

Genetic Disorders Testing

Genes are the hereditary units that express proteins in a cell to carry out body metabolism and overall growth and survival. They are the functional units on DNA that is packaged inside the nucleus within chromosomes. Humans consist of 23 pairs of chromosomes where 22 are autosomes and one is sex chromosome. These chromosomes have been studied to comprise of around 25,000 protein coding genes. There are continuous phenomenon of changes and aberrations occurring in bases and chromosomes that alter its composition and sequences. These changes or mutations can be harmful if it modifies the gene information as a whole and can be neutral if it does not do so. In the former cases, there are several genetic diseases that can arise.

Why Genetic Testing?

Many of the genetic diseases are hereditary that are inherited from carrier or affected parents. These diseases cannot be prevented or cured but can only be diagnosed with the latest screening tests. Ideally, it should be done before planning for pregnancy but if not possible then it should be done during early pregnancy so as to decide upon the continuation of pregnancy in case any genetic disorder is detected. Genetic testing also let a couple know whether they are carriers (25% chances of having affected child) or either one is a carrier or none is a carrier (no chances of disease in child).

Genetic Tests

The genetic testing identifies any conditions prevailing with the genes and chromosomes of an individual as well as an unborn child. Diagnostic and prenatal testing is done to eliminate

the presence of a particular genetic disorder and is performed in unborn as well as adults (NIH, 2016). Whereas, newborn testing is done on newborns to test for any genetic disorders such as phenylketonuria, Down's syndrome, Edward's syndrome, Patau's syndrome, etc. A genetically diseased child can be avoided by carrier testing on parents who tend to have family history of an autosomal recessive disease. The in vitro fertilization cases can go for pre-implantation testing to test for any genetic diseases in the in vitro developed embryo.

References

NIH. What are the Types of Genetic Tests? 2016. Accessed at <https://ghr.nlm.nih.gov/primer/testing/uses> on 22 April 2016.