Misinterpreting the Results: Patient Misconceptions about Genetic Cancer Risk after Obstetrical Carrier Screening Shortened Title: Perceptions of Genetic Cancer Risk in Prenatal Carrier Screening

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Short Report

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Abstract

Obstetric carrier screening (OCS) is recommended for all individuals during pregnancy by leading professional societies. However, patient understanding of the scope and limitations of OCS remains poorly characterized, especially with regards to inclusion of cancer genes on OCS panels. This quality improvement initiative evaluated pregnant patients' knowledge of their OCS results. We contacted 100 pregnant patients who had recently completed OCS and participated in a structured telephone interview following physician disclosure of results. When asked about the content of OCS, 52% of patients were unsure or incorrectly believed that cancer-related genes were included on the panel. After clarification of the specific genes and syndromes tested, 73% of patients reported that they would have elected to undergo hereditary cancer screening had it been offered concurrently with OCS. These findings reveal substantial gaps in patient comprehension of OCS and suggest that many pregnant patients incorrectly assume that cancer susceptibility genes are included in their testing. The high level of interest in hereditary cancer screening following clarification underscores pregnancy as a unique window of opportunity to expand access to cancer genetics. Integrating cancer risk assessment into obstetric care may improve uptake of preventive strategies and broaden the impact of genomics on women's health.

Introduction

Obstetric carrier screening (OCS) traditionally involves genetic testing to identify single autosomal recessive variant alleles whereby, if two copies are inherited by offspring, there can be phenotypic disease.[1] Leading professional societies, including the American College of Obstetricians and Gynecologists and the American College of Medical Genetics and Genomics, recommend that OCS be offered to all individuals during or prior to pregnancy.[1, 2] Currently, approximately 40% of pregnant women in the United States complete OCS, and uptake continues to grow. This trend reflects rising public interest in genetic information, as demonstrated by the increasing popularity of direct-to-consumer genetic and ancestry testing.[3–5] Despite this uptake, little is known about patient understanding of the scope and limitations of OCS. In particular, as OCS panels expand and other forms of testing (such as hereditary cancer screening (HCS)) become increasingly available, patients may confuse one form of testing for another. This misunderstanding could lead patients to assume they have been screened for cancer risk when they have not. This quality improvement initiative aimed to evaluate patient understanding of OCS through structured post-test telephone interviews.

Methods

This quality improvement initiative was guided by the Model for Improvement (MFI) framework.[6] Monthly stakeholder meetings included obstetricians, oncologists, genetics clinicians, genetic counselors, and patient representatives. Retrospective review of patient demographics was approved by the Weill Cornell Medicine Institutional Review Board. Methods and outcomes were reported according to quality improvement guidelines.[6]

Population

Consecutive pregnant patients who completed OCS at three obstetrical practices affiliated with a single academic institution between July 2024 and November 2024 were eligible.

Data Collection

Patients were contacted by telephone 6–8 weeks after OCS sample submission, following results disclosure by their clinical team. A structured interview script, designed by the stakeholder team (Table 1, Fig. 1), assessed patient understanding of OCS, including whether they believed cancer genes were included.

Variables

Patient demographics were abstracted from the medical record. The primary outcome was patient understanding of the scope of their OCS testing.

Results

During the study period, 201 patients completed OCS across the three practice sites. Telephone contact was attempted with all patients, and 100 (50%) were successfully reached and agreed to a telephone interview.

Demographics

Median patient age was 34 years (range 18–43); median gestational age was 20 weeks (range 10–37). Self-reported race was White (52%), Asian (18%), Black (13%), Other (11%), and 6% declined. Fourteen percent of patients identified as Hispanic. Insurance coverage was public for 21% and private for 79%.

Understanding of OCS:

- 53% of patient reported knowing which genes and conditions were included on their OCS panel.
- 19% of patients incorrectly believed OCS included hereditary cancer syndromes.
- 33% of patients were uncertain whether cancer genes were included.

Interest in Combined Testing:

Following questions regarding understanding of OCS, patients were informed by the research team that standard OCS does not include cancer genes. Patients were then asked about inclusion of cancer-related genes on OCS panels. 61% of patients stated that hereditary cancer screening should be offered at the time of OCS. Furthermore, 73% of patients reported that if HCS had been offered at the same time as OCS, they would have elected to complete both tests (Table 1).

No significant differences in responses were observed based on patient age, race/ethnicity, or insurance status or practice setting.

Discussion

This study highlights a significant gap in patient understanding of OCS, with more than half of surveyed patients either uncertain or incorrectly believing that cancer-related genes were included. This misinterpretation is clinically meaningful: patients may assume they have been tested for hereditary cancer risk when they have not, potentially delaying appropriate preventive care. At the same time, our findings reveal high patient interest in hereditary cancer testing during pregnancy. Nearly three-quarters of participants indicated they would accept combined OCS and HCS if offered. These results align with prior studies suggesting strong patient demand for cancer risk assessment during pregnancy.[7, 8] Existing modeling analyses have further demonstrated that adding BRCA testing at the time of OCS is cost-effective, with the potential to prevent cancer cases and deaths.[9]

Clinical Implications

Pregnancy represents a unique window of opportunity for genetic testing. Nearly all women engage with the healthcare system during this period and many undergo some form of genetic screening. Integrating hereditary cancer testing into OCS could: 1) Increase identification of high-risk individuals at a life stage where preventive actions are most impactful. 2) Facilitate early initiation of enhanced surveillance, chemoprevention, or risk-reducing surgery. 3) Offer critical information during family planning and reproductive decision-making

Comprehensive pre-test counseling is essential to ensure patients understand the scope of OCS, particularly as panels expand. Addressing misconceptions proactively can prevent false reassurance and build patient trust in genetic services.

Strengths and Limitations

Strengths of this study include its diverse patient sample across multiple practices within an academic institution and its direct engagement with patients to assess understanding. Limitations include the single-institution design, potential recall bias from surveying patients several weeks after results disclosure, voluntary uptake of OCS introducing selection bias, and a 50% response rate. While we did not identify any patient characteristics that impacted response to survey questions, the study had a small sample size and was not powered to address this question.

Future Directions

Our findings highlight the need for further research on implementing combined OCS/HCS in preconception and prenatal care across diverse healthcare settings. Future studies should evaluate how

to optimize counseling and address potential stress or anxiety associated with cancer genetic testing in pregnancy, while also ensuring equitable access to these services.

Conclusions

This quality improvement initiative demonstrates that a substantial proportion of pregnant patients misunderstand the scope of OCS, with many incorrectly believing hereditary cancer genes are included. At the same time, there is strong patient interest in combined OCS and HCS, with 73% stating they would have accepted such testing if offered. These findings underscore both a gap in patient education and an opportunity to integrate hereditary cancer testing into prenatal care. Leveraging the unique window of pregnancy could expand access to lifesaving prevention and shift the paradigm of cancer genetics nationwide.

Declarations

Conflict of interest disclosure:

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Competing Interests

Melissa K. Frey was supported by the following grants:-American Board of Obstetrics and Gynecology / American Association of Obstetricians and Gynecologists Foundation (ABOG/AAOGF) Scholar AwardMelissa K. Frey reports consultation work for WndrHLTH.Jessica Scholl serves as the Weill Cornell Medicine site principal investigator for Natera's EXpanding Prenatal cell free DNA screening Across moNogenic Disorders (EXPAND) (Protocol ID: 23-075-WH)

Author Contribution

AR: Investigation, Data Curation, Formal Analysis, Writing – original draft, SI: Writing – Reviewing and Editing, SD: Project administration, Writing- Reviewing and Editing, RK: Project administration, Writing- Reviewing and Editing, JS: Project administration, Writing- Reviewing and Editing, MP: Conceptualization Resources, Project administration, ALO: Conceptualization Resources, Project administration, RS: Supervision, Writing – reviewed & editing, MF – Conceptualization, Methodology, Supervision, Project Administration, Writing – Reviewing & Editing. All authors reviewed the manuscript.

Data Availability

All data supporting the findings of this study are available within the paper and its Supplementary Information.

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Tables

Table 1
Survey exploring patient understanding of obstetric carrier screening.

Question	N / %
Did you receive counseling about obstetric carrier screening prior to the testing?	
Yes	94
No	4
Uncertain	2
Do you know what genes/conditions are evaluated with obstetric carrier screening?	
Yes	53
No	34
Uncertain	13
Do you think that the obstetric carrier screening you completed included cancer-related genes?	
Yes	19
No	48
Uncertain	33
Do you think cancer-related genetic testing should be offered with obstetric carrier screening?	
Yes	61
No	15
Uncertain	24
If screening for cancer genes had been offered to you with your obstetric carrier screening, would you have been interested in completing both types of genetic testing?	
Yes	73
No	16
Uncertain	11

Figures

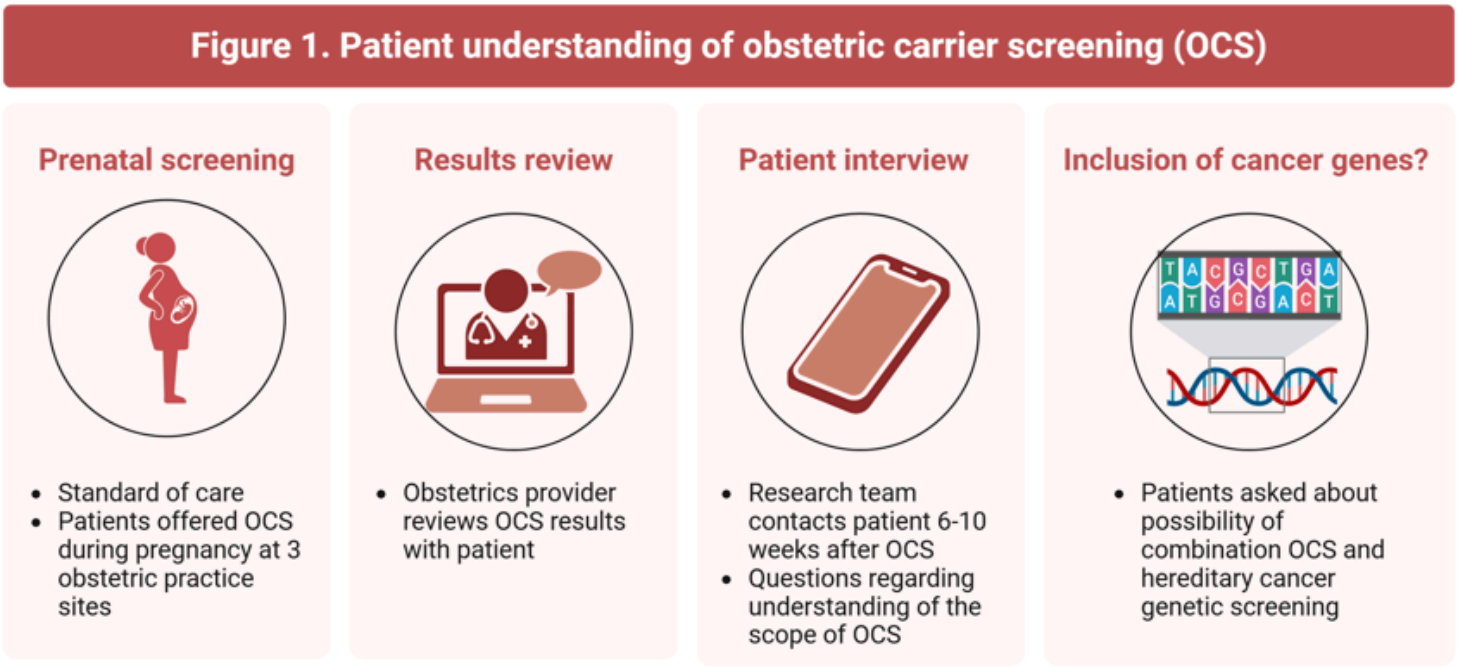

Figure 1

Patient understanding of obstetric carrier screening