FACT SHEET

MYOTONIC MUSCULAR DYSTROPHY

MUSCULAR DYSTROPHY NSW BUILDING STRENGTH REACHING POTENTIAL

Myotonic Muscular Dystrophy (MMD) is a common form of muscular dystrophy occurring in children and adults. In children it is called congenital myotonic dystrophy.

INCIDENCE: 1 : 8,000 live births

AVERAGE AGE OF DIAGNOSIS: birth, teens or older depending on severity

OCCURRENCE: males and females equally affected. Passed from parent to child

LIFE EXPECTANCY: is dependent on the age of diagnosis. The younger someone is diagnosed with MMD the more severe the symptoms usually are, and can be life limiting. People with MMD can have a life expectancy similar to that of the general population unless medical complications arise

Myotonic Muscular Dystrophy (MMD) is a genetic condition that occurs when a section of the faulty gene is larger than it should be and this affects the messages being sent to the muscle cell. This leads to muscle weakness. The genetic change is present from birth, but may not cause symptoms until later in life. MMD affects many organs of the body and varies between individuals even within the same family. The condition tends to be more severe in successive generations, particularly when passed on from a mother.

MMD particularly affects the muscles supporting the skeleton, hands, feet, heart, eyes and can result in breathing difficulties, day time sleepiness, night time breathing problems, digestive problems leading to incontinence, constipation and issues with speech and swallowing. Issues with heart rhythm can result in sudden collapse or death. Most people with MMD will have some form of cognitive difficulty, ranging from very mild to severe. People with MMD have increased risk of reacting to the anaesthetic drugs, they must let the anaesthetic doctors know about their diagnosis. People with MMD often have 'myotonia' - involuntary muscle movements which cause the muscle to tighten and not relax, which can effect function and be distressing.

Infants born with congenital MMD have very distinct features at birth such as weakness and floppiness, facial weakness with a "tented" upper lip, breathing and feeding difficulty, poor cry and turned in or club feet. These problems can be life threatening.

There is no current cure for MMD and treatment is focused on symptom management and reducing medical complications. Medication may be used to help control the involuntary muscle movements and to help reduce day time sleepiness especially in children as it can affect concentration and learning. The earlier a child presents with MMD usually the more severe the condition. Children with this condition will likely require support with learning. Adults with MMD may have subtle features of developmental delay and require assistance with complex paperwork if they cannot read or comprehend information.

Treatments for MMD focus on regular monitoring and the reduction of medical complications. Monitoring of the heart is vital and annual heart checks are recommended. A large proportion of adults with MMD will go on to have a heart pace maker. The muscles of breathing also need to be checked regularly as weakness will lead to lack of efficient sleep and an increased risk of pneumonia. Occupational therapy improves the skills of everyday living and focuses on equipment to enhance mobility and independence. The majority of people with MMD maintain the ability to walk independently well into late adulthood unless complicated by foot deformities.

Planning ahead for services and equipment is essential, particularly given the progressive nature of MMD

As children become adolescents and adults they will have goals similar to their peers. This may include relationships, work, study or planning for holidays and so on. Access and inclusion are paramount to ensure independence, quality education and community involvement. Young people are also challenged by puberty issues especially if there is a degree of cognitive delay which reduces their understanding of changes in their body. Issues related to puberty, relationships and sexuality may require professional intervention.

As people with MMD age their needs will change, and the amount of support and care will increase. Regular assessment is important to plan future medical, physical, social and psychological supports. Medical complications can progress; therefore, the following areas should be monitored: respiratory care; sleep studies and non-invasive mechanical respiratory support; annual heart review; speech, swallowing and eating; continence issues,

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cataracts, diabetes, feeding devices and equipment; foot deformities and related surgeries.

People with MMD may have a distinctive appearance as the weakness of the facial muscles cause their faces to look long and expressionless. The weakness prevents blinking and shutting eyes when sleeping is difficult which can cause eye injuries. Closing of the mouth and speech becomes difficult. Dry mouth, bad breath and difficulty with dental care are issues for people with MMD.

An unusual appearance may cause people to be bullied and withdraw socially. Psychological support is paramount. A person with MMD may rely on others for a certain level of care support, which may effect family dynamics. Enough support needs to be provided to the person with MMD so that their carer can have a break. Independent living is challenging but can be achieved with the right supports in place.

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

Genetic testing and counselling should be offered to people with MMD, 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 appropriate support, people will be empowered to make the right decisions for themselves. Despite the challenges that a person with MMD faces, there are many opportunities available to support people to live fulfilled 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, heart physicians
- Support coordination for complex care needs
- Physiotherapy eg. hydrotherapy, breathing exercises maintaining strength and mobility
- Occupational therapy eg. to support activities of everyday living, increase independence
- Speech therapy for swallowing and speech problems
- Assistive technology eg. walker, scooter, manual wheelchair, voice command devices
- Orthotics eg. ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. may need medication to support normal sleep patterns
- Continence management eg. nappies, incontinence sheets
- Nutritional support eg. feeding pumps & consumables, high calorie drinks
- Opportunities to socialise and be away from home eg. MDNSW Camps
- Assistive ventilation devices eg. CPAP or BiPAP machines
- Home modifications eg. ramps, lifts, bathrooms, bedrooms, kitchen, yard, pool
- Adapted sport and recreation eg. boccia equipment
- Transport eg. taxis to and from school, work and the community
- Medications eg. for cramps or daytime sleepiness which can impair normal activities
- Surgery and hospital admission planning eg. implantable heart devices, foot surgery, cataract surgery
- Support workers eg. showering, toileting, dressing, eating, meal preparation, accessing the community
- Psychological support eg. counsellor or clinical psychologist
- Future goal planning eg. independent living, recreation, study and work
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au
MDNSW NDIS Toolkit www.mdnsw.org.au/ndis
Myotonic Dystrophy Foundation www.myotonic.org

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