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Clinical features and diagnosis of congenital myasthenic syndrome at older age: A case report

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Background & Aim: Congenital myasthenic syndrome is an uncommon cause of neuromuscular junction disorders caused by genetic defect in pre-synaptic, synaptic basal lamina or post synaptic component of neuromuscular junction with no involvement of immune system. In this study a case of congenital myasthenia diagnosed by history, physical exam and electrophysiological study of an old male at age of 19 is described. The objective is to describe a case of congenital myasthenia graves diagnosed at age of 19 years old male.

Case Report: 19 years old male with kyphoscoliosis presented with bilateral symmetrical lid ptosis since birth with evidence of proximal muscle weakness mainly when he tries to stand from sitting position, SOB on exertion and hypophonic voice on prolonged talking. He denies diplopia, dysarthria, dysphagia, sensory symptoms or sphincter dysfunction. Neurological exam showed symmetrical lid ptosis with limitation in lateral gaze in both eyes only; there were no signs of other ocular or bulbar deficit or proximal muscle weakness in time of examination. Ice back test showed improvement in bilateral ptosis. Basic labs including, CBC, KFT, LFT, electrolytes and TFT, LDH, CK were all normal. CT chest was done also and it was unremarkable, anti-acetyl choline receptor and anti-musk abs still pending. The patient trial of pyridostigmine after that he showed worsening in ptosis with limitation of gaze directions and worsening proximal muscle weakness. Next day repetitive nerve stimulation test was positive for neuromuscular junction disorder defect with characteristic after discharge that attenuated with repetitive stimulation.

Conclusion: The diagnosis of congenital myasthenia graves should be suspected in setting of fatigable weakness affecting mainly ocular and bulbar muscle with onset at birth to early childhood, nonetheless, some types of CMS present later in life which could be sometimes difficult to diagnose.

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