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Dysferlinopathy misdiagnosed as inflammatory myopathy

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A 20-year-old man presented with low back pain and lower limb pain then slowly progressive weakness in lower limbs from 2 years ago and exacerbated from 4 months ago. He complained myalgia and cramp but without ptosis, ophthalmoparesis, dysphagia and cardiac problems but he had mild dyspnea during walking. Before referring to our neuromuscular clinic, treatment with prednisolone, methotrexate and azathioprine was started from two months ago with diagnosis of inflammatory myopathy and because of the exacerbation of weakness, he stopped this treatment arbitrary. His medical, developmental and family history was unremarkable. His parents are relatives.

In neurological examination, cranial nerves were intact without facial weakness or gaze palsy. Neck flexor and extensor strength was 5/5. There was no scapular winging. Muscle strength in upper limb was 5/5 and in lower limb was: hip flexion 2/5, quadriceps 0/5, dorsiflexion 3/5, and plantar flexion 2/5. Atrophy of calves was seen. DTR was absent in lower limb but present in upper limb. Sensory and cerebellar examination were normal. He had a waddling gait and had difficulty in walking on his heels and toes. Gower's sign was positive. The electrodiagnostic study revealed irritative myopathy. Lab data showed elevated CPK level (11791). Muscle biopsy was obtained. Gene analysis identified one heterozygous disease-causing mutation in [c.3059C>T] of the dysferlin protein. According to these findings, the patient was diagnosed as limb-girdle muscular dystrophy type 2B (LGMD R2).