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Limb girdle muscular dystrophy type 2E: A case report

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In this case report a 23 years' boy from consanguineous marriage was referred to our hospital (Isfahan Alzahra hospital muscles). He is wheelchair bound and also has a cousin with signs and symptoms similar to him. She achieved normal motor milestones in childhood and had normal growth parameters. She started frequent falling from5yrs and she had difficulties in walking from 9yrs and from 10yrs she became wheelchair bound. Genetic test for Duchene muscular dystrophy was negative. He had frequent falling from 4yrs. He had slowly progressive proximal lower limb weakness at childhood (from 7 years old) and then distal lower limb and then upper limb weakness at 14 years old. He was completely disabled and became wheelchair bound at age 10 years. At present he has dysarthria and head drop. He complained from recurrent shoulder and hip dislocation.

In neurological examination he had normal mental activity. Multiple skeletal deformities such as severe lordosis, Achilles, knee and elbow contracture, pes cavus was detected. His force of the muscles was decreased in both upper limbs (particularly proximal muscles) and lower limbs (particularly proximal muscles) DTRs were absent Babinski was negative. Sensory exam was normal. EMG-NCV reported generalized non irritative myopathy. The ejection fraction was 35%. CPK :847. Gene analysis was sarcoglycanopathy (LGMDE2), SGCB gene.