



DON'T FALL FOR IT: A MISDIAGNOSED CASE OF PAROXYSMAL LEG STIFFNESS





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CLINICAL HISTORY

Female, 29yo.

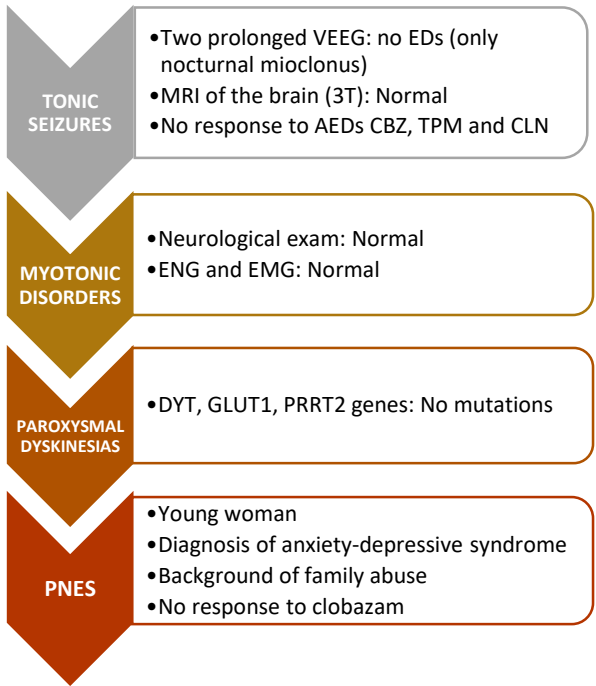
 **Perinatal** distress, at birth “**hypertonic syndrome**” with **seizures**, treated with phenobarbital for 3y. At the beginning of deambulation, around 1yo, **paroxysmal** episodes of **stiffness in the lower limbs** with contextual **falls**, without loss of consciousness.

 **Prodromal features:** the pt reports feeling stiffness in the AAll, which tends to intensify over a few days, with cramp-like pain, reaching its peak with the fall.

 **Episode features:** During the episodes she emits a ‘choked’ sound, falls forward ‘like a statue’ without protective reflexes, with occasional sphincter release.

 **Frequency and triggers:** multimonthly, associated with traumas. Triggered by external auditory or tactile stimuli, or by intense negatives emotions. Spontaneous episode-free period from 14 to 18 years of age.

DIFFERENTIAL DIAGNOSIS



FINAL DIAGNOSIS

We found a heterozygous mutation in the **GLRA1 gene**, pathogenic for **hyperekplexia 1 (Startle Syndrome - SS)**.

She was shifted from clonazepam to higher doses of clobazam, with benefit.

CONCLUSION:

Startle Syndrome (SS) is characterized by sudden jerks with paroxysmal stiffness that can results in falls, usually promoted by external stimuli¹. We describe this case to demonstrate that even cases with atypical presentations, negative instrumental examinations and a positive history of psychiatric disorders are sometimes worthy of further investigations. Although the treatment can sometimes be the same, the impact of the wrong diagnosis can deeply affect the patient.

¹ Saini AG, Pandey S. Hyperekplexia and other startle syndromes. J Neurol Sci. 2020 Sep 15;416:117051. doi: 10.1016/j.jns.2020.117051. Epub 2020 Jul 20. PMID: 32721683.



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