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Clinical Features of Congenital Myasthenic Syndrome in Japan

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Objective: We examined the clinical and neurophysiological features of Japanese patients with congenital myasthenic syndrome (CMS).

Method: Subjects were five patients who were diagnosed with CMS on the basis of clinical course, repetitive nerve stimulation (RNS), and genetic analysis.

Results: Four patients manifested motor retardation within one year of birth, while one manifested motor intolerance at three years of age. The most characteristic symptom observed in all the patients was fluctuating muscle weakness, which varied on a daily basis or continued for several days after the late infancy period. Only one patient manifested daily fluctuation of muscle weakness. RNS of the accessory nerve evoked a decrementing response in three patients who were examined; however, RNS of the median, ulnar, and tibial nerves (one patient each) did not evoke such responses. After the edrophonium chloride test, no improvement was seen even if the patients manifested ptosis. For judgment of this test, improvement in decrementing rate observed while performing RNS was useful. All five patients who were administered medication based on the results of genetic analysis demonstrated an improvement in their symptoms.

Conclusion: We suggest that CMS can be diagnosed based on careful examination and electrophysiological results. CMS is a treatable disorder, and therefore, correct diagnosis is important.

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