Natural history of Ullrich congenital muscular dystrophy.

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Abstract

OBJECTIVE: To describe the course, complications, and prognosis of Ullrich congenital muscular dystrophy (UCMD), with special reference to life-changing events, including loss of ambulation, respiratory insufficiency, and death.

METHODS: Review of the case notes of 13 patients with UCMD, aged 15 years or older at last visit, followed up at a tertiary neuromuscular centre, London, UK, from 1977 to 2007. Data collected were age at onset of symptoms, presenting symptoms, mobility, contractures, scoliosis, skin abnormalities, respiratory function, and feeding difficulties.

RESULTS: The mean age at onset of symptoms was 12 months (SD 14 months). Eight patients (61.5%) acquired independent ambulation at a mean age of 1.7 years (SD 0.8 years). Nine patients (69.2%) became constant wheelchair users at a mean age of 11.1 years (SD 4.8 years). Three patients continued to ambulate indoors with assistance. Forced vital capacity (FVC) values were abnormal in all patients from age 6 years. The mean FVC (% predicted) declined at a mean rate of 2.6% (SD 4.1%) yearly. Nine patients (69.2%) started noninvasive ventilation at a mean age of 14.3 years (SD 5.0 years). Two patients died of respiratory insufficiency.

CONCLUSION: In Ullrich congenital muscular dystrophy (UCMD), the decline in motor and respiratory functions is more rapid in the first decade of life. The deterioration is invariable, but not always correlated with age or severity at presentation. This information should be of help to better anticipate the difficulties encountered by patients with UCMD and in planning future therapeutic trials in this condition.

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