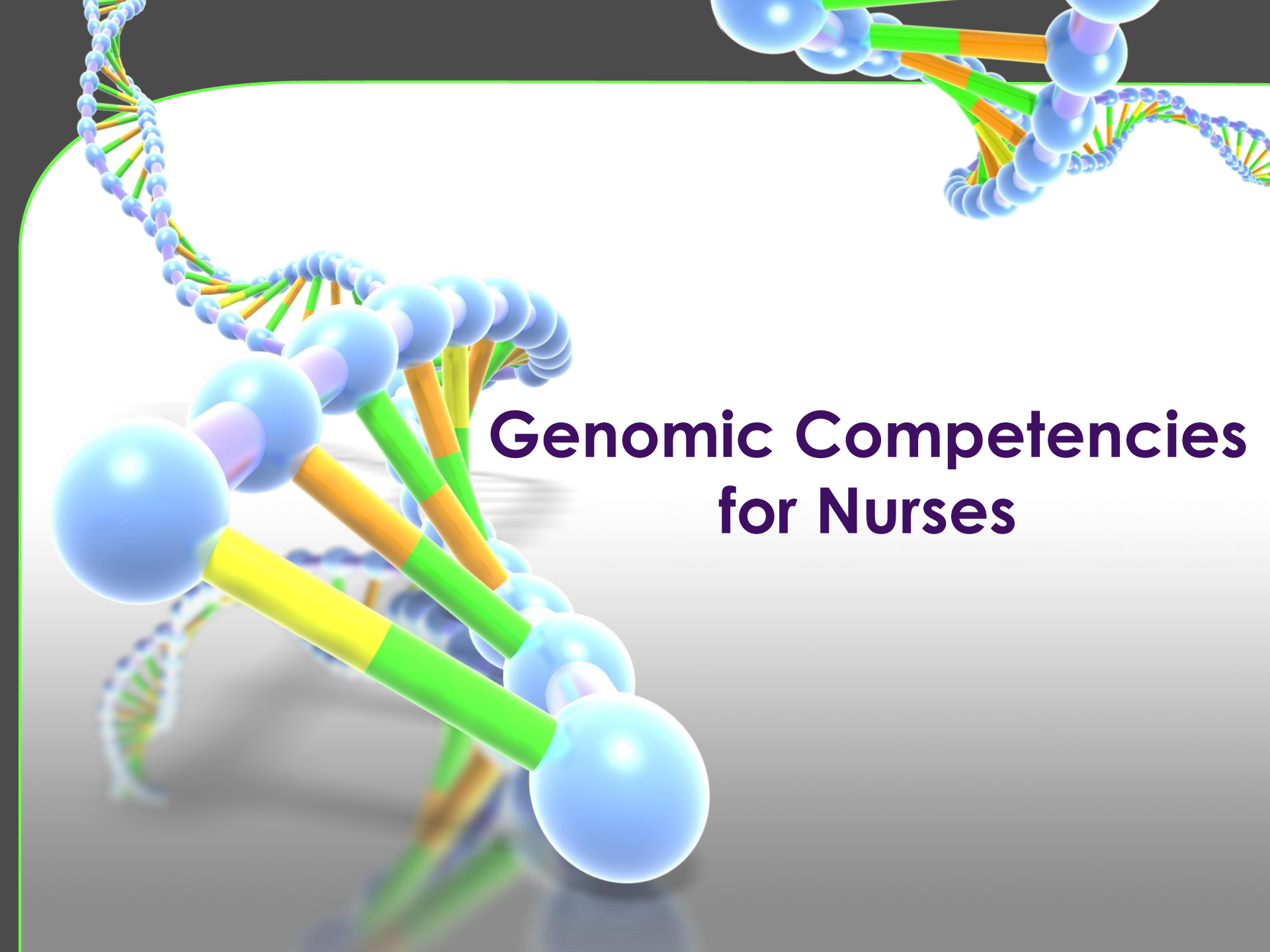
- Has expanded to include any whole genome analysis such as
  - Whole genome sequencing
  - Whole exome sequencing
  - RNA and RNAi sequencing
  - Whole genome SNP analysis
- Consideration for incidental findings
  - Previously unknown information
    - Clinical and analytic validity of finding
    - Immediacy and seriousness of risk
    - Actionable finding

Genomic Healthcare Applications



The Quest for Personalized Health Care

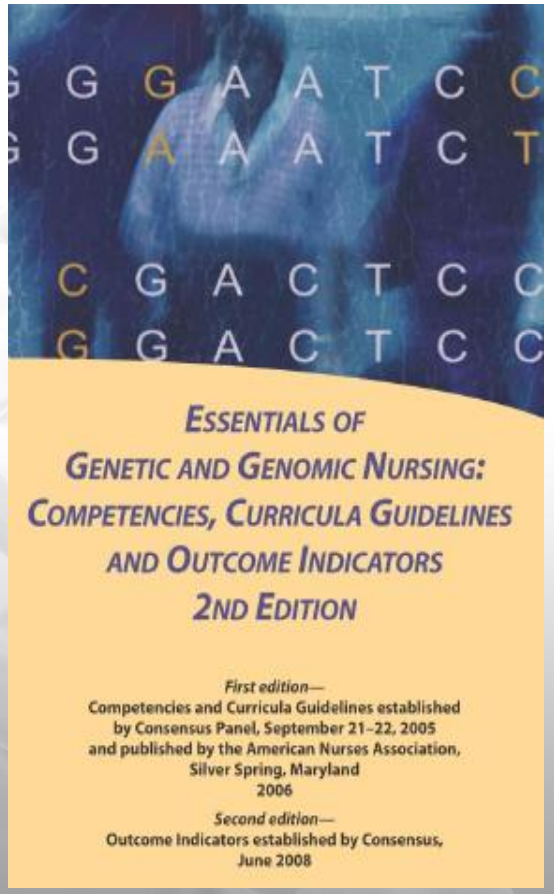
- Use of an individual's genetic/genomic information In addition to traditional health information to guide health care decision-making
- Disease prevention, risk reduction, diagnosis, treatment, symptom management and palliative care
  - Pharmacogenomics
    - Medication selection
    - Dose selection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



Genomic Competencies for Nurses

Essentials of Genetic and Genomic Nursing

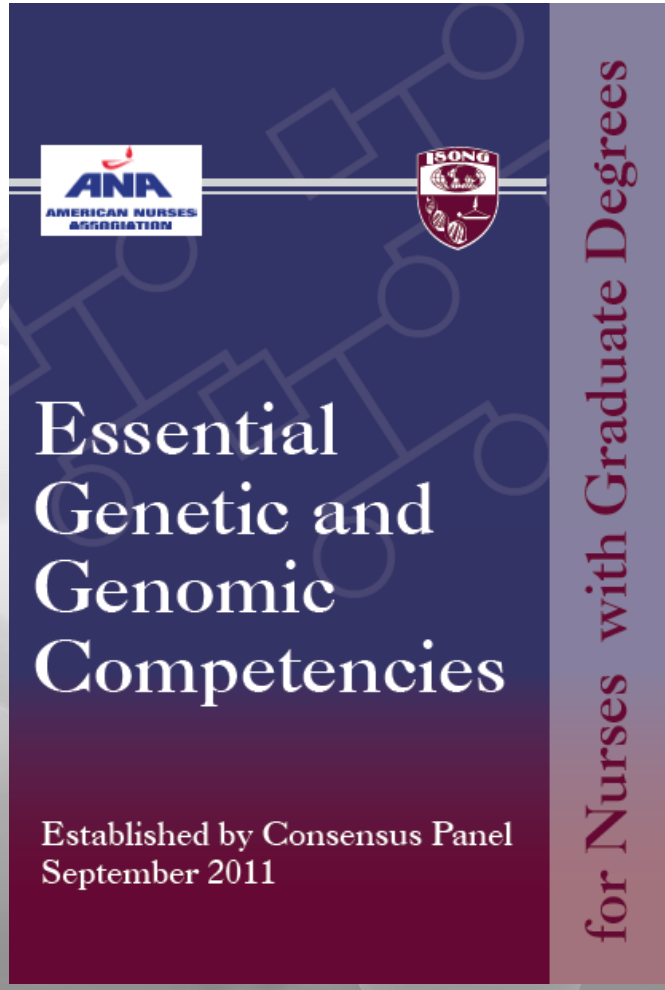
- Define essential genetic and genomic competencies for **ALL** nurses regardless of level of academic preparation, practice setting or specialty.
- Endorsed by 50 nursing organizations
- October 22-24 2006 Strategic Implementation Meeting
- 2nd Edition incorporated Outcome Indicators
  - Specific Areas 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-Graduate-Degrees.pdf

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:

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Heather Skirton PhD, RGN, RGC, QMW
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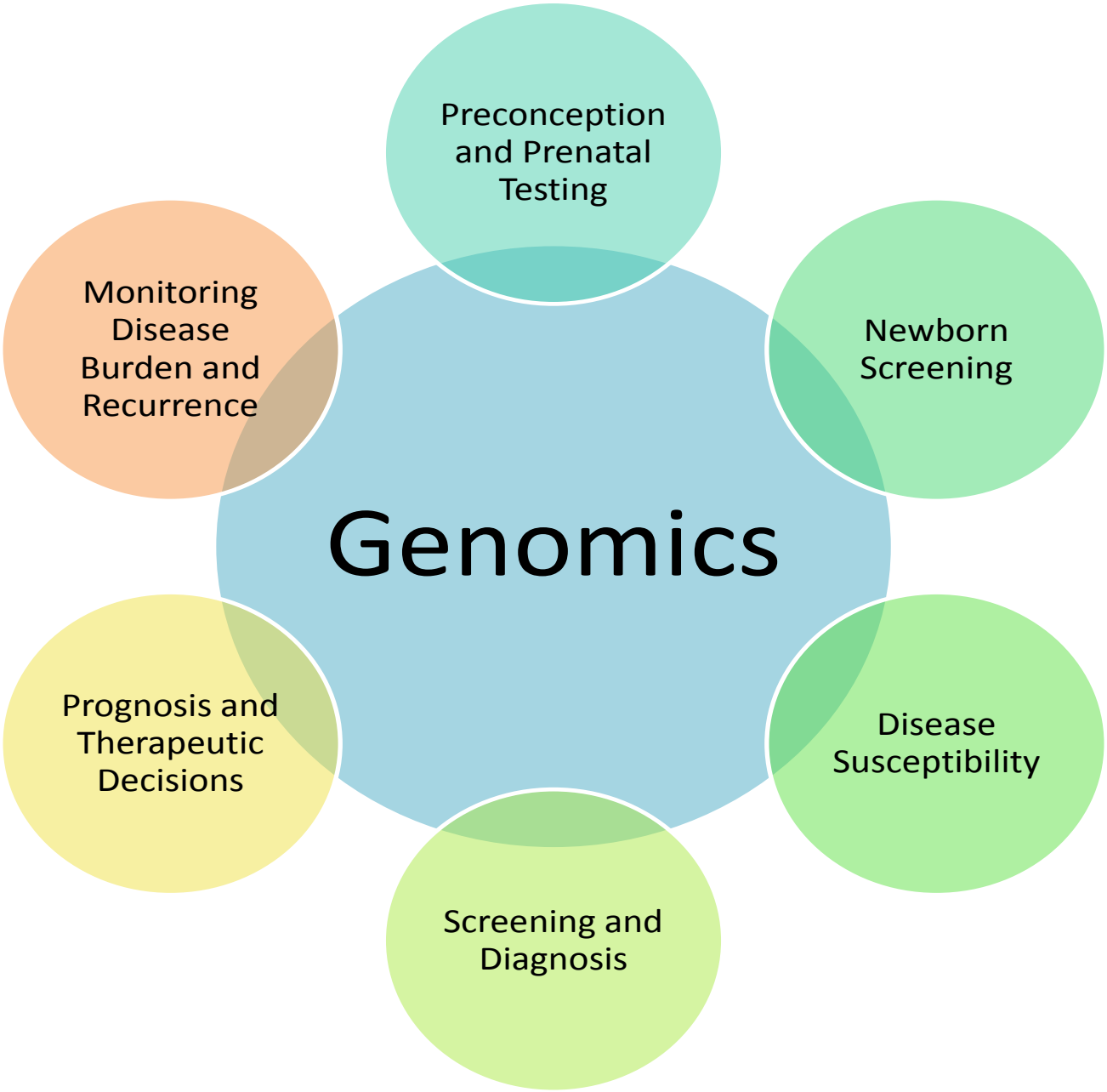
Editorial


Preconception and Prenatal Testing
Monitoring Disease Burden and Recurrence
Newborn Screening
Genomics
Prognosis and Therapeutic Decisions
Disease Susceptibility
Screening and Diagnosis

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
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    Uniformed Services University of the Health Sciences
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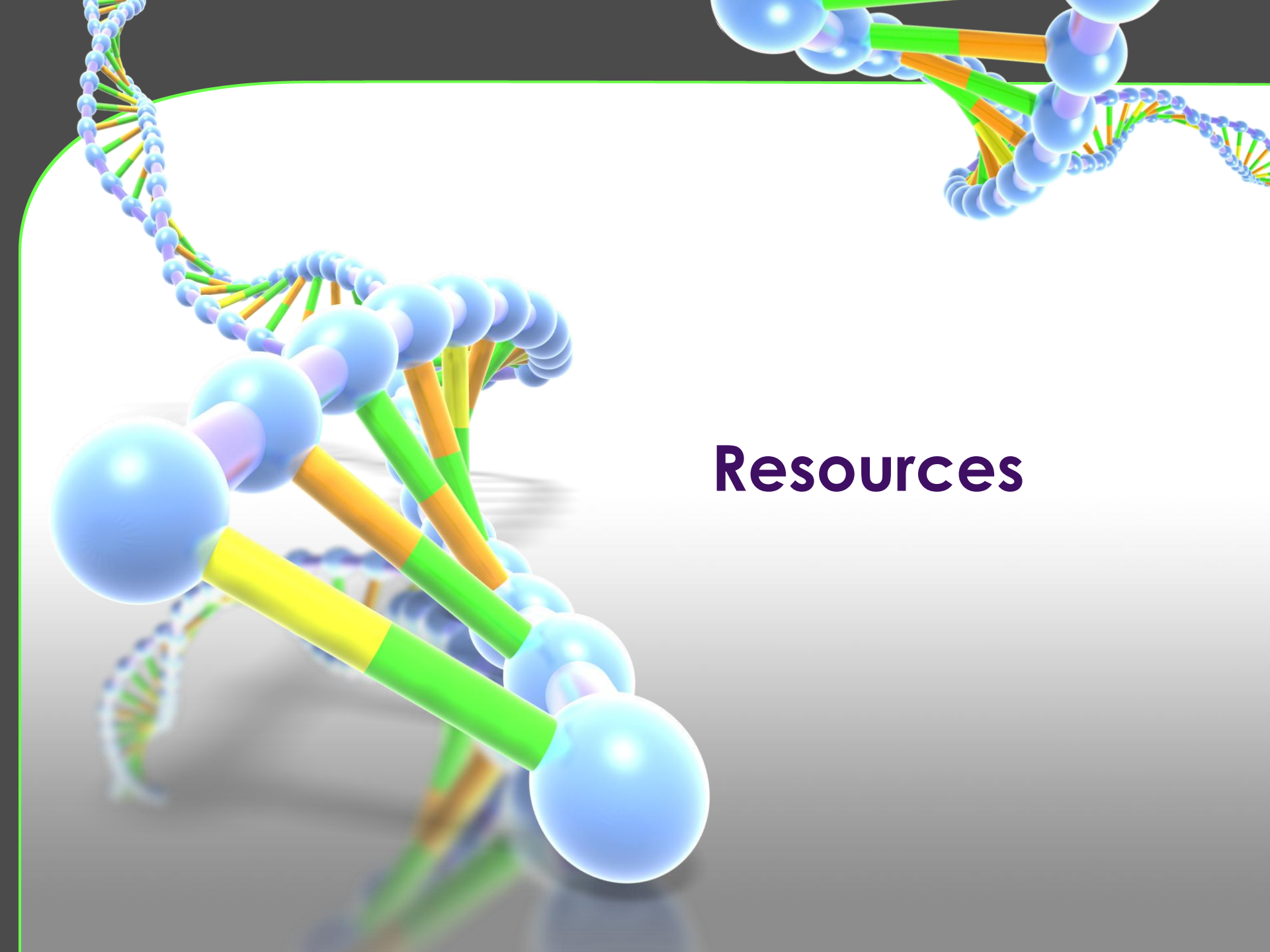
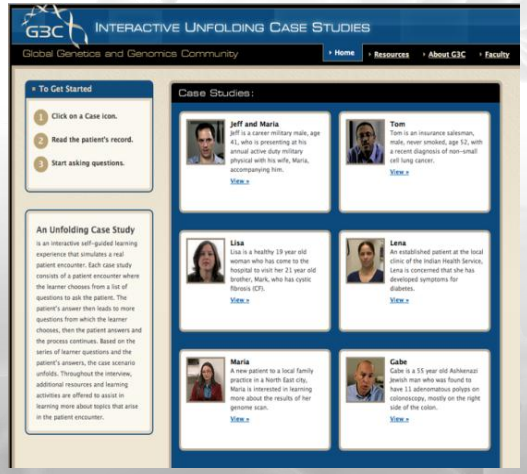
    National Human Genome Research Institute



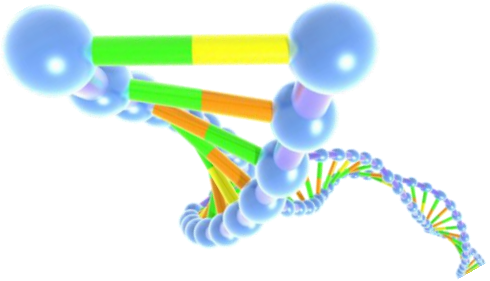
Resources

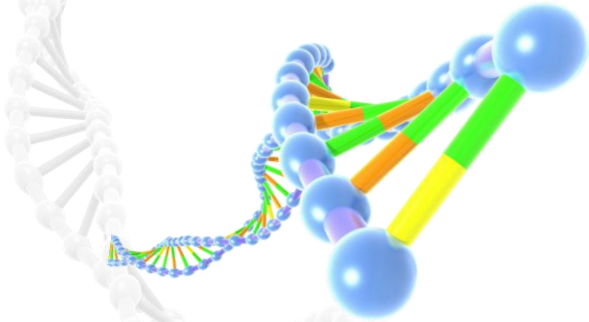
Other Genomic Resources

- 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**

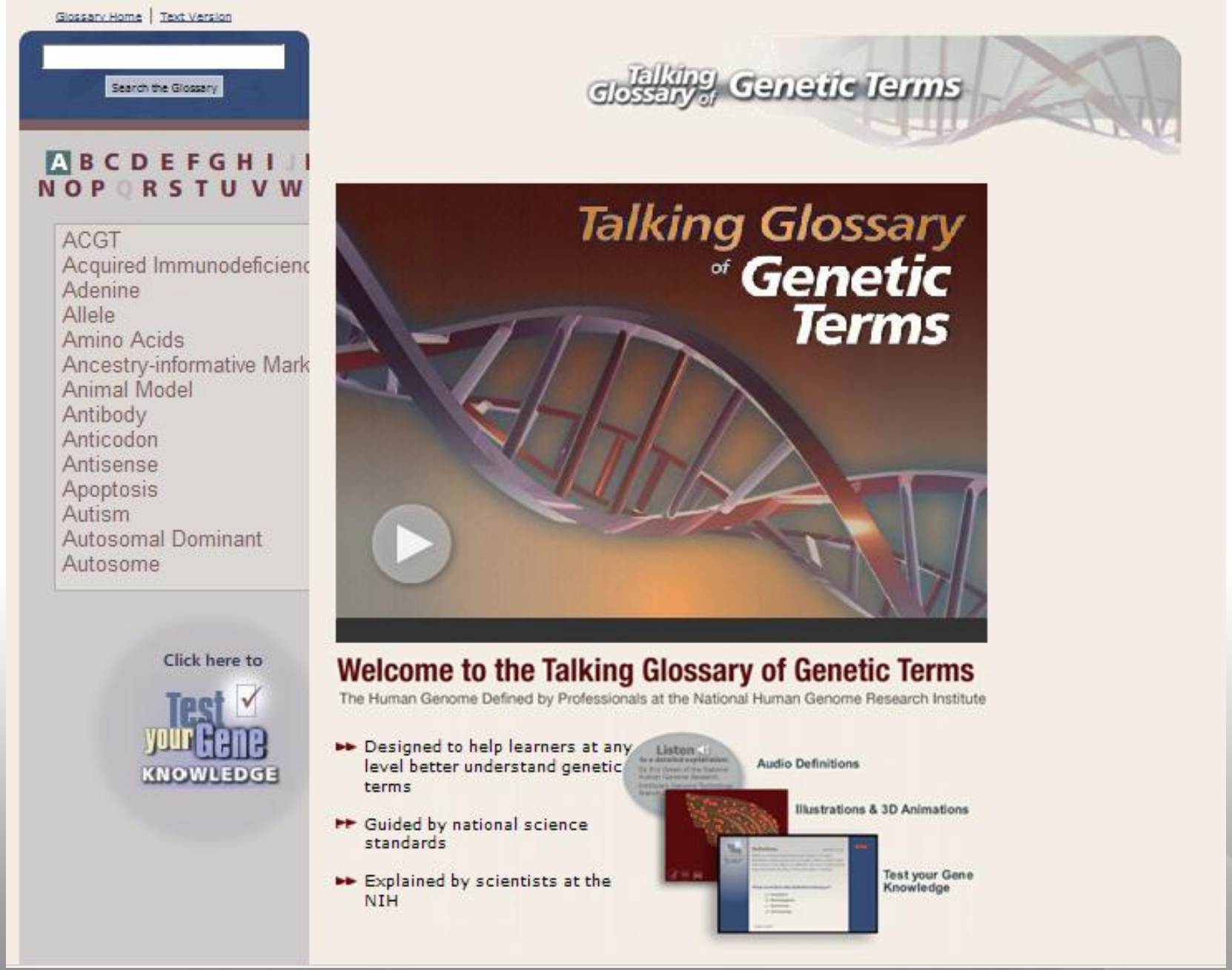


Publications





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



http://www.genome.gov/glossary

Surgeon General Family History Tool

https://familyhistory.hhs.gov/

Competency focus

ISSUE 6 | VOLUME 1 | YEAR 2012

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

Essential Genetic and Genomic Competencies for Nurses With Graduate Degrees

Competency focus

ISSUE 4 | VOLUME 3 | YEAR 2009

NURSING COMMUNITY ON THE IMPLEMENTATION OF
NURSING COMPETENCIES AND CURRICULA
NETICS AND GENOMICS.

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Second Edition

Competency focus

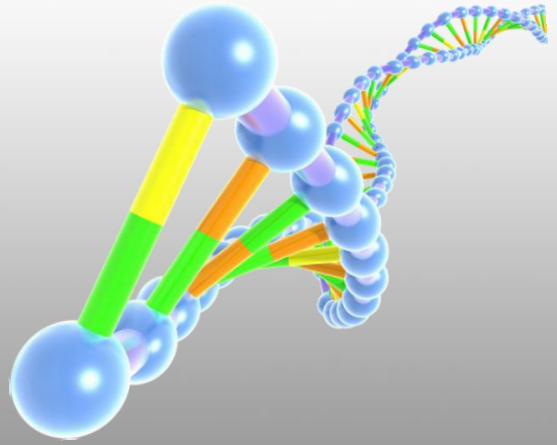
ISSUE 1 | VOLUME 1 | YEAR 2007

Implementation
Are We Now?

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

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