STUDY BACKGROUND
Many couples planning a pregnancy want to find out if they carry genes for conditions that could affect their child. Traditionally, carrier tests have included only a single condition, such as cystic fibrosis, or a small number of genes, such as the Ashkenazi Jewish panel test. With advances in technology, however, couples may soon be offered expanded carrier testing, which can test for hundreds of conditions.

We conducted a randomized clinical trial within the Kaiser Permanente Northwest health plan to learn about how health plans might deliver this new service. We offered some participants and their partners expanded carrier testing, and other participants usual care—standard testing. We identified which carrier test results were meaningful enough to report, tested participants, disclosed test results to them, and evaluated participants and genetic counselors’ experiences with the process.

We gathered qualitative data to discover whether participants would find expanded carrier screening acceptable, understandable, valuable, and useful for family planning. We also wanted to learn what it would take for a health plan to implement such screening.

QUALITATIVE METHODS
We conducted a variety of qualitative methods at different times and for different purposes. We held focus groups at the beginning of the study, gathered process data throughout the study, and conducted evaluative interviews with participants at the end.

Focus Groups
We held focus groups to assess participants’ attitudes and receptivity toward expanded carrier testing using genome screening and to get their feedback on study recruitment materials and consent forms. We also asked for their opinions of the categories of test results we planned to offer participants. Attendees recommended offering couples choices about the kind of information they want from expanded carrier screening.

Process Data
We collected process data to assess barriers to implementing such screening and to gauge participants’ reactions to getting tested and finding out results. In monthly journal entries, genetic counselors and project staff reflected on issues arising in their work—these entries might describe an ethical question, or a concern about how to offer the best care given time constraints. We also documented weekly case conferences where genetic counselors discussed test results amongst themselves prior to giving results to patients—these often touched on how to address sensitive issues, for example how to deliver test results to a participant who was pregnant. Case conferences were audio recorded, transcribed, and
FEATURED QUALITATIVE STUDY

Next Gen: Clinical Implementation of Carrier Testing Using Next Generation Sequencing

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reviewed by qualitative research staff. Research staff also observed visits when genetic counselors disclosed test results to participants and took field notes; the visits were also audio recorded. After these results visits, we held short 15-minute debriefing interviews separately with both counselor and patient and/or partner, for a subset of participants.

Evaluative Interviews
About one year after participants received their test results, we conducted in-depth interviews with the same sub-set of participants that we observed and debriefed previously to understand how they incorporated this information into their lives, and whether they found it valuable. Was any temporary anxiety outweighed by the value of knowing the information? Did they take any actions based on the information?

KEY FINDINGS FROM QUALITATIVE METHODS
Different streams of data all helped to shape our study. The study team reviewed journal entries regularly to scan for issues that concerned genetic counselors; this uncovered a need for more frequent and more formal case conferences, which we implemented. Together, the journal entries and case conferences shaped the questions the study team asked during debrief interviews. What we learned from the debrief interviews in turn informed the questions we asked in our evaluative interviews.

In our evaluative interviews, we asked how participants were affected by results indicating they had a less than 25% risk of having a child with a specific genetic condition. We found that these results had little lasting impact on participants. More than half (61%) said they did not change their family plans due to their results; the remaining 39% said their results made them more confident in going forward.

We also assessed patients’ willingness to pay for expanded carrier screening by combining participant surveys with qualitative interviews. Most women and couples planning a pregnancy were willing to pay for expanded carrier screening. High-income participants were willing to pay more than lower-income participants, however.

Our qualitative research methods yielded important information. Our focus groups suggested health care providers may need to tailor patient materials, depending on patients’ desire for information. Our evaluative interviews showed that low-risk results had little lasting negative effect on our participants. Finally, our surveys and interviews indicated that access to this new service could depend on income. These findings and other lessons from our study can inform health systems as they consider offering expanded carrier testing.


