Exploring the Role of Diversity and Health Disparities in Consumer Genomics

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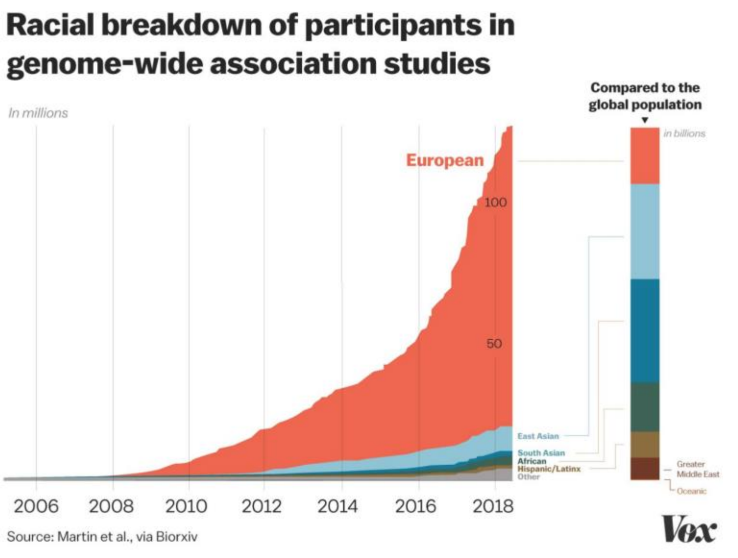
DTC Genetic Testing - Increase in Innovation?

- Analysis of rare variations is more powerful when sample sizes increase and become more diverse
- *Bias is exacerbating disparity*



Racial breakdown of participants in genome-wide association studies

Source: Martin et al., via Biorxiv

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

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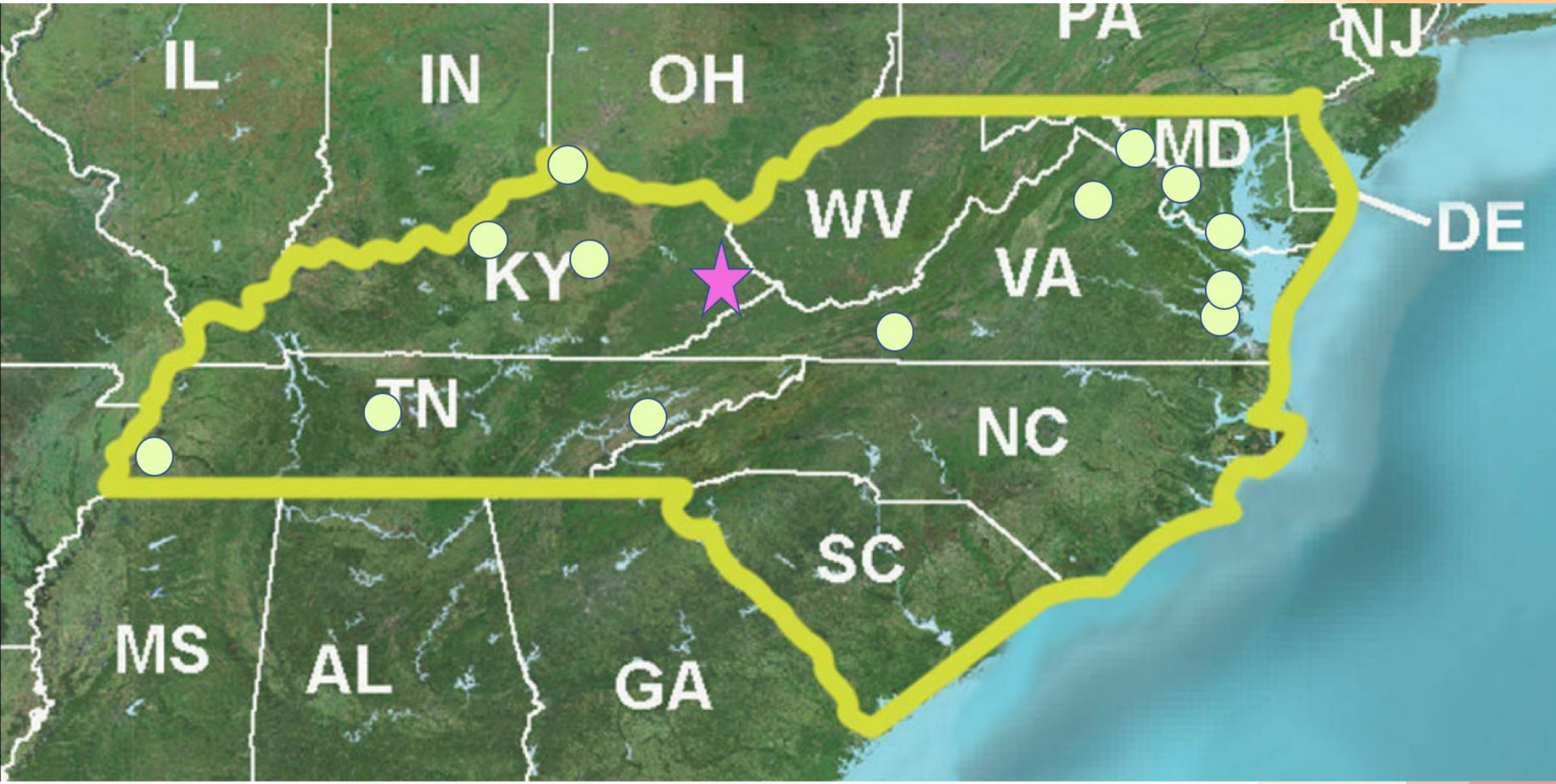
Rural & Underserved Populations and Engagement with Genomics Services

- 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)

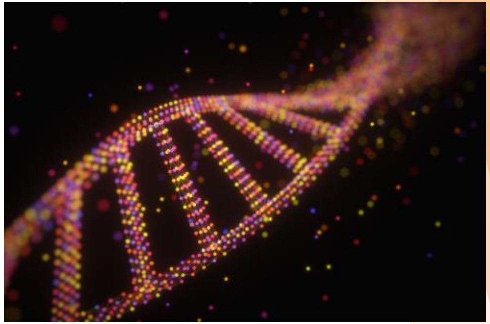


Source: www.nsgc.org

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

Central Appalachia
Health Wagon

Leonard Lawson Cancer Center
Together. We Fight Cancer.

Remote Area Medical
Volunteer Corps

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