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Genetic Carrier Screening: Historical Perspective and Overview

GENETIC CARRIER SCREENING AND counseling is an important part of preconception and prenatal care.[1] As genetic testing technology rapidly evolves, clinicians often face the question regarding the most appropriate testing methods to use for their patients. In addition, prospective mothers often have questions about whether to undergo a particular screening procedure. This article, the first of a 3-part series, reviews the purpose of genetic carrier screening and summarizes current preconception and prenatal screening guidelines.

Inheritance of Autosomal Recessive Disorders

Genetic conditions are inherited in one of several patterns.[2] In autosomal recessive disorders, both copies of the gene need to carry the mutation for a patient to have the disorder.[2] Heterozygous carriers have 1 copy of the mutated gene, do not show symptoms of the condition, and may be unaware of their carrier status.[1,2] If 2 reproductive partners are both heterozygous carriers, the affected offspring has a 1 in 4 risk of inheriting the mutation in both gene copies, 1 from each parent.[1,2] This child will be homozygous for the mutation and will be affected by the condition.

Purpose of Carrier Screening

Carrier screening allows individuals without a known personal or family history of recessive disorders to learn about potential risks that may affect their offspring.[3] If screening reveals carrier status in both reproductive partners, they can receive information about the disease's natural history and management, as well as available reproductive options.[1] In this way, couples can make autonomous choices.[3] Genetic screening and counseling is an important part of preconception and prenatal care.[1]

If a patient is a carrier for a genetic condition, the reproductive partner should also be tested. Results acquired from the test can lead to more accurate information regarding potential reproductive outcomes.[4] Genetic counseling should be available for couples in which both partners have been identified as carriers of a genetic disorder.[4] Reproductive options to reduce the risk of an affected offspring should be discussed.[4] In addition, if an individual is found to be a carrier for a genetic disease, the individual should be encouraged to inform his or her relatives because these relatives are also at risk of carrying the same genetic mutation.[4]

Carrier screening and counseling performed before pregnancy allows couples the opportunity to consider their best reproductive options.[4] The range of reproductive possibilities include the choice to not attempt conception, use of reproductive technologies such as donor gametes and preimplantation genetic diagnosis, and other family building options such as adoption.[1,4] When genetic screening is performed during pregnancy, knowledge of carrier status allows patients to consider pregnancy management options, including early prenatal diagnosis, pregnancy termination, or planning for the birth of an affected offspring.[1]