

C G T A C G T A
A C G T A C G T

Returning Genomic Findings to Research Participants: Attitudes and Outcomes

NHANES Workshop on Returning Individual Genomic Results

Julie C. Sapp, ScM, CGC

Genetic Counselor, Center for Precision Health Research

December 2, 2022



National Human Genome
Research Institute



Disclaimer

I am a Federal Government employee; no conflicts.

Outline

- How research participants think about and react to options for learning genomic results
- What happens after participants learn their genomic findings

Attitudes

Outcomes

Outline

- How **people** think about and react to options for learning genomic results
- What happens after individuals learn **actionable** genomic findings

Attitudes

Outcomes

Attitudes toward return of results

- Thoughts
- Opinions
- Motivations
- Intentions
- Preferences

of

- Research participants
- Public
- Specific groups
 - Parents
 - Patients
 - Sub-studies

Overwhelming preference for return^{1,2}

- Up to 100% want “all”
- ↑ actionability = ↑ preference for return
- “Normal” findings³ and raw data are also desired⁴
- Judged of interest by researchers⁵
- Very small minority want none or primary only¹

Why?

- Empowering^{1,2}
- Disease-prevention^{3,4}
- Benefit self and/or family^{5,6}
- Curiosity^{2, 5, 7-9}
- Self-exploration/ancestry¹⁰

Why?

- Empowering^{1,2}
- Disease-prevention^{3,4}
- Benefit self and/or family^{5,6}
- Curiosity^{2, 5, 7-9}
- Self-exploration/ancestry¹⁰

Concerns

- Privacy
- Burden of knowledge
- Adverse effects on life

**Factored in but do not
override preference
for return¹**

Ownership and entitlement¹

- Control over “their” data
- Participants have highest investment
- Others may not determine “best interests”
- “Right to know” and autonomy
- Deliberation and restriction viewed as paternalistic

How?

- After broad consent^{1,2,3}
- According to plan and to foster engagement⁴
- Genetic counseling
- Face-to-face

Refusers' attitudes

- Minority¹
- Many studies report hypothetical attitudes^{2,3}
- Refusal may not be durable⁴
 - Of 8,843 enrolled, 165 (1.8%) refused secondary findings
 - 50% initial refusers contacted changed their minds
 - Most (75%) of these believed they HAD agreed

Actionable findings

Attitudes : Outcomes Ratio

A C G
C G T
A C G

Attitudes

Outcomes

Psychological

- Emotions include surprise, relief, and sadness¹
- Regret reported 0-5%²
- Depression/anxiety unchanged³
- Some forget⁴

Family and provider communication

- High numbers report sharing their results with family, especially close relatives^{1,2}
- Disclosure to primary care and specialists common but not universal^{1,3,4}

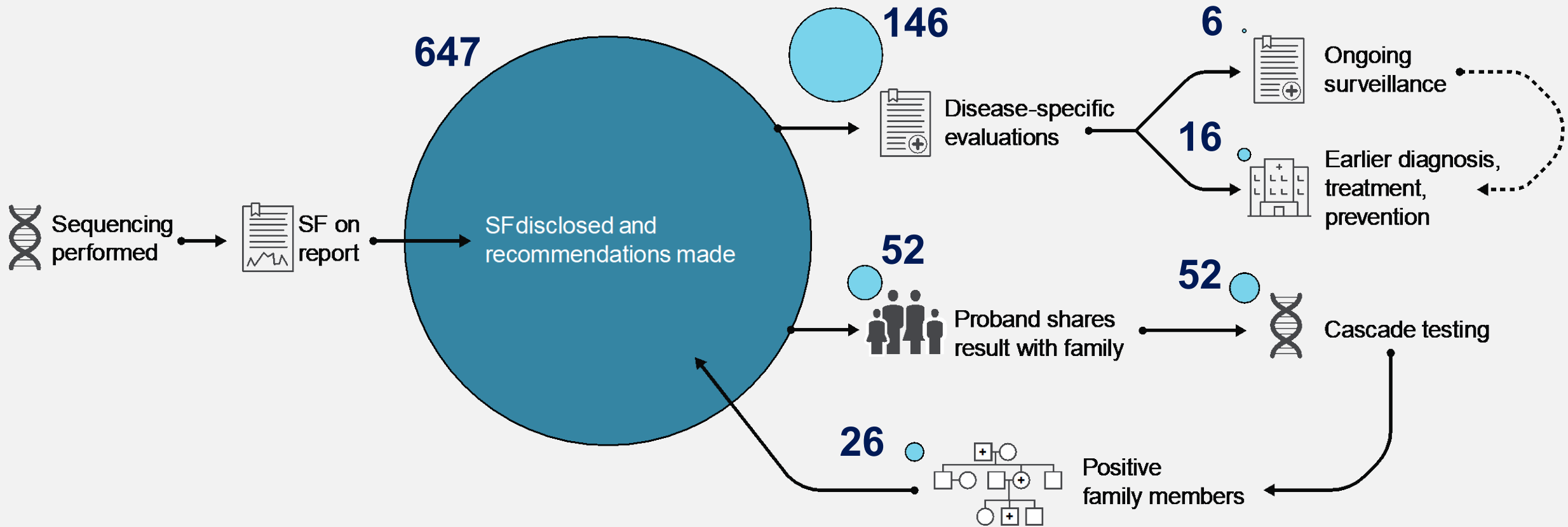
Precision medicine surveillance

- 55%-94% adherence to recommendations¹⁻³
- Genetic counseling and specialist referrals⁴
- Costs lower than expected^{1,4}

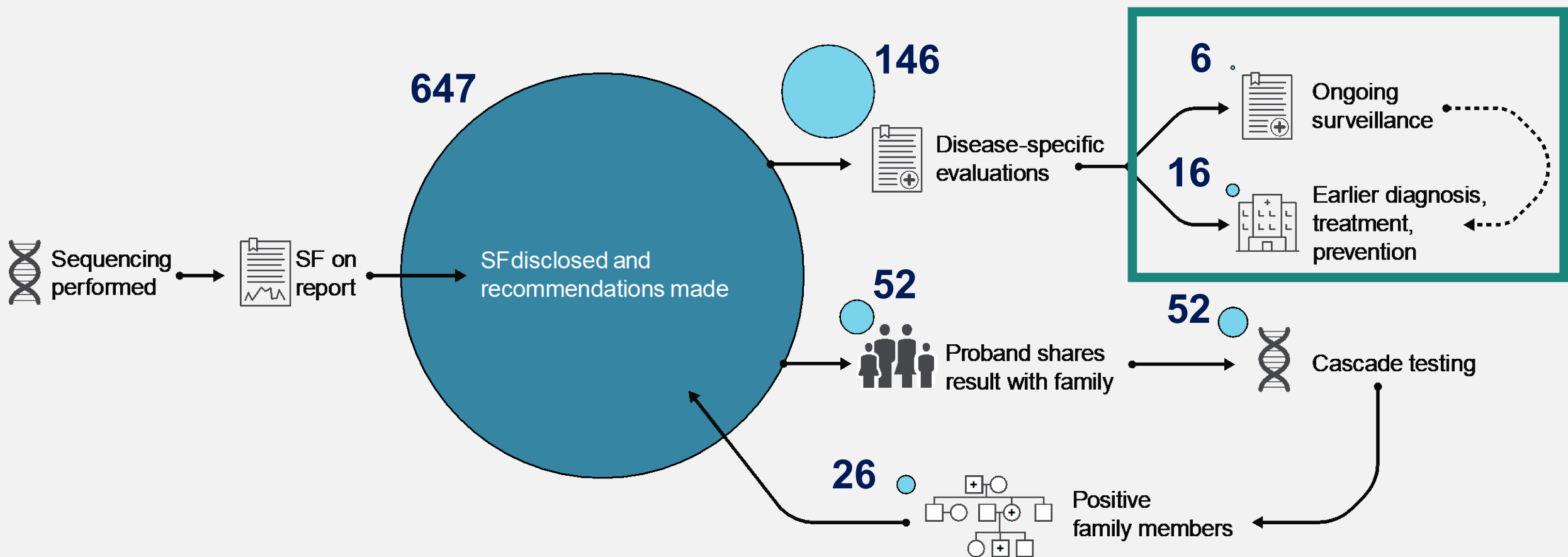
Reports of life-saving treatment

- Medication to avoid QT interval prolongation¹
- Targeted therapy for familial hypercholesterolemia²
- Early identification of lesions by imaging^{2,3}
- Prophylactic surgeries to reduce cancer risk^{1,4-7}

Actionable findings recipients understudied¹

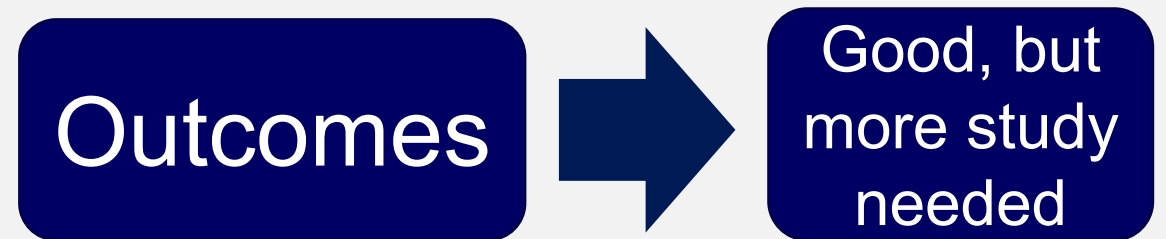
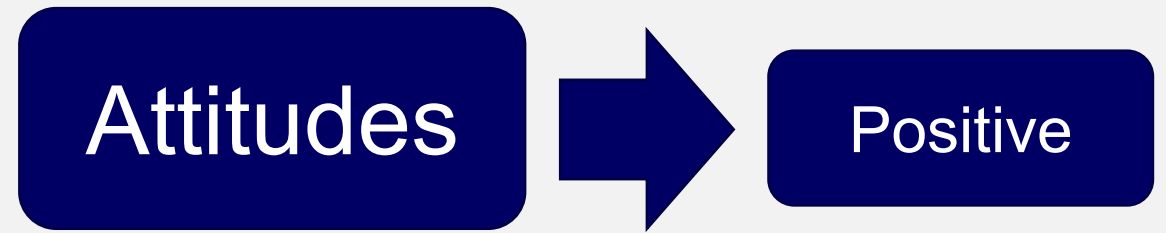


Health outcome data lacking



Conclusion

- How **people** think about and react to options for learning genomic results
- What happens after individuals learn **actionable** genomic findings



A C G
C G T
A C G

Acknowledgments

Leslie G. Biesecker

Katie L. Lewis

Flavia M. Facio

Jennifer J. Johnston

Emily Modlin

Alana Davidson

Alexis Darling

Linton Early

Sophia Hernandez

Nancy Beltran

References

A C G
C G T
A C G



My NCBI » Collections > Sapp NHANES References 2022

✓ Collection permission update successful.

The public URL for your collection is <https://www.ncbi.nlm.nih.gov/sites/myncbi/julie.sapp.1/collections/62402829/public/>

<https://www.ncbi.nlm.nih.gov/sites/myncbi/julie.sapp.1/collections/62402829/public/>