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Congenital muscular dystrophies

What is congenital muscular dystrophy?

The congenital muscular dystrophies (CMD) are a group of conditions which 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 arthrogyposis). Contractures occur because the baby has not had the muscle strength to move around freely in the womb. Some babies with CMD may also have respiratory problems owing to 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 up, or delays in learning how to sit unaided, stand or walk.

What causes congenital muscular dystrophy?

CMD covers a very varied group of conditions. These are generally grouped into two main types:

1. children who have muscle weakness involving all muscles, and who 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 into 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 the genetics of congenital muscular dystrophies, which have resulted in a better understanding of this group of disorders.

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 CMD, also known as 'merosin-deficient CMD' affects approximately 25 percent of the children with congenital muscular dystrophy. More recently, further genes causing CMD have been identified, and to date we know of at least 19 genes responsible for separate forms of CMD. In the UK, the most common type of congenital muscular dystrophy is Ullrich congenital muscular dystrophy, and merosin-deficient CMD is the second most common individual condition. Various other subtypes are less common.

Some cases of CMD do not have a genetic cause yet 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 CMD is usually first diagnosed as a 'floppy baby'. Doctors can see the symptoms described above, but as these could be because of 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 or CK level). In approximately 40 percent of cases of congenital muscular dystrophy, this level is between five and 20 times higher than normal. An **electromyography** (EMG) test may also be performed, where 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. This test is usually not necessary in children who have markedly elevated serum CK.

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

A muscle biopsy can help to identify the subtype of CMD 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 CMD in which the gene defect has been identified, **genetic tests** can provide a definitive diagnosis.

Is there a treatment or cure?

At the moment there is no cure for the CMD, but there are ways, described below, 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 CMD to be regularly seen by a paediatric neurologist with expertise in neuromuscular conditions, working as part of a multidisciplinary 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 CMD 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 CMD should ideally be seen 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 legs and arms is concerned, and often the child appears to gain strength in the first decade of life. In several forms of CMD, the acquisition of new skills with time is possible although some motor difficulties, dependent on how severe the condition was at presentation, will always be present.

In many CMD 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 CMD, it is important not to assume that certain motor developments will or will not take place, but rather to work with children with CMD so that they can achieve the goals which are within their ability.

Some children will walk, but sometimes this can be delayed until five years of age 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 never be able to walk.

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

As the muscles are weak and mobility is limited, the child may 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 healthy child. 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 CMD but are less frequent in others.

In some subtypes of CMD, 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. This usually will be evident in the first year or life, and its severity can vary considerably. However, this complication will not develop at a later stage in children whose intellectual function is not affected.

Other related publications

This factsheet is to be used alongside the following publications:

- Congenital muscular dystrophies
 - MDC1A (merosin-deficient congenital muscular dystrophy)
 - SEPN1-related myopathy
 - Ullrich congenital muscular dystrophy
 - Bethlem myopathy
- Carrier detection tests and prenatal diagnosis of inherited neuromuscular conditions
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders

The friendly staff in the care and support team at the Muscular Dystrophy Campaign's London office are available 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 people who can help.

Contact our Freephone helpline on 0800 652 6352 or info@muscular-dystrophy.org

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