

## **Congenital fibre type disproportion**

Congenital fibre type disproportion falls under the umbrella of congenital myopathies, a group of conditions characterised by muscle weakness and wasting. It is a rare condition and the genetic basis of it is not yet known. Common features of the condition include poor muscle tone, delay in achieving motor milestones, breathing problems, and possible bone deformities. There is currently no cure, or effective treatment, but management of the condition is very important.

### **What is congenital fibre type disproportion?**

Congenital fibre type disproportion falls in the category of congenital myopathies. This is a group of conditions characterised by muscle weakness and wasting. The condition is so named because of the appearance of muscle fibres under a microscope. Muscle tissue has two different types of fibre; type 1 and type 2. In normal muscle these are approximately the same size, and the proportion of them is even. In muscle of people affected by congenital fibre type disproportion, type 1 fibres are visibly smaller than type 2, and there are often more type 1 fibres than type 2.

### **What causes it?**

Congenital fibre type disproportion is an inherited condition. The genetic cause is unknown but probably it reflects a group of different conditions which have the same appearance on muscle biopsy. In most instances the myopathy is thought to be passed on in an autosomal recessive pattern, meaning that a child must inherit one copy of the genetic error from each parent, although neither of them usually has any symptoms. There have, however, been cases reported of autosomal dominant inheritance, where only one copy of the error is needed to have the condition. In such cases, all children of an affected parent have a 50% chance of also having the condition. For both patterns of inheritance, both males and females can be affected. Many sporadic cases have also been seen where there has been no previous family history.

More information on genetic inheritance is available from the Information and Support Line. (Contact details shown below.)

### **What are the common features?**

Congenital fibre type disproportion becomes apparent at birth or in early childhood, and the initial signs are floppiness (hypotonia) and motor delay such as delay in crawling and walking. Generalised muscle weakness is seen and sometimes children are born with dislocated hip(s). Breathing problems can occur and these vary in severity, but can be life-threatening in some cases. Generally this is more serious during the first year of life and improvement is seen after the first year. Other characteristic features include curving of the spine (scoliosis), tightening of joints (contractures), deformities of the feet and a high arched palate. Problems with the heart are only rarely seen.

### **How is it diagnosed?**

Many of the features of congenital fibre type disproportion are also associated with other conditions, in particular other congenital myopathies. For this reason, a diagnosis of congenital fibre type disproportion must be confirmed through a **muscle biopsy**. In a muscle biopsy, a sample of muscle is taken, and examined under a microscope. This is done in one of two ways: either a small piece of muscle is taken under general anaesthetic or a needle biopsy is performed to remove a small sample. As mentioned earlier, muscle from people with congenital fibre type disproportion has a characteristic pattern when viewed under a microscope. A factsheet on *Muscle biopsies* is available from the Information and Support Line.

### **What other tests are available?**

Since the gene or genes involved in congenital fibre type disproportion have not yet been identified, a genetic test is not available for this condition. This also means that prenatal diagnosis and carrier testing are not currently available.

### **How will it progress?**

Some people with congenital fibre type disproportion show an initial progression, but it is very rare that any progression will be seen after 2 years of age. Unlike some other congenital myopathies, congenital fibre type disproportion becomes static, and some individuals may even show improvement over time.

### **Is there a treatment?**

There is currently no treatment for congenital fibre type disproportion, but management of the condition is very important for maintaining quality of life.

- **Physiotherapy** - The primary aim of an individual with a neuromuscular disorder is to increase or at least maintain function and mobility. Physiotherapy can assist in doing this, and it can also maintain breathing capacity, delay the onset of curvature of the spine (scoliosis), and help prevent the development of contractures. It is important that the physiotherapist involved is familiar with the treatment of people with neuromuscular disorders.
- **Exercise** - There is debate over whether people with neuromuscular disorders should undertake strenuous physical exercise. Some say that putting additional strain on already weakened muscles will cause additional harm, whilst others believe that the exercise may increase muscle strength. Insufficient evidence exists to support either, but it is believed that moderate non-weight bearing exercise such as swimming, walking or peddling may be the best solution. This sort of aerobic exercise helps to maintain a healthy cardiovascular system and a steady weight. It is however, important that this is discussed fully with a clinician.
- **Night time ventilation** - Breathing problems are common with congenital fibre type disproportion, and thus respiratory function should be regularly monitored. A decrease in oxygen intake can lead to, among other things, headaches, breathlessness, poor appetite and disturbed sleep. Night time ventilation involves the use of a face mask attached to a

small machine, which assists in breathing. This aids the muscles that control breathing, and allows a greater intake of oxygen. Night time ventilation may be beneficial to people with congenital fibre type disproportion, but this should be discussed fully with a consultant to determine whether it is appropriate. If there is a tendency to chest infections it is worth considering pneumovax and the flu vaccine.

- **Corrective surgery** - Scoliosis, or curvature of the spine, is common with congenital fibre type disproportion. Spinal surgery aims to correct the posture by realigning the spinal column, and involves the insertion of rods, screws or wires. There are benefits and risks associated with this surgery, and more information is available from the Information and Support Line. As with other treatments, it is very important that the options are discussed fully with a consultant or specialist, before a decision is made. In young children a spinal brace may be used and in children who do not walk moulded seating is used.

### Other things to consider

**Anaesthetics** - It has now been recognised that the use of general anaesthetics in people with neuromuscular disorders, can cause a variety of different problems. Although anaesthetics are generally well tolerated by people with congenital fibre type disproportion, due to the nature of the anaesthetic drugs used, problems can include dysfunction of the heart, and relaxation of the muscles round the lungs causing problems with breathing. Generally if a patient is properly assessed and monitored, the risks associated with anaesthetic use are low, but it is very important that the medical professionals involved are fully aware of the muscle condition.

**Medical alert card** - It is very important that health professionals are aware of your condition should you require treatment. There are often issues they will have to consider. Many companies are able to provide a Medic Alert Card, which can be carried to advise of any medical condition. These come in the form of bracelets, pendants etc and carry information such as the name of the condition in the case of congenital fibre type disproportion and if the child has breathing problems this can be included. Medic alert cards are not essential but some families choose to have them for their children. Please contact the Information and Support Line for details of the companies that provide alert cards.

### Is there a cure?

Currently there is no cure for the congenital fibre type disproportion although much research is currently being conducted into the myopathies. Although there is no effective treatment to halt the progression, there are different ways to manage the symptoms of congenital fibre type disproportion and these are outlined above.

### What research is currently being done?

Researchers world-wide are exploring many avenues in an attempt to develop more effective treatments and hopefully a cure. The research department at the Muscular Dystrophy Campaign, regularly monitors research advances in the congenital myopathies, and produces releases which are sent to members when significant scientific advances occur.

### **Planning for the future?**

Since congenital fibre type disproportion is generally a non-progressive condition, the needs of affected individuals will not change greatly. Depending on the severity of the condition there are things which may have to be considered, such as education, adaptations and holidays. Further information on these subjects can be obtained from the Information and Support Line.

### **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 Support 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**

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:**

- Myopathy
- Congenital myopathies
  - Central core disease
  - Minicore (multicore) myopathy
  - Myotubular (centronuclear) myopathy
  - Nemaline (rod) myopathies
- Mitochondrial myopathies
- Ocular myopathies
- Inheritance and the muscular dystrophies
- Muscle biopsies
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders

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