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CASE REPORT
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Septo-optic dysplasia plus diagnosed in a middle-aged woman

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Septo-optic dysplasia is a congenital anomaly with diverse phenotypes from normal to mixtures of visual abnormality, endocrine dysfunction, psychomotor retardations and epileptic seizures. It is characterized by optic atrophy, pituitary dysfunction and midline structure abnormalities in corpus callosum or septum pellucidum. Diagnosis of septo-optic dysplasia plus is made when cortical malformations accompanied. Here we report a middle-aged woman with septo-optic dysplasia plus having unilateral optic atrophy, agenesis of septum pellucidum and cortical malformations.

Key words: Septo-optic dysplasia; Optic atrophy; Congenital abnormalities

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Septo-optic dysplasia (SOD) is a congenital anomaly and distinctive of optic nerve dysplasia, pituitary dysfunction, and brain anomalies in midline structures such as corpus callosum and septum pellucidum.[1] In most cases, SOD is diagnosed during the investigation of strabismus, seizures, developmental delay and cerebral palsy in infancy.[2] The diagnosis of SOD-plus can be made when SOD accompanied by cortical malformations.[3,4] Patients with SOD-plus are more likely having focal neurologic deficits and seizures than SOD.[4,5] The authors experienced a middle-aged SOD-plus patient complaining headache, dizziness and having complex partial seizures with unilateral optic atrophy, agenesis of septum pellucidum and cortical malformations such as schizencephaly, lissencephaly, and gray matter heterotopia.

CASE

A 48-year-old woman came to the hospital with a headache and dizziness from a month ago. The pain was squeezing, moderate to severe intensity (Numerical Rating Scale, 6), and localized in the left temporomandibular region. It came ten or more times per day, and it lasted for 10-20 minutes. Dizziness, which was non-rotatory and non-positional was asso-

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ciated with a headache occasionally, but nausea, vomiting, photophobia or phonophobia was not. Physical exertion did not worsen a headache.

In medical history, she had seizures in her childhood but she denied of family history in her parents, little sister, and two sons. She also denied history of developmental delay. Her highest level of education was high school. During interview she answered to physician's questions fluently with appropriate manner. She had taken antiepileptic drugs irregularly and stopped medication at nineteen years old by herself. The patient did not know whether she had taken brain images in childhood. Although she had seizures occasionally in her twenties, she has not received treatment. The patient reported that there was no clinical seizure in recent years, but a son of the patient said that she had occasionally become unresponsive for a while. She stated that she had a low visual acuity of the right eye since early childhood but did not complain another visual symptom. Neurological examination revealed suspicious temporal hemianopsia and relative afferent pupillary defect in the right eye. We referred her to ophthalmology department for further evaluation. The visual acuity without correction was 0.3 in the right eye and 0.4 in the left eye. Optic disc was atrophic in the right eye (Fig. 1A). Optical coherence tomography revealed a decrease in the thickness of the regional nerve fiber layer in the right eye, which suggested right optic nerve dysplasia. There was no strabismus or nystagmus. Humphrey visual field test was unreliable considering her high fixation losses during test but it suggested right temporal hemianopsia. Visual evoked potential (VEP) test showed bilateral P100 latency prolongation in pattern VEPs suggested bilateral visual pathway dysfunction. Brain magnetic resonance imaging

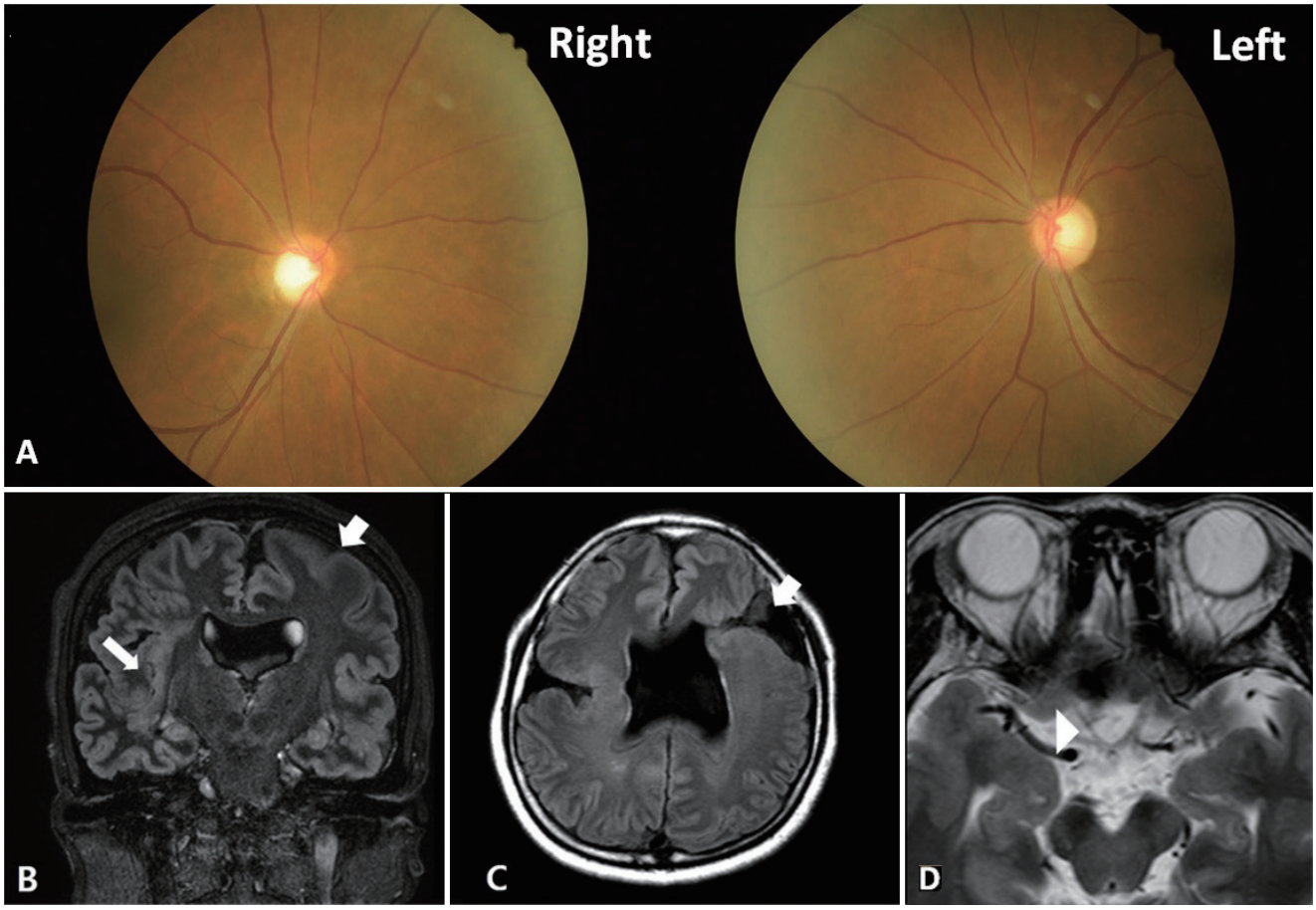

Fig. 1. Fundus photo (A) and brain MRI (B-D). (A) Relative disc pallor in the right eye compared to the left. (B) Lissencephaly (thick arrow), gray matter heterotopia (thin arrow) and absent of septum pellucidum on coronal short tau inversion recovery fluid-attenuated inversion recovery MRI. (C) Schizencephaly (thick arrow) on axial fluid-attenuated recovery MRI. (D) Hypoplasia of right optic nerve (arrow head) on T2 axial MRI. MRI, magnetic resonance imaging.

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showed agenesis of septum pellucidum, schizencephaly, lissencephaly, and gray matter heterotopias (Fig. 1B, C). Right optic atrophy was also detected (Fig. 1D). Electroencephalography showed intermittent slowings in the right anterior temporal region.

We concluded the patient had SOD-plus and added controlled-release carbamazepine to control the seizure. We recommended hormone testing, but she and her family refused further evaluation and just wanted symptom control. A several months later, she notified us the results of some laboratory workup done in another hospital including complete blood count, serum electrolyte, glucose, liver function, renal function and hormonal function including adrenocorticotrophic hormone and thyroid stimulating hormone, which were within normal range. After antiepileptic therapy (controlled-release carbamazepine 200 mg twice a day), our patient had no seizure attack, but her symptoms of a headache or dizziness have been unchanged.

DISCUSSION

SOD is a rare congenital central nervous system malformation, and the incidence of SOD is estimated at 1 in 10,000 live births.[2] Although SOD may show a variety of neurological development from normal to autism, and epileptic seizures,[5] most patients with SOD are diagnosed in infancy and childhood. Developmental disruption of the forebrain in the embryonic period of 4-6 weeks of gestation is considered to cause SOD.[2] SOD is sporadic, but rarely familial SOD has been reported.[1] Especially, SOD-plus patients accompanied by cortical malformations are more likely to have neurological manifestations such as psychomotor developmental delay, motor deficits or seizure.[4,5] Therefore, a few have reported incidental diagnoses made as SOD-plus after brain image in adulthood showing subtle neurological symptoms.

Our patient had decreased visual acuity and seizures since childhood but had no evident sign of psychomotor delay, motor deficit, and hormonal abnormalities. We can diagnose the patient as SOD-plus only after the brain MRI revealed the characteristics brain abnormalities such as agenesis of septum pellucidum and cortical malformations. Our patient visited the hospital because of a headache and associated dizziness, but in the literature, there is a lack of reporting main symptoms like a headache and dizziness in SOD or SOD-plus.

It is more likely that patients have no developmental delay if there is unilateral optic nerve hypoplasia or absent pituitary dysfunction.[6] Moreover, the literature suggests patient with SOD-plus show more heterogeneous neurodevelopmental outcomes than SOD.[5] Our patient had multiple cortical malformations including schizencephaly, lissencephaly, and gray matter heterotopy but she had no developmental problems and evident endocrinological abnormality. Although drug-resistant epilepsy may be related to cortical malformations of SOD-plus,[7] seizures of our patient had been wax and waned even without antiepileptic medication and have responded excellently to a low dose of controlled-release carbamazepine. The patient with SOD-plus is more likely to have various epileptic prognosis but less likely to have endocrinological abnormality than SOD.[5,7] SOD-plus is a rare and highly heterogeneous disorder that can be diagnosed in an adult.

There are some points to address in this case. We examined her hormonal function incompletely and failed to evaluate her cognitive function with a proper neuropsychological test. We overlooked the necessity of cognitive evaluation because she showed fluency in the expression of her symptoms and did not complain about cognition and psychomotor function. Therefore it is possible that we have missed a subtle hormonal and neuropsychological dysfunction.

In conclusion, the present case report demonstrates that a patient with SOD-plus may have subtle manifestations when a patient has unilateral optic nerve hypoplasia and no evident endocrine dysfunction. As a diagnosis of SOD is mainly dependent on clinician's suspicions, various combinations of symptoms such as poor visual acuity, history of developmental delay, endocrine disorders, psychomotor retardations, and epileptic seizures should be assessed radiologically.

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