Congenital Myopathies (CM)

Including 5 related types:

- Central core myopathy
- Centronuclear myopathy
- Congenital fibre-type disproportion myopathy
- Multi mini-core myopathy
- Nemaline myopathy

Processing a new diagnosis

A diagnosis of CM can be a huge shock for parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your family's life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and your family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at **https://youtu.be/A9XM71knDVw.**

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A **psychologist** is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing.
- A **counsellor** is a trained professional who can help you talk about and work through problems.
- A **social worker** can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma.
- A **genetic counsellor** can help you understand how CMD is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required.
- Your **state or territory neuromuscular organisation** can provide support, advice and information about living with CM.
- Your GP can talk to you about a **mental health plan** and how you can use this to help you access the support and services you need.
- Other people and families living with CM have also experienced the emotional rollercoaster that comes with a diagnosis of CM and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CM in **The Loop Community**.

How to talk to your child and others about a new diagnosis

Having a conversation with a loved one about a diagnosis of CM is not easy. In fact it will probably be one of the hardest talks you will have. But, like all difficult conversations it is important and necessary.

There are many reasons why people may not want to have this conversation, mostly centred on wanting to protect and not upset their loved ones and themselves.

But avoiding the topic is not helpful. Children are often aware of the differences between themselves and their siblings and/or peers and may hear their condition being discussed. At some point your child with CM will have questions and it's important

you answer these as honestly as possible, reassuring them that they have done nothing wrong.

So it's crucial that you have a role in your loved one learning about a CM diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. Together you can learn about the journey ahead.

It will also allow you to

- provide answers to questions in an age appropriate way
- correct any misinformation your child has heard or read
- show your child that you have faith in their ability to handle difficult conversations.

Ultimately, these conversations are an important step in providing your child with the tools to succeed, navigate their world and develop independence and self-advocacy skills.

There is no right time to start this conversation. Research suggests the earlier you do so, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don't

- Do be positive but also realistic
- Do reassure them that they have done nothing wrong and this is not their fault
- Do tell them they will do many wonderful things in their lives – they may just do them differently
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to
- Don't avoid answering questions and don't shut the conversation down

Remember, it is okay to be upset during the conversation but try to avoid breaking down as this will only cause your child greater distress. Children are resilient and they generally handle information of this nature far better than adults.

Understanding CM and how it's diagnosed

What causes CM?

CM is caused by a change (also called a mutation) in a gene (the parts of the cells that tell the cells what to do). The faulty genes cause muscles to be built incorrectly or not work properly, leading to the symptoms of CM. Different types of CM can be caused by a fault in just one gene or in multiple genes. Mutations in the same gene can cause a range of changes in the muscles.

What are the types of CM?

There are many different types of CM which are grouped into different subtypes. Subtypes are named in 2 ways:

1. by the abnormal or damaged areas of the muscle cells that can be seen under a microscope, such as cores, nemaline rods or central nuclei
2. by the specific genetic mutation that causes the condition

Common CM subtypes are:

- Nemaline myopathy
- Central core myopathy
- Multi mini-core myopathy
- Centronuclear myopathy
- Congenital fibre-type disproportion myopathy

How is CM diagnosed?

CM might be suspected when doctors notice a baby is "floppy" at birth or when parents or healthcare workers notice that the

infant is not meeting normal developmental milestones, such as lifting their head during tummy time, rolling or sitting.

The child needs to be assessed by a Paediatric Neurologist (a doctor specialising in muscle conditions in children) who will then decide to do some or all of the following tests to to try to make a diagnosis, including:

- Genetic testing looking for changes in one of the genes that cause congenital muscular dystrophy. This is usually done by sending a neuromuscular panel, which looks at a number of genes causing muscle disorders. Further information about genetic testing can be found on the **Healthdirect website**.
- Electromyography (EMG), a test that involves inserting tiny needles into a muscle to record its electrical activity. An EMG can show if there is any abnormal electrical activity in the muscles or the nerves that control them. Children with an elevated CK level do not usually require an EMG.
- MRI of the muscles to look at the pattern of muscle damage for clues to the type of congenital myopathy.
- Muscle biopsy can be used to narrow down the type of congenital myopathy. A doctor removes a tiny piece of muscle for examination under a microscope to see if there are abnormalities of the muscle fibres. It is minor surgery. Specific muscle proteins can also be studied under the microscope to try to identify the cause of congenital myopathy.

The genetics of CM

The genetics of CM is very complex, with some of the subtypes able to be passed down from parents to their children by more than one method. Inheritance pattern can either be:

- Autosomal recessive – this means both parents must be carriers of the faulty gene and both must pass on this gene to their child. If both parents are carriers, the likelihood of a child inheriting the condition is 25 percent, or one in four pregnancies. Carriers do not show signs and symptoms of the condition. Often a family has no idea that some members are carriers until a child is born with CM.
- Autosomal dominant – this means only one copy of the faulty gene, inherited from either parent, is enough to cause the condition. The condition is so dominant it overrides the normal functioning copy from the other parent. On average, half the children from a parent with the faulty gene will inherit the condition.
- X-linked – Males are born with an X chromosome and a Y chromosome. Females are born with two X chromosomes. When the faulty gene occurs on the X chromosome and is passed on to an offspring:
    - male offspring (sons) will develop the condition, as they only have one X chromosome
    - female offspring (daughters) will not usually inherit the condition as they have two X chromosomes. The normal copy of the gene will compensate for the faulty copy. These females will then be carriers and may pass the condition on to their children.

Genetic counselling

If your child has been diagnosed with CM, your family will usually be referred for genetic counselling. Genetic counsellors are medical professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand CMD and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. A genetic counsellor can also explain family planning options to reduce the risk of passing on the condition to future children. Visit our page on **Genetic Counselling** for more information.

Long-term outlook

Living with CMD usually means life, and the future, looks different from what you had planned. However children and adults with CMD can lead fulfilling, rewarding lives just like everyone else. School, friendships, sports and recreation, arts, university, work and travel are all possible when you have CMD. The long-term outlook for people living with CMD varies and can be difficult to predict. It depends on which genes are involved but can even vary between people with exactly the same condition. Most forms of CMD are slowly progressive or may not change at all over time. Some types of CMD affect the breathing muscles and can shorten lifespan due to breathing complications that develops over time. Generally, speaking unless there is severe respiratory (breathing) failure during infancy, most children with CM will live into adulthood.

Understanding and planning for changes

Learning about CM and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person’s journey with CM will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of CM?

CM usually presents at birth or soon after. Babies with CM are often “floppy”, have weak muscles, no muscle reflexes and experience delays in reaching their motor milestones such as rolling over, sitting up and crawling. They may also have the appearance of a long expressionless face, due to weakness of the facial muscles. As CM is a group of conditions, many subtypes share some common symptoms, though not everyone will experience all of the symptoms listed below.

Common symptoms of CM:

- Respiratory (breathing) problems – Some subtypes of CM may experience weakness within the respiratory muscles. This can increase the risk of lung infections.
- Nutrition and growth – children with CM don’t follow the normal growth patterns. Warning signs of an eating problem include frequent choking or coughing when eating or drinking, vomiting, reflux and constipation.
- Growth and development – children with CM have difficulty achieving motor milestones such as rolling over, sitting up and crawling.
- Skeletal system – common problems for children with CM include scoliosis (curvature of the spine) and contractures (stiffness and shortening of the joints)
- Ear, nose and throat – fluid can often build up and be trapped within the ear canal causing discomfort, mild hearing loss and ear infections
- Heart – most CM subtypes are not associated with heart (cardiac) problems, though in some rare cases they can be. Heart problems are usually due to severe lung problems and therefore should be monitored.
- Pain – the exact cause of pain is not known but may be caused by the stiffness in the joints, thinner bones, or joint deformities that form over time. Muscle soreness is commonly the result of physical exertion such as standing or writing for too long.
- Fatigue – people living with CM often experience fatigue, this could be due to the fact that people with CM burn more calories than people without CM due to differences in their muscle function.
- Orthopaedic complication – contractures, scoliosis and dislocation of the hips are common in children with CM, due to weakness of the supporting muscles
- Reflexes – deep tendon reflexes can be low (known as hyporeflexia) or absent (known as arefelxia).
- Weakness in the eye/eye lids – can occur in some types of CM, however most children will not develop a vision problem as the brain will adapt over time.

Source: ***Consensus Statement on Standard of Care for Congenital Myopathies from Journal of Child Neurology*** *accessed from* **CureCMD**

CM impacts both males and females and occurs in about 1 in 50,000 live births.

For more information about the specific symptoms of the subtypes of CM:

- **The Care of Congenital Myopathy: A Guide for Families** accessed from **Cure CMD**
- **Consensus Statement on Standard of Care for Congenital Myopathies** from Journal of Child Neurology accessed from **CureCMD**

How does CM progress?

The progression of CM differs from person to person, even within the same subtype or between people in the same family with the same genetic mutation. There are a number of factors, including the age of onset of symptoms, the impact on other body systems and how the condition is managed, that play a role in the effects of CM.

What we do know about the course of CM:

- Most cases of CM do not dramatically worsen over time. Many people actually find their function improves throughout childhood and they maintain their muscle strength into adulthood. Children with CM who can walk do not usually lose this ability later in childhood.
- Muscles weakness can change over time, either improving or worsening. In general, after the few first years of life, gradual improvement in muscle strength can be seen, and sometimes maintained into adulthood. This improvement is more likely to occur in those who participate in physiotherapy.

- In mid to late adulthood, muscle weakness may gradually worsen, as it does with everyone as they age. Walking may become tiring or difficult but is usually maintained until much later in life. Other complications such as breathing problems and scoliosis may also develop.
- Walking can sometimes be affected by a rapid growth spurt, such as during puberty.

Life stuff

To find out more about living with a neuromuscular condition and to access stories and peer-advice from the community, visit our **Living Life** section.

Where to find more information about living with CM

Congenital Myopathies fact sheet from Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

Congenital Myopathies from Muscular Dystrophy Association – US based organisation. This fact sheet outlines symptoms, causes, progression and status of research into CM.

Standards of Care

CureCMD has information about CM, particularly **The Care of Congenital Myopathy: A Guide for Families** which provides information to help you understand CM and the medical care needed for children and adults with CM.

Consensus Statement on Standard of Care for Congenital Myopathies, published in the Journal of Child Neurology and available from **CureCMD website**, provides detailed care guidelines for the treatment of CM. Although this is written in medical terms it may be useful for discussions with specialists, doctors, nurses and allied health professionals.

Treatment and management

At the moment there is no cure for CM however research has led to a greater understanding of the condition and what you can do to improve your quality of life. Being a rare disease, it is often not well understood by the general medical community and you may find you need to work in partnership with, and help educate, your healthcare team.

A **Standard of Care for CM** was developed by international experts to set a standard for the diagnosis and clinical care of people affected by CM. The Standard details the medical care required for people with CM, including respiratory (breathing), cardiac and neurological care. These standards are further explained in **The Care of Congenital Myopathy: A Guide for Families**.

Treatment and management of CM should be delivered by a multi-disciplinary care team, made up of a range of specialities such as orthopaedics, respiratory, neurology, cardiac and physiotherapy. Depending on how a person is affected by CM, treatments can include:

- Respiratory (breathing) – it is crucial to prevent infections, such a pneumonia, as they can hinder growth and lead to hospitalisation. Regular monitoring of lung health and function and the annual flu and pneumonia vaccination are recommended. Some children with CM may need to use devices such as ventilators to support their breathing.
- Growth and development – physiotherapy, occupational therapy and speech pathology can help address developmental difficulties, such as problems with movement, self-care and daily tasks, and communication. Monitoring growth is important to recognise the signs of failure to thrive or early signs of obesity. Support from a dietitian can assist people with nutritional issues.
- Skeletal system – treatments include an exercise program, with stretching exercises developed by a physiotherapist, braces or orthoses to assist with scoliosis or contractures, and surgery to correct scoliosis. It may also be recommended that vitamin D and calcium be taken for bone health.
- Ear, nose and throat – for many people living with CM, it is important to monitor their ears as fluid can build up and be trapped causing discomfort, mild hearing loss and ear infections. If this is an ongoing problem, a referral to an ENT specialist may be required.
- Cardiac (heart) – heart problems are rare and usually related to severe lung problems. Regular monitoring for heart problems can be done with an ECG test.
- Pain and soreness – can be treated by regular stretching, manual mobilisation and massage, and medications.
- Fatigue – supportive mobility devices such as scooters, walkers and wheelchairs may assist in reducing fatigue

- Oral care – the health of your teeth and mouth will have an effect on your overall health. Without good oral care, the bacteria in your mouth can reach high levels and cause infections. It is recommended that people living with CM regularly see a dentist.

Family planning

If CM runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. Identifying the faulty gene(s) is the first step in family planning as this will provide an estimate of the level of risk for each pregnancy. If the risk is substantial, a genetic counsellor will discuss pre-pregnancy options such as IVF (in vitro fertilisation). You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests. For more information visit our page on **Genetic Counselling**.

Malignant hyperthermia

Some subtypes of CM are at an increased risk of malignant hyperthermia, an increased body temperature to dangerously high and sometimes even fatal levels. In CM, this can result as a reaction to some anaesthetic medications used during surgery. Before undergoing surgery, it is important to discuss CM with the surgeon and anaesthetist. It is also recommended that people with CM wear a medical alert bracelet in case of unexpected or emergency surgery.

Registry

The Congenital Muscle Disease International Registry (CMDIR) raises awareness, develops standards of care and can help you register to participate in clinical trials, finding treatments or cures for muscle diseases. Joining the registry is voluntary and you can learn more by visiting the **CMDIR website** or registering **here**.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s.**

- Your **state or territory neuromuscular organisation** can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them in our pages about **Counselling**, **Social Work**, **Psychology** and **Wellbeing**
- Living with CM may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support in our **Disability Support Workers** page.
- Other people and families living with CM have also experienced the emotional rollercoaster that comes with a diagnosis of CM and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CM in **The Loop Community**.

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Education

About CM

Congenital Myopathies
Fact sheet from Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

Congenital Myopathies
From Muscular Dystrophy Association – US based organisation. This fact sheet outlines symptoms, causes, progression and status of research into CM.

Standard of Care and Guide for Families

Consensus Statement on Standard of Care for Congenital, Journal of Child Neurology
This statement was developed by international experts to set a standard for the diagnosis and clinical care of people affected by CM. The Standard details the medical care required for people with CM, including respiratory (breathing), cardiac and neurological care.

The Care of Congenital Myopathy: A Guide for Families
A comprehensive care guide developed for families and people with congenital myopathy (CM) in response to community requests for useful information to help guide them.

CM Registry

CMDIR Registry
The Congenital Muscle Disease International Registry (CMDIR) raises awareness, develops standards of care and can help you register to participate in clinical trials, finding treatments or cures for muscle diseases. Joining the registry is voluntary and you can learn more by visiting the **CMDIR website** or registering **here**.