Compound heterozygous CHAT gene mutations, a missense and a splice site variant, in two siblings with congenital myasthenic syndrome

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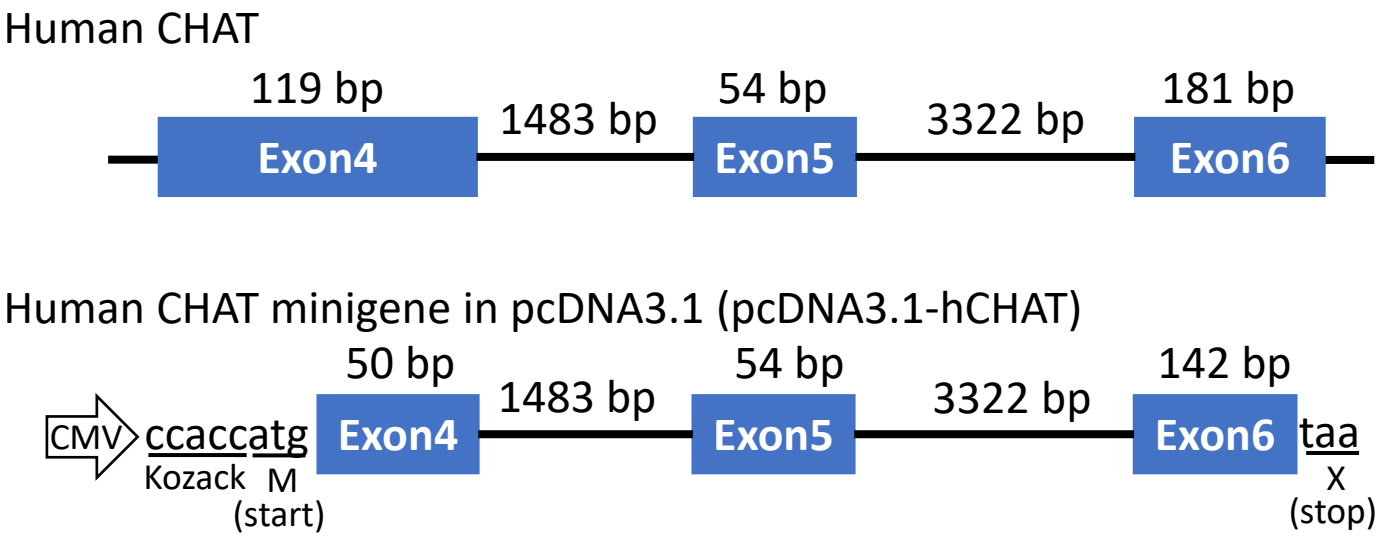

Fig. S1

The upper panel is a part of *CHAT* gene (Human CHAT). The lower panel is mini-gene image built in pcDNA3.1 (pcDNA3.1-hCHAT). CMV, cytomegalovirus promoter; Kozack, Kozack sequence; M, methionine (start codon); X, stop codon.