FACT SHEET

DUCHENNE MUSCULAR DYSTROPHY

MUSCULAR DYSTROPHY NSW BUILDING STRENGTH REACHING POTENTIAL

Duchenne Muscular Dystrophy (DMD) is the most common form of childhood muscular dystrophy.

INCIDENCE: 1 : 5,000 live births

AVERAGE AGE OF DIAGNOSIS: 2 - 5 years of age

OCCURRENCE: mostly in boy's due to its inheritance pattern. There are some girls with symptoms of DMD. It is not uncommon for there to be more than one sibling affected

LIFE EXPECTANCY: usually in the mid 20's depending on severity of complications

Duchenne Muscular Dystrophy (DMD) is a genetic condition that the child is born with but they may not show symptoms until a few years of age. DMD is caused by a lack of a protein in the muscle cell which causes the muscle to breakdown over time. The muscle cell cannot repair itself. The damage in the muscle leads to significant progressive weakness of the muscles that support the spine and the body such as the arms, legs, the breathing muscles and the heart muscle. Due to the progressive nature of DMD those affected will most likely lose the ability to walk around age 12 years. They will then use a power wheelchair for mobility and will have a shortened life expectancy.

There is currently no cure for DMD. Most children with DMD take daily medication called "glucocorticoid steroids" that can help to decrease inflammation of the muscle cell and slow weakness. The aim is to keep the child walking longer and support their breathing and heart muscles. A proportion of children will also have speech delay and learning difficulties.

Treatments for DMD focus on keeping the child healthy and active for as long as possible. Regular physiotherapy aims to prolong walking and maintain the muscles used for breathing. Occupational therapy improves the skills of everyday living and focuses on equipment to enhance mobility. The person with DMD will need to be engaged with an experienced physiotherapist and occupational therapist to ensure their needs are being met.

As DMD progresses, needs will change, and the amount of support and care the person needs will increase. It is important that they are assessed at regular intervals to plan their future medical, physical, social and psychological supports.

As children become adolescents and adults they will have goals similar to their peers. This may include relationships, work options, study or planning for holidays and so on. Access and inclusion are paramount to ensure independence for education and community involvement. Young people are also challenged by puberty issues especially as the steroid medication can delay pubertal progression and they may be on hormone replacement. Issues related to puberty, relationships and sexuality may require professional intervention.

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

In adulthood, the weakness associated with DMD progresses considerably and it is a time of enormous transition from school to study and/or work. Adults with DMD can have difficulty accessing the wider community and are often at risk of social isolation. It is important for them to get support to overcome barriers such as physical access, transport, social exclusion, financial constraints, health risks, low self- esteem and complicated daily support needs. Given their high support needs, people with DMD can become isolated and withdraw from society, so it is important that a variety of social opportunities are encouraged, for example accessible sport and recreation. A person with DMD relies heavily on others for a significant level of care support, which may effect family dynamics. Enough support needs to be provided to the person with DMD so that their carer can have a break. Independent living is challenging but can be achieved with the right supports in place.

As a person with DMD ages, medical complications can progress, therefore the following areas should be monitored: respiratory care including prevention and treatment of chest infections; sleep studies; non-invasive

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mechanical respiratory support; annual heart reviews; speech, swallowing and eating; constipation, artificial feeding devices and equipment; continence issues and associated equipment needs; curvature of the spine and possible surgery; tightening of the joints, foot deformities and related surgeries; and psychological issues as they grapple with the loss of independence and increased reliance on others such as family, friends or paid carers.

People with DMD and their families experience enormous grief and loss, both in the dealing with the changes in function but also coping with the deaths of friends with DMD. There may come a time for referral to palliative care services for symptom management of pain and discomfort. The need for "end-of-life" care planning should be discussed when deemed appropriate.

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 DMD, parents and family members for future pregnancy planning. Families and individuals affected by chronic life limiting conditions need to be empowered to have control over their life choices. Through seeking information about what is available, people will be empowered to make the right life choices. Despite the challenges that a person with DMD 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 due to progressive nature of DMD and complex care needs
- Physiotherapy eg. hydrotherapy, stretching, chest physio
- Occupational therapy eg. to increase independence, equipment assessment
- Support for children with developmental delay eg. speech therapy
- Assistive technology eg. scooter, manual wheelchair, power wheelchair, shower chair, hoist, slings, voice command devices
- Orthotics eg. leg splints for night-time and ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. air pressure relieving mattress, electronic bed with high/low options
- Continence management eg. urinary bottle
- Nutrition support eg. straws, feeding equipment
- 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, yard, pool
- Adapted sporting and recreation equipment eg. boccia equipment, sports wheelchair
- Vehicle modifications/transport eg. taxis to and from school or work and the community
- Support workers eg. showering, toileting, meal preparation, feeding, accessing community
- Professional psychological support eg. counsellor or clinical psychologist
- Future goal planning eg. independent living, study, work choices
- Surgical interventions and admission to hospital planning
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au
MDNSW NDIS Toolkit www.mdnsw.org.au/ndis
Save Our Sons Duchenne Foundation www.saveoursons.org.au

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