Genetic Inheritance Worksheet

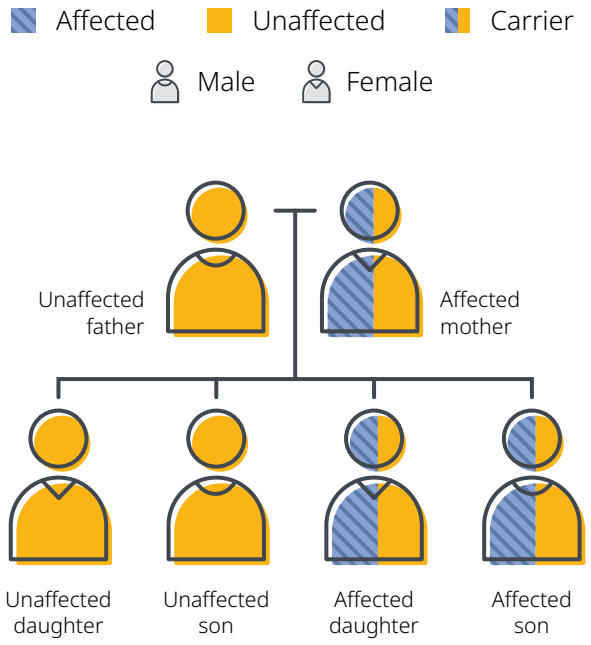
#RAREis...

Some genetic conditions are caused by variances or mutations in a single gene.[1] Read below and complete the worksheet to understand how genetic disorders can be passed down through families in an autosomal dominant, autosomal recessive or X-linked manner. For a list of rare genetic disorders, visit globalgenes.org/rare-list.

Autosomal Dominant Inheritance

An autosomal dominant inheritance pattern includes one mutated copy of the gene in each cell. In some cases, an impacted person can inherit a genetic disorder from an affected parent. In other cases, the genetic disorder may be a new mutation with no family history of the genetic disorder.[1]

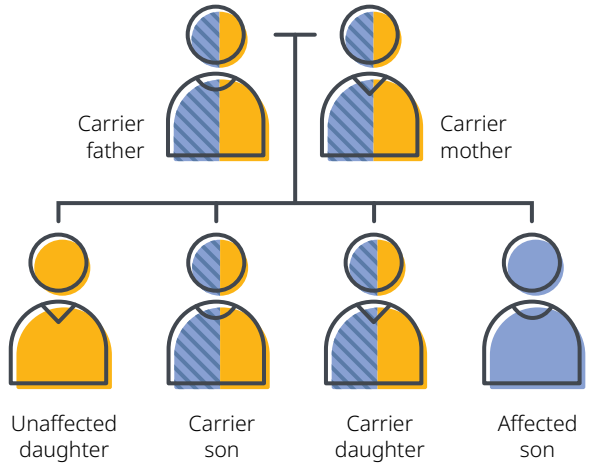
Examples of Autosomal Dominant genetic diseases include Huntington Disease and Marfan Syndrome.[1]



Autosomal Recessive Inheritance

An autosomal recessive genetic disorder may occur if both parents carry one copy of the mutated or variant gene and pass it on to their child. Often, parents will not show any signs or symptoms of the genetic disorder and therefore not be aware they are carriers of a genetic disorder.[1]

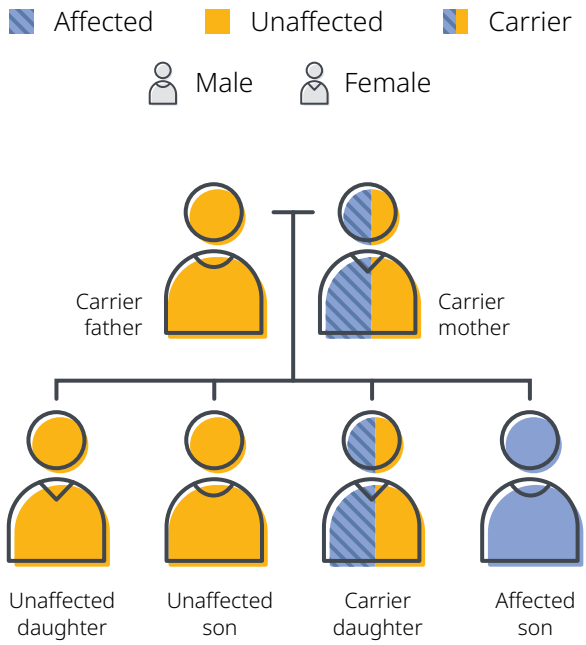
Examples of Autosomal Recessive genetic disorders include Cystic Fibrosis and Friedreich Ataxia.[1,2]



X-Linked Inheritance

An X-linked disorder is caused by mutations or variants in genes on the X chromosome, one of the two sex chromosomes in each cell. In women (who have two X chromosomes), a mutation or variant in one of the two copies of the gene may cause a genetic disorder. In men (who have one X chromosome), a mutation or variance in the only copy of the gene may cause a genetic disorder. Many women who have inherited an X-linked disorder are referred to as "carriers" because it is unlikely that women will have inherited two altered copies of the mutated gene. Males are more likely to be affected by X-linked disorders because they only inherit one of the X genes. Fathers cannot pass X-linked traits to their sons, but they can pass them to their daughters.

Examples of X-linked genetic disorders include Hemophilia and Fabry Disease.[1]



Other Family Members Who Could Be Affected

If you inherit a genetic condition from one of your parents, here is a list of others on that side of your family who should be tested:

- Your siblings
- Your siblings' children (your nieces and nephews)
- Your aunts and uncles
- Your cousins
- Your cousins' children

More information and resources on genetic testing can be found on RAREisCommunity.com.

References

[1] Genetics Home Reference. What are the different ways in which a genetic condition can be inherited? Available at: https://ghr.nlm.nih.gov/primer/inheritance/inheritancepatterns. Last accessed October 2020.

[2] Genetics Home Reference. Friedreich Ataxia. Available at: https://medlineplus.gov/genetics/condition/friedreich-ataxia/#inheritance. Last accessed November 2020.

Now, create your own family tree.

Fill out the family tree below with information from your parents, your partner and your children to better understand genetic inheritance.