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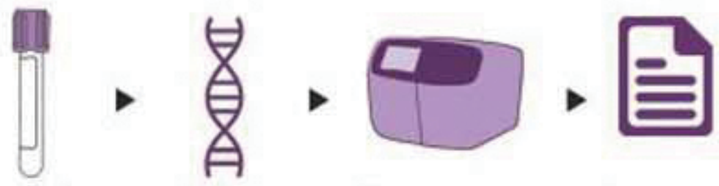
Who should consider carrier screening ?

Carrier screening can provide important information for people who are :

- Planning a pregnancy
- Currently pregnant
- Either partner is affected by a hereditary disorder
- Consanguineous marriage
- High-risk ethnic group
- Family history of a genetic disorder

Methodology



Special Features of Genetic Carrier Screening testing

- ✓ This test covers more than 400 genetically inherited as well as congenital disorders.
- ✓ Advantages include precision testing and timely report delivery.
- ✓ Detection of all known common and rare disease-causing mutations.

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Interpretation of Results

There are around 20,000 genes in our body. The genes many times show variations in individual cases. These variations are very common. For each characteristic, there are two gene types called alleles.

- **Pathogenic mutations** (variations) : if mutations are identified in both the alleles, then they may lead to a diseased condition.
- **Carrier State :** if only one of the two alleles shows mutation (variation) then it is called as a carrier state (not diseased).
- **Likely pathogenic mutation/ variant** : A likely pathogenic variant can probably contribute to the development of disorder.
- **No mutation/ variant :** No pathogenic or likely pathogenic variants identified from the genes analyzed.

Limitation

- This screening method does not detect large deletions/duplications, triplet repeat expansions and epigenetic changes.
- The classification and interpretation of all the variants in this assay reflects the current state of scientific understanding.
- A negative screening result does not eliminate the risk of being a carrier for the conditions screened but does reduce that risk.

References

- The Burden of Genetic Disorders in India and a Framework for Community Control I.C. Verma S. Bijarnia. Community Genet 2002;5:192–196
- Expanded carrier screening in pregnant women and women planning pregnancy :Britton D Rink, MD, MS, Louise WilkinsHaug, MD, PhD, Vanessa A Barss, MD, FACOG. Jan 2019 Gregg, A.R., Aarabi, M., Klugman, S. et al. Screening for autosomal recessive
- and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med 23, 1793–1806 (2021). https://doi.org/10.1038/s41436-021-01203-z

About Greenarray

Greenarray is a molecular diagnostic laboratory. We offer diagnosis of infectious diseases, genetic testing and healthcare information to improve health and wellness. Our goal is to provide high quality affordable and accessible services.

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Genetic Carrier Screening

Know your genes for wellness of your future family

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Genetic Carrier screening is a test used to determine whether one is a carrier of genetic disorder.

This test can help you to find out if your baby is likely to inherit any genetic condition.

What is carrier screening ?

We all have two copies of genes for any given trait. Each parent passes one copy of the gene to their child. A person is called a carrier when one of his gene carries mutation which may be passed on to the offspring.

Carriers themselves are usually not affected in most cases (unless it is an X-linked gene mutation).

Carriers however can pass gene mutations to their children causing genetic abnormalities. Genetic Carrier screening is a test used to screen more than 400 genes.

This test helps to indicate any possibility of passing on the genetic disorder to the offspring. This way the test helps in making informed decisions regarding reproductive options and specialized prenatal care for an individual.

Source : Centre of medical genetics, New Delhi,2011

What are the Disorders Screened ?

Common Autosomal Recessive Disorders

- Beta Thalassemia
- Sickle Cell Anaemia
- Cystic Fibrosis
- Spinal Muscular Atrophy
- Congenital Adrenal Hyperplasia
- Congenital hypothyroidism

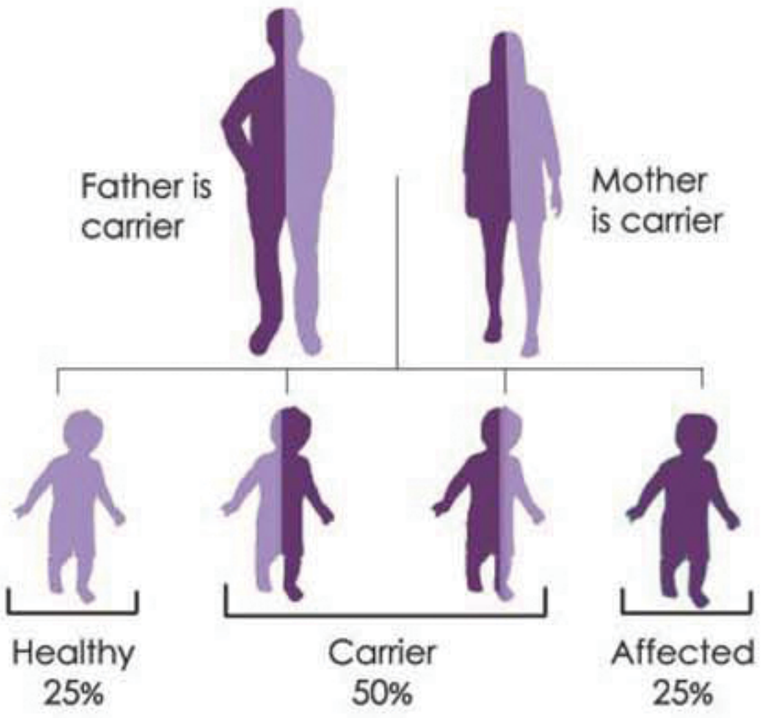
X-linked Recessive Disorders

- Duchenne Muscular Dystrophy
- Haemophilia A/B
- Hunter Syndrome
- G6PD Deficiency
- X-Linked Mental Retardation

More than 400 inherited and congenital disorders

What is Autosomal Recessive disorder ?

Autosomal recessive disorders occur when a person has defects in both copies of gene present on autosomes.



What is X Linked Recessive disorder ?

X-linked recessive inheritance is a mode of inheritance in which the gene causing the trait or the disorder is located on the X chromosome .

X-linked recessive genes are expressed in females if the gene is present on both the X chromosomes however, for a male only one copy of an X-linked recessive gene is enough to express the disorder.