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Journal of Clinical Neuromuscular Disease. 20(1):14–27, SEPTEMBER 2018
DOI: 10.1097/CND.0000000000000222, PMID: 30124556
Issn Print: 1522-0443
Publication Date: September 2018

Print

Congenital Myasthenic Syndrome: Spectrum of Mutations in an Indian Cohort

Pavalan Selvam;Gautham Arunachal;Sumita Danda;Aaron Chapla;Ajith Sivadasan;Mathew Alexander;Maya Thomas;Nihal Thomas;

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Abstract

Objectives:

To investigate the mutational spectrum and genotype–phenotype correlation in Indian patients with congenital myasthenic syndrome (CMS), using next-generation sequencing of 5 genes.

Methods:

CHRNE, *COLQ*, *DOK7*, *RAPSN*, and *GFPT1* were sequenced in 25 affected patients.

Results:

We found clinically significant variants in 18 patients, of which variants in *CHRNE* were the most common, and 9 were novel. A common pathogenic COLQ variant was also detected in 4 patients with isolated limb-girdle congenital myasthenia.

Conclusions:

Targeted screening of 5 genes is an effective alternate test for CMS, and an affordable one even in a developing country such as India. In addition, we recommend that patients with isolated limb-

girdle congenital myasthenia be screened initially for the common *COLQ* pathogenic variant. This study throws the first light on the genetic landscape of CMSs in India.

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