

Clinical Severity Scoring:

We are currently using the age-adjusted (see below) 10-grade clinical severity scale (CSS) developed by Ricci et al. (1). This severity score takes into account the extent of weakness in various body regions and considers the descending spread of symptoms from face and shoulders to pelvic and leg muscles typical of FSHD (Table below). Higher scores are assigned to patients with involvement of pelvic and proximal lower limb muscles. Weakness of such muscles always follows that of facial and shoulder muscles and Ricci et al contend that it represents an unequivocal index of disease progression.

Though, a reasonable argument could be made that a descending progression is typical for most patients with FSHD, it is not always the case.

Since age at disease onset is very variable in FSHD, van Overveld et al. (2) describe a severity scale that adjusts for the patient's age at examination:

$$\text{Age-corrected CSS} = ((\text{CSS} \times 2) / \text{age at examination}) \times 1000$$

The severity score is multiplied by two to generate whole numbers and then multiplied by 1000 to improve interpretation of the results and visualization in graphs.

Disease Severity Scale:

0.5 Facial weakness

1 Mild scapular involvement without limitation of arm abduction; no awareness of disease symptoms is possible

1.5 Moderate involvement of scapular and arm muscles or both (arm abduction >60° and strength ≥ 3 in arm muscles); no involvement of pelvic and leg muscles

2 Severe scapular involvement (arm abduction <60° on at least one side); strength <3 in at least one muscular district of the arms; no involvement of pelvic and leg muscles

2.5 Tibioperoneal weakness; no weakness of pelvic and proximal leg muscles

3 Mild weakness of pelvic and proximal leg muscles or both (strength ≥ 4 in all these muscles); able to stand up from a chair without support

3.5 Moderate weakness of pelvic and proximal leg muscles or both (strength ≥ 3 in all these muscles); able to stand up from a chair with monolateral support

4 Severe weakness of pelvic and proximal leg muscles or both (strength <3 in at least one of these muscles); able to stand up from a chair with double support; able to walk unaided

4.5 Unable to stand up from a chair; walking limited to several steps with support; may use wheelchair for most activities

5 Wheelchair bound

References:

1. Ricci E, Galluzzi G, Deidda G, Cacurri S, Colantoni L, Merico B, Piazzo N, Servidei S, Vigneti E, Pasceri V, et al. Progress in the molecular diagnosis of facioscapulohumeral muscular dystrophy and correlation between the number of KpnI repeats at the 4q35 locus and clinical phenotype. *Ann Neurol* 1999; 45(6): 751-757.
2. van Overveld PG, Enthoven L, Ricci E, Rossi M, Felicetti L, Jeanpierre M, Winokur ST, Frants RR, Padberg GW, van der Maarel SM. Variable hypomethylation of D4Z4 in facioscapulohumeral muscular dystrophy. *Ann Neurol* 2005; 58:569-76.

FIELDS
FSHD & Neuromuscular Research
CENTER