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SCIENTIFIC LETTER

Benign Infantile Epilepsy Mimicking Reflex Anoxic Seizures in an Infant with *PRRT2* Gene Mutation

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To the Editor: A 12-mo-old Caucasian girl presented with recurrent seizures since the age of nine months. She was the first child of non-consanguineous parents. She would become vacant with facial pallor, occasional lips cyanosis and isolated upper limb jerky movement. A tonic stiffening of upper limbs, myoclonic movements of fingers and facial musculature contraction would follow after five minutes. She would then fall asleep for two hours. The triggers were difficult to identify, but speculated to include frustration, fright or a feeling of abandonment.

Neurological examination was normal. Routine, sleep-deprived and 24-h electroencephalography (EEG) telemetry study were all reported as normal. Brain MRI did not reveal any pathological findings and cardiac investigations were also normal. Given the unusual presentation, a genetic cause was suspected. Targeted sequence analysis of the *PRRT2* gene, the leading cause for paroxysmal disorders, detected the exon 2 c.649dupC p.(Arg217Profs*8) *PRRT2* variant in the heterozygous state. The father was found to share the same mutation.

PRRT2 encodes for the proline-rich transmembrane protein 2 in the developing nervous system and interacts with synaptosomal associated protein 25 (SNAP-25), involved in the interaction of Ca2 + −triggered release of neurotransmitters at the presynaptic terminals [1]. To date, some 1500 patients with approximately seventy different *PRRT2* mutations have been reported. Of these, 5.5% (79/1444) occurred de novo, while 87.1% (1258/1444) are familial in origin [2]. *PRRT2* mutations have been mainly associated with Benign Familial Infantile Epilepsy, Paroxysmal Kinesigenic Dyskinesia and Infantile Convulsions and Choreoathetosis [2] and lately with benign myoclonus of early infancy [3] and paroxysmal hypnogenic dyskinesia (PHD) [4].

At 25 mo of age, the child has been seizure free with a normal psychomotor development and no anticonvulsant medications. Though her presentation was highly evocative of reflex anoxic seizures, her clinical phenotype and the *PRRT2* loss of function mutation corroborated the diagnosis of a benign form of epilepsy, according to ILAE classification of 2010 [5]. Currently, *PRRT2* physiological function remains unclear, even though a marked pleiotropy and variable penetrance of its mutations are well-described. Our case herein concurs in expanding the clinical spectrum of *PRRT2* related disorders.

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Compliance with Ethical Standards

Conflict of Interest None.

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