Clinical commentary

Epileptic Disord 2017; 19 (3): 339-44

Pharmacoresistant epileptic eyelid twitching in a child with a mutation in *SYNGAP1*

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Received February 28, 2017; Accepted April 20, 2017

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- *SYNGAP1* gene mutation has been reported to result in a phenotype of intellectual disability, associated with generalized epilepsy, which is often pharmacoresistant (Berryer et al., 2013; Mignot et al., 2016) .

- Mignot *et al*. described three patients with eyelid myoclonia among 16 with *SYNGAP1* gene mutations (Mignot et al., 2016).

- The characteristics of eye phenomena may provide useful diagnostic information for patients with *SYNGAP1* mutations.

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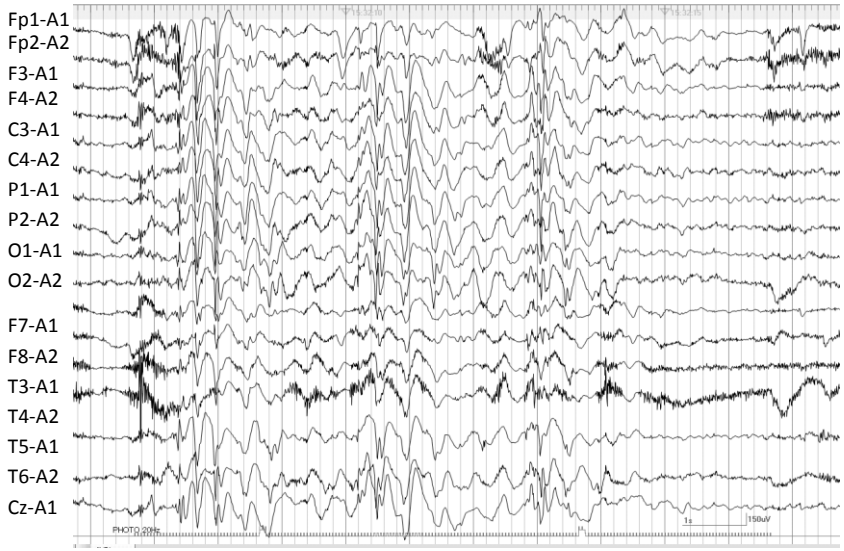

Ictal EEG of the present patient during photic stimulation at a frequency of 20 Hz revealed diffuse spike-and-wave activity, simultaneous with motion arrest, unresponsiveness, upward eye deviation, and intermittent eyelid twitching, lasting for several seconds.

Note the posterior predominance of spike-wave bursts, which is in contrast to the frontal predominance of ictal spike-wave bursts in childhood absence epilepsy (CAE) and eyelid myoclonia with absences (EMA).

References

Mignot C, von Stülpnagel C, Nava C, et al. Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. J Med Genet 2016; 53: 511–22.

Berryer MH, Hamdan FF, Klitten LL, et al. Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. Hum Mutat 2013; 34: 385–94.

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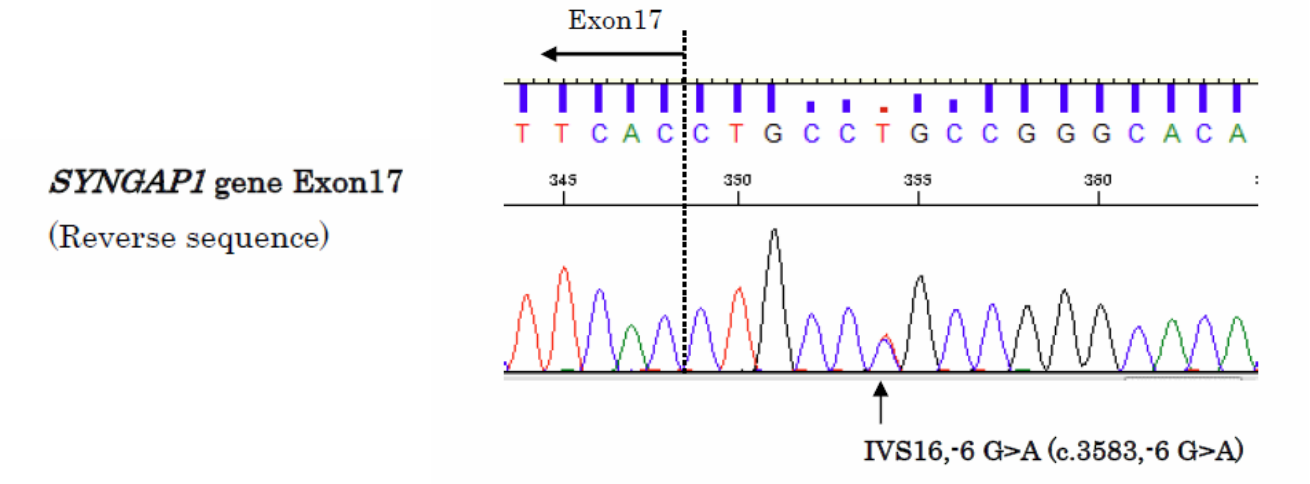
・Epilepsy appears more pharmacosensitive in patients with mutations in exons 4-5 than in patients with mutations in exons 8–15, possibly related to a residual action of an isoform lacking exons 4-5 (Mignot *et al.*, 2016).

・The c.3583-6 G>A mutation in our patient was located in intron 16,
in agreement with this hypothesis, which may further expand the pharmacoresistant locus at the 3′ end.

・Seizures were refractory to carbamazepine and levetiracetam, but were reduced in frequency by ethosuximide and lamotrigine administration in the present patient.



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References
Mignot C, von Stülpnagel C, Nava C, et al. Genetic and neurodevelopmental spectrum of SYNGAP1-associated intellectual disability and epilepsy. J Med Genet 2016; 53: 511–22.