Congenital myasthenic syndromes (CMS) are a group of hereditary disorders affecting the neuromuscular junction. Here, we present clinical, electrophysiological and genetic findings of 69 patients from 51 unrelated kinships from Turkey. Genetic tests of 60 patients were performed at Mayo Clinic. Median follow-up time was 9.8 years (range 1-22 years). The most common CMS was primary acetylcholine receptor (AChR) deficiency (31/51) and the most common mutations in AChR were c.1219+2T>G (12/51) and c.1327delG (6/51) in CHRNE. Four of our 5 kinships with AChE deficiency carried p.W148X that truncates the collagen domain of COLQ, and was previously reported only in patients from Turkey. These were followed by GFPT1 deficiency (4/51), DOK7 deficiency (3/51), slow channel CMS (3/51), fast channel CMS (3/51), choline acetyltransferase deficiency (1/51) and a CMS associated with desmin deficiency (1/51). Distribution of muscle weakness was sometimes useful in giving a clue to the CMS subtype. Presence of repetitive compound muscle action potentials pointed to AChE deficiency or slow channel CMS. Our experience confirms that one needs to be cautious using pyridostigmine, since it can worsen some types of CMS. Ephedrine/salbutamol were very effective in AChE and DOK7 deficiencies and were useful as adjuncts in other types of CMS. Long follow-up gave us a chance to assess progression of the disease, and to witness 12 mainly uneventful pregnancies in 8 patients. In this study, we describe some new phenotypes and detail the clinical features of the well-known CMS.

**KEYWORDS:** Congenital myasthenic syndromes; Genetic; Myasthenia; Turkey

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