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# Congenital ataxia, hemiplegic migraine due to a novel mutation of CACNA1A

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## **POSTER PRESENTATION**

### **Open Access**

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

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*From* Abstracts from the 1st Joint ANIRCEF-SISC Congress Rome, Italy. 29-31 October 2015

#### 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**

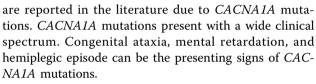
An 8-year-old boy was admitted to our neurological unit due to an acute onset of left hemiparesis developed after a febrile episode. He also complained of headache with migraine characteristics. Brain MRI showed right hemispheric oedema. The hemiparesis disappeared completely after 1 week, and after 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 *CAC*-*NA1A* gene has not been reported in the literature. Similar cases of a relatively long history of cerebellar ataxia, cognitive impairment and paroxysmal episodes

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

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