FACT SHEET

CONGENITAL MYOPATHIES

MUSCULAR DYSTROPHY NSW
YOUR NEUROMUSCULAR SUPPORT COMMUNITY

Congenital Myopathy (CM) is a term given to a group of rare inherited congenital muscle disorders that present at birth or soon after.

INCIDENCE: 1 : 50,000 live births
AVERAGE AGE OF DIAGNOSIS: birth to 6 months. Some rarer forms in adults
OCCURRENCE: affects both males and females
LIFE EXPECTANCY: depends upon type and severity of complications

Congenital Myopathies (CM) are grouped into types depending on what the problem is with the proteins or chemicals that cause the muscle cell to become abnormal and not contract or relax properly. The muscle cells are looked at under a microscope and some are given a description such as rods, cores or holes. The tone and contraction of the fibres is affected and leads to weakness of the skeletal muscles. Over time, the weakness usually remains fairly stable and some infants/children can get stronger. Myopathies usually have no effect on intelligence.

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

- Nemaline Myopathy
- Central Core Myopathy
- Minimulticore Myopathy
- Centronuclear Myopathy
- Congenital Fibre Type Disproportion Myopathy
- Myotonia Congenita
- Paramyotnia Congenita

Infants with a congenital myopathy present usually at birth with muscle weakness, floppiness, no muscle reflexes, and experience delays in reaching motor milestones such as rolling, crawling and sitting. Some people may have a long expressionless looking face due to the weakness of the facial muscles. There is currently no cure for this group of myopathies, although research is leading to a greater understanding about congenital myopathies, and this will lead to improved diagnosis and treatments.

Infants born with the severe form of congenital nemaline myopathy (approx. 20%) present very floppy and have very little spontaneous movement. Some may have a shortened life expectancy and not live beyond two years of age. Others may be able to walk independently. It is a very variable and complex condition.

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

No specific treatment exists for people with a myopathy. Treatment is supportive and directed toward the specific symptoms that are apparent in each individual. Infants may benefit from a program involving mild-to-moderate, low-impact exercise, massage, and stretching techniques. Such therapy is aimed at preserving muscle strength and function and to prevent the development of contractures.

Weakness in congenital myopathies can be severe and debilitating. Children may never walk and will use a power wheelchair for mobility. They are at risk of developing complications such as a sway back or curvature of the spine due to weakness of the muscles that support posture; breathing and lung complications such as aspiration pneumonia; sucking, swallowing and reflux problems leading to weight loss. Due to the progressive nature and severity of physical symptoms, social and psychological impacts exist. Regular multidisciplinary medical assessments are important and should include clinical psychology and counselling. Heart and lung function assessment is recommended at least annually.

Enough support needs to be provided to the person with CM so that their carer can have a break.

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Parents and families may experience enormous grief and loss in the dealing with the diagnosis and change in function of their child with a congenital myopathy. 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.

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 CM, 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 a child with a congenital myopathy may experience there are many opportunities available to support them to live fulfilling and productive lives. Goal setting and positive attitudes contribute to achieving success in many facets of life.

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, respiratory care
- Occupational therapy eg. to increase independence, equipment assessment
- Speech therapy for swallowing and speech problems
- Assistive technology eg. scooter, manual wheelchair, power wheelchair, voice command devices
- Orthotics eg. ankle-foot orthoses (AFO's), spinal braces
- Comfort/sleeping eg. beds, mattresses
- Nutrition 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. taxis to school, work and the community
- Adapted sporting and recreation equipment eg. boccia equipment
- Support workers for in home care needs eg. personal care, cleaning
- Professional psychological support eg. counsellor, 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

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