X-Linked Recessive Inheritance

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X-linked inheritance refers to the pattern of inheritance of a faulty (mutated) gene that is located on the X chromosome (X-linked genes). Males have one Y chromosome and one X chromosome and therefore only have one copy of the genes that make up the X-chromosome. Females have two copies of the X-chromosome genes (Female=XX, Male =XY). Shortly after conception, one of the X-chromosome copies in each female cell is randomly inactivated. This means that both males and females only have one active copy of the X-chromosome in each cell.

X-LINKED RECESSIVE INHERITANCE

The pattern of inheritance of a condition due to a recessive mutation in a gene that is located on the X chromosome is called X-linked recessive inheritance. A female who has a mutation in a gene on one of her X chromosome copies but a correct copy of the gene on the other X chromosome, is a "carrier" of the gene mutation (X-linked genetic carriers). Females who are "X-linked genetic carriers" will therefore usually have only half of her cells containing the information for the correct gene product. Males who have the faulty gene on their X chromosome are usually affected by the condition due to the faulty gene.

What is the risk of having a child affected with an X-Linked recessive disorder?

Figure 1 illustrates the risk of having children affected by an X-Linked recessive disorder if the mother is a carrier. These risks apply to every pregnancy. The risk, in every pregnancy of a carrier mother, that a boy will be affected is 50%. However, there is also a 50% chance that her son will inherit the correct copy of the gene and not be affected. The daughters of a carrier each have a 50% risk of being a carrier (but not being affected) and a 50% chance of not inheriting the faulty gene (thus not being a carrier).

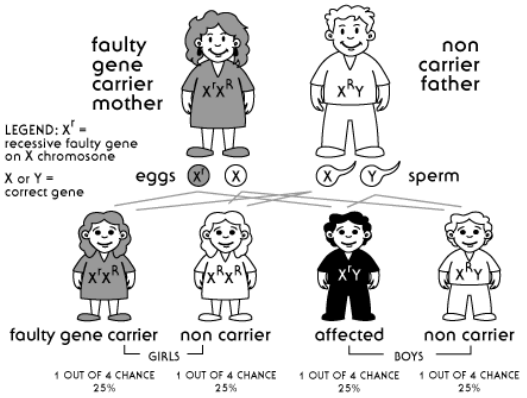

Figure 1: The faulty copy of the gene containing a recessive mutation is shown by "r". The correct copy of the gene is shown by "R". **(Picture from the Centre for Genetics Education website: http://genetics.com.au)**

A man with an X-linked disorder cannot pass the "faulty" gene to his sons because he gives his son a Y chromosome, not an X chromosome. But all his daughters will receive the "faulty" gene and be carriers for the disorder, because each daughter inherits her father's only X chromosome. This is illustrated in figure 2.

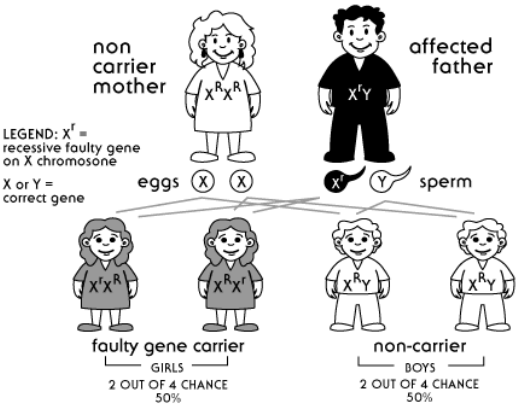

Figure 2: The faulty copy of the gene containing a recessive mutation is shown by "r". The correct copy of the gene is shown by "R". (Picture from the Centre for Genetics Education website: http://genetics.com.au)