Two homozygous *KIF1C* mutations in a Turkish family with cerebellar dysfunction and spastic paraparesis

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KIF1C mutation is genetic defect which is observed with hereditary spastic paraparesis and cerebellar dysfuntion. In this report, we analyzed the Turkish family who had spastic paraparesis and cerebellar dysfunction with KIFC mutation. There were paraparesis, ataxia, dysarthria, tremor in brothers whom parents were second degree relatives and asymptomatic. Father and mother's ages were 65 and 62 and brothers were 44, 42, 36. Their complaints appeared with tremor in their hands in childhood. In the following terms, the other complaints began to reveal. There were no point in family history. Patients' neurologic examination: their speech were dysarthric, biletaral dysmetria and dysdiadokokinezi in all. Intention tremor could be seen in all brothers' heads and extremities. The patient's, who were 44 years old, spastic paraparesis is more serious than the other two and he were walking by using one crutch. The other two brothers', who are 42 and 36 years old, spastic paraparesis were less serious and they could walk without using any crutch. General medical tests such as routine biochemistries, hemograms, hormones, B12, VDRL, HIV, vitamin A and E, thyroid hormones, plasma ceruloplasmin and copper were normal. In Cranial-Spinal MR, there were remarkable cortical and cerebellar atrophie. Electrocardiographies, electromyographies and odiographies were normal. They had adequate IQ scores, which were among 80-100 scores. In whole exome sequencing two variant mutation were identified in their *KIF1C* genes. The parents are heterozygote and brothers are homozygote. On the basis of clinical and genetical analyzies, autosomal recessive spastic paraparesis and ataxia were considered due to mutation in *KIF1C*.

Biography

Güllü Tarhan is a 26 year old and working as a Neurology resident in the Gaziosmanpasa Taksim Training and Research Hospital. She has completed Istanbul Faculty of Medicine two years ago.

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