MFM & GENETICS
OF ADVANTIA

PATIENT INFORMATION

CARRIER SCREENING

What is carrier screening?

Carrier screening is a type of genetic testing performed on a blood or saliva sample that helps determine if you carry a change in a gene for certain inherited conditions that could be passed on to your child. This test can be performed prior or during a pregnancy.

How does carrier screening work?

- Every person inherits two copies of most genes—one from each parent. Females (XX sex chromosomes) inherit one X chromosome from each parent. Males (XY sex chromosomes) inherit an X chromosome from the egg and a Y chromosome from the sperm.
- A "carrier" has one normal copy of a gene and one altered (changed) copy of a gene. In most situations, carriers are healthy and show no symptoms of the condition. Most carriers will have no history of the condition in their families.
- For an autosomal recessive condition, if both parents are carriers of a change in the same gene, there is a 25% chance in each pregnancy that both changed genes would be inherited and a baby would have the associated condition.
- X-linked conditions involve genes on the X chromosome. If an individual with two X chromosomes is a carrier of an X-linked condition, there is a 50% chance that the changed gene will be inherited. X-linked conditions are more likely to affect males who typically only have a single copy of the X chromosome.

What conditions are screened for?

Screening panels can vary from limited to expanded based on patient preference, and focus on childhood-onset conditions that may affect quality of life, life expectancy, or recommended medical treatment. Some common conditions include:

- **Cystic Fibrosis:** Affects breathing and digestion.
- **Spinal Muscular Atrophy:** Causes muscle weakness and mobility issues.
- **Fragile X Syndrome:** Associated with intellectual disability and autism, more commonly in males.

Who should consider carrier screening?

- Anyone planning a pregnancy or currently pregnant.
- Those with a family history of genetic conditions.
- People of specific ethnic backgrounds at higher risk for certain conditions (e.g., Ashkenazi Jewish, Mediterranean, African, or Southeast Asian heritage).
- Couples using egg or sperm donors.

How is the screening done?

1. **A blood draw or saliva sample is collected.**
2. **Screening Results:** If your results are negative.low risk, no further testing is needed.
    - If you are a carrier for a recessive condition, your partner would be tested to assess the pregnancy's risk.
    - Fox an X-linked condition, testing of your partner may not be needed to determine the risk for a pregnancy.

What happens if both parents are carriers?

If both parents are carriers for the same recessive condition, or an individual carries an X-linked condition that puts a pregnancy at risk, a genetic counselor and your Doctor will review results and guide you through next steps. Additional testing options may include:

- **Diagnostic testing during pregnancy:**
    - **Chorionic Villus Sampling (CVS):** Performed in the 1st trimester.
    - **Amniocentesis:** Performed in the 2nd trimester and beyond.
- **Preimplantation Genetic Testing (PGT):** For couples undergoing IVF, it may be possible to screen embryos for the condition before transfer.

Benefits of carrier screening

- Helps you make informed decisions about your pregnancy and family planning.
- Provides peace of mind if no significant risks are identified.

Limitations of carrier screening

- Only screens for the conditions included in the panel - does not detect all genetic conditions, does not evaluate all genes and cannot detect all carriers.
- A negative result reduces, but does not eliminate, the possibility of being a carrier.
- Not all conditions screened have available treatments.
- Limited information may be available about what a condition could mean for an at risk individual.
- You may receive unexpected information about your own health, which can have implications for life insurance, disability and long-term care insurance.