■ Case vignette

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Intractable startle epilepsy in Schuurs–Hoeijmakers syndrome

Kento Ohta[1], Tohru Okanishi[1,2], Sotaro Kanai[1], Tetsuya Okazaki[3], Ayataka Fujimoto[2], Yoshihiro Maegaki[1]

[1] Division of Child Neurology, Department of Brain and Neurosciences, Faculty of Medicine, Tottori University, Yonago, Japan
[2] Comprehensive Epilepsy Center, Seirei Hamamatsu General Hospital, Hamamatsu, Shizuoka, Japan
[3] Division of Clinical Genetics, Tottori University Hospital, Yonago, Japan

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Case

- A 31-year-old male presented with severe global developmental delay. His morphological features included a down-slanted palpebral fissure, a thin upper lip, and large hands.
- He had developed epilepsy with generalized tonic seizures at the age of 4 months, and the seizures resolved without antiepileptic drugs at the age of 3 years.
- At around 10 years of age, he developed atonic seizures, which involved knee buckling in response to sudden unexpected noise or touch.
- Seizures gradually became intractable to antiepileptic drugs.
- A heterozygous *de novo* c.607C>T (p.Arg203Trp) mutation in *PACS1* was found, and he was diagnosed with Schuurs–Hoeijmakers syndrome.

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Discussion

- Schuurs–Hoeijmakers syndrome is a rare disorder characterized by dysmorphic facial features, intellectual disability and various physical malformations.

Schuurs-Hoeijmakers JH, et al. Am J Hum Genet. 2012; 91: 1122-7.

- More than half of the cases are associated with epilepsy.

Schuurs-Hoeijmakers JH, et al. Am J Med Genet A. 2016; 170: 670-5.

- The aetiology of startle epilepsy is heterogeneous. Startle epilepsy has been reported in patients with perinatal and postnatal factors.

Yang Z, et al. Clin Neurophysiol. 2010; 121: 658-64. Tibussek D, et al. Epilepsia. 2006; 47: 1050-8. Aguglia U, et al. Epilepsia. 1984; 25: 712-20.

- Based on intracranial electroencephalography, magnetoencephalography and functional neuroimaging studies, the origin of startle epilepsy is considered to involve supplementary motor area and primary motor and sensory cortex.

Saeki K, et al. Epilepsia. 2009; 50: 1274-9. Fernández S, et al. Epilepsia. 2011; 52: 1725-32. García-Morales I, et al. Epilepsy Behav. 2009; 16: 166-71.

Discussion

- The startle epilepsy in our patient was resistant to antiepileptic drugs. No previous cases of startle epilepsy have been reported in this syndrome, and the refractory nature was considered a unique characteristic of our case.
- Our patient presented with intractable epilepsy, severe intellectual disability and diffuse brain abnormalities. Cerebral atrophy, which involves the auditory and primary sensory areas and is a relatively profound abnormality in this syndrome, might be associated with auditory and tactile-induced startle seizures in our patient.

Conclusion

- We demonstrate a case of Schuurs–Hoeijmakers syndrome with refractory startle epilepsy.
- This report adds a new seizure type to this syndrome.
- In addition, epilepsy in this syndrome may be intractable in some patients.

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