Inheritance and the muscular dystrophies

This leaflet provides a brief summary of the genetics of the muscular dystrophies. An understanding of their inheritance patterns makes it possible for families to know what the risk is of the dystrophy happening again.

If, after reading this, you feel that any member of your family may be an unrecognised carrier of muscular dystrophy, or may have the condition themselves, do get expert advice from a genetic centre. New genetic tests can help trace, within families, those members with the abnormal gene for many muscular dystrophies.

How are the muscular dystrophies inherited?

The way in which our bodies grow and work is controlled by structures called genes. Genes are parts of chromosomes, which are strands of biochemical material transmitted from every cell to its offspring. We have two genes for every hereditary characteristic and function, one inherited from each parent. Sometimes a gene is faulty in part of its structure with the result that a certain bodily process is impaired and this fault may then be passed on to the children.

There are three main types of inheritance, and when a gene is faulty the fault is inherited in one of these ways with differing result.

1) X-linked or sex-linked recessive

Including: Duchenne muscular dystrophy
           Becker muscular dystrophy
           Scapuloperoneal muscular dystrophy

These conditions are determined by genes which are carried on one of the chromosomes which control the sex of a child. The result is that only boys are affected and they inherit the disease from their mothers who are known as carriers. Carriers usually show no sign of the disease (though sometimes trivial or even unrecognised symptoms are present) but they are capable of passing the condition on to their own sons. Each son of a carrier has a 50% chance of having muscular dystrophy and each daughter has a 50% chance of being a carrier (see figure 1):
Unaffected boys cannot transmit the disease; however, the daughters of a man with any X-linked dystrophy are all carriers (see figure 2):
Genetic mutation
This is an unpredictable change in the structure of a gene causing a different characteristic to appear for the first time in a family. In about a third of those who have Duchenne muscular dystrophy, it is due to genetic mutation.

2) Autosomal recessive
Including:  
Most 'Limb girdle' types  
Scapulohumeral muscular dystrophy  
Recessive childhood muscular dystrophy  
Congenital muscular dystrophy  
and most types of Spinal muscular atrophy.

These conditions become apparent only if both parents carry a faulty gene but the parents themselves do not manifest any symptoms. Each child of such parents has a 25% chance of carrying both abnormal genes and therefore of being affected. Either sex can have the condition (see figure 3):

The children of an affected individual are usually unaffected but cousin marriages between affected or unaffected members of such families greatly increase the risk that they will have affected children. Limb girdle dystrophy is a term that is becoming out-dated because it is thought to include several different forms of muscular dystrophy inherited in different ways. Some people who have been diagnosed as having limb girdle muscular dystrophy in the past have turned out later to have entirely different non-genetic disorders. Furthermore many cases of the Becker type have been called Limb girdle muscular dystrophy in the past and occasionally women with 'limb girdle muscular dystrophy' turn out really to be carriers of the Duchenne gene. If concerned about the inheritance of their limb
girdle muscular dystrophy, it is important people are reassessed to make sure of the diagnosis, this would be done as part of a referral for genetic counselling.

3 Autosomal dominant
Including: Facioscapulohumeral muscular dystrophy
Myotonic dystrophy
Some oculopharyngeal/ocular muscular dystrophy
and some limb girdle dystrophies
Here the condition becomes apparent even though the affected person has only one abnormal gene. Either sex can have the condition and each child of an affected parent has a 50% chance of being affected (see figure 4):

However, the severity of the condition may vary considerably in different individuals. Some people are so mildly affected that they may not have recognised that they have the condition but they may then have a more severely affected child. So examination of all the family members is important as part of genetic counselling.

Can muscular dystrophy be identified in the unborn child tested for in pregnancy?
Research on gene probes has made the diagnosis of some forms of muscular dystrophy possible in early pregnancy. Although these tests of the unborn child can be done from the tenth week of pregnancy onwards, investigations of some family members may be required before the prenatal tests are done. (Please see the leaflet in this series entitled Carrier detection and prenatal diagnosis of inherited dystrophies).
Where can advice be obtained?
The various tests can be carried out and genetic advice given at a number of genetic centres throughout the country. Your family doctor can make the appropriate referral for you and any of your relatives who are worried. You can also contact the Information Officers at the Muscular Dystrophy Campaign headquarters for a list of such centres.