Exploring the Role of Diversity and Health Disparities in Consumer Genomics

Sadie P. Hutson, PhD, RN, WHNP-BC, FAANP
DTC Genetic Testing - Increase in Innovation?

- Analysis of rare variations is more powerful when sample sizes increase and become more diverse
- **Bias is exacerbating disparity**
Lack of Diversity and Relationship to Health Disparities

- Under-representation of non-European and rural/underserved populations in genomic databases is problematic
- Limited generalizability of findings from genomic research
- Limited evidence base for translation of findings into clinical care, particularly for diverse populations

Rural & Underserved Populations and Engagement with Genomics Services

- Interest in participating in genetics/genomics studies
- Influence of environment on health (mining)
- Divided interest between those felt genetics studies are not risky vs those who had a fear of the unknown
- Concern that knowledge about disease risk would not translate into evidence based strategies to prevent disease occurrence
- Mistrust of healthcare provider knowledge

Mallow et al., 2017
Factors Affecting Use of Consumer Genomics Services

- Lack of awareness of DTC
  - Awareness of DTC lower among rural residents and racial/ethnic minorities (Salloum, et al., 2018)
  - Awareness of genetic testing can vary by acculturation, racial and ethnic identity (Vadaparampil, et al., 2006; Sussner et al., 2009)
- Lack of awareness of genetic counseling (Personalized Medicine Coalition, 2018)
- Lack of access to genetic counseling (Fogelman et al., 2019)
Factors Affecting Use of Consumer Genomics Services

- **Cost** (Personalized Medicine Coalition, 2018)
- **Privacy Concerns** (Personalized Medicine Coalition, 2018)
- **Differences in discussions about genetic testing by healthcare providers** (Goldsmith et al., 2012)
- **Sociodemographic Factors** (Agurs-Collins, 2015)
- **Family Communication**
- **Fear of Discrimination** (Hull & Vassey, 2018)
- **Barriers can change over time**
Advantages of DTC Testing for Underserved Populations

- Patient empowerment
- Affordability (relative)
- Sample collection is non-invasive
- Increased accessibility to genetic testing
- Results may prompt adoption of healthier lifestyle changes
- Increased awareness of diseases that have a genetic component
- Increased patient engagement may lead to improved genetics literacy
Challenges of DTC Genetic Testing for Underserved Populations

- Questions about the accuracy of results in underrepresented populations
- Results of DTC testing are not definitive; additional testing may be needed if there is a clinical indication of increased risk or to confirm identified variants
- Important decisions about treatment and disease management may be made based on incomplete, inaccurate, or misunderstood information
Challenges of DTC Genetic Testing for Underserved Populations

- DTC tests do not achieve improved access to those with barriers to access for general healthcare needs
- Availability of test does not provide assurance that health outcomes will change for patients
- Unintended psychosocial consequences
- Genetic privacy may be compromised if companies use results in an unauthorized manner (use of “wrap contracts” by DTC companies)
Opportunities to Increase Reach

- Genetic Testing
  - DTC, consumer-initiated, provider-initiated
- Acceptability
- Genomic Literacy
- Affordability
Structure of Genetics Services

- Model at Pikeville Medical Center (Testing, Affordability & Acceptability)
  - Oncology & genetics nurse navigators
  - Health Wagon/Invitae
  - Remote Area Medical
- Telemedicine
- Faith-based initiatives
Acceptability of Genomics Services

- Meeting people where they are at:
  - Communication and Time (Williams et al., 2019)
  - Genetics Literacy (what is your understanding of why you were referred?)
Implications for Patients

- Test results can often be confusing
- Conflicting information from multiple companies
- Communication with providers regarding testing and subsequent results may not be happening
- Family implications resulting from testing
Implications for Providers

- The implications for patients are inherently the implications for providers
- Landscape of genetic and genomic testing is rapidly evolving
- Providers of all types must be informed about all of these tests and take the time to seek resources for their practices and for their patients
  - Ex] Germline vs. Somatic cell testing
Case Study - Direct to Consumer

- 31 year-old female- lives 2 hours away in WV, came with her sister for disclosure of genetic test results
- Mother diagnosed with breast cancer at 49
- Sister diagnosed with ovarian cancer at 44
- Maternal grandmother diagnosed with breast cancer at 55
- 23 & Me test revealed "No Variants Detected"
"I feel so blessed that I did not have that breast cancer gene and I can live my life without this worry."
How Can We Achieve Balance?

- Patient Autonomy
- Technological Innovation
- Clinical Utility & Ensuring Patient Safety