

Emery-Dreifuss muscular dystrophy

This form of muscular dystrophy was so named after Professor Emery in the UK and Professor Dreifuss in the United States, who together first described the disorder nearly 40 years ago.

What is it?

Like other muscular dystrophies it is a wasting disease of muscle. It usually begins in childhood or adolescence. The features, which make it unique and different from other muscular dystrophies, are the early development of muscle contractures, the distribution of muscle weakness, and the fact that the heart may be affected in a particular way.

What are muscle contractures?

This is a tightening and shortening of certain muscle groups so that the joints, which are involved, become increasingly difficult to move. Such contractures are common at a late stage in most muscle wasting diseases and result from inactivity. But in this disease muscle contractures develop very early or before there is any marked muscle weakness.

What effect do contractures have?

In this condition they limit elbow straightening so that the arms are often held in a semi-flexed way, they result in a tendency to walk on the toes, and they limit forward bending of the neck.

Which muscles are affected?

In the upper limbs weakness affects mainly the shoulders and upper arms. In the lower limbs, unlike most other dystrophies, weakness affects the lower legs first. This distribution of muscle weakness is sometimes referred to as 'scapulo-humero-peroneal'.

At first there is difficulty in raising the arms above the head and lifting heavy objects, and a tendency to trip over carpets. Later on the hip and thigh muscles also become affected so that climbing stairs becomes increasingly difficult, as does rising from a chair without assistance.

How is the heart affected?

This is affected in a way that is unusual for muscular dystrophy, rather than affecting the heart muscle, it is the electrical wiring (called the cardiac conduction system) that controls the rate at which the heart beats that is mainly involved and is referred to as "heart block". The heart rate is often abnormally slow, palpitations may occur (which feel like "fluttering" in the chest - this is not uncommon in normal individuals and alone would not be a cause for concern), as well as attacks of giddiness and fainting spells. Increasing tiredness and breathlessness may also occur.

What can be done if the heart is affected?

If the heart becomes affected, and not every person is affected in this way, the doctor may recommend the insertion of a pacemaker. This small gadget is inserted just below the skin of the chest and prevents further problems arising by ensuring that the heart thereafter beats normally.

How severe is the disease?

In general the condition is less severe than many other forms of muscular dystrophy and though life expectancy may be shortened, many affected individuals can expect to reach middle age or later. However, it is essential that affected individuals be checked at frequent intervals, say every 12 months, to ensure that the heart is not affected. There is evidence that a more severe recessive form also exists. Here weakness is present and progresses rapidly from early childhood, however this is very rare.

Can it be treated?

Unfortunately there is as yet no cure or effective treatment apart from the insertion of a heart pacemaker when this is necessary. However, having an adequate diet and maintaining good general health are very important as in all muscular dystrophies.

Can I improve muscle strength?

Regular gentle exercise, which is tolerated without causing stress, is beneficial. But hard physical exercise (weight training for example) should be avoided. It is essential to eat a well balanced diet, to include plenty of roughage and to avoid becoming overweight since this will only overburden the already weakened muscles.

Can surgery help?

Division of the heel cords can be useful in helping walking. Other operations can be indicated in individual cases where expert advice from a neurologist and orthopaedic surgeon should be sought. Because the heart may be affected and could complicate an operation, the anaesthetist must be told of the diagnosis before any operation is undertaken.

Will I become disabled?

The condition progresses very slowly over the years (except for the very rare recessive form), and it may be that later in life a wheelchair may be required.

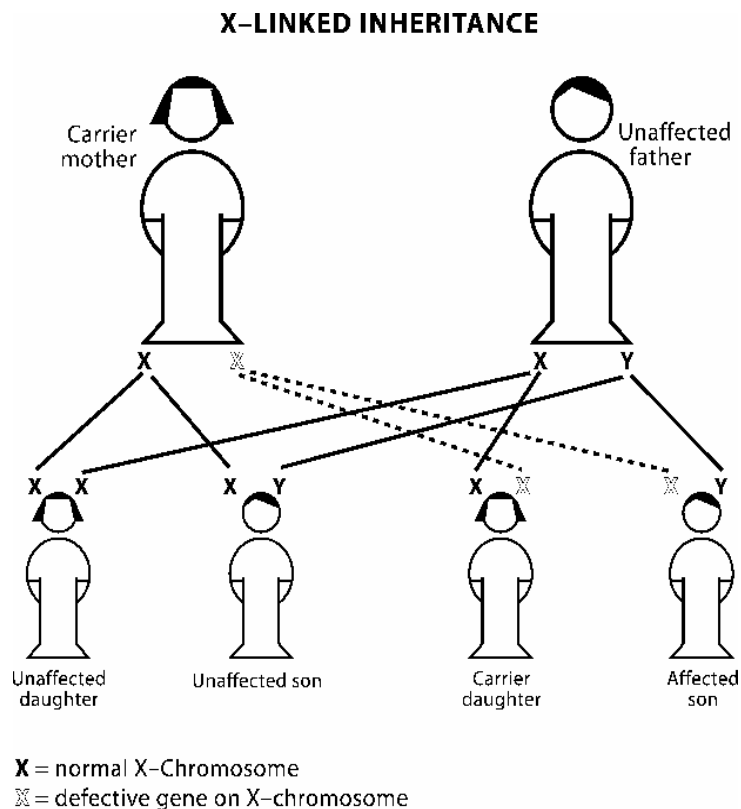
Job prospects

As in other slowly progressive muscle diseases, in the early stages most occupations could be considered (a driving licence however maybe subject to a medical examination). But since physical disability increases with age, some form of sedentary occupation is preferable in the long term.

Can it affect my children?

The condition is inherited and can therefore affect other members of the family. In many families it is inherited as a **sex-linked (X-linked)** trait and therefore only affects males and is carried by unaffected females. All the sons of an affected male will be unaffected but all his daughters will be carriers. With regard to the offspring of a woman who is a carrier, on average each of her daughters has a 50:50 chance of also being a carrier, and on average each of her sons has a 50:50 chance of being affected. The condition can also be inherited as an **autosomal dominant trait**, which affects both males and females. Here the genetic risks are different. As in all autosomal dominant disorders on average each son or daughter of an affected parent has a 50:50: chance of also becoming affected. In the case of the **recessive**

form, both parents are normal but there is a 1 in 4 chance of any future children also being affected.



To make matters even more complicated, sometimes in x-linked and dominant cases there is no family history of the disorder, only one individual in the family being affected. In these cases the disease has arisen as the result of a new mutation in the affected individual but who may then later transmit the disorder to his or her children. For these various reasons it is very important to seek the professional advice of a neuromuscular specialist or medical geneticist if you are at all concerned about the risks to your children and other relatives.

What of the future?

This is much more hopeful than in the past. Not only through the use of pacemakers in cases with heart disease, but the genes responsible both the autosomal dominant (codes for lamin A/C) and the X-linked forms of Emery-Dreifuss muscular dystrophy (codes for emerin) have been identified. This information is vital in the search for an effective treatment and also for prenatal diagnosis.

SOME OTHER SIMILAR CONDITIONS

Rigid spine syndrome

In this disease, which begins in childhood, muscle weakness is usually slight, the main problem being contractures of the neck muscles, elbows and knees. The heart is not affected. The condition is very heterogeneous being associated with various dystrophies. Expert advice is therefore necessary concerning the precise diagnosis and mode of inheritance in each case.

Rare benign sex-linked muscular dystrophies

Apart from Becker muscular dystrophy (which is dealt with in a separate leaflet) and Emery-Dreifuss muscular dystrophy, other benign sex-linked dystrophies are very rare and have only been described in occasional families.

Limb –girdle dystrophies

In the late stages of Emery-Dreifuss muscular dystrophy the distribution of muscle weakness may resemble that in limb-girdle dystrophy. Again expert advice is required to confirm the precise diagnosis.

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Author: Professor Alan E.H. Emery for the Muscular Dystrophy Campaign

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