

Research in follow-up to testing: uptake of interventions,
cascade testing, transportability throughout the lifespan

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Uptake of Interventions

- How to set realistic expectations – consent process and community engagement
 - Risk of false reassurance – clinical testing vs. screening
- Generation of data on penetrance – and define penetrance for what
- Is there something to be learned from longitudinal follow-up of persons found to be at risk?
- Can population screening be modeled using All of Us dataset?
- Cost/value analysis – is it a moving target?
- Need for standardization of outcome measures
 - Does knowledge of risk improve medical outcomes?
- Point-of-care decision support
 - Genetic counseling
 - Integration into EHR
 - AI systems

Cascade Testing

- How to facilitate communication of information through the family?
- Effects on family relationships
- What accounts for families with higher or lower penetrance?
- Should All of Us encourage family enrollments and linkages between family members?

Transportability through Lifespan

- Reanalyze vs. resequence
 - Changes in technology
 - Fragmentation of health care
- Where does sequence data reside?
 - In health system
 - In research enterprise
 - With individuals
 - In a "DNA data bank"
 - With commercial entities

