UNDERSTANDING GENETIC SCREENING
AND MATERNAL HEATH CARE

Society for Women's Health Research

Genetic screening can offer soon-to-be parents and those considering parenthood information about the risk of passing on certain conditions to their child, allowing women and their partners to make informed decisions about planning their family, pregnancy management, and future child-rearing.

CARRIER SCREENING

A carrier is someone who possesses a recessive trait or genetic mutation that does not cause them to display symptoms of the associated disease. Carrier screening is a type of genetic test that allows individuals to assess the chance of having a child with certain inherited genetic conditions for which they may be a carrier.

This type of screening is ideally performed before pregnancy, and has traditionally been offered based on an individual's ethnicity because some heritable conditions occur more commonly in specific ethnic groups. Expanded carrier screening (ECS) tests for a wider array of recessive genetic conditions than traditional carrier screening without regard to ethnicity.

NONINVASIVE PRENATAL SCREENING (NIPS)

NIPS is used to assess the risk of **chromosomal aneuploidy** – when there are extra or missing chromosomes in fetal DNA. NIPS uses a blood sample from the pregnant mother to examine the fetal component of cell-free DNA (cfDNA) from the placenta, which is found in the mother's blood stream. NIPS (also referred to as cfDNA screening) is considered noninvasive because the required blood draw poses minimal to no risk to mother or baby.

	ECS	NIPS
Purpose	Determine risk of passing on certain recessive genetic disorders	Determine risk of fetal aneuploidy
Process	Blood or saliva sample	Blood sample
Common Conditions Screened	Up to hundreds of inherited genetic conditions	Down syndrome (trisomy 21) Edward syndrome (trisomy 18) Patau syndrome (trisomy 13)
Timing	Before or during pregnancy	During pregnancy (as early as 10 weeks)
Can Provide Diagnosis	No	No

Screening is Not Diagnostic Testing

It is important to remember that ECS and NIPS only provide information about risk for certain genetic conditions. Results cannot provide definitive diagnoses.

KEY CONSIDERATIONS FOR PROVIDERS & PATIENTS:

Informed Consent and Shared Decision-Making: The consensus among experts is that all pregnant women should be offered the opportunity to pursue both ECS and NIPS. It is important to provide adequate education about the process and potential outcomes, and to communicate that every patient has the freedom to decide whether or not they would like to be screened.

Insurance Coverage: Access to genetic screening and/or genetic counselor services may be limited due to insurance coverage. However, there are **resources** to assist with identifying additional support for maternal health care.

Health Equity: Equity and inclusion are essential for providing optimal genetic health care. Consider how cultural diversity affects potential genetic conditions to screen, patient-provider interactions, and the overall health care experience.

WHAT TO DO AFTER SCREENING:

- Waiting for results can be challenging for patients. Continue to engage in materials and resources to learn more about genetic screening and women's health, and try to remain positive and pragmatic about the next steps – regardless of the test results.
- If the screening results indicate certain chromosomal abnormalities, parents should be offered and seek counseling and guidance for next steps. Additional tests, such as amniocentesis and chorionic villus sampling, can be used to help diagnose a genetic condition of a baby in utero.

For additional resources, visit **www.swhr.org** to download the **SWHR Genetic Screening Roadmap: A Clinician's Guide to Providing Quality Maternal Health Care.**

SWHR thanks the following sponsors for their support of this educational work:

illumina INVITAE myriad WOMEN'S HEALTH

Published January 2022