**IVD5**

A case of chromosomal 15q proximal tetrasomy associated with infantile spasms and partial seizures

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We report a 6 month old girl of 15q proximal tetrasomy with infantile spasms (IS) and cerebral cortex atrophy. She was born from healthy parents, after 39 weeks of gestation, with a birth weight of 2358g. The perinatal period was uneventful. Milestones were delayed, with head control at 5 months and rolling at 6 months. When she was 6 months old, her parents brought her to our clinic because of several series of tonic spasms. On physical examination, she showed no neurologically abnormal signs, and was without general muscle hypotonia. EEG examination showed hypsarrhythmia. The brain MRI and CT showed diffuse cortical atrophy. No abnormal findings were observed in CBC, blood examination, cerebrospinal fluid, and serological test for congenital viral infection of the central nervous system. High resolution analysis of 15th chromosome revealed 15q proximal tetrasomy (47,XX,+idic(15)(q13).

Before the treatment, DQ score was 62. Tonic spasms have disappeared using Zonisamide mono therapy without ACTH treatment. However, soon after the cessation of the spasms, she showed partial seizures, which were controlled by Carbamazepine. Since then, no seizure recurrence has been observed during a 2 yr follow-up. The present total DQ score is 52. Three cases of 15q proximal tetrasomy with infantile spasms have previously been reported. Inverted duplication of chromosome 15 in the above three cases was at 15pter-q13.

**IVD6**

How SCN1A mutations contribute to Dravet syndrome - a comparison of clinical and EEG features between Dravet syndrome with and without SCN1A mutations

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[目的] We studied the clinical and EEG differences between Dravet syndrome with or without mutations of the gene encoding the alpha1 subunit of the Na channel, SCN1A to confirm how the SCN1A mutations are responsible for this epilepsy syndrome.

[被験者] Subjects were 28 patients with Dravet syndrome including 9 patients with SCN1A mutations and 19 patients without SCN1A mutations. The details of the method of analyzing SCN1A mutations were described elsewhere. The onset of epilepsy, seizure types (with or without myoclonic and absence seizures), family history of convulsive disorders within third degree relatives, photo-pattern sensitivity and prognosis were compared between the patients with and without SCN1A mutations.

[結果] There were no clinical or EEG differences between 9 patients with SCN1A and 19 without SCN1A mutations (p>0.05). Furthermore, there were no significant differences in the ratio of SCN1A mutations between 12 patients with and 16 patients without myoclonic and absence seizures (42%: 25%) (p>0.05).

[結論] The absence of clinical and EEG differences between Dravet syndrome with and without SCN1A mutations suggests genetic heterogeneity. Either unknown genes or allelic mutations in SCN1A would play a role in producing myoclonic and absence seizures generated by diffuse spike-wave complexes as well as EEG photo-pattern sensitivity in this syndrome.