Attention deficit hyperactivity disorder and neuronal ceroid lipofuscinosis type 1 - case report

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**Introduction:** Attention deficit and hyperactivity disorder (ADHD) is a neurobiological syndrome, diagnosed in approximately 5% of children and adolescents. It is characterized by distractibility, hyperactivity and impulsivity. The boys affect 3 – 6 times more often than girls. The causes of ADHD may be characterized as idiopathic, symptomatic and secondary to a brain structural abnormality or familial and presumed genetic. A majority of cases of ADHD are idiopathic or of uncertain cause.

**Materials and methods:**
We present an 8-year-old boy with mild developmental delay (IQ=73), speech and motor deterioration, hyperactivity, attention deficit, memory disturbances, facial dysmorphism (deep set eyes, retrognathia), obesity and visual failure appeared between the age of 7 – 8 years. The disorder gets worse over time. The ADHD was the leading reason for targeting the patient in Clinical genetics department.

**Results:** The molecular genetic analysis by NGS revealed homozygous pathogenic variant – c.541G>T (p.Val181Leu), rs14842181 in exon 6 in PPT1 gene. This variant confirms the diagnosis neuronal ceroid lipofuscinosis type 1.

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Discussion: The neuronal ceroid lipofuscinoses (NCLs) are a group of severe primarily autosomal recessive childhood lysosomal disorders. They have a combined incidence of 1:12,000 live births. The distinct forms of NCLs are caused by mutations in different genes. NCL type 1 is caused by homozygous or compound heterozygous pathogenic variants in NCL1 encoding the palmitoyl protein thioesterase 1 enzyme, resulting in deficient PPT1 production. In the classic form of NCL1 disease symptoms begin during infancy, additional phenotypes are with late infantile, juvenile and adult onset. The symptoms of NCL 1 are vision loss, motor dysfunction, cognitive dysfunction, seizures and early death. Our patient has the most of the typical symptoms of NCL 1 (not seizures), with beginning on the preschool age.

Conclusions: Neuronal ceroid lipofuscinosis type 1 is a rare neurodegenerative lysosomal storage disease, caused by an enzyme deficiency of palmitoyl-protein thioesterase 1. Presently no disease-modifying therapies are available. Although screening for metabolic disorders is not recommended in all children with ADHD, selective metabolic testing should be done in the presence of suggestive clinical findings for IEM. In some patients, early diagnosis of the metabolic disorders and proper therapeutic interventions may significantly improve the cognitive and behavioral outcome.

References:

The authors declare that there is no conflict of interest