P-006 - A novel homozygous frameshift SPTBN2 gene mutation associated with Spinocerebellar Ataxia-14

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Spinocerebellar ataxia-14 caused by a homozygous mutation in the SPTBN2 gene, is a neurologic disorder characterized by delayed psychomotor development, severe early-onset gait ataxia, eye movement abnormalities, cerebellar atrophy on brain imaging, and intellectual disability. Unlike many other spinocerebellar ataxia, it is infantile onset. Whole-Exom Sequencing is performed by using an isolated DNA from peripheral blood sample of a female patient who had psychomotor retardation, behavior anomalies, delayed speech and cerebellar atrophy on cranial MR. We detected a homozygous frameshift deletion, c.3427delC, also confirmed via Sanger squencing, on SPTBN2 gene. This variant was also observed heterozygous state in her mother and father who had relationship between them.As far as we know, this frameshift deletion is not found any clinical databases (ClinVar or Human Genome Database HGMD ect.). It could alter gene function by causing an early termination of gene expression. According to the guideline of American College of Medical Genetics (ACMG) (PVS1, PM2, PP3) it is classified as 'pathogenic'. As a conclusion, with this mutation that we found, we added a new one to the variants detected in the SPTBN2 gene. Functional studies are needed to contribute to the determination of the pathogenicity of this variant.

KEYWORDS: Spinocerebellar Ataxia, SPTBN2 gene, c.3427delC, Next Generation Sequencing

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