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We report on the clinical features of a distinct group of DOK 7 negative limb girdle congenital myasthenic syndrome (LG-CMS), the cause of which has only recently been unravelled. Congenital myasthenic syndromes are a rare group of inherited neuromuscular disorders associated with distinct clinical and genetic abnormalities, in which neuromuscular transmission is impaired. An interesting and often difficult group to recognise is the LG-CMS. This typically manifests with shoulder and pelvic girdle muscle weakness with or without additional features including ocular and bulbar involvement. Until recently, DOK 7 gene mutations have been the only recognised genetic cause of this phenotype and were found in half of all LG-CMS patients. Mutations in a novel CMS gene (GFPT1) were recently implicated in an undiagnosed, DOK 7 negative, LG-CMS cohort of 24 patients. All patients had proximal limb weakness with no ocular or bulbar features and showed a positive response to pyridostigmine. Although age of onset of disease was usually in early childhood, a great number of patients presented to medical attention well into adulthood. Furthermore, tubular aggregates arising from the sarcoplasmic reticulum were seen in the majority of patients.

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1106 **THE CLINICAL PHENOTYPIC SPECTRUM OF GFPT1 ASSOCIATED CONGENITAL MYASTHENIC SYNDROME**

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