CASE REPORT

ALLGROVE SYNDROME WITH A NOVEL MUTATION - CASE REPORT IN TWO SIBLINGS

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Abstract: *Allgrove syndrome (AS/Triple A syndrome) is a rare, familial, multisystem, potentially fatal autosomal recessive disorder characterized by achalasia, alacrimia and ACTH-resistant adrenal failure. There is significant heterogeneity in the clinical features and the types of mutations reported in families with Allgrove syndrome. Two siblings (ten- year-old girl and her six-year-old brother) presented with adrenal insufficiency, hyperpigmentation and alacrimia. Genetic exome sequencing revealed a homozygous variant of uncertain significance in exon 6 of the Triple A syndrome (AAAS) gene in the proband which was further confirmed by Sanger validation.*

The parents were found to be heterozygous, and the sibling homozygous for the tested variant of the Achalasia, Adrenocortical insufficiency, Alacrimia Syndrome AAAS gene. There was good response to replacement therapy with hydrocortisone.

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