Multilevel barriers and facilitators to widespread use of preconception carrier screening in the United States

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Supplemental Materials: Table of Contents

Study Fact Sheet

Below is a copy of the Study Fact sheet sent to participants at recruitment, which provides information about the information the informants had about the study and the research team.

Elucidating the Barriers and Facilitators to Implementation of Widespread Preconception Genetic Carrier Screening: Study Fact Sheet

Principal Investigator: Leland E. Hull, MD

What is the purpose of this research study?

• This research study has two purposes:

o First, we are interviewing key informants with diverse areas of expertise about their perceptions of the barriers and facilitators to implementation of population-based preconception genetic carrier screening to better understand the multi-level factors involved in implementation of a screening program.

o Second, we hope to identify different attributes of how preconception carrier screening could be offered to patients that might impact whether or not they would participate in screening. These data will be used to inform the design of a patient-facing survey used to elicit patient preferences for how carrier screening is performed.

• We are asking you to take part in this study because you have been identified by as a key informant whose perspective would be valuable in achieving the above goals.

What is involved with this research study?

• If you are interested in joining the study, you will be asked to participate in a video interview lasting approximately 30 minutes. There will not be any follow-up required outside of the interview.

• Verbal consent to participate will be obtained at the start of the interview.

• Your interview will be recorded and transcribed. The research team will review the results of the different interviews together, looking for common themes. Any personal identifiers will be removed from the transcripts.

What are the risks and possible discomforts?

• Interview questions may make some people feel uncomfortable. Participation in the interview is voluntary and you can stop at any time. You may also skip any question(s) that you wish.

What about privacy and confidentiality?

• There is a risk of a loss of privacy associated with being part of this study.

• To mitigate this risk, you will be assigned a participant identification number. Your transcript will be scrubbed to ensure that it does not contain any personal identifiers. The key linking your participant identification number to your personal information will be stored in secure electronic folder. Your de-identified information will not be used or shared with other researchers outside of this study.

• Please note that we are asking you to participate in this interview as an individual, and not as a representative of any organization or company. In the presentation of results, we will describe your role in general terms only to protect your identity (e.g. obstetrician involved in clinical guideline development, genetic testing laboratory leadership team member, genetic counselor, policy maker,

ethicist, etc). However, it may still be possible that a reader could deduce your identity, given your prominence in the field.

Is there any remuneration associated with this study?

• You will receive a $50 Amazon gift card after the interview for participating in this study.
• The gift card will be sent to you electronically through your preferred email address within 1 month following your interview, along with an email from our team prompting you to check for this.

Who is doing this study?

Leland Hull, MD at Massachusetts General Hospital is doing this study as part of her NHGRI-funded career development award (Project Number: K08 HG012221-01). You can contact her 24/7 at [redacted] or email her at [redacted]. If you have questions about the interview schedule, email the study's clinical research coordinator [redacted]. If you'd like to speak to someone not involved in this research about your rights as a research subject, or any concerns or complaints you may have about the research, contact the Partners Human Research Committee at [redacted].

[HIPAA Privacy Notice to follow]

Key Informant Interview Guide

Introduction and Fact Sheet Review:

[This section is not audio-recorded. Please read verbatim to the participant]:

Thank you for agreeing to meet with me today. Before we get started, I just wanted to ask, did you receive and review the confirmation email and the attached study fact sheet?

[Wait for response. If they have not reviewed the Study Fact sheet, ask them to take the time to review it.]

Do you have any questions before we get started?

[Wait for response.]

So, we wanted to ask you to describe the work you do before we turn on the recording?

[Wait for response.]

Thank you. We have recorded this information for our own internal records, but we will only use generalized information about our study participants in the presentation when we present the results. If you would like, we can contact you prior to the public presentation of any results to ensure that you are comfortable with the generalizations we've made. Would you like for us to recontact you before we present any results publicly?

[Wait for response.]

Okay, let's get started. We will be video recording from this point forwards as we describe the study and review verbal consent language with you. Please refrain from this point on from referring to yourself or any organizations you may be associated with by name from to minimize privacy risks.

I will now turn on the recorder.

Introduction and Recording Consent:

[Please start by stating interviewer name, participant ID number, and the date for the record.]

[Please read verbatim to the participant]:

Thank you for again. The purpose of today's interview is to better understand your perceptions of reproductive genetic carrier screening, variation in how carrier screening can be delivered to patients, and the barriers to broadening access to preconception screening.

Your participation in this interview is voluntary. The interview will be video recorded and transcribed. The risks and benefits of participating and study procedures were laid out in the Study Fact Sheet you reviewed. You may refuse to answer any questions and you can discontinue your participation at any time by asking me to stop.

Do I have your permission to proceed with the interview?
[Response is audio-recorded]

[For the Interviewer: Please read verbatim statements in bold. For each question, there are core questions with optional italicized prompts. Please use the core questions and prompts as you see fit based upon the key informant's background and whether the topic has or has not already been covered. Additional probes and follow-up questions may be used at the interviewer's discretion]

Section I: Introduction & General Perceptions

[Please read verbatim]:
Thank you again. Let's go ahead and get started.

Question and Prompts	**CFIR Domain**	**CFIR Construct**
• **To start broadly, what do you think are some important priority areas in reproductive health? Do any of these involve genetic testing?** • **For this interview, I'd like to focus on reproductive genetic carrier screening. C*ould you please describe what reproductive genetic carrier screening means to you?*** o *What do you see as the goals of this screening?*	Characteristics of Individuals	Knowledge & Beliefs about the Intervention
• **How important is reproductive carrier screening to you, the patients you serve, or your organization?** o *Who should be screened? When should it be used?*	Intervention Characteristics	Relative Priority
• What advantages does preconception screening have compared to screening performed in the prenatal period? • What disadvantages? o *What kind of supporting evidence or proof would be needed about the effectiveness of the intervention to recommend wider implementation of preconception carrier screening?*	Intervention Characteristics	Relative Advantage Evidence Strength and Quality (prompt)
• How well and in what ways do you think preconception carrier screening would meet the needs of the patient population you/your organization serves? o *To what extent do you think the individuals/patients served by your organization will respond to preconception carrier screening programs?*	Outer Setting	Patient Needs & Resources

○ *What barriers will the individuals served by your organization face to participating in screening?* ○ *What would make it easier for them to get screened?*		
• What changes must be made to increase access to preconception carrier screening [for the broader U.S. population]? ○ *Individual/micro level changes? Broader systems or policy changes?*	Intervention characteristics	Adaptability
• What kinds of initiatives or activities are already happening that you are aware of (in your organization or elsewhere) to increase access to carrier preconception screening? ○ *Are you specifically focusing on preconception screening?* ○ *What is the relative priority of facilitating implementation of preconception carrier screening, compared to other initiatives to improve reproductive care?*	Inner Setting	Relative Priority
• What kinds of changes in how preconception care is delivered will be needed to improve access to preconception carrier screening? ○ *Changes in scope of practice? Changes in formal policies? Changes in information systems or electronic records systems?* ○ *What kinds of approvals will be needed? Who will need to be involved?* • Can you describe the process that will be needed to make these changes? ○ *How will the infrastructure of the U.S. healthcare system affect the broad implementation of preconception carrier screening programs? How will it facilitate/hinder implementation?*	Inner Setting	Structural Characteristics
• Please describe if any materials, resources, training, and/or education is needed to facilitate implementation of preconception carrier screening programs.	Inner Setting	Available Resources Access to Knowledge & Information
• **Can you tell me what you know about any organizations such as health care systems, non-profits, or commercial entities that have implemented preconception carrier screening programs or found ways to widen access to this service successfully?** ○ *Can you tell me more about [program/service]?*	Outer Setting	Peer Pressure

• **What kind of local, state, or national performance measures, policies, regulations, or guidelines could impact the implementation of preconception carrier screening programs?** o *What kind of financial or other incentives influence the decision to implement preconception carrier screening programs?*	Outer Setting	External Policies and Incentives

Thank you so much for answering my questions so far.

There is a lot of variation in how preconception carrier screening can be done. In this next set of questions, I'd like for you to take me through the process of how carrier screening could be performed, describing different options for how this testing could be done. For example, who might provide the screening and/or manage the results? Who might be tested? When would carrier screening be done? In what setting/where might it be performed? What different options for testing might be available? What might they cost? How might the process of being screened work?

If you could walk me through the diverse ways that this screening could be performed, that would be helpful. I will make some notes as we discuss this.

[Prompt: Pause and try to prompt patient to think about more variables. Also, the interviewer could ask for more detail about how different steps in this process might vary where they see fit. For example, prompts could include: When might be good potential times to offer preconception carrier screening? What might screening cost a patient?]

What aspects of this process do you think might influence whether a <u>patient</u> would be willing to participate in preconception carrier screening? [Can review list of variables with the participant.]

In your opinion, which of these things do you think would be most important to patients who are considering whether to pursue preconception carrier screening? Please limit to 1-2 items.

Thank you. We've come to the end of our prepared questions. At this time, I'd like to ask if there are any other reflections you would like to share.

COREQ 32-Item Checklist[1]

Topic	Item No.	Guide Questions/Description	Reported on Page No.[a]
Domain 1: Research team and reflexivity			
Personal characteristics			
Interviewer/facilitator	1	Which author/s conducted the interview or focus group?	7
Credentials	2	What were the researcher's credentials? E.g. PhD, MD	Title Page. Study Fact Sheet (supplement)
Occupation	3	What was their occupation at the time of the study?	Title Page, Study Fact Sheet (Supplement)
Gender	4	Was the researcher male or female?	7
Experience and training	5	What experience or training did the researcher have?	7
Relationship with participants			
Relationship established	6	Was a relationship established prior to study commencement?	6
Participant knowledge of the interviewer	7	What did the participants know about the researcher? e.g. personal goals, reasons for doing the research	Study Fact Sheet (Supplement)
Interviewer characteristics	8	What characteristics were reported about the interviewer/facilitator? e.g. bias, assumptions, reasons and interests in the research topic	Study Fact Sheet (Supplement)
Domain 2: Study design			
Theoretical Framework			
Methodological orientation and Theory	9	What methodological orientation was stated to underpin the study? e.g. grounded theory, discourse analysis, ethnography, phenomenology, content analysis	6
Participant selection			
Sampling	10	How were participants selected? e.g. purposive, convenience, consecutive, snowball	7
Method of approach	11	How were participants approached? e.g. face-to-face, telephone, mail, Email	7
Sample size	12	How many participants were in the study?	7

[1] Developed from: Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *International Journal for Quality in Health Care*. 2007. Volume 19, Number 6: pp. 349 – 357.

Non-participation	13	How many people refused to participate or dropped out? Reasons?	7
Setting			
Setting of data collection	14	Where was the data collected? e.g. home, clinic, workplace	7
Presence of non-participants	15	Was anyone else present besides the participants and researchers?	7
Description of sample	16	What are the important characteristics of the sample? e.g. demographic data, date	8-9, Table 1
Data collection			
Interview guide	17	Were questions, prompts, guides provided by the authors? Was it pilot tested?	6-7, Supplement
Repeat interviews	18	Were repeat interviews carried out? If yes, how many?	7
Audio/visual recording	19	Did the research use audio or visual recording to collect the data?	7
Field notes	20	Were field notes made during and/or after the interview or focus group?	7
Duration	21	What was the duration of the interviews or focus group?	7
Data saturation	22	Was data saturation discussed?	7
Transcripts returned	23	Were transcripts returned to participants for comment and/or correction?	No
Domain 3: analysis and findings			
Data analysis			
Number of data coders	24	How many data coders coded the data?	8
Description of the coding tree	25	Did authors provide a description of the coding tree?	8
Derivation of themes	26	Were themes identified in advance or derived from the data?	8
Software	27	What software, if applicable, was used to manage the data?	8
Participant checking	28	Did participants provide feedback on the findings?	Yes, they were offered opportunity
Reporting			
Quotations presented	29	Were participant quotations presented to illustrate the themes/findings? Was each quotation identified? e.g. participant number	9-16, Table 2
Data and findings consistent	30	Was there consistency between the data presented and the findings?	9-16
Clarity of major themes	31	Were major themes clearly presented in the findings?	9-16
Clarity of minor themes	32	Is there a description of diverse cases or a	9

		discussion of minor themes?	

[a]Page numbers reflect the numbers in the original manuscript file