FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY: A COLLABORATIVE STUDY ON CLINICAL COURSE

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FacioScapuloHumeral Muscular Dystrophy (FSHD), an autosomal dominant disease linked to a 4q35 deletion, is characterized by a variable clinical progression. However studies focused on clinical course are not reported. Preliminary data among our series of 142 FSHD cases confirmed the usual benign clinical course, with evidence of 14% of patients wheel-chair-bound after 40 years of age. We present data on disease progression concerning 48 subjects, evaluated during a mean period of 7 years (range 4-10). They were 24 males and 24 females, with a mean age of 50 years (19-76). The mean EcoRI fragment was 24 Kb (range: 15-35). The investigation was carried out by a standardized protocol that included Manual Muscle Testing (MRC) and four Functional Tests (walking, Gowers' manoeuvre, arms-raising, arms-forwards). General muscular ability was graded by a Disability Scale ranging I (mild) to VII grade (severe weakness). We identified worsening by at least one Functional Test in 34 cases (71%): "walking test" was the most sensitive to changes. Variation in MRC score was more evident for tibialis anterior and brachial biceps. Disability Scale variation was evident in 56% of our cases, appearing very slow in most (42%) and very fast in few (14%). A static clinical course characterized an evident part of them (44%). On the whole, our investigation on the FSHD disease course indicates that weakness progression is very variable among patients. Worsening of Disability Scale was evident in 56%, while singular Functional Tests appeared more sensitive (71%), with particular evidence for the "Walking test".