Precision Health Genetic Screening: Where is the Pipeline Leaky?

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Background

Offering: Precision Health Genetic Screening (PHGS) is offered through the Program for Precision Medicine in Healthcare (PPMH)
- Determining whether genetic screening should be part of routine healthcare in the future

Purpose: Screens 11 total genes associated with 3 inherited conditions
- Hereditary Breast and Ovarian Cancer Syndrome (*BRCA1*, *BRCA2*)
- Lynch Syndrome (*MLH1*, *MSH2*, *MSH6*, *PMS2*, *EPCAM*)
- Familial Hypercholesterolemia (*APOB*, *LDLR*, *LDLRAP1*, *PCSK9*)

Reason for selected genes: CDC[1] designates these as Tier 1 genomic applications due to the capacity for early detection and intervention

Eligibility criteria:
- Over 18 years of age
- Established UNC Health patient
- No personal or family history requirements
- No federally funded insurance policy
  **due to Beneficiary Inducement Law*

Goals

- After completing the questionnaire, are there demographic differences between individuals that complete the PHGS process and those that do not?
- If we can identify differences, how can we improve retention rates in PHGS by identifying and addressing any areas of inequity that disproportionately affect certain groups?

Methods

1. Pulled patient questionnaire responses since February 2022 from Epic medical records system
2. Sorted data into two categories: those who completed screening, and those who did not complete screening
3. Analyzed data from each category for age, sex, and self-reported ancestry

Citations

1. Abul-Husn NS, Soper ER, Braganza GT, Rodriguez JE, Zeid N, Cullina S, Bobo D, Moscati A, Merkelson A, Loos RJF, Cho JH, Belbin GM, Suckiel SA, Kenny EE. Implementing genomic screening in diverse populations. Genome Med. 2021 Feb 5;13(1):17. doi: 10.1186/s13073-021-00832-y. PMID: 33546753; PMCID: PMC7863616.

Workflow

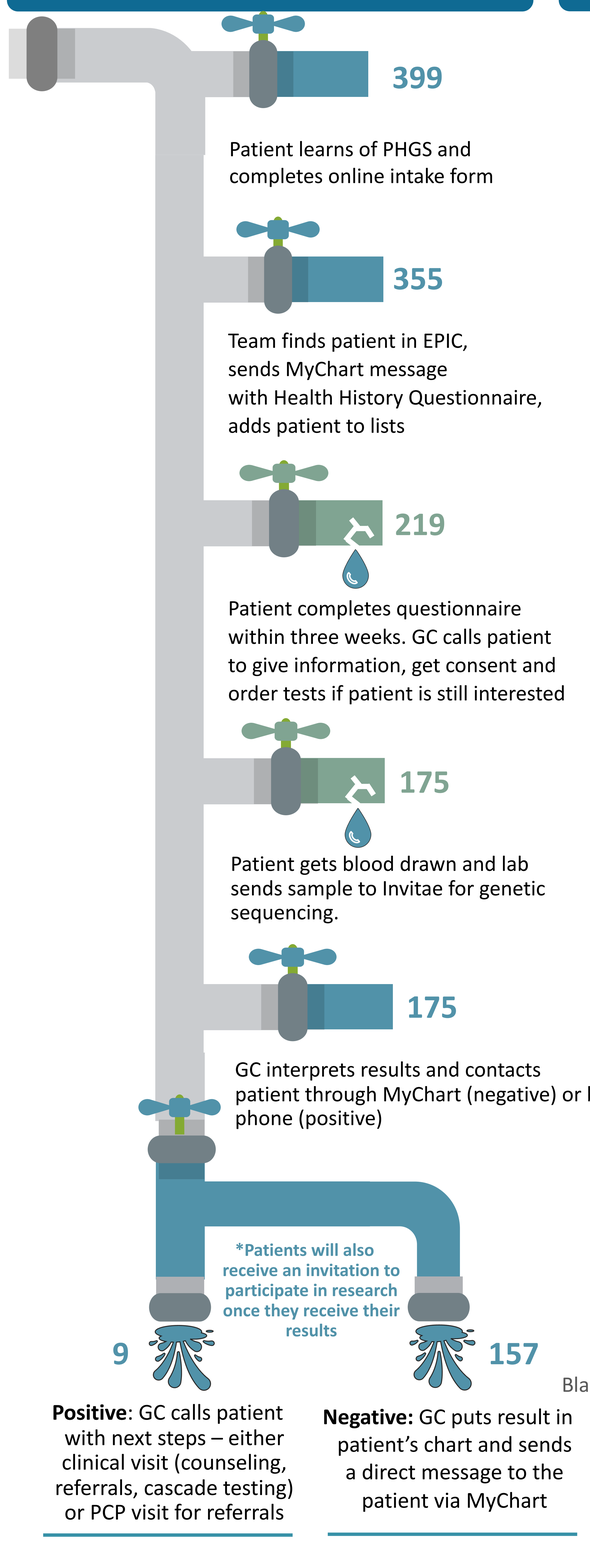

Figure 1. *Outline of the PHGS workflow process from initial distribution of the intake form through documentation of lab results by a genetic counselor. 9 results still pending.*

Results

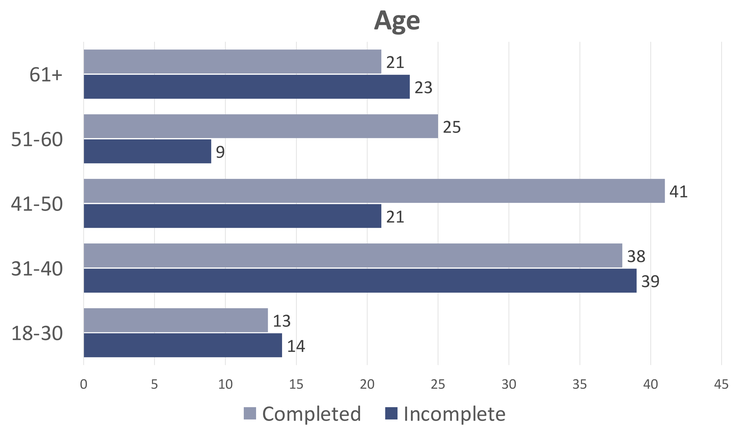

Figure 2. *Graph comparing the ages of participants as of February 2022 who completed labs for PHGS, and those who did not complete the process.*

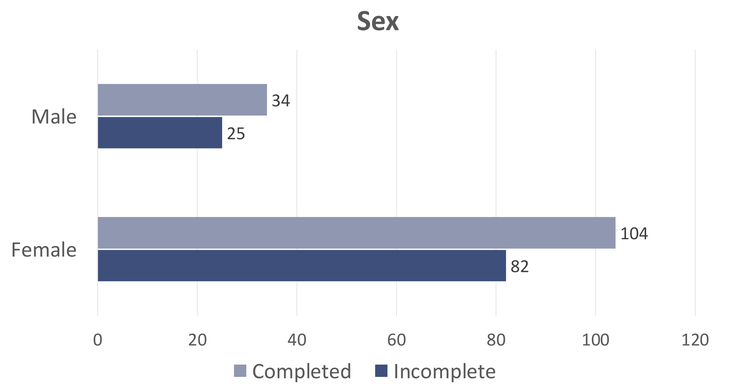

Figure 3. *Graph comparing the sexes of participants as of February 2022 who completed labs for PHGS, and those who did not complete the process.*

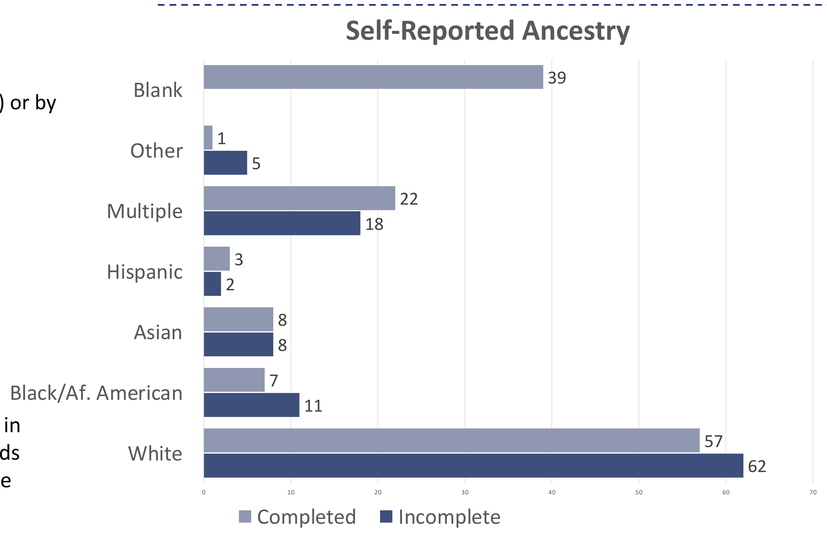

Figure 4. *Graph comparing the self-reported ancestry of participants as of February 2022 who completed labs for PHGS, and those who did not complete the process.*

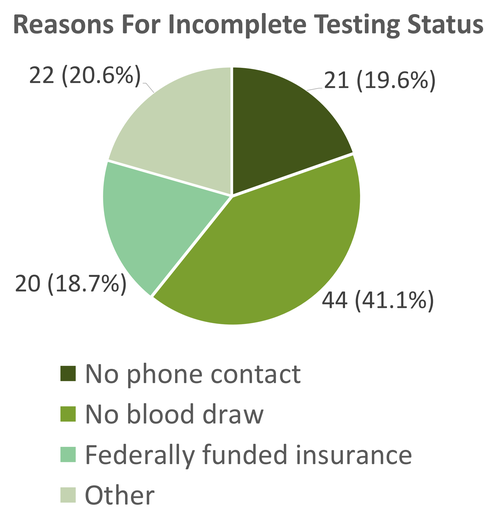

Figure 5. *Comparison of the reported reasons why applicants were given the status of "declined" or did not complete PHGS labs. N=107*

Conclusions

- No substantial differences between the quantity of complete and incomplete participation within sex and ancestry subsections
- Age groups 18-30, 31-40, and 65+ showed similar numbers for completion and noncompletion
- Age groups 41-50 and 51-60 showed far greater numbers for completion than noncompletion
- Most common reason for an incomplete participation status was no blood drawn

Future Directions

- Identify reasons why applicants pass the consent stage but do not complete the labs by reaching back out to individuals
- Address the subsection of applicants who do not answer their phone by possibly exploring alternative initial contact methods
- Collaborations with primary care offices to offer in-office blood draws to minimize the inconvenience of a separate appointment

Acknowledgements

- Funding from the Program for Precision Medicine in Healthcare (PPMH)
- The PHGS team, especially Kriste Kuczynski and Kim Foss
- The PHGS participants who made this possible