Climb

National Information Centre for Metabolic Disease

What is a Metabolic Disease?

supporting families, changing lives

Climb Building, 176 Nantwich Road, Crewe, Cheshire, CW2 6BG, UK

Freephone: **0800 652 3181**

Climb is the only charity in the United Kingdom that provides support on all Metabolic Diseases with links Worldwide

Email: **info.svcs@climb.org.uk**
Website: **www.climb.org.uk**
Family Services: **0845 241 2172**
Development Services: **0845 241 2173**
Facsimile: **0845 241 2174**

Climb
Children Living with Inherited Metabolic Diseases

Registered Charity No. 1089588. Company Limited by Guarantee No. 4267454
Patron: Her Grace The Duchess of Westminster.
President: Phil Collins L.V.O.
Executive Director : Mr Steven Hannigan MIOD. ACMI.

What is a Metabolic Disease?

Metabolism: - "The sum total of all the chemical reactions that take place in the body."

Most of us think of metabolism in terms of the rate at which we burn off food to make us fatter or thinner than we want to be.

Our body is in fact a highly tuned machine with chemical changes constantly taking place, some breaking down substances (catabolism), some recycling substances and some creating new substances (anabolism).

All these changes are programmed by different genes which direct different enzymes to carry out the work. Enzymes are proteins which speed up chemical changes. In a healthy individual all these processes occur automatically and we tend to take our finely tuned "human machine" for granted - until something goes wrong.

Metabolic Diseases are caused by a defect in one of the genes. This may disrupt the break down or build up of components because it is unable to pass important messages on to the appropriate enzyme. This may mean that the enzyme is not present or it may not function correctly. Due to the body's chemistry being so complicated, if one chemical in the body does not work properly, a number of other defects may occur because of this. In these cases there can be an accumulation of toxic chemicals or a reduction in important substances which can be harmful to the body and potentially fatal without the correct treatment.

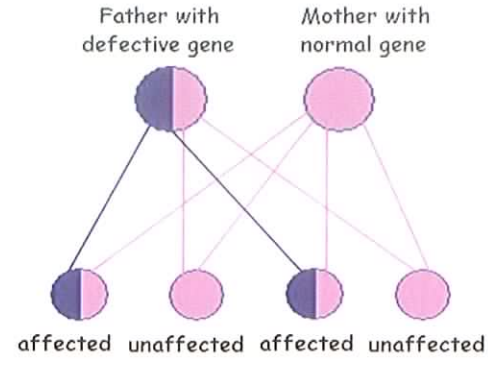
Metabolic diseases affect each individual differently. The symptoms are dependant on which gene is defective. Some gene defects can cause serious illness; others mean the person becomes dependant on a special diet or certain prescribed drugs. In some cases others do not appear to cause any symptoms (asymptomatic). Symptoms can appear at any time from birth to adulthood. Some disorders are tested for at birth.

How Do you get a Metabolic Disease?

In rare cases, metabolic disease can be caused by the individual being exposed to certain drugs or foods which trigger their illness, others may be because they have eaten too much of a certain type of food but most often the fault has occurred because of incorrect genetic instructions that are inherited from the parents.

When the disorder is inherited it can be passed down in many different ways depending where the gene is located on the chromosome and the type of defect. We always recommend that you should be referred to a genetic counsellor. Below are the ways that disorders can be inherited.

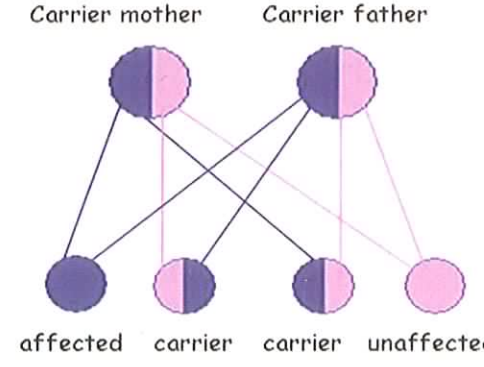
Autosomal Dominant Inheritance



One set of genes comes from the mother and one set of genes comes from the father. This method of inheritance is when a single copy of the diseased gene will dominate the other normal gene. Therefore, if a defective gene is inherited from either parent the child will be affected with the disorder. This means for each pregnancy if either of the parents has a defective gene, there is a 50% chance of a child being affected by the disease.

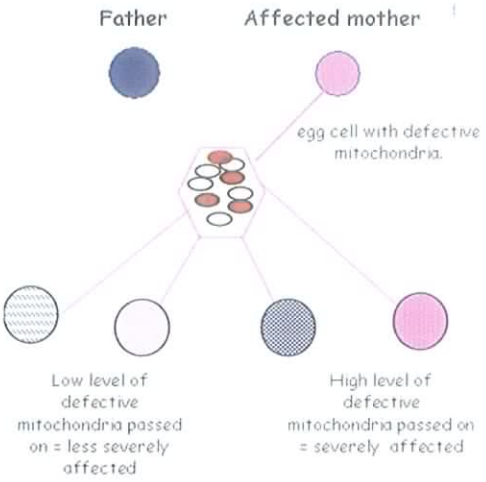
Autosomal Recessive Inheritance

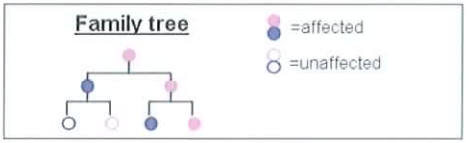
A person who has one normal gene and one gene for the disease is termed a carrier for the disease and does not show any symptoms. The condition arises when a child inherits a gene for the disease from both parents. The risk to the offspring of a couple who are both carriers is 25%. There is a 50% chance that their child will be a carrier. There is a 25% chance that the child will not carry the abnormal gene. This risk is the same for each pregnancy.

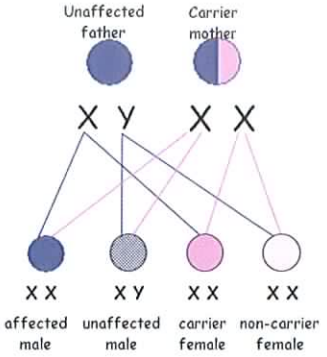


Mitochondrial Inheritance

Children inherit Mitochondrial DNA (mtDNA) from the maternal line. The defective mitochondria is passed down to children from their mothers and not from their fathers. The mother can pass the disorder on to male and female children but the males will never pass it on to their children. The level of defective mitochondria passed on causes the severity of the disorder







X-Linked Inheritance

This occurs when the defect is on the X-chromosome. Females have two X-chromosomes and any disease traits that are on one of the X chromosome's are usually masked by the other normal X chromosome. X-linked Recessive disorders only affect females when there are two copies of the gene, one on each X chromosome.

Is there a treatment?

Some disorders can be treated through a special diet or medication. Others may be so rare that a treatment has not yet been found. However, recent advances in research mean that for a handful of conditions treatment is on the horizon using gene therapy, so there is a brighter future for many children where there was no hope before. **Climb** has contributed towards research programmes that have located the genes which will make gene therapy a real possibility.