소아재활

게시일시 및 장소 : 10 월 19 일(토) 08:30-12:30 Room G(3F)

질의응답 일시 및 장소 : 10 월 19 일(토) 11:00-11:30 Room G(3F)

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STXBP1 mutation associated ataxia-tremor-retardation syndrome : Case report

Su jin Choi[1*], Jong Weon Lee[1], Ha Ra Jeon[1], Seong Woo Kim[1], Minji Chae[1†]

National Health Insurance Service Ilsan Hospital, Department of Physical Medicine and Rehabilitation[1]

Introduction

De novo mutations in STXBP1 have been found in a group of patients with early infantile epileptic encephalopathy (EIEE), the so-called Ohtahara syndrome. The Ohtahara syndrome is characterized by a severe early infantile epileptic encephalopathy. All patients suffer from severe developmental delay and severe neurologic pathology, such as muscular hypotonia, spasticity, or motor asymmetries. The prognosis is poor with a high mortality rate during infancy. Also, a variety of associated movement disorders like ataxia, tremor, and head tremor are described in a few patients with STXBP1 mutations. Here, we report on one female patient with ataxia-tremor movement pattern and intellectual disability caused by a de novo STXBP1 mutation.

Case

A 2year 6 months year old girl was delivered via normal spontaneous vaginal delivery at IUP 41 weeks and birth weight of 2.9kg. She was born without any medical history or family history. At 3 days after birth. However, she demonstrated seizure and underwent neurosonography, which showed mild subdural hemorrhage. Though electroencephalogram(EEG) did not detect any irregular activities, she was prescribed phenobarbital for 1 month. At 2 months after birth, she again showed seizure activity and was admitted to our hospital for further evaluation. She was started on antiepileptic drug (AED) regimen. Although brain MRI did not show a delay in maturation or any abnormality in shape, size, and signal of the cerebellum or the cerebrum, gene study revealed mutation in STXBP1 gene. After 7 months of AED therapy, EEG returned to normal and we discontinued AED. All growth parameters were within normal range. However, this patient displayed dysmorphic features, such as almond-shaped eyes and low nasal bridge. Furthermore, she showed global development delay in gross motor function, fine motor function, language, and cognitive function. For example, she was not able to stand with holding until 18 months. When she was able to ambulate independently at a delayed 24 months, her gait pattern was ataxic. She tumbled quite often due to imbalance, but ataxic gait pattern did not exacerbate. Trunk tone was hypotonic and ataxic movements made it quite difficult to change position from supine to sit and from sit to stand. This patient was not able to perform mature pincer grasp until 24 months. Even immature grasps were

difficult to execute due to ataxia and tremor. This patient displayed no oculomotor abnormalities like nystagmus, slowing of saccades. Reflexes were normal. There was also no clinical indication of neuropathy or muscle weakness.

Conclusion

We have diagnosed a patient with an ataxia-tremor-retardation syndrome without epilepsy caused by a de novo mutation in the STXBP1 gene. The vast majority of patients carrying a STXBP1-mutation have an early onset epilepsy. But we can herewith document that the phenotypical spectrum of STXBP1 mutations is much broader than described before.