

CONGENITAL MUSCULAR DYSTROPHY

Congenital Muscular Dystrophy (CMD) refers to a group of muscular dystrophies present at birth or within the first six months of life.

INCIDENCE: 1 : 15,000 - 1 : 20,000 births

AVERAGE AGE OF DIAGNOSIS: birth onwards

OCCURRENCE: both males and females can be affected

LIFE EXPECTANCY: infancy to early adulthood. Some will have a shortened life depending on age of diagnosis, severity of symptoms and medical complications

Congenital Muscular Dystrophy (CMD) covers at least 33 different types of muscular dystrophies. CMDs are grouped depending on which protein within the muscle is affected. These proteins are important for the survival and regeneration of the skeletal muscle cells. Muscle weakness and cell damage occurs when these proteins are absent, made in excessive amounts, not made correctly and don't work efficiently. Only about 25% of CMDs have an identified gene cause. Many people with CMD remain genetically unclassified but are classified due to signs and symptoms they present with clinically. Most of these conditions are recessive which means both parents carry a copy of the gene but do not have the condition themselves.

Most types progress slowly and children may even gain the ability to walk independently but at a slower rate and pace compared to peers. Others may never walk and will use mobility aids such as a power wheelchair. Some people will have a shortened life expectancy due to severe heart and breathing problems. There is currently no cure for any types of CMDs, although research is leading to a greater understanding about CMD and this will lead to improved diagnosis and treatments.

Signs and symptoms can include the following and vary in severity depending on which gene and protein are affected: brain abnormalities; intellectual disability; unusual thumbs or joints; paralysed eye muscles or involuntary eye movements; heart problems (enlarged heart muscle or rhythm disturbances); central nervous and peripheral nerve problems; poor muscle function; breathing problems and respiratory failure; poor muscle tone (floppy and flexible), increased muscle tone (rigid spine and joint contractures); speech and swallow problems; skin abnormalities; hearing loss; seizures; spinal curvatures (scoliosis); and delays in achieving motor milestones (inability to ever walk).

This group of disorders includes but is not limited to the following:

- Ullrich / Collagen 6 MDs
- Merosin-Deficient MDs
- Laminopathies
- Dystroglycanopathies
- Titinopathies
- Selenoproteinopathies

Usually, CMD progress slowly and the level of weakness eventually seems to plateau. As children/adults with CMD age their needs will change, and the amount of support and care can increase. Medical complications can arise, therefore the following areas should be monitored: respiratory care and sleep studies; annual heart reviews; speech, swallowing and eating; feeding devices and equipment if required in severe cases.

Planning ahead for services & equipment is essential, particularly given complex & varied nature of CMD in children

CMD presents at birth or early infancy and weakness can be severe and debilitating. Children with CMD may develop a 'sway back' or curvature of the spine due to weakness of the muscles that support posture; they can have hearing loss and may require the use of hearing aids; swallowing and speech difficulties leading to weight loss and frustration of not being understood clearly. Due to the progressive nature of CMD and the severity of physical symptoms, social and psychological impacts may exist. Regular multidisciplinary medical assessments are important and should include clinical psychology and counselling. Heart and lung function assessment is recommended at least annually. Interventions for learning may need implementing in those children with intellectual delay.

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Children and adults with CMDs often rely on carers for a high level of care support, which may affect family dynamics. Enough support needs to be provided to the person with CMD so that their carer can have a break. Independent living can be achieved with the right supports in place.

It is important that service provision is centered around quality of life outcomes & meets the needs of the individual

Genetic testing and counselling should be offered to people with CMDs, parents and family members for future pregnancy planning. Families and individuals affected by chronic conditions need to be empowered in order to have control over their life choices. Through seeking out what support is available, people will be empowered to make the right decisions to achieve their goals. Despite the challenges that a person with a CMD faces, there are many opportunities available to support people to live fulfilling and productive lives. Goal setting and positive attitudes contribute to achieving success in many facets of life.

Parents and families experience enormous grief and loss in the dealing with the diagnosis and change in function of their child with a severe CMD. The knowledge that their child may have a shortened life expectancy can cause parental anxiety and counselling may be helpful for families. The need for "end-of-life" care planning should be discussed when deemed appropriate and referral to palliative care services made.

CONSIDERATIONS FOR PLANNING SHOULD INCLUDE BUT ARE NOT LIMITED TO THE FOLLOWING:

- A consistent General Practitioner (GP)
- Specialists for monitoring and medical care planning eg. neurologist, respiratory, sleep physicians
- Support coordination for complex care needs
- Physiotherapy eg. hydrotherapy, stretching, chest physio
- Occupational therapy eg. to increase independence, equipment assessment
- Speech therapy for swallowing and speech problems
- Assistive technology eg. walker, scooter, manual wheelchair, power wheelchair, voice command devices, alternative augmentative communication devices (AAC)
- Orthotics eg. ankle-foot orthoses (AFO's), spinal braces
- Comfort/sleeping eg. beds, mattresses
- Continence management eg. urinary bottle
- Nutritional support eg. dietician, high calorie drinks
- Opportunities to socialise and be away from home eg. MDNSW Camps, accessible travel
- Assistive ventilation devices eg. CPAP or BiPAP machines, cough assist machines
- Home modifications for access to all areas eg. ramps, lifts, bathrooms, bedrooms, kitchen, outdoor areas, pool
- Vehicle modifications/transport eg. steering wheel conversion, taxis to school, wheelchair ramps
- Adapted sporting and recreation equipment eg. boccia equipment
- Support workers for in home care needs eg. personal care, cleaning
- Professional psychological support eg. counsellor or clinical psychologist
- Future goal planning eg. independent living, study, work choices
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au

MDNSW NDIS Toolkit www.mdnsw.org.au/our-services/support-coordination-your-choice/ndis-faqs

The Loop - Your Neuromuscular Resource Hub www.theloopcommunity.org