



## Congenital muscular dystrophies

### What is congenital muscular dystrophy?

The congenital muscular dystrophies are a group of conditions which share early presentation and a common muscle pathology. Congenital means 'from birth' and in the great majority of cases of congenital muscular dystrophy the initial symptoms are present at birth or in the first few months.

Babies with congenital muscular dystrophy often have hypotonia (low muscle tone or floppiness), and may have reduced movements. Other common signs are contractures (tightness) in the ankles, hips, knees and elbows. The contractures can sometimes be severe and affect several joints (known as arthrogryposis). They happen because the baby has not had the muscle strength to move freely enough in the womb. Some of these babies may also have respiratory problems because of weakness of breathing muscles.

In some children who do not have contractures the first problems are only noted after a few months because of difficulties in holding the head or delay in learning how to sit unaided, stand or walk.

### How many people are affected by congenital muscular dystrophy?

A precise and recent study on the frequency of this disorder in UK is not available, but we estimate that 1 baby every 20,000-50,000 is born with congenital muscular dystrophy.

### Is congenital muscular dystrophy inherited?

Yes. The pattern of inheritance is known as 'autosomal recessive'. This means that both parents are carriers of the condition (although clinically unaffected) and they have a risk of 25%, or 1 in 4, in each pregnancy of passing the condition on to their children. Occasionally a case may be 'sporadic' which means is a one-off with little risk of recurrence in other children. There is no accurate way of predicting who is and who is not a carrier.

### How many forms of congenital muscular dystrophy exist?

Congenital muscular dystrophy is a very heterogeneous group of conditions. These are generally grouped under two main types:

1. Children who only have muscle weakness involving all muscles but have normal intelligence
2. Children who have muscle weakness and learning difficulties, with or without seizures. Learning difficulties may be subtle, moderate or severe.

While this classification is helpful in most cases, an overlap between different categories can occur. A lot of effort has gone into identifying separate entities within each group and in locating the gene responsible for each form. A number of specific conditions can now be recognised but for others a final diagnosis is still not possible.

**Genetic advances.** There have been recent developments in the genetics of congenital muscular dystrophy which have resulted in a better understanding of this group of disorders.

The first gene abnormality to be discovered was that of the LAMA2 (laminin alpha-2 chain) gene, the gene responsible for merosin. This form affects approximately 40% of the children with congenital muscular dystrophy. More recently several other abnormal genes have been identified and today we know nine genes responsible for separate forms of congenital muscular dystrophy, each of them with specific clinical features.

A number of cases do not have any of these forms and more research into these conditions is needed.

### **How is congenital muscular dystrophy diagnosed?**

A baby with congenital muscular dystrophy is usually first diagnosed as a 'floppy baby'. Doctors can see the symptoms described above, but as these could be due to a number of different conditions, they have to conduct a series of tests to try to make an accurate diagnosis.

Firstly a **blood test** is taken and the level of a muscle enzyme assessed (the creatine kinase or CK level). In approximately 40% of cases of congenital muscular dystrophy this level is 5-20 times higher than normal.

**Muscle ultrasound** may also help to detect abnormalities of the muscle. The technique is very simple, similar to the ultrasound studies carried out in pregnancy and may provide further evidence of the involvement of the muscle.

An **electromyography** (EMG) test may also be done. A small needle is inserted into muscle and the electric activity recorded. This test may provide evidence of an abnormal pattern of electric activity in the muscle.

At this stage however even in the cases with high CK levels, abnormal muscle ultrasound and EMG, an additional test which is required in almost every case is a muscle biopsy.

**Muscle biopsy** can help to identify the subtype of congenital muscular dystrophy to provide a precise diagnosis in several ways:

- When the muscle is studied under the microscope, it will show variation in the size of muscle fibres and that some of these fibres are replaced by fat and fibrous tissue.
- In addition, the production of individual components of the muscle fibre can be studied in detail with specialised tests. This greatly helps to narrow down the diagnostic possibilities.

In the forms of congenital muscular dystrophy in which the gene defect has been identified, **genetic tests** will provide the ultimate diagnosis.

**Prenatal diagnosis** is possible in several types of congenital muscular dystrophy. It is based on the ability to detect the genetic abnormality in the developing foetus. This however can only be used in the forms of congenital muscular dystrophy associated with a recognised gene defect or a specific protein deficiency.

### **Is there a treatment or cure?**

At the moment there is no cure for congenital muscular dystrophy, but there are ways, described below, of helping to alleviate the effects of the condition.

### **Is congenital muscular dystrophy progressive and is it life threatening?**

The condition is fairly stable and the child appears to gain strength as he or she gets older. In several forms therefore acquisition of new skills with time is possible although difficulties will always be present. If the condition is not progressive, it is possible to live a normal lifespan.

In some conditions the muscle weakness however becomes worse with time and can lead to respiratory problems. This may happen in children of any age.

### **Can a child with congenital muscular dystrophy learn to walk?**

The severity of this condition varies greatly from person to person. As the severity varies so much sometimes even within the same form of congenital muscular dystrophy, it is important not to assume that certain developments will or will not take place, but to work with the child to achieve the goals which are in his or her power.

Some children will walk but sometimes this can be delayed until five years of age or older. Leg splints (callipers) are often used to assist a child to walk. Children who have successfully walked may lose the ability later on because as they grow older and heavier, the muscles are unable to cope with a greater strain. Other children never achieve walking.

### **What other physical effects might congenital muscular dystrophy have on a child?**

The presence and the severity of other problems depend on the form of congenital muscular dystrophy. Some features however are generally found in many children with congenital muscular dystrophy, irrespective of the form.

As the muscles are weak and mobility is limited, the child may develop or be born with 'contractures'. This means that the muscle tendons tighten up and the child is unable to move the limbs or the joints as freely as a healthy child. Physiotherapy can help prevent this and a programme of exercises should be worked out with a physiotherapist very soon after diagnosis. Even a very young baby can be helped to maintain suppleness. Hips are commonly affected and if they are dislocated this may require treatment with a splint or sometimes surgery.

Breathing and feeding problems are commonly observed in some forms of congenital muscular dystrophy but are less frequent in others.

As these complications can be helped by timely recognition and professional advice and intervention, it is advisable for individuals with congenital muscular dystrophy to be regularly followed by someone with expertise in neuromuscular disorders. There are several examinations that might be needed, such as over night sleep studies to monitor the breathing quality during sleep. In some children it is also advisable to reduce the risk of chest infections, performing flu jabs and other vaccinations.

### **Where can I get help?**

#### **Muscular Dystrophy Campaign**

61 Southwark Street

London SE1 0HL

Tel: **020 7803 4800** (all departments)

Free phone: **0800 652 6352**

Email addresses:

Information and Advice Line: [info@muscular-dystrophy.org](mailto:info@muscular-dystrophy.org)

Research: [research@muscular-dystrophy.org](mailto:research@muscular-dystrophy.org)

#### **Contact a Family**

209-211 City Road,

London EC1V 1JN

Tel: **020 7608 8700**

Fax: **020 7608 8701**

Helpline: **0808 808 3555** or

Textphone: **0808 808 3556**

Freephone for parents and families (10am-4pm, Mon-Fri)

Email: [info@cafamily.org.uk](mailto:info@cafamily.org.uk)

Web: [www.cafamily.org.uk](http://www.cafamily.org.uk)

#### **Other MDC factsheets that may be useful:**

- Congenital muscular dystrophies
  - MDC1A (merosin-deficient congenital muscular dystrophy)
  - Rigid spine syndrome
  - Ullrich congenital muscular dystrophy
- Carrier detection tests and prenatal diagnosis of inherited neuromuscular conditions
- Inheritance and the muscular dystrophies
- Muscle biopsies

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