Investigator-Initiated Research on Genetic Counseling Processes and Practices

May 18-19, 2020

Heather Colley, Lucia Hindorff, Dave Kaufman, Nicole Lockhart, Ebony Madden

NIH NHGRI

Reasons for Genetic Counseling

Family planning

Risk assessment

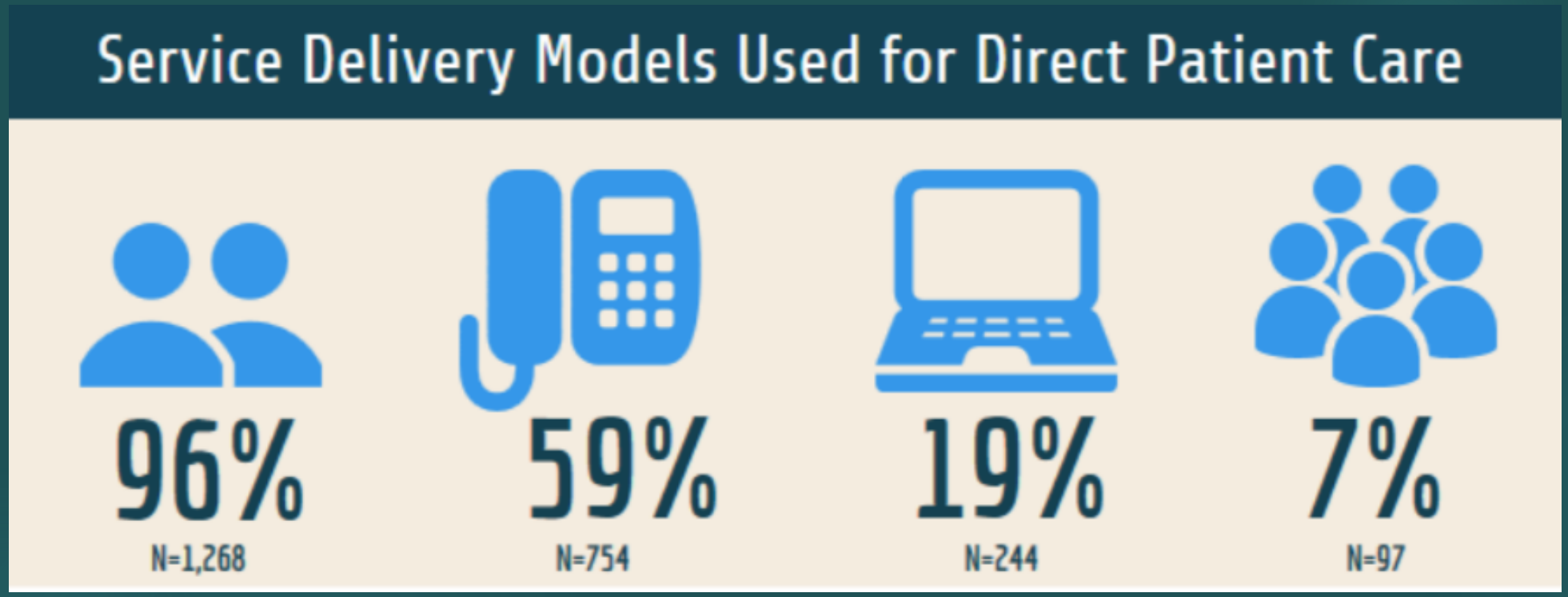
Understanding phenotype

Health management

Era of Genomic Counseling

- Genetic test results have moved from single gene tests to include small risk changes for common diseases and pharmacogenetics
- American College of Medical Genetics and Genomics (ACMG) recommends that genetics experts be made available for patient test result consultations
- Only ~4,200 genetic counselors and ~1,300 clinical geneticists currently employed in the US
- Insufficient to meet current and potential future demand

McGrath, S.P., Walton, N., Williams, M.S. *et al.* Are providers prepared for genomic **medicine:** interpretation of Direct-to-Consumer genetic testing (DTC-GT) results and genetic self-efficacy by medical professionals. *BMC Health Serv Res* **19,** 844 (2019).



Taken from 2018 NSGC professional status survey

- Need more efficient strategies for genetic counseling to provide genomic tests results
- Predicted growth in telemedicine post-COVID
- Continue to provide emotional support as genetic information is translated into healthcare decisions

https://www.nsgc.org/page/healthcareteam

Concept: Investigator-Initiated Research on Genetic Counseling Processes and Practices

Purpose:

- **Assess, innovate, scale, and/or research the implementation of novel genetic counseling practices for genomic medicine**
- **Support investigator-initiated research (R01, R21, R41 & R43) on how to optimize the genetic counseling processes including, but not limited to, the communication of genomic results in the context of limited resources**

Research topics on various approaches to genetic counseling in genomic medicine could include:

- Developing and evaluating processes to triage communication of clinical genomes
- Assessing alternatives to in-person genetic counseling, including IT solutions
- Developing and assessing methods to increase capacity for genetic counseling in underserved areas
- Evaluating and improving strategies to communicate genomic findings and update variant reclassifications
- Understanding needs of patients and stakeholders and the impact of genetic counseling processes on patient outcomes
- Evaluating strategies for including genetic counseling processes in clinical and research workflows

Responsive applications would include:

- Research personnel with experience identifying and overcoming challenges in genetic counseling

- Projects broadly applicable to genomic medicine; projects studying a specific disease area would have to yield generalizable findings.

Current Activities

- NHGRI's genomic medicine consortia (ClinGen, CSER, eMERGE, UDN) have Working Groups which are relevant to genetic counseling practices.

- *All of Us* Research Program recently funded a Genetic Counseling Resource to provide genetic counseling services to enrolled study participants.

- NCI* supports research related to genetic counseling, cancer risk communication and testing strategies to improve inherited cancer syndrome case ascertainment (RFA-CA-17-041, RFA-CA-19-017 and RFA-CA-20-006)

*Collaborating

Mechanism & Funds Anticipated

- R01s up to $500,000 direct costs/year; project period up to 4 years
- R21s up to $200,000 direct costs/year; maximum of $400,000 for grant; project period up to 3 years
- R41s and R43s up to $200,000 direct costs for up to 1 year
- Two receipt dates to allow chance for resubmissions
- Planning for 7 – 9 awards across all mechanisms
- Total cost: up to $5 M per year

Proposed Timeline

- Applications due: November 2020
- Council review: May 2021
- Second round/resubmissions due July 2021
- Second Round Council February 2022

Thank You

Questions?
Suggestions?