Novel de novo heterozygous mutation on SYNGAP1 from the Indian population

CURRENT STATUS: POSTED

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Subject Areas

Internal Medicine

Keywords

SYNGAP1, autism spectrum disorder, intellectual disability, epilepsy
Abstract
Background: Exome sequencing is a prominent tool to identify novel and deleterious mutations which could be nonsense, frameshift, and canonical splice-site mutations in a specific gene. De novo mutations in SYNGAP1, which codes for synaptic RAS-GTPase activating the protein, causes Intellectual disability (ID) and Autism Spectrum Disorder (ASD). SYNGAP1 related ASD/ID is one of the rare diseases that is detrimental to the normal neuronal developmental and disrupts the global development of a child.

Results: We report a case of a child of 2-year old with global developmental delay, microcephaly subtle dysmorphism, absence seizures, disrupted sleep, delay in learning a language, and eating problems. Upon further validation, the child has a few traits of ASD. Here, based on focused exome sequencing, we report a de novo heterozygous mutation in SYNGAP1 exon 11 with c. 1861 C>T (p.arg612ter). Currently, the child is on atorvastatin and has shown considerable improvement in global behaviour and cognitive development. The long-term follow up of the child’s development would contribute to the already existing knowledge of the developmental trajectory in individuals with SYNGAP1 heterozygous mutation.

Conclusion: In this report, we discuss the finding of a novel mutation in one of the genes, SYNGAP1, implicated in ASD/ID. In addition, we discuss the current treatment prescribed to the patient and the progress of global developmental of the child.

Full-text
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