

Facioscapulohumeral
DYSTROPHY

Clinical Consensus on
Diagnosis & Management



In 2015, the Foundation brought together a group of the world's leading experts on FSHD to develop the first clinical consensus on the diagnosis and management of FSHD.

This Consensus provides evidence based advice on diagnostics, rehabilitation and management of complications such as breathing problems. The Consensus also provides advice about what an effective neuromuscular care team might look like to provide the best level of care to people with FSHD.

The Foundation has prepared a suite of materials to support this statement and to help people in Australia who are living with FSHD have access to the best care available.

To view the published journal list online visit [http://www.nmd-journal.com/article/S0960-8966\(16\)30096-7/abstract](http://www.nmd-journal.com/article/S0960-8966(16)30096-7/abstract)



Clinical practice points

Diagnosis

Genetic testing should be considered for all individuals displaying signs and symptoms of FSHD in the absence of a first degree relative with genetically confirmed FSHD.

Genetic testing of D4Z4 repeat length and/or SMCHD1 mutations is recommended for patients suspected of having FSHD who have a family history or who are displaying traditional symptoms.

Lifestyle interventions

Aerobic exercise is probably not harmful and may be beneficial for people with FSHD. Consider referral to rehabilitation specialists with experience in neuromuscular disease.

To date, there are no dietary interventions that modify the progression of FSHD.

Rehabilitation

Orthotics may be helpful to prevent falls and aid in completion of activities of daily living. Consider referral to a rehabilitation specialist, physiotherapist, or an occupational therapist and/or assessment by an orthotist.

Mobility aids may help maintain independence. Consider referral to a rehabilitation specialist for assessment.

Surgery

Scapular fixation therapy may be effective in stabilising the shoulder joint and assisting in movement of the upper body. Consider referral to an orthopedic surgeon with experience in neuromuscular conditions.

Pain management

Pain is a common presentation in FSHD. Consider routinely enquiring about pain. Options for management include physical therapy and bracing. Pharmacological options may include nonsteroidal anti-inflammatory medications and adjuvant agents such as tricyclic antidepressants or antiepileptics.

Bone health

FSHD is associated with poor bone health which can make people with FSHD vulnerable to fracture. Consider assessment of bone density using dual-energy

Xray

Absorptiometry (DEXA) in people who meet screening guidelines and managing osteoporosis according to guidelines for the general population.

Respiratory management

Respiratory insufficiency due to respiratory muscle weakness occurs in one in ten people with FSHD.

Sleep disorder

Breathing due to upper airway obstruction may occur in people with FSHD.

Regular monitoring of respiratory function is recommended in all people with FSHD.



The FSHD Global Research Foundation is committed to advancing global medical research, education and collaboration to improve quality of life and ultimately find a cure for Facioscapulohumeral Dystrophy. Through transparency, accountability, good governance and pure passion we aim to achieve results as quickly as possible.



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