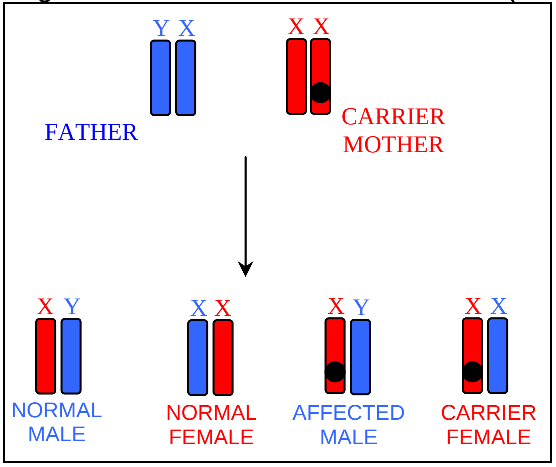
Duchenne muscular dystrophy (DMD) is an inherited disorder characterized by progressive muscle weakness which starts in the legs and pelvis and later affects the whole body. DMD is the most common form of muscular dystrophy. It usually affects only boys, but in very rare cases it can also affect girls. It is an X-linked recessive inherited disease.

What is X-linked recessive Inheritance?

The word X-linked refers to the fact that the condition is caused by a gene that is on the X chromosome (the 23rd chromosome pair consists of the X and the Y-chromosomes). Females inherit two X-chromosomes, one each from the mother and father. Males inherit one X chromosome (from the mother) and one Y chromosome (from the father). Recessive inheritance occurs when "both" genes of a pair have a defect in order to produce the condition. Since a female has two X chromosomes, if she has a mutation or a defect on one of her X chromosomes but a normal or working gene on her other X chromosome, she will not have the condition. A female in this situation would be considered a "carrier". Since a male has only one X chromosome, he has only one copy of each gene on that chromosome. Therefore, if one of the genes on his X chromosome has a mutation or a defect, he will be affected.



What is the cause of DMD?

DMD is caused by fault in the genes present on the X chromosome. This faulty gene causes defect in a single important protein in muscle fibers called Dystrophin. DMD is X-linked, that means it affects only boys while females are carriers. Since males have one X and one Y chromosome, so any fault on the X chromosome would make him affected. In many cases, there are other family members who have a similar illness. Sometimes it appears in a family, which has no previous history and is due to a fresh mutation (defect) in the gene.

What are the clinical features and problems of DMD?

Babies usually appear normal during early infancy. Most affected boys develop first signs of difficulty in walking at the age of 2-3 years. As the disease progresses they become more and more clumsy in walking, have difficulty in running, climbing stairs and getting up after a fall, calf muscles start to look abnormally large. The muscle weakness gets progressively worse and the children assume a typical posture in trying to stand up from a lying down position. In most cases contractures (tight joints) develop in ankles, knees and hip. By about 10-12 years the children are unable to walk and have to use a wheel chair. Eventually respiratory muscles become weak and these boys have repeated chest infections. Heart failure can also develop. Obesity is a common problem as the disease advances due to lack of physical activity.

How to diagnose / confirm DMD?

Diagnosis is based on:

1. The family history
2. Clinical examination of the pattern of muscle weakness, age of onset, how it progressed, any enlargement of muscles
3. Creatine kinase (CPK) level is important as the value is very high in person affected with DMD

4. An Electromyographic (EMG) study is also required to find out that basic pathology is in the muscle and not in the nerve cells. A small needle is inserted in the muscle and a recording is made of its electrical activity. Distinct patterns are seen if the primary disease is in the muscles or nerves.
5. Muscle biopsy is required in most cases and should be done in the best laboratory in the area as along with routine histological studies, and special (dystrophin) staining is required. These tests are available only in the advanced laboratories.
6. Gene studies are carried out on the DNA sample of the child to find out which parts of the gene are missing (deleted) or have other defects. Even if there is no deletion of the gene, it is important to store the DNA of the affected child. DNA is extracted from blood.

What is the Treatment for DMD?

Unfortunately there is no cure at present. The gene has been identified, as well as the protein it makes (called dystrophin). Active research is being done to find ways to induce the muscles to form dystrophin .

Numerous other therapies have been tried in the past e.g. Vitamin E, amino acids, Growth Hormone inhibitor, nucleotides, vasodilators, Vitamin B6 and zinc, but none have shown consistent benefit.

What about Steroids?

Steroids are not a cure. If started early, when patients are still able to walk on their own, it tends to stabilize the disease or slow the progression of the disease. Steroids are ineffective when given to children who are already wheelchair bound or non-ambulatory. Steroids have several side effects such as weight gain, behavior changes acne, hair-growth, hypertension. Therefore regular follow-up is a must.

What should be told to the affected child?

It is important to decide in advance how you will answer possible questions. The information you will give will depend on the child’s age and understanding. Answer the questions on the child’s own terms, never tell anything untrue, without going beyond the literal question asked.

Try to encourage the child to talk about the muscle problem. You will find that probably you are far more nervous about the situation than the child. Do not be evasive as the child will seek information elsewhere.

So what should be done for the affected child?

Active exercise strengthens normal muscle fibres. Try to keep as fit and active as possible. Regular daily exercise is better than occasional sudden bouts of exertion. Plastic splints can be used to maintain a gentle stretch of the calf muscles overnight. Sessions of calf muscle massage may also be useful.

In the latter stages a wheelchair is likely to be needed to get about independently. There is a great deal of other helpful equipments that may be useful to individuals, and much can be done to help both at home and at work to make certain tasks easier by careful choice of furniture, bathroom equipment etc . As overweight is a common problem in later years, and is best prevented by establishing sensible eating habits .Regular medical supervision will become increasingly important as the years go by. Early identification of contractures and spinal curvature will allow these to be treated more effectively, and indeed preventive treatment may be recommended. The physiotherapist will play an important part in this, and can advise about exercises. In some cases surgical treatment for contractures and spinal deformity may be considered.

What about School?

Most children with muscular dystrophy cope pretty well in the local nursery school and their first few years at junior school. If walking becomes precarious or access to classroom or toilet becomes too difficult special arrangements will have to be made. If the school is too unsuitable, an alternative school for disabled children will provide the answer.

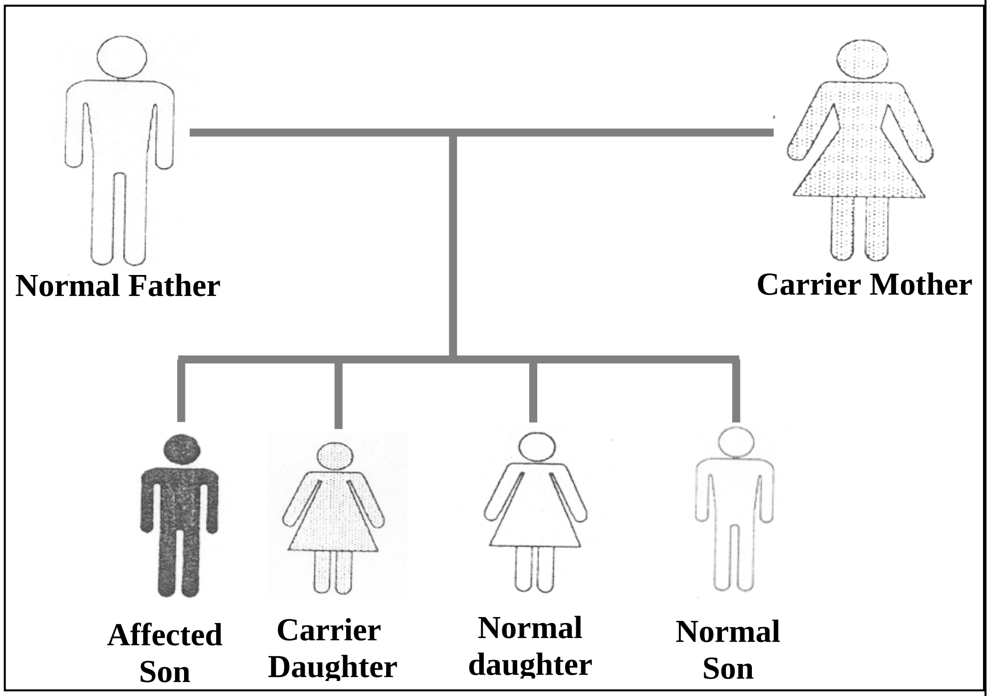
Some boys with Duchenne muscular dystrophy are found also to have learning difficulties. Language and communication skills are often the main difficulty. Manual skills, design sense and imaginativeness are often excellent. Parents should work with the school to discover and develop the child's best talents, as well as helping him to learn and to cope with tasks he finds difficult.

What can the parents do to help the child with DMD?

They should learn all about muscular dystrophy. This will give them confidence, and help them to foresee and prevent problems and to make balanced decisions.

Help the child to enjoy active exercise so that it becomes a life-long habit. Active exercise means making the muscles work hard. Games, swimming and walking for pleasure are some of the best ways to start. Passive exercise means stretching and it becomes necessary a bit later to prevent contractures. Encourage his education, his skills and his independence. Make sure that other children in the family are also involved in his care and neither should feel neglected.

What is the risk of having another child with DMD if there is one in the family?



DMD has X-linked recessive inheritance. This means if the mother is a carrier, there is a 50% chance of disease in the case of sons, while a 50% chance of carrier status in the daughters who are born.

The mother is a definite carrier if she has a son with DMD, as well as a brother with DMD.

Carrier status in the mother can be determined by :

- Family history.
- Careful clinical examination, as some carriers may show mild manifestations.
- Estimation of Creatine-Kinase (CK) in the blood. The CK is raised in about two-thirds of carriers. CK estimation should be done before pregnancy, as the value falls during pregnancy. Moreover, before the blood is withdrawn for the CK test, the subject should not have undertaken any physical exertion as this raises the value.
- Genetic studies.

If a woman has only one affected son, and no other affected son or brother, the affected child is called a sporadic case. In 2/3 of these cases the mother is likely to be a carrier of the abnormal gene while in 1/3 of cases the mother is not a carrier. Ordinarily in ‘sporadic’ cases where mother is not a carrier, there should be very low risk to future offspring.

How can the birth of another child with DMD be prevented?

If you wish to know your risk of having a child affected with muscular dystrophy you should seek genetic counseling. The genetic counselor records the family history, and carries out any laboratory or histological tests that are necessary, and provides the risk of recurrence, and advises on how to reduce or cope with that risk.

No couple who has a child affected with muscular dystrophy would like to have another affected child. Therefore prenatal diagnosis of muscle dystrophies and the option to abort in case the fetus is affected, offers an opportunity to prevent the birth of other affected children. If the fetus is normal, the pregnancy is continued. This approach to avoid birth of affected children has been made possible with the genetic technology.

At 10-12 weeks of pregnancy chorionic villus samples are obtained. These are parts of the fetus, through which the fetus is attached to the mother’s womb. Tests are carried out on the CVS (by DNA technique) to look for the defects which were present in the affected child. In this way prenatal diagnosis of DMD can be made. In those cases where no deletion is present, one has to carry out studies to determine which of the X chromosomes of the mother, whether the fetus has inherited the affected or the unaffected one. The chances of error in these tests is very low (about 2%).

What does the future promise?

Currently there is no satisfactory treatment for muscular dystrophies. However, the experiments on gene therapy and myoblast transfer are on.

Let us hope and pray that a cure will be found. Concentrate on encouraging the child’s skills and interests and give him the best we can in every way.

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