Whole Genome Sequencing and Nursing Science among Minority Populations

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# Faculty Disclosure

<table>
<thead>
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<tbody>
<tr>
<td>Conflicts of Interest</td>
<td>None</td>
</tr>
<tr>
<td>Employer</td>
<td>Yale University School of Nursing</td>
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<tr>
<td>Sponsorship/Commercial Support</td>
<td>None</td>
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Goals and Objectives

**Session Goal:**
- Explicate how nurse scientists can improve understanding of DNA sequencing data and translational genomics

**Learner Objectives:**
1. The learner will be able to articulate the current state of the science for nurses and genome sequencing.
2. The learner will be able to understand differences in mass spectrometry and genome sequencing in new born screenings in various regions.
Overview

- Introduction
- Framework for Genome Based Population Screening
- Current Policy Landscape
- Implications for Nurses and Nurse Scientists
Introduction

- DNA sequencing can dramatically improve health outcomes
- Raw Sequence data
  Clinical Utility*

*Image from: https://www.genome.gov/health/
Introduction
Framework for Genome Based Population Screening

Mass Spectrometry vs. Genome Sequencing

**MASS SPECTROMETRY**

1. Cell or tissue → Protein mixture → Enzymatic digestion into peptides
2. Peptide mixture → Liquid chromatography (peptide separation)
3. Ionization of peptides → Electrospray ionization
4. Ion-peptide mixture → Neutral gas
5. Mass analyzer → Fragmentation by collision
6. Product ions → Mass analyzer
7. Signal detection → Peptide sequence

**DNA SEQUENCING**

1. DNA extraction
2. DNA fragmentation
3. Clone into vectors
4. Transform bacteria, grow, isolate vector DNA
5. Sequence the library
6. Assemble contiguous fragments

Guiding Principals: Newborn Screening

- Responsibility to save & improve lives
- Actionable variants

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<tr>
<th>Category</th>
<th>Definition</th>
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<tr>
<td>Pathogenic</td>
<td>Previously reported and recognized as cause of the disorder.</td>
</tr>
<tr>
<td>Likely Pathogenic</td>
<td>Previously unreported but expected to cause the disorder.</td>
</tr>
<tr>
<td>Variant of Unknown Significance</td>
<td>Previously unreported and may or may not cause the disorder.</td>
</tr>
<tr>
<td>Likely Benign</td>
<td>Previously unreported and probably not causative of disease</td>
</tr>
<tr>
<td>Benign</td>
<td>Previously reported and is recognized as a neutral variant.</td>
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<tr>
<td>Variant of unknown significance- suspicious</td>
<td>Not known or expected to be causative of disease, but is found to be associated with a clinical presentation.</td>
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Current Policy Landscape

AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMIC

- Trained Healthcare Professional
- Informed of 56 “actionable” variants
  ✦ Opportunity to opt-out
- No current recommendation for incidental findings

ASSOCIATION OF GENETIC NURSES AND COUNSELLORS

- Similar to ACMG with 2 differences:
  1. Refers to sequencing screenings as “opportunistic”
  2. Does not recommend screening of children for adult onset conditions
Genomic Sequencing in the Clinic & Community

DNA testing companies

- 23andMe (adoption, deep ancestry, ethnicity, genealogy, health)
- African Ancestry (deep ancestry)
- AfricanDNA (FTDNA affiliate) (deep ancestry, ethnicity, genealogy)
- AncestrybyDNA (deep ancestry, ethnicity)
- AncestryDNA, a subsidiary of Ancestry.com (adoption, ethnicity, genealogy)
- BritainsDNA (formerly Ethnoancestry) (deep ancestry, ethnicity)
- CymruDNAWales - see BritainsDNA
- DNA Ancestry and Family Origin (FTDNA affiliate in the Middle East) (adoption, deep ancestry, full mtDNA sequencing, genealogy)
- DNA Consultants (deep ancestry, ethnicity)
- DNA Spectrum (ethnicity)
- DNA Tribes (ethnicity)
- DNA Worldwide (FTDNA partner) (deep ancestry, ethnicity, genealogy, paternity, relationship)
- Ethnoancestry - see BritainsDNA
- Family Tree DNA (adoption, deep ancestry, full mtDNA sequencing, genealogy, identity, relationship, Y chromosome sequencing)
- Full Genomes Corporation (whole genome sequencing, Y-chromosome sequencing)
- Futura Genetics (health)
- Gene by Gene - the parent company of Family Tree DNA which now incorporates the companies previously known as DNA Traits, DNA DTC and DNA Findings (research, health, exome sequencing, whole genome sequencing)
- Genebase (deep ancestry, genealogy)
- Genographic Project (deep ancestry, ethnicity)
- iGENE (FTDNA affiliate) (deep ancestry, genealogy)
- IrelandaDNA - See BritainsDNA (formerly Ethnoancestry)
- MyDNA Global - a new name for BritainsDNA
- Oxford Ancestors (deep ancestry, genealogy)
- Roots for Real (deep ancestry, ethnicity, genealogy)
- ScotlandsDNA - (formerly Ethnoancestry) (deep ancestry, ethnicity)
- Sorenson Genomics (laboratory services)
- Sure Genomics (whole genome sequencing)
- Centrillion Biosciences (aka TribeCode) (deep ancestry, ethnicity)
- YorkshiresDNA - See BritainsDNA (formerly Ethnoancestry)
- YSEQ (custom Y-SNPs, Y-STRs, SNP panels)

List retrieved from the International Society of Genetic Genealogy Wiki: http://isogg.org/wiki/List_of_DNA_testing_companies
Proposed role of Nursing in DNA Sequencing

**Prepare**
- Incorporate clinical utility of DNA sequencing technology into nursing curricula

**Participate**
- Remain current on procedures, policies and clinical implications of DNA sequencing
- Engage in policy-making and generate research with clinical utility

**Inform**
- Ensure patients receive accurate information prior to testing using DNA sequencing
- Teach patients implications of results and refer to specialists as needed
- Inform research subjects benefit of research may not benefit them, but help future generations

**Taylor, JY., Wright, ML., Hickey, K.,Housman, D., (under review) Genome Sequencing Technologies and Nursing: What is the role of Nurses and Nurse Scientists?**
Acknowledgements

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Thank you