P016. Congenital ataxia, hemiplegic migraine due to a novel mutation of CACNA1A: a case report

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Background  
The CACNA1A gene encodes the pore forming alpha-1A subunit of neuronal voltage-dependent P/Q-type Ca (2+) channels. Mutations in this gene result in clinical heterogeneity, including hemiplegic migraine, episodic ataxia, or progressive chronic conditions.

Case report  
An 8-year-old boy was admitted to our neurological unit due to an acute onset of left hemiparesis developed after a febrile episode. He also complained of headache with migraine characteristics. Brain MRI showed right hemispheric oedema. The hemiparesis disappeared completely after 1 week, and after steroid treatment. The patient was already known to our clinic since he was 2 years old when he was referred to us for a motor and cognitive developmental delay and for a cerebellar syndrome diagnosed as congenital ataxia. In the past all metabolic, biochemical and genetical analyses resulted negative. Serial brain MRI showed a progressive cerebellar atrophy. A CACNA1A gene mutation was hypothesised and sequence analysis revealed a heterozygous mutation c.4013C>T (p.I1338T) affecting the S4 segment and potentially damaging to the protein. This was a de novo mutation because it was not found in either parent.

Conclusions  
To the best of our knowledge this mutation of the CACNA1A gene has not been reported in the literature. Similar cases of a relatively long history of cerebellar ataxia, cognitive impairment and paroxysmal episodes are reported in the literature due to CACNA1A mutations. CACNA1A mutations present with a wide clinical spectrum. Congenital ataxia, mental retardation, and hemiplegic episode can be the presenting signs of CACNA1A mutations.

Written informed consent to publish was obtained from the patient(s).

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