

# **Facioscapulohumeral dystrophy: A pediatric perspective**

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# Infantile FSHD Case Definition

Facial weakness < age 5

Shoulder girdle weakness < age 10

Other working definitions:

Wheelchair use > 50% by age 18

EcoRI fragments < 15kb

# CNS manifestations of iFSHD

Retinal vasculopathy  
Cochlear dysfunction

Epilepsy  
Intellectual disability

# **CINRG\* Early Onset FSHD Study**



\*Cooperative International Neuromuscular Research Group

# Objective

To describe motor function associations with age, sex, and D4Z4 repeats among participants with early onset FSHD type I.

# Inclusion Criteria

Participants had a genetically confirmed contraction of the *D4Z4* repeat array, ranging from 1–10 units on chromosome 4q35.

Additionally, they met criteria for early onset FSHD based on symptoms or signs of facial weakness before 5 years of age and/or shoulder girdle weakness before 10 years of age.

# Methods

Standardized motor function assessments, including:

- Manual muscle testing (MMT)
- Quantitative muscle testing (QMT)
- Timed functional evaluations
- FSHD clinical severity scores (CSS)

# FSHD Clinical Severity Score

Lamperti et al 2010

Facial muscles

Scapular girdle muscles

Upper limbs

Lower limbs

Pelvic girdle muscles

Abdominal muscles



# Results:

**53 participants (22 males; 31 females)**



# Results (I)

Weakness was most pronounced in the shoulder and abdominal musculature.

Older enrollment age (and shorter D4Z4 repeats) was associated with greater disease severity (as measured by CSS,  $p=0.003$ ) and slower timed function velocities.

# Results (2)

QMT measurements of strength confirmed weakness of shoulder abduction and adduction, elbow flexion, and knee extension.

Side-to-side asymmetry in muscle strength was noted, unrelated to hand dominance.

# Results (3)

Among 36 patients who were able to complete the 6MWT, their mean z-score for the total distance was -1.98 (SD 1.91) when compared with reference data from healthy age- and sex-matched controls.

# Results (4)

When adjusted for enrollment age, sex, and *D4Z4* repeats, younger age at onset of facial weakness was associated with more severity, slower velocities in timed function tests, and lower MMT scores ( $p < 0.05$ ).

# Conclusion

Significant clinical variability was observed in early onset FSHD

Earlier age at onset of facial weakness was associated with greater disease severity.

Other longitudinal studies are ongoing to determine the rate of disease progression in this population.

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