FACT SHEET

FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY (FSHD)

MUSCULAR DYSTROPHY NSW
YOUR NEUROMUSCULAR SUPPORT COMMUNITY

Facio-Scapulo-Humeral Muscular Dystrophy (FSHD) is the most prevalent form of muscular dystrophy that affects primarily the muscles of the face, shoulder area and upper arms. FSHD also affects other muscles of the body but at a slower rate.

INCIDENCE: 1 : 8,300 people
AVERAGE AGE OF DIAGNOSIS: 20 - 49 years (some in childhood)
OCCURRENCE: males and females
LIFE EXPECTANCY: normal life expectancy depending on age at diagnosis and severity

Facioscapulohumeral Muscular Dystrophy (FSHD) is a complex genetic condition caused when a section of DNA on chromosome 4 is too short and this affects its ability to stop certain proteins being “turned off” within the cell. There is currently no cure for FSHD and treatment evolves around physical and supportive therapies. FSHD can run in families therefore genetic testing and counselling should be offered to all parents of an FSHD affected child, and adults with FSHD for future pregnancy planning.

The average age of onset, severity and rate of progression in FSHD varies greatly. If symptoms are noticed in early childhood the more severe the weakness and complications. Symptoms usually become noticeable in their early 20’s or even well into the persons 40’s. People with FSHD may have neck weakness, droopy eyelids, a straight smile, prominent shoulder blades and difficulty lifting their arms up to put on clothing over their heads.

Usually, FSHD progresses slowly and the level of severity eventually seems to plateau. People affected by FSHD of average severity usually retain the ability to walk and have a normal life span. As people with FSHD 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.

If FSHD presents in childhood, weakness can be severe and debilitating. Children with FSHD may develop a ‘sway back’ or lordosis of the spine due to weakness of the muscles that support posture; they can have hearing loss requiring the use of hearing aids; swallowing and speech difficulties leading to weight loss and frustration of not being understood clearly. It is important to protect the eyes from injury as weakness of the eye muscles may prevent them from shutting properly especially during sleep. Due to the progressive nature of FSHD and the 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. Intellectual function is not affected.

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

People with FSHD can have a distinctive appearance as the weakness of the facial muscles may cause their faces to look relatively expressionless. Not being able to express one’s self through facial cues can be very difficult socially.

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Children and adults with severe FSHD 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 FSHD 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 FSHD, 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 for themselves. Despite the challenges that a person with FSHD 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, sleep physicians
- Support coordination for complex care needs
- Physiotherapy programs eg. hydrotherapy, stretching, breathing exercises
- Occupational therapy to support activities of daily living, increase independence
- Speech therapy for swallowing and speech problems
- Assistive technology eg. walker, scooter, manual wheelchair, power wheelchair, voice command devices
- Orthotics eg. ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. beds, mattresses
- Nutritional support eg. dietician, high calorie drinks
- Opportunities to socialise and be away from home eg. MDNSW Camps
- Assistive ventilation devices eg. CPAP or BiPAP machines
- Home modifications for access eg. ramps, lifts, bathrooms, kitchen, outdoor areas
- Vehicle modifications eg. steering wheel conversion
- Adapted sporting and recreation equipment eg. boccia equipment
- Transport eg. taxis to school, work and the community
- 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

Fact sheets produced with funding from Family and Community Services NSW

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