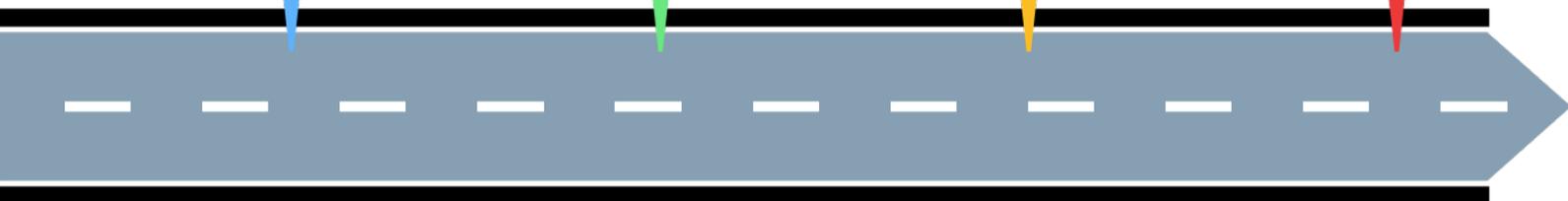




GENETIC SCREENING ROADMAP: A Clinician's Guide to Providing Quality Maternal Health Care



ABOUT SWHR

The Society for Women's Health Research (SWHR) is a national nonprofit and thought leader dedicated to promoting research on biological sex differences in disease and improving women's health through science, policy, and education. Founded in 1990 by a group of physicians, medical researchers, and health advocates, SWHR is making women's health mainstream by addressing unmet needs and research gaps in women's health. Thanks to SWHR's efforts, women are now routinely included in most major medical research studies and more scientists are considering sex as a variable in their research. Visit www.swhr.org for more information.

SWHR's Genetic Screening Programs

SWHR Science Programs identify research gaps and address unmet needs in diseases and conditions that exclusively affect women or that disproportionately or differently affect women. The Genetic Screening programs in **Expanded Carrier Screening** and **Noninvasive Prenatal Screening** were launched in 2021 to address barriers to access and reduce health disparities related to preconception and prenatal genetic testing for women. The programs engage genetic counselors and clinicians, medical professional society leaders, patients and patient advocates, diagnostic company scientists, and health care decision-makers to explore strategies to address knowledge gaps, unmet patient needs, and relevant policies that present barriers to equitable and quality care for women surrounding genetic screening.

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INTRODUCTION

Genetic screening exemplifies the translation of advances in human genomics research into early interventions that promote individual public health benefits. A variety of tools and methods are available to screen adults, fetuses, and newborns — all with a goal of gaining insight into the potential outcome of certain genetic conditions. Genetic screening can help women make informed decisions about their reproductive health and pregnancy management, and may improve maternal and fetal outcomes by allowing preparation to care for children who may have a genetic disorder. However, there is limited independent and culturally-inclusive research assessing the public health implications of preconception and prenatal screening, resulting in significant challenges to integrating research and development advances into existing health care and regulatory systems, as well as gaps in patient care and inequities between populations. The SWHR Genetic Screening Roadmap was developed to raise awareness, expand education, and improve implementation of genetic screening among women and their families, health care providers, and policy stakeholders.

Expanded Carrier Screening

Carrier screening allows individuals to assess the chance of having a child with certain inherited genetic conditions prior to conception or during pregnancy. It can determine the carrier status of reproductive partners for some recessive and/or X-linked genetic conditions that can be inherited by their children. Carrier screening traditionally has been offered based on a patient's ethnicity, as some heritable conditions occur more commonly in specific ethnic groups. However, self-reported ethnicity may not accurately reflect a patient's unique ancestry. As families and societies grow more diverse, ethnicity-based screening has become more challenging, with 20-40% of Americans unable to accurately report their ethnicity. Additionally, research continues to evolve our understanding of genetic etiologies of conditions compared to when practice guidelines were initially developed. Clinical guidelines today include a combination of population and ethnicity-based recommendations and individualized approaches to determine appropriate screening for each patient.

Expanded carrier screening (ECS) tests for a much wider panel of conditions, without regard to ethnicity. While family medical history and ethnicity are often used to guide targeted screening decisions, ECS can be useful for identifying genetic risks if ethnicity is difficult to determine and family history is unknown or insufficient due to the autosomal recessive or X-linked nature of these conditions. However, formal guidelines as to who should be offered carrier screening (all patients or only those of certain ethnicities), the timing of testing (preconception or during the prenatal period), and how to determine which conditions should be included in targeted and expanded screening panels have not yet reached widespread consensus and implementation.

Noninvasive Prenatal Screening

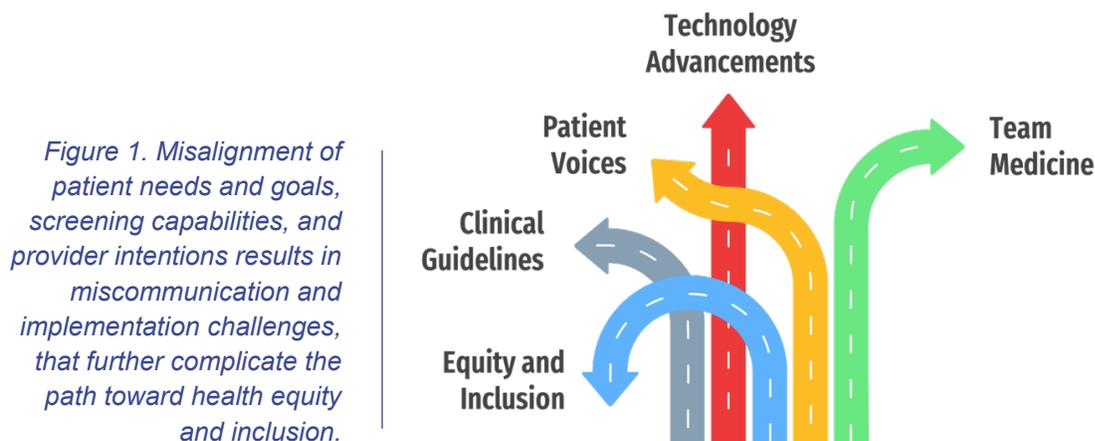
Noninvasive prenatal screening (NIPS) uses a blood sample taken from the mother to examine cell-free DNA (cfDNA) from the placenta that circulates in the mother's blood to determine if the pregnancy is at risk for a chromosomal aneuploidy. NIPS is also referred to as noninvasive

prenatal testing (NIPT) or cfDNA screening. This technology is commonly used to screen for chromosomal conditions such as Down syndrome; however, it is also capable of detecting a number of aneuploid conditions, fetal sex, and some microdeletions and copy number variants. NIPS panels that include single gene disorders are also becoming more readily available. NIPS provides a screening alternative to traditional maternal serum screens to assess the chance for fetal chromosomal conditions during pregnancy. Although NIPS can tell parents whether the fetus is at an increased risk for certain chromosomal conditions, positive results require amniocentesis or chorionic villus sampling to provide a definitive diagnosis.

SWHR GENETIC SCREENING ROADMAP

Genetics can provide keys to advance care for women as they consider incorporating genetic screening in their health and the health of their babies. However, women from diverse backgrounds are often not empowered to receive quality care, and without equitable access to genetic health care, health disparities among women will continue to persist.

Key components – from bench to bedside – must be addressed with a cohesive strategy. Often, scientific and technological advancements in genetic screening move ahead of clinical guidelines. Policies and insurance coverage do not always support access to comprehensive care with the necessary providers that support informed patient decisions. Misalignment of patient needs and goals, screening capabilities, and provider intentions results in miscommunication and implementation challenges. Moreover, attempts at equity and inclusion, if present, often lag or run in a separate direction. See Figure 1.



SWHR convened a cross disciplinary working group of women’s health experts to discuss how to eliminate barriers to access and reduce health disparities related to genetic screening – with a focus on [expanded carrier screening](#) and [noninvasive prenatal screening](#). While a direct path that aligns all goals of access to care is desired, a more realistic description of the path to address knowledge gaps, patient needs, and relevant policy issues is a windy road that requires

strategic turns and detours to reach the destination of equitable, inclusive, and improved health outcomes for expecting mothers and their babies through genetic screening (Figure 2). This path incorporates science, education, and policy to equip providers, engage policymakers, and empower patients along the journey.



Figure 2. Improving health disparities and outcomes through genetic screening is complex, however, the regardless of the path taken, it must incorporate key principles, such as collaboration, science-based policies, education, equity, protecting patient rights.

Primary care physicians or obstetrician/gynecologists are often the providers that will first engage women in their pre-conception or initial pregnancy care. However, other professionals, including nurses, phlebotomists, midwives, and medical assistants, will interact with expectant mothers during health care appointments and could benefit from additional education on the topic. It is important to ensure the various providers in the maternal health ecosystem are equipped to advocate for and meet the specific needs of women and families planning to become pregnant.

SWHR has created a roadmap to assist clinicians in framing and implementing genetic screening across the women’s health continuum by providing: **educational materials and resources** to support genetic screening in clinical practice and **discussion guidance for counseling patients**, including self-assessments of patient and provider values, checklists for identifying complementary resources, and tools to promote cultural competency.

This roadmap is meant to be an educational tool for clinicians. SWHR does not make medical recommendations nor endorse or promote specific screening or diagnostic tests for patients, as listed in this resource or elsewhere.

Education Materials and Resources to Support Clinical Practice

Pre-pregnancy general awareness and education about genetic screening provides a foundation to introduce women about the purpose and limitations of screening tests vs. diagnostic tests. Education among health care providers, patients, and their families also promotes more informed and shared decision-making for optimal health outcomes.

Resources to assist clinicians in understanding the scope of preconception and prenatal testing:

- [CDC Genetic Counseling and Testing](#)
- [AMA Precision Medicine and Genomics: Understanding Implications and Applications](#) (self-paced CME courses)
- [NHGRI Understanding Human Origins and Ancestry](#)
- [NIH Genetic Testing Registry](#)
- [GeneReviews](#) covers diagnosis, management, and counseling for patients concerning single genes, phenotypes, or genetic causes of common conditions

Resources that provide guidance to clinicians on implementing genetic testing:

- Professional Medical Society Recommendations
 - [ACOG Guidance on Carrier Screening for Genetic Conditions](#)
 - [ACMG Practice Resource: Screening for Autosomal Recessive and X-linked Conditions](#)
 - [Expanded Carrier Screening in Reproductive Medicine — Points to Consider](#) (Joint Statement of ACMG, ACOG, NSGC, PQF, and SMFM)
 - [ACOG NIPS Summary of Recommendations](#)
- [Precision Medicine for Your Practice: Expanded Carrier Screening](#) (self-paced online CME course offered by The Jackson Laboratory)
- [Precision Medicine for Your Practice: Prenatal Cell-Free DNA Screening](#) (self-paced online CME course offered by The Jackson Laboratory)
- [Find a Genetic Counselor Directory](#) hosted by the National Society of Genetic Counselors

Discussion Guidance for Counseling Patients

A critical barrier to implementing genetic screening involves patient access to conversations about the purpose and process of incorporating screening into their reproductive health plan. It is important that every patient is provided opportunities for individualized genetic screening. Further, patients should be reassured that they have options and the freedom to choose if they want to be screened and how to navigate the process. Providers should provide additional resources to support informed decision-making.

Below are some recommendations for clinicians to help identify access points and opportunities to engage women patients and their families about genetic screening:

- [ACOG Guidance on Counseling about Genetic Testing and Communication of Genetic Test Results](#)
- [JAX Pretest Counseling Recommendations](#)
- [Noninvasive Prenatal Testing: How Can You Apply New Screening Methods and Updated Guidance for their Use in Your Clinical Practice? \(self-paced online CME course offered by Med Learning Group\)](#)

Engaging in patient conversations:

- [JAX Communicating Genetic Risk](#)
- [JAX Informed Consent Checklist](#)
- [JAX Genetic Information Nondiscrimination Act \(GINA\) Discussion Guide](#) about how GINA protects individuals from misuse of genetic information in health insurance and employment
- [JAX Accessing Genetic Services](#) provides talking points to assist patients in the referral process to secure services

Everyone faces the challenge of suppressing personal biases and values when engaging individuals from different backgrounds. Even the most well-intentioned providers must periodically take a self-inventory to ensure their communication and engagement is culturally sensitive and appropriate to the patients they are serving. Cultural and personal values also affect how patients, in turn, interact with their health care providers and how they will view genetic screening. It is important to adopt patient-centered practices that take into consideration these factors.

Addressing cultural competency:

- [NCCC Cultural and Linguistic Competency for Personnel Providing Health Care Services Checklist](#)
- [Ethnic-Sensitive Inventory](#) counselor self-assessment tool for practitioners working with ethnic minority clients
- [CDHPD Addressing Current Gaps in the Provision of Prenatal Testing to Improve Patient Support and Present Disabilities More Equitably](#)

Below are resources for patients and providers to get additional information and support, especially as a follow-up to positive screening results.

Genetic Condition Organizations:

- [Cystic Fibrosis Foundation](#)
- [The Marfan Foundation](#)
- [National Fragile X Foundation](#)
- [National Tay-Sachs and Allied Diseases Association, Inc.](#)
- [Osteogenesis Imperfecta Foundation](#)
- [Sickle Cell Disease Association of America](#)
- Spinal Muscular Atrophy: [Cure SMA](#)

Patient Support Organizations:

- [Expecting Health](#)
- [Genetic Support Foundation](#)
- [Jewish Genetic Disease Consortium](#)
- [Lettercase: National Center for Prenatal and Postnatal Resources](#)
- [Little People of America](#)
- [Remember the Girls](#) for females with X-linked disorders

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