Prenatal Genetics Referrals: When to Refer to Genetics

- Genetic risk assessment and testing can be helpful in aiding in the care of individuals with a personal or family history of genetic conditions.
- A genetics referral can be considered for **individuals/couples who are currently pregnant** or for **individuals/couples who are planning to become pregnant**.
- Some genetic conditions can be inherited through either a **mother or father**, while others can be inherited through **both parents** or can be **new in an individual**.
- Those with an unknown or limited family history can still be referred to genetics for evaluation.
- Prenatal genetic risk assessment and testing can:
  - Be performed as part of routine prenatal screening for aneuploidy and open neural tube defects
  - Screen for and diagnose genetic conditions prenatally
  - Impact treatment and management recommendations
  - Inform individuals of possible risks to future children

General Referral Guidelines for Prenatal Indications

A genetics evaluation should be offered to all women interested in prenatal genetic screening or testing. Women who are pregnant or planning to become pregnant who meet any of the following guidelines may be at an increased risk and a genetics referral should be considered: *(adapted from NSGC.org)*

- A personal or family history of a **known pathogenic variant/mutation**
- A personal or family history of a known or suspected **genetic disorder, birth defect, or chromosomal abnormality**
- **Consanguinity** (mother and father are related by blood)
- Women who will be **35 years of age or older** at the time of delivery
- Women with **positive or abnormal results following maternal serum screening**
- Women with **abnormal results following a CVS or amniocentesis**
- **Fetal anomalies** identified via ultrasound
- **Exposure to potential teratogens during pregnancy**, including certain prescription medications, maternal infections, recreational drugs, or radiation
- A history of **stillbirth, SIDs, or 3 or more recurrent miscarriages**

Carrier Screening

Carrier screening may be appropriate for individuals or couples who are interested in learning if they carry a gene for certain genetic conditions. In some cases, it takes two genetic variants for a person to develop a disorder, and through carrier screening, couples who are carriers can be counseled about their risk of having a child with that genetic condition.

- **Carrier screening for Cystic Fibrosis (CF) and Spinal Muscular Atrophy (SMA) should be offered to all pregnant women.**
- Carrier screening is recommended for **individuals from certain ethnic backgrounds** that are associated with a higher carrier frequency. **This includes individuals with Ashkenazi Jewish, African American, French-Canadian, Mediterranean, and/or Asian ancestry**.

Full referral guidelines are referenced on the back of this handout and are periodically updated. If you are concerned about a patient's personal or family history, a genetics professional can help determine if further risk assessment and/or genetic testing is indicated.

Prenatal Screening and Testing Overview

Test Name & Type of Test	Timing	Description
Carrier Screening *Screening Test*	Preconception or any time during pregnancy	Genetic testing performed in one or both parents in order to determine risk for child to be affected with certain recessive or x-linked genetic conditions.
Maternal Serum Screening *Screening Test*	First Trimester: 10 to 13 weeks Second Trimester: 15 to 22 weeks	First trimester: Maternal blood test paired with ultra-sound exam to screen for trisomy 21 and trisomy 18. Second trimester: Maternal blood test to screen for trisomy 21, trisomy 18, and neural tube defects.
Non-Invasive Prenatal Screening/ Testing (NIPS/NIPT) *Screening Test*	10 weeks and beyond	Maternal blood test to screen for trisomy 21, trisomy 18, trisomy 13, and sex chromosome abnormalities.
Chorionic Villus Sampling (CVS) *Diagnostic Test*	10 to 13 weeks	Analysis of fetal cells from chorionic villi sample which can diagnose chromosomal abnormalities and other genetic conditions.
Amniocentesis *Diagnostic Test*	15 to 20 weeks	Analysis of fetal cells from an amniotic fluid sample which can diagnose chromosomal abnormalities, neural tube defects, and other genetic conditions.

Take Action

If your patient meets any of these criteria:

- Talk to your patient about recommendations for a genetics referral
- For assistance locating the nearest genetics service provider, in the New York-Mid-Atlantic Consortium (NYMAC) Region, please contact the **Genetic Services Referral Call Center** at **1-833-545-3218** or visit our website.

Guidelines and Recommendations (Links)

The Professional Practice and Guidelines Committee of the American College of Medical Genetics and Genomics (ACMG) Indication for genetic referral: a guide for healthcare providers

The American College of Obstetricians and Gynecologists (ACOG): Prenatal Genetic Screening and Diagnostic Testing

National Society of Genetic Counselors (NSGC) Practice Guideline: Prenatal Screening and Diagnostic Testing Options for Chromosome Aneuploidy

Resources (Links)

- **March of Dimes**
  A non-profit organization focused on education about and improving the health of mothers and babies
- **National Society of Genetic Counselors (NSGC) Find a Genetic Counselor**
  A tool developed by NSGC for patients and providers to locate genetic counseling services in North America (U.S. and Canada)
- **GeneReviews through the NIH**
  Resource for providers about the diagnosis and management of patients with genetic conditions

NYMAC REGIONAL GENETICS NETWORK

NEW YORK STATE Department of Health | Wadsworth Center

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