

Facioscapulohumeral muscular dystrophy. Report of seven patients Distrofia muscular facioescapulohumeral en Chile: Presentación de serie en hospital de referencia terciario

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© 2015, Rev Med Chile. All rights reserved. Background: Facioscapulohumeral muscular dystrophy is the third most common muscular dystrophy with an estimated prevalence of 1 per 20.000 and a normal life expectancy in the majority of patients. However, approximately 15% of patients become wheelchair bound in the course of their life. It is a hereditary autosomal dominant disease with high (95%) penetrance by the age of 20, but with variable degree of phenotypic expression even in the same family group. Symptoms frequently start in the second decade of life, with facial and scapular weakness. Aim: To report the clinical features of seven patients with the disease, seen at a public hospital. Material and Methods: Analysis of seven patients with genetic study seen in a public Hospital in Santiago. Results: The age of patients fluctuated from 18 to 61 years and four were females. The mean age at onset of symptoms was 29 years and four had a family history of the disease. The usual presenting