



Congenital muscular dystrophy

What is congenital muscular dystrophy?

The congenital muscular dystrophies are a group of conditions that share an 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) of the ankles, hips, knees and elbows. Rarely, contractures can be severe and affect several joints (known as arthrogryposis). Contractures occur because the baby has not had the muscle strength to move around freely in the womb. Some babies with congenital muscular dystrophy may also have respiratory problems owing to weakness of breathing muscles.

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

What causes congenital muscular dystrophy?

The congenital muscular dystrophies are a varied group of conditions. They are generally grouped into two main types:

- children who have muscle weakness involving all muscles, but have average intellectual function
- 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 and mutations responsible for each form. A number of specific conditions are recognised, but for others a final genetic diagnosis may not be possible.

Genetic advances. There have been recent developments in research into the genetics of congenital muscular dystrophies, which have resulted in a better understanding of this group of conditions.

This factsheet is under review, due for updating later in 2017. If you have any queries, please contact us.

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The first mutation to be discovered was in the LAMA2 (laminin alpha-2) gene, the gene which carries the information to make a protein called merosin. This form of congenital muscular dystrophy, also known as 'merosin-deficient congenital muscular dystrophy' affects approximately 25 percent of the children with congenital muscular dystrophy. More recently, further genes have been identified, and to date we know of at least 19 genes responsible for different forms of congenital muscular dystrophy. In the UK, the most common type of congenital muscular dystrophy is Ullrich congenital muscular dystrophy, followed by merosin-deficient congenital muscular dystrophy. There are various other less common subtypes.

Some cases of congenital muscular dystrophy have genetic causes that have not yet been identified.

However, the availability of new genetic technologies has increased the probability that the genes for these conditions will be found.

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 owing 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 protein measured – the creatine kinase (CK) level. In approximately 40 percent of cases of congenital muscular dystrophy, this level is five to 20 times higher than normal.

An **electromyography** (EMG) test may also be performed. A small needle is inserted into muscle and the electric activity recorded. This test, which may provide evidence of an abnormal pattern of electric activity in the muscle, is usually not necessary in children who have markedly elevated serum CK.

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

A **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
- muscle proteins can be studied in detail with specialised tests. This greatly helps to narrow down the diagnosis.

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

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Is there a treatment or cure?

There is currently no cure for the congenital muscular dystrophies, but there are ways of helping to manage the effects of the condition.

As this condition can be managed by timely recognition, professional advice and intervention, it is advisable for individuals with congenital muscular dystrophy to be regularly followed by a paediatric neurologist with expertise in muscle-wasting conditions, working as part of a multi-disciplinary team. Review should include monitoring weight, respiratory function, muscle strength and joint range. There are several additional examinations that might be needed, such as overnight sleep studies to monitor the breathing quality during sleep and, in some subtypes, a yearly echocardiogram. In children with respiratory impairment, it is also advisable to reduce the risk of chest infections by giving annual flu immunisations and other vaccinations.

Children and adults with congenital muscular dystrophy should ideally be followed regularly in a specialist neuromuscular clinic, with access to physiotherapy, orthotic, respiratory, orthopaedic, spinal and genetic specialists as needed.

What is the prognosis?

The condition is usually fairly stable as far as the muscle power in the legs and arms is concerned, and often the child appears to gain strength in the first decade of life. In several forms of congenital muscular dystrophy, the acquisition of new skills – over time – is possible, although some motor difficulties will always be present, depending on how severe the condition was at presentation.

In many congenital muscular dystrophy subtypes, muscle weakness can increase with time and can lead to respiratory problems. This may happen in children of various ages and is potentially a very serious complication which, if not recognised, can be life-threatening.

The severity of this condition varies greatly from person to person. As the severity varies even within the same form of congenital muscular dystrophy, it is important not to assume that certain motor developments will or will not take place, but rather to work with children so they can achieve the goals which are within their ability.

Some children will walk, but sometimes this can be delayed until the age of five or older. Leg splints are often used to assist a child to walk. Some children who have achieved independent walking may lose this ability later on because, as they grow older and heavier, the muscles are unable to cope with a greater strain. Other children may not be able to walk at all.

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

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As the muscles are weak and mobility is limited, the child may either be born with – or develop – joint ‘contractures’ or tightness. This means that the muscle tendons tighten up, and the child is unable to move the limbs or the joints as freely as a non-affected child can. Physiotherapy can help to prevent or slow the progression of contractures, therefore a programme of exercises should be established with the help of a physiotherapist soon after diagnosis. Hips are commonly affected with contractures and may sometimes be dislocated, which may require treatment with a splint or surgery.

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

In some subtypes of congenital muscular dystrophy, the function of the brain can be affected and give rise to different degrees of learning difficulties. This complication occurs only in specific subtypes and is not progressive. It will usually be evident in the first year of life, and its severity can vary considerably. However, this complication will not develop at a later stage in children who have average intellectual function.

We’re here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child’s muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it’s like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you’re entitled to.

If you have feedback about this factsheet please email info@muscular dystrophyuk.org.

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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK’s London office are available on **0800 652 6352** or info@muscular dystrophyuk.org from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can’t help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org

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