Congenital Muscular Dystrophy (CMD)

Including 8 related types:

- Bethlem myopathy
- Fukuyama CMD (Dystroglycanopathy)
- Laminin a2 related dystrophy
- Merosin-deficient CMD
- Muscle-eye-brain disease (Dystroglycanopathy)
- SEPN1 related myopathy or rigid spine muscular dystrophy
- Ullrich congenital muscular dystrophy
- Walker-Warburg Syndrome (Dystroglycanopathy)

Processing a new diagnosis

A diagnosis of CMD 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 CMD.
- 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 CMD have also experienced the emotional rollercoaster that comes with a diagnosis of CMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CMD in **The Loop Community**.

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

Having a conversation with your child or a loved one about a diagnosis of CMD 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 parents may not want to have this conversation, mostly centred on wanting to protect and their

loved ones.

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 CMD 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 CMD 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 CMD and how it's diagnosed

What causes CMD

CMD is caused by changes (also called mutations) in genes (the parts of the cells that tell the cells what to do). The faulty genes affect the production and function of proteins, the building blocks of muscles. Proteins are important for muscles to work, heal and repair. The proteins affected by CMD are either missing, produced in large amounts, not made properly or don't work correctly, leading to muscle weakness and damage.

Different forms of CMD are grouped together depending on which protein within the muscle is impacted. In some CMDs the affected proteins, such as collagen and laminin, provide structural support for the muscles. Other types of CMD are caused by problems with a protein called alpha-dystroglycan. This protein normally combines with a sugar molecule to form an important link between the muscle cells and their surroundings. In CMD, the protein and sugar molecules aren't able to combine, meaning the protein can't perform its linking role properly.

Only about 25% of CMDs have an identified genetic cause.

What are the types of CMD?

There are at least 33 different types of CMD. They are grouped together depending on which protein within the muscle is impacted.

Types of CMD include:

- Collagen VI related myopathy – these conditions are caused by changes in one or more of the collagen VI genes. The faulty genes result in very little or no collagen produced, or the production of abnormal collagen proteins. Without collagen the structure of the muscle cell weakens. Subtypes include:
  - Ullrich congenital muscular dystrophy –Muscular Dystrophy Australia has a factsheet with more information about **Ullrich CMD**
  - Bethlem myopathy – For more information see Muscular Dystrophy Australia's **Bethlem myopathy factsheet**
- Laminin α2 related dystrophy – is the result of a genetic mutation of the laminin apha-2 gene that makes a protein called merosin. Merosin is important for maintaining muscle cell structure. Without merosin the muscle cell weakens.
  - Merosin-deficient CMD - **Cure CMD** has more information about merosin-deficient CMD on
- Dystroglycanopathies – also known as alpha-dystroglycan related dystrophy. The genes involved in this form of CMD affect a protein called alpha-dystroglycan. This protein is important in helping connect the cell to the structures outside and around the cell. Subtypes of this group of conditions include:
  - Fukuymama CMD
  - Walker-Warburg Syndrome
  - Muscle-eye-brain disease
  - **Cure CMD** has more information about these types of CMD.
- SEPN1 related myopathy – also known as rigid spine muscular dystrophy or multi-minicore disease. The protein affected in these types of CMD is called selenoproteinN1. **Cure CMD** have more information about this group of conditions.

How is CMD diagnosed?

CMD 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.

CMD can be difficult to diagnose and a series of tests are often used to try to make a diagnosis, including:

- A blood test to measure the levels of creatine kinase(CK) in the blood. CK often leaks from damaged muscle cells into the blood. High levels of CK in the blood can suggest a muscle problem but does not confirm CMD.
- 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 scans of the brain can show changes that can be seen in the brain with congenital muscular dystrophy or help to rule out other conditions that cause similar symptoms.
- A muscle biopsy is used to narrow down the type of CMD. 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 CMD. This test is being done less frequently with improved genetic testing.

The genetics of CMD

Most forms of CMD are inherited in an autosomal recessive pattern. 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 CMD.

LMNA-related CMD, Bethlem myopathy and sometimes Ullrich CMD are inherited in a different way, called an autosomal dominant pattern. This means only one copy of the faulty gene, inherited from either parent, is enough to cause the disorder. On average, half the children from a parent with the faulty gene will inherit the condition.

Genetic counselling

If your child has been diagnosed with CMD, 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 and proteins 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 heart or breathing muscles and can shorten lifespan due to breathing complications and heart muscle weakness that develops over time.

Understanding and planning for changes

Learning about CMD 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 CMD 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 CMD?

CMD affects people in different ways and with different severity, depending on which gene and protein are involved. Early signs of CMD include floppiness (low muscle tone, known as hypotonia), delays in achieving milestones such as holding their head up and sitting, contractures (shortened and tightened muscles and joints) and breathing and feeding problems.

It's important to note that not all people with CMD will experience all the symptoms and complications listed here and each person's journey with CMD will be unique. But some of the common symptoms of CMD include:

- Neurological:
  - learning, concentration and memory difficulties
  - seizures (fits) – usually associated with children with CMD who have some brain involvement. Seizures can start at any age.
- Respiratory (breathing) – all types of CMD can affect the breathing muscles. Signs of respiratory problems include:
  - a weak cry
  - ineffective cough
  - repeated chest infections
  - irregular breathing patterns
  - choking during feeding, eating or drinking or on their own secretions
  - weight loss or poor weight gain (failure to thrive)
- Gastrointestinal (feeding and nutrition problems):
  - gastroesophageal reflux (GER)
  - aspiration (breathing food or saliva into the airways)
  - constipation
  - speech and swallowing difficulties
  - poor bone health
  - difficulties with oral and dental hygiene

- Cardiac (heart) – with some forms of CMD, heart problems are likely to develop and require regular cardiac screening. Potential heart problems include arrhythmias (abnormal heart rhythm) and cardiomyopathy (enlarged, poorly functioning heart muscle). Signs of cardiac problems include:
  - fatigue (tiredness)
  - shortness of breath
  - paleness of the skin, lips, nose, gums
  - heart palpitations or irregular heartbeat
  - loss of consciousness (fainting)
  - light-headedness
  - dizziness
- Orthopaedic (bone, joint and muscle problems):
  - problems present at birth
    - arthrogryposis (joint contractures or stiffness)
    - hypotonia (very weak muscles)
    - torticollis (neck muscle tightness that causes tilting and restricted movement of the head)
    - hip dislocation
    - scoliosis (curvature of the spine)
    - clubfoot (twisted foot)
  - problems that develop when a child is older
    - development of contractures (tightening and stiffening of joints
    - scoliosis (curvature of the spine)

Breathing problems often start at night, when people breathe more shallowly. The age at which breathing problems develops varies but begin between the ages of 8 to 15 years.

Cure CMD's **Family Guide** provides more in-depth information about the range of CMD signs and symptoms.

How does CMD progress?

CMD is an extremely variable condition and it is difficult to predict how each child will be affected. Some children walk by themselves or with supports, other children learn how to walk but then become weaker and stop walking, and others may never walk at all. The progression of the condition varies between subtypes and even from person to person with the same type of CMD.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at **https://youtu.be/_6oJ9Mc0UO8.**

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 CMD

CureCMD
Has a range of care guidelines for people living with CMD, their families and health professionals who support them, including respiratory care, cardiac care and an excellent family guide. The guidelines are also available in other languages.

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

Congenital Muscular Dystrophy information page
From Muscular Dystrophy Australia provides a range of CMD information, including fact sheets on some individual subtypes

International organisations

Muscular Dystrophy Association (US)
This website has excellent information regarding CMD. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

Treat-NMD – Neuromuscular Network
This international network has information about CMD, care standards and research registries.

Muscular Dystrophy UK
UK based site that has useful information regarding CMD.

Treatment and management

At the moment there is no cure for CMD 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.

It is important to note that not everyone living with CMD will require the treatments that are listed below.

Current treatment focuses on the management of symptoms and reducing the risk of medical complications. Treatment options include:

Neurological management

Depending on the types of symptoms, treatment/management strategies could include:

- specialised school programs
- working with a **speech pathologist** to improve communication strategies
- medication, particularly if your child has been diagnosed with epilepsy.

Respiratory management

Your child may undergo a number of breathing tests including:

- lung function testing such as spirometry – these tests measure how the lungs are functioning
- sleep study – helpful in monitoring night-time respiration and sleep apnea, if it is present.

Depending on the test results, a range of treatments might be recommended including:

- a cough assist machine
- breath stacking techniques
- chest **physiotherapy**
- the use of non-invasive ventilation or ventilator equipment.

The yearly flu and pneumonia vaccination are also recommended. Visit our page on **Respiratory Care** for more information.

Gastrointestinal management

Feeding problems can be managed with the support of a team, including your doctor, **dietitian**, **speech pathologist**, dentist and **occupational therapist**. Treatments can include:

- medications and antacids to manage reflux
- positioning during feeding
- changing the texture of food if swallowing if an issue

- increasing fibre content
- increasing the amount of water/fluids
- regular dental hygiene from a paediatric dentist.

Cardiac management

Medications, such as ACE inhibitors and beta-blockers, or an implanted defibrillator may be required if your child's heart muscle is affected. Your can learn more at our **Cardiology** page.

Orthopaedic management

Management of problems of the joints and muscles can include:

- daily stretching can help in the management of contractures
- orthoses (splints or braces) can assist with contractures
- spinal bracing may assist with scoliosis
- using mobility equipment such as crutches, walking frames, standing frames, scooters and wheelchairs
- advice from an **occupational therapist** about seating at home and school
- sometimes surgery is required for hip instability, knee contractures, ankle contractures and scoliosis.

Pain and fatigue management

Pain can be a significant and under-recognised problem for people living with CMD. Pain can have many different causes, including both physical and emotional problems. Helping your child manage their pain requires:

- comprehensive assessment of their symptoms
- understanding factors that contribute to pain and things that may help provide relief
- learning ways to deal with the emotional impacts of living with CMD, such as depression, anxiety, fear and stress.

Fatigue is also a problem for many children with CMD. Work with your child's healthcare team to overcome or manage factors that can cause or worsen fatigue, such as their breathing, sleep habits, activity level and various medications. Find out more about how an **occupational therapist**, **physiotherapist** or **psychologist** can help.

CMD treatment resources

Cure CMD has more information on CMD treatment and management, including:

- **Family Guide**, which explains treatment guidelines for CMD based on recommendations by a group of 82 international experts
- **Proactive cardiac care in LMNA - CMD**
- **Hypoglycaemia in CMD**

CMD Registries

You may be interested in registering with the **Congenital Muscular Dystrophy International Registry (CMDIR).** This patient registry is a database that contains information about patients with a particular condition. Clinical trial organisers and other researchers use this information to learn more about the conditions and plan clinical trials. They are also a useful source of information for patients and their families as regular newsletters are sent out.

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 **Psychology**, **Wellbeing**, **Counselling** and **Social Work** pages.
- Living with CMD 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.
- Other people and families living with CMD have also experienced the emotional rollercoaster that comes with a diagnosis of CMD and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with CMD 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 CMD

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

Congenital Muscular Dystrophy information page
From Muscular Dystrophy Australia provides a range of CMD information, including fact sheets on some individual subtypes

There are at least 33 different types of CMD. Here you will find useful resources specific to the following types:

- **Ullrich CMD**

  Muscular Dystrophy Australia has a factsheet with more information.

- **Bethlem myopathy factsheet**
  Muscular Dystrophy Australia has a factsheet with more information.

- **Merosin-deficient CMD - Cure CMD**
  Cure CMD has more information about merosin-deficient CMD.
- **Dystroglycanopathies, including Fukuymama CMD, Walker-Warburg Syndrome and Muscle-eye-brain disease - Cure CMD**
  Cure CMD has more information about these types of CMD .
- **SEPN1 related myopathy - Cure CMD**
  Cure CMD has more information about this Group of Conditions.

Treatment Guidelines

Cure CMD has more information on CMD treatment and management, including:

- **Family Guide**, which explains treatment guidelines for CMD based on recommendations by a group of 82 international experts
- **Proactive cardiac care in LMNA - CMD**
- **Hypoglycaemia in CMD**

Support & Education

CureCMD
Cure CMD has a range of care guidelines for people living with CMD their families and health professionals who support them, including respiratory care, cardiac care and an excellent family guide as listed above. The guidelines are also available in other languages.

CMD Education Series features a series of videos about CMD, ask the expert videos, care planning and overcoming common challenges

International Organisations

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This website has excellent information regarding CMD. You can find a full list of neuromuscular conditions, symptoms, causes and care options, as well as use the clinical trial finder tool.

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