Genomics and Leadership

Kathleen Calzone, PhD, RN, APNG, FAAN
Center for Cancer Research, Genetics Branch
National Cancer Institute
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
Extent of Genetic and Genomic Testing

- 9/5/2011, 2430 diseases with available genetic tests
  - 2168 clinical
  - 262 research
- 8/23/2012, 2735 diseases with available genetic tests
  - 2487 clinical
  - 248 research
- 2/19/2013, 2951 diseases with available genetic tests
  - 2702 clinical
  - 249 research

GeneTests: Growth of Laboratory Directory

The Race for the $1000 Genome

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

Inquiries about NHGRI’s program interests should be addressed to the Division of Program Office.

Mardis Genome Medicine 2010, 2:84
http://genomemedicine.com/content/2/1/184

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
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 at 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.
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
Genomic Healthcare Applications

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

The Cancer Exemplar

- Risk Assessment
- Screening
- Therapy Selection
- Diagnosis/Prognosis

Symptom Management
Risk Assessment

- More than 55 hereditary cancer syndromes have been identified
- The most common syndromes are those associated with breast, ovarian, and gastrointestinal cancers
  - Tumor features at diagnosis are now being used as an indication for genetic assessment
- Germline cancer susceptibility gene testing
  - Relevant to individuals diagnosed with cancer whose cancer management may be altered
  - Individuals unaffected with cancer who could benefit from mutation specific cancer risk management
  - At-risk family members
Screening

- Genetic information is being used to personalize cancer screening recommendations
- SNP test results are being studied as a means to increase the specificity of cancer 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
Diagnosis/Prognosis

- Establish an accurate diagnosis
- Tumor profiling is being used to identify recurrence risk to guide adjuvant therapy


Therapy Selection

The use of therapies targeted to proteins encoded by mutated cancer genes

Pre and Post PET Scan using Targeted Treatment for V600E *BRAF* Mutation in Melanoma

<table>
<thead>
<tr>
<th>Gene</th>
<th>Genetic Alteration</th>
<th>Tumor Type</th>
<th>Therapeutic Agent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Receptor tyrosine kinase</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>EGFR</td>
<td>Mutation, amplification</td>
<td>Lung cancer, glioblastoma</td>
<td>Gefitinib, erlotinib</td>
</tr>
<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, B1BF-1120</td>
</tr>
<tr>
<td>FGFR2</td>
<td>Amplification, mutation</td>
<td>Gastric, breast, endometrial cancer</td>
<td>PKC412, B1BF-1120</td>
</tr>
<tr>
<td>FGFR3</td>
<td>Translocation, mutation</td>
<td>Multiple myeloma</td>
<td>PKC412, B1BF-1120</td>
</tr>
<tr>
<td>PDGFRα</td>
<td>Mutation</td>
<td>Glioblastoma, gastrointestinal stromal tumor</td>
<td>Sunitinib, sorafenib, imatinib</td>
</tr>
<tr>
<td>PDGFRβ</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>
</tr>
<tr>
<td>c-MET</td>
<td>Amplification</td>
<td>Gefitinib-resistant non–small-cell lung cancer, gastric cancer</td>
<td>Crizotinib, XL184, SU11274</td>
</tr>
<tr>
<td>IGF1R</td>
<td>Activation by insulin-like growth factor II ligand</td>
<td>Colorectal, pancreatic cancer</td>
<td>CP-751,871, AMG479</td>
</tr>
<tr>
<td>c-KIT</td>
<td>Mutation</td>
<td>Gastrointestinal stromal tumor</td>
<td>Sunitinib, imatinib</td>
</tr>
<tr>
<td>FLT3</td>
<td>Internal tandem duplication</td>
<td>Acute myeloid leukemia</td>
<td>Lestaunitinib, 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>
<td></td>
</tr>
<tr>
<td>ABL</td>
<td>Translocation (BCR-ABL)</td>
<td>Chronic myeloid leukemia</td>
<td>Imatinib</td>
</tr>
<tr>
<td>JAK2</td>
<td>Mutation (V617F), translocation</td>
<td>Chronic myeloid leukemia, myelo-proliferative disorders</td>
<td>Lestaunitinib, INCB018424</td>
</tr>
<tr>
<td>SRC</td>
<td>Overexpression</td>
<td>Non–small-cell lung cancer; ovarian, breast cancer; sarcoma</td>
<td>KX2–391, dasatinib, AZD0530</td>
</tr>
<tr>
<td>Serine–threonine–lipid kinase</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>BRAF</td>
<td>Mutation (V600E)</td>
<td>Melanoma; colon, thyroid cancer</td>
<td>SB-590885, PLX-4032, RAF265, XL281</td>
</tr>
<tr>
<td>Aurora A and B kinases</td>
<td>Overexpression</td>
<td>Breast, colon cancer; leukemia</td>
<td>MK-5108 (VX-689)</td>
</tr>
<tr>
<td>Polo-like kinases</td>
<td>Overexpression</td>
<td>Breast, lung, colon cancer; lymphoma</td>
<td>BI2536, GSK461364</td>
</tr>
<tr>
<td>mTOR</td>
<td>Increased activation</td>
<td>Renal-cell carcinoma</td>
<td>Temsirolimus (CCI-779), BEZ235</td>
</tr>
<tr>
<td>PI3K</td>
<td>PIK3CA mutations</td>
<td>Colorectal, breast, gastric cancer; glioblastoma</td>
<td>BEZ235</td>
</tr>
<tr>
<td>DNA damage or repair</td>
<td></td>
<td></td>
<td></td>
</tr>
<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.
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 (i.e. anti-convulsants)
      - Selecting medications to control symptoms such as hot flashes, nausea/vomiting
      - Use of over the counter medications like St. Johns’ Wort
      - Consumption of certain foods or supplements like grapefruit/grapefruit juice
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
AONE Nurse Executive Competencies

Leadership

- Communication & Relationship Management
- Professionalism
- Knowledge of Health Care Environment
- Business Skills and Principles

http://www.aone.org/resources/leadership%20tools/PDFs/AONE_NEC.pdf
# Genomics and the Nursing Workforce

<table>
<thead>
<tr>
<th>Study</th>
<th>N</th>
</tr>
</thead>
<tbody>
<tr>
<td>National Nursing Workforce Study in collaboration with ANA (NNWF)</td>
<td>619</td>
</tr>
<tr>
<td>ANA House of Delegates (HOD)</td>
<td>244</td>
</tr>
<tr>
<td>National Coalition of Ethnic Minority Nurses (NCEMNA)</td>
<td>392</td>
</tr>
<tr>
<td>Expanding RN Scope of Practice: A Method for Introducing a New Competency into Nursing Practice (MINC)</td>
<td>7347</td>
</tr>
<tr>
<td>MINC Admin Only</td>
<td>439</td>
</tr>
</tbody>
</table>
Domain: Professionalism

- Ethics
  - Articulate the application of ethical principles to operations
- Evidence-Based Clinical and Management Practice
  - Advocate for use of documented best practices
  - Teach and mentor others to routinely utilize evidence-based data and research
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
  - Timing
  - Confirmation in CLIA approved laboratory
Potential Policy Implications

➢ Regulatory
  • Guidance to IRBs and researchers using whole genome analysis

➢ MINC Participant Policy Initiatives
  • Genetic education, counseling and informed consent for genetic tests
  • Pathways for referrals to genetic services
  • Documentation of family history
  • Genomic Nursing Competency

➢ MINC Existing Policies
  • Genomic Advanced Directives
Domain: Business Skills and Principles

- Influencing Behaviors
  - Develop, communicate and monitor behavior expectations

- Medical /Staff Relationships
  - Collaborate with medical staff leaders in determining needed patient care services

- Academic Relationships
  - Identify educational needs of existing and potential nursing staff
Pharmacogenomic Influences

Efficacy

Toxicity
- inducers
- inhibitors

Pharmacodynamics

Pharmacokinetics

Target

PK = absorption, distribution, metabolism and excretion
PD = mechanism of action, drug concentration and effect
Polymorphisms and Phenotype

• **UM-Ultrarapid Metabolizer**
  - Unusually high activity of a drug metabolizing enzyme (DME) or drug transport protein (DTP)
  - Limited response to recommended doses

• **EM-Extensive Metabolizer**
  - Wild-type (normal activity) form of a DME or DTP
  - Expected efficacy at recommended doses

• **IM-Intermediate Metabolizer**
  - Reduced activity of a DME or DTP
  - Some decreased efficacy at recommended doses

• **PM-Poor Metabolizer**
  - Very low or no activity of a DME or DTP
  - Increased toxicity
  - Decreased efficacy at recommended doses

Inhibitors and Inducers

- **Inhibitors**
  - Reduce the drug metabolizing enzyme or drug transport protein

- **Inducers**
  - Increase the drug metabolizing enzyme or drug transport protein
# P450 Drug Interaction Table

## Indiana University

### SUBSTRATES

<table>
<thead>
<tr>
<th>A2</th>
<th>B6</th>
<th>C8</th>
<th>C9</th>
<th>C19</th>
<th>D6</th>
<th>E1</th>
<th>3A4,5,7</th>
</tr>
</thead>
<tbody>
<tr>
<td>amitriptyline</td>
<td>bupropion</td>
<td>cyclophosphamide</td>
<td>efavirenz</td>
<td>ifosfamide</td>
<td>methadone</td>
<td>sorafenib</td>
<td>torsemide</td>
</tr>
<tr>
<td>caffeine²</td>
<td>amodiaquine²</td>
<td>cerivastatin</td>
<td>paclitaxel</td>
<td>repaglinide</td>
<td>sorafenib</td>
<td>tamsulosin</td>
<td></td>
</tr>
<tr>
<td>clomipramine</td>
<td>diclofenac</td>
<td>ibuprofen</td>
<td>lornoxicam</td>
<td>meloxicam</td>
<td>S-naproxen-Norpiroxicam</td>
<td>suprofen</td>
<td></td>
</tr>
<tr>
<td>clozapine</td>
<td>lansoprazole</td>
<td>omeprazole²</td>
<td>pantoprazole</td>
<td>rabeprazole</td>
<td>tamoxifen</td>
<td>TAMOXIFEN GUIDE</td>
<td></td>
</tr>
<tr>
<td>cyclobenzaprine</td>
<td>diltiazem</td>
<td>lorazepam</td>
<td>pantoprazole</td>
<td>rabeprazole</td>
<td>beta-blockers</td>
<td></td>
<td></td>
</tr>
<tr>
<td>estradiol</td>
<td>carvedilol</td>
<td>S-metoprolol</td>
<td>propafenone</td>
<td>timolol</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>fluvastatin</td>
<td>acetaminophen→NAPQI</td>
<td>acetylsalicylic acid</td>
<td>ibuprofen</td>
<td>indomethacin</td>
<td>CYP3A4 inhibitors</td>
<td></td>
<td></td>
</tr>
<tr>
<td>fluoxetine</td>
<td>amitriptyline</td>
<td>clomipramine</td>
<td>desipramine</td>
<td>fluoxetine</td>
<td>imipramine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>haloperidol</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>imipramine N-DeMe</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>mexiletine</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>naproxen</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>olanzapine</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>ondansetron</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>phenacetin¹</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>acetaminophen→NAPQI</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>propranolol</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>riluzole</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>ropivacaine</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>tacrine²</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>theophylline²</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>tizanidine</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>verapamil</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(R)-warfarin</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>zileuton</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
<tr>
<td>zolmitriptan</td>
<td>amitriptyline</td>
<td>citalopram</td>
<td>clomipramine</td>
<td>paroxetine</td>
<td>venlafaxine</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### Oral Hypoglycemic Agents:
- tolbutamidé¹
- glipizide

### Angiotensin II Blockers:
- losartan
- irbesartan

### Sulfonylureas:
- glyburide
- glimepiride
- tolbutamidé

### Antipsychotics:
- haloperidol
- perphenazine
- risperidone→9-OH
- thioridazine
- zuclopenthixol

### Antidepressants:
- amitriptyline
- clomipramine
- desipramine
- fluoxetine
- imipramine
- paroxetine
- venlafaxine

### Beta Blockers:
- carvedilol
- S-metoprolol
- propafenone
- timolol

### Anti-arrhythmics:
- quinidine→3-OH
- (not 3A4)

### Immune Modulators:
- cyclosporine
- tacrolimus (FK506)

### Macrolide Antibiotics:
- clarithromycin
- erythromycin² (not 3A4)

### HIV Antivirals:
- indinavir
- nelfinavir
- ritonavir
- saquinavir

### Prokinetic:
- cisapride

---

1. *CYP2C9 substrate*
2. *CYP2C19 substrate*
Pharmacogenomics Uses

- Drug Selection
- Dose Selection
- Avoidance of Inducers and Inhibitors
- Insurance Coverage for Specific Agents
- Financial Plan for Genomic Integration
  - 2013 Centers for Medicaid and Medicare Services physician payment rules
    - Transitional Healthcare Care Bundles
  - Affordable Care Act penalties for high readmission rates

Bindman et al. (2013). Medicare’s Transitional Care Payment-A step toward the Medical Home. NEJM 368, 692-694
New Labeling Information for Warfarin (marketed as Coumadin)

- FDA News
- Coumadin Labeling approved 8/16/2007
- Questions and Answers
- FDA Critical Path Initiative

PDF requires the free Adobe Acrobat Reader

Date created: August 16, 2007
Questions and Answers on New Labeling for Warfarin (marketed as Coumadin)

What is warfarin and the recently approved change in the label for the drug?

Warfarin is a drug prescribed to help prevent blood clots. Following oral ingestion, it acts by blocking the action of certain proteins that cause blood to clot. Since warfarin acts to keep blood from clotting, one of the side effects of the drug is an increased risk for bleeding. This risk increases importantly if patients receive too much warfarin. Patients frequently differ in the dose of warfarin necessary to prevent blood clots and also differ in the risk for bleeding.

The recently approved change in the warfarin label provides information on how people with certain genetic differences may respond to warfarin. Specifically, people with variations in two genes may need lower warfarin doses than people without these genetic variations. The two genes are called CYP2C9 and VKORC1. The CYP2C9 gene is involved in the breakdown (metabolism) of warfarin and the VKORC1 gene helps regulate the ability of warfarin to prevent blood from clotting.

How will this new warfarin label information benefit patients?

This information will benefit patients because it will describe why patients with a variation in the CYP2C9 and/or VKORC1 genes may need a lower warfarin dose than patients with the usual forms of these genes. For patients with these genetic variations, the new label information may help physicians prescribe the correct warfarin dose and also encourage them to give increased attention to how these patients respond to warfarin. Identification of patients with these
Emergency Department Visits


Adverse drug events (ADE) defined as undesirable pharmacologic or idiosyncratic effects from medications administered at correct dosages.

- From 5077 cases there were 99,628 hospitalizations as a result of ADE’s in people ≥65
- 33% of ED visits that resulted in hospitalization were associated with ADE’s from warfarin.

Domain: Knowledge of the Healthcare Environment

- Clinical Practice Knowledge
  - Maintain knowledge of current nursing practice and the roles and functions of patient care team members
  - Ensure that written organization clinical policies and procedures are reviewed and updated in accordance with evidence-based practice

- Evidence-Based Practice/Outcome Measurement
  - Utilize research findings for the establishment of standards, practices and patient care models in the organization

- Quality Improvement Metrics
  - Determine patient care quality improvement goals and objectives
<table>
<thead>
<tr>
<th></th>
<th>In the prior three months nurses seeing patients who RARELY OR NEVER assessed a family history.</th>
<th>Took family history: Assessed age at dx</th>
<th>Took family history: Assessed maternal and paternal lineages</th>
<th>AGREED OR STRONGLY AGREED that family history taking should be a key component of nursing care</th>
</tr>
</thead>
<tbody>
<tr>
<td>NNWFS</td>
<td>67%, (n=288/510)</td>
<td>41% (n=200/483)</td>
<td>66% (n=320/484)</td>
<td>84% (n=369/442)</td>
</tr>
<tr>
<td>HOD</td>
<td>58% (n=59/102)</td>
<td>51% (n=116/227)</td>
<td>75% (n=168/224)</td>
<td>91% (n=219/242)</td>
</tr>
<tr>
<td>NCEMNA</td>
<td>Not Done</td>
<td>64% (n=231/363)</td>
<td>78% (n=280/361)</td>
<td>Not assessed</td>
</tr>
<tr>
<td>MINC</td>
<td>69% (n=3270/4774)</td>
<td>29% (n=1564/5348)</td>
<td>53% (n=2850/5336)</td>
<td>71% (n=4051/5701)</td>
</tr>
<tr>
<td>MINC Admin</td>
<td>63% (n=91/294)</td>
<td>34% (n=142/421)</td>
<td>64% (n=267/420)</td>
<td>80% (n=347/436)</td>
</tr>
</tbody>
</table>

**Legend:**
- **NNWFS:** North West Florida Superintendents
- **HOD:** Hospital Directors
- **NCEMNA:** National Cancer Education and Management Network
- **MINC:** Medical Institute for Nursing Care
- **Admin:** Administrative Staff
Family History in Nursing Practice

“It’s one of those times in your life that you are grateful you had the knowledge.”

Quote from: Barbara Ganster, RN, BSN
Breast Cancer Case Manager
National Naval Medical Center
# Total Knowledge Scores

<table>
<thead>
<tr>
<th>Total knowledge score was calculated from 12 knowledge questions</th>
<th>CORRECTLY answered question about whether genomic risk (as indicated by Fm Hx) has clinical relevance for coronary heart disease</th>
<th>INCORRECTLY stated that diabetes and heart disease are caused by a single gene variant</th>
</tr>
</thead>
<tbody>
<tr>
<td>NNWFS</td>
<td>8.99/12, Range 1-12, SD 1.69</td>
<td>99% (n=437/442)</td>
</tr>
<tr>
<td></td>
<td><strong>INCORRECTLY stated that diabetes and heart disease are caused by a single gene variant</strong></td>
<td>61% (n=268/442)</td>
</tr>
<tr>
<td>HOD</td>
<td>9.24/12, range 3-12, SD 1.50</td>
<td><strong>CORRECTLY answered question about whether genomic risk (as indicated by Fm Hx) has clinical relevance for coronary heart disease</strong></td>
</tr>
<tr>
<td></td>
<td>98% (n=216/220)</td>
<td>62% (n=137/220)</td>
</tr>
<tr>
<td>NCEMNA</td>
<td>Not Done</td>
<td>74% (n=275/274)</td>
</tr>
<tr>
<td></td>
<td><strong>INCORRECTLY stated that diabetes and heart disease are caused by a single gene variant</strong></td>
<td>66% (n=92/138)</td>
</tr>
<tr>
<td>MINC</td>
<td>8.09/12, range 0-12, SD 1.62</td>
<td>82% (n=4116/5118)</td>
</tr>
<tr>
<td></td>
<td><strong>INCORRECTLY stated that diabetes and heart disease are caused by a single gene variant</strong></td>
<td>71% (n=3580/5008)</td>
</tr>
<tr>
<td>MINC Admin</td>
<td>Not Done</td>
<td>89% (n=386/434)</td>
</tr>
<tr>
<td></td>
<td><strong>INCORRECTLY stated that diabetes and heart disease are caused by a single gene variant</strong></td>
<td>76% (n=330/435)</td>
</tr>
</tbody>
</table>
Domain: Leadership

- Foundational Thinking Skills
  - Maintain curiosity and an eagerness to explore new knowledge and ideas
  - Provide visionary thinking on issues that impact the healthcare organization

- Change Management
  - Serve as a change agent, assisting others in understanding the importance, necessity, impact and process of change
# Genomic Attitudes

<table>
<thead>
<tr>
<th></th>
<th>Reported it was SOMEWHAT OR VERY IMPORTANT for nurses to become more educated about genetics of common disease</th>
<th>Believe senior staff see genetics as an IMPORTANT part of the survey respondent’s personal role</th>
<th>WOULD attend a genetics course on their own time</th>
</tr>
</thead>
<tbody>
<tr>
<td>NNWFS</td>
<td>92% (n=572/607)</td>
<td>Not assessed</td>
<td>73% (n=368/506)</td>
</tr>
<tr>
<td>HOD</td>
<td>98% (n=239/244)</td>
<td>Not assessed</td>
<td>75% (n=182/240)</td>
</tr>
<tr>
<td>NCEMNA</td>
<td>97% (n=374/385)</td>
<td>24% (n=87/359)</td>
<td>Not Assessed</td>
</tr>
<tr>
<td>MINC</td>
<td>89% (n=5992/6741)</td>
<td>26% (n=1302/5110)</td>
<td>64% (n=3248/5087)</td>
</tr>
<tr>
<td>MINC Admin</td>
<td>93% (n=406/438)</td>
<td>27% (n=118/431)</td>
<td>68% (n=294/433)</td>
</tr>
</tbody>
</table>
Diffusion of Innovations

Clues to Educational Needs

Most:

- Indicate a potential disadvantage to integrating genomics into practice was that it would increase insurance discrimination.
- Felt that genetics could increase patient anxiety about risk, despite behavioral studies in many conditions indicating that most patients do well with genetic information.
- Felt genetics is not reimbursable or too costly.
- Feel genetics is important BUT do not think that senior staff feel it is important to their role.
- Are willing to learn more, and are willing to do so on their own time.
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 may be published in 2013 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
Domain: Communication and Relationship Building

- Influencing Behaviors
  - Develop, communicate and monitor behavior expectations

- Medical/Staff Relationships
  - Collaborate with medical staff leaders in determining needed patient care services
  - Collaborate with physicians to develop patient care protocols, policies, and procedures

- Academic Relationships
  - Identify educational needs of existing and potential nursing staff
  - Collaborate with nursing programs to provide required resources
Personalized Health Care Requirements


Published by AAAS
Challenges and Opportunities

- Funding
- Capacity to Understand Relevance
- Lack of Outcome Evidence
- Faculty Capacity to Teach Genomics
- Size of Workforce
- Diversity of Workforce
Additional Nursing Bottlenecks

- Insufficient numbers of faculty AND continuing education educators prepared to teach this content
  - Assessment is still needed to assess impact of CCNE accreditation standard changes
  - Some academic accrediting bodies do not consider genomics in their evaluations
- Institutional accrediting bodies do not consider genomics in their evaluations
- NCLEX and State Boards of Nursing do not require genomic competency for licensure or re-licensure
- A financial planning guide justifying genomics investment not yet established
Summary

- Recognize the relevancy and value of genomics to your responsibilities.
- Utilize your leadership and skills to be a change agent/champion in the healthcare environment.
- Recognize policy opportunities to ensure safe, effective and efficient translation of genomic clinical care.
- Think creatively and be innovative about designing services, staff education, clinical infrastructure (i.e., EHR) that facilitates adoption of genomics in care.
- Visualize how you can leverage interprofessional teams to assure that our healthcare providers are adequately prepared to be able to transform healthcare delivery.
Resources

- Journal of Nursing Scholarship Genomic Special Issue
  Webinar Series with Issue Authors
  http://www.genome.gov/27552312

- Genetics/Genomics Competency Center for Education (G2C2)
  http://www.g-2-c-2.org

- CDC Public Health Genomics
  http://www.cdc.gov/genomics/

- Genomic Competency Listserv
  Leave business card or email: calzonek@mail.nih.gov
Questions/Discussion

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