# Atypical Hemifacial Spasm and Myoclonus Related to AIFM1 Variant

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We report the case of a 7-year-old boy affected by AIFM1 variant[1,2] who progressively developed severe ataxia at 5 years of age. A brain magnetic resonance imaging (MRI) showed vermian atrophy and electromyography revealed four limbs polyneuropathy. Genetic analysis showed a maternally inherited variant in AIFM1 gene (c.470C > T[p.Ala157Val]).

At 7 years of age, he presented with atypical hemifacial spasm developed over a few days. In addition to cerebellar ataxia and dysarthria, clinical examination identified severe and disabling right hemifacial spasm (►**Video 1**; available in the online version), characterized by irregular and unusually fast clonic movements of the facial muscles,[3] combined with mild right facial paresis, and with left laterocolis and right torticolis, indicating an implication of the 11th cranial nerve. Electroencephalogram (EEG) was normal and brain MRI remained unchanged.

**Video 1**

Part 1: cranial nerve examination showing severe, irregular and fast clonic movements of the right part of the face, right torticolis and left laterocolis. Speech (in French) was also impaired, suggesting larynx involvement. Part 2: upper limbs examination revealing fast myoclonus of the right upper limb in various situations (posture, rest, etc.). Part 3: gait examination, showing cerebellar ataxia. Online content including video sequences viewable at: https://www.thieme-connect.com/products/ejournals/html/10.1055/s-0042-1744159.

Over the following months, myoclonus appeared in his right upper limb, whereas hemifacial spasm spontaneously disappeared, as did myoclonus later.

Myoclonus has only been described once in a patient with AFM1 variant, affecting the upper limb, then spreading to the other limbs, as well as the nostrils and larynx.[2] The authors suggested brainstem originated myoclonus in that case. Hemifacial spasm, considered as a peripheral myoclonus, is in the same clinical spectrum, and we suspect a similar origin in our patient.

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**Conflict of Interest**
None declared.

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