How is carrier screening helpful?

Carrier screening can let a couple or individual know if they are at an increased chance to have a child with an inherited genetic condition. By assessing specific genes through a blood or saliva sample, carrier screening can identify if certain genes are not functioning as expected, potentially causing a genetic condition if passed down. Carriers, typically, do not have any symptoms of the condition.

If someone or a couple are identified as carriers, then they may have a 25% - 50% chance to have a child with that condition.

With this knowledge, individuals can explore options like diagnostic testing during pregnancy or in-vitro fertilization (IVF), and learn about the necessary treatment and care for a child with the condition.

Modern Reproduction's mission is to facilitate the discussion on the current technologies and genetic testing options available prior and during pregnancy by educating, empowering, and embracing modern reproduction. This handout does not serve as medical advice. Please refer to Modern Reproduction's Disclaimer and Terms and Service on their website.

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Carrier Screening

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What Conditions Are Assessed?

Carrier screening focuses on assessing the certain inherited genetic conditions, including Sickle Cell Disease, Cystic Fibrosis, and Spinal Muscular Atrophy. It does not cover all possible genetic conditions, The conditions that are tested can have two types of inheritance patterns or ways the conditions are passed on in the family: autosomal recessive and x-linked.

For autosomal recessive inheritance, both members of a reproductive pair are carriers of the same condition. There is a 25% chance to conceive a pregnancy with the genetic condition.

For x-linked inheritance, genetic females have a 50% chance to pass on their nonworking copy of the gene to a pregnancy. If they are having a male baby, then there is a 50% for him to have the condition. Females can have symptoms of the condition as well, however, and may need additional evaluations and monitoring.

What Options Are There?

If a person or reproductive pair are identified as carriers for a specific genetic condition, there are several options available:

- In-Vitro Fertilization (IVF) with genetic testing of the embryos: This allows the selection and implantation of embryos that unlikely have the genetic condition.
- IVF with donor sperm or egg: Using a donor who is not a carrier can eliminate the risk of passing on the genetic condition.
- Adoption or donor embryo: Choosing to adopt or use a donated embryo can provide the opportunity to build a family without the genetic risk.
- Conceive a pregnancy and undergo diagnostic testing: Diagnostic testing can be performed to determine if the baby has the genetic condition. Based on the results, options such as adoption, termination, or preparing for parenting and treatment can be considered.
- Choosing to be childfree: Given the increased chances of having a child with the genetic condition, some individuals or couples may decide not to pursue pregnancy
- Conceive and test the baby after birth

What Next?

Deciding whether to undergo carrier screening is a highly personal choice. While some individuals find the information empowering and valuable for their reproductive planning, others may feel overwhelmed or face barriers to accessing options based on the results. Genetic counselors play a crucial role in assisting individuals in making informed decisions about testing, determining which conditions to assess, and providing guidance throughout the process. If you would like more information about carrier screening, you can reach out to genetics@modernreproduction.org.