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Case Report

Dandy-Walker Malformation Associated with Neurocutaneous Melanosis

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Neurocutaneous melanosis associated with Dandy-Walker malformation is a rare dysmorphogenesis that is associated with single or multiple giant pigmented cutaneous nevi and diffuse involvement of the central nervous system. In this article, we present a 2-month-old patient with neurocutaneous melanosis associated with Dandy-Walker malformation. In addition, we reviewed the literature and discussed the pathogenesis based on the preferred hypotheses.

Key Words : Dandy-Walker malformation · Neurocutaneous melanosis · Hydrocephalus.

INTRODUCTION

The association of congenital abnormalities of the skin and the central nervous system is well recognized. Neurocutaneous melanosis is a rare dysmorphogenesis that is associated with single and multiple giant pigmented cutaneous nevi and the involvement of benign and/or malignant melanocytic tumors of the leptomeninges. After the first description by Rokitansky[17], around 100 cases of neurocutaneous melanosis have been reported in the literature[1-3,5,10,11,13-16,18,19]. Eight to 10% of these were associated with Dandy-Walker malformation, indicating a common origin of these developmental abnormalities [1-3,7,10,11,13,14,18,19].

We report the clinical features, neuroimaging findings, and therapeutic management of a rare case of neurocutaneous melanosis associated with Dandy-Walker malformation.

CASE REPORT

A two-month-old female patient was first admitted to our hospital with complaints of vomiting and a gradually enlarging head circumference. She did not have an eventful history or pregnancy except for multiple melanocytic nevi over her whole body. She had attained normal neurodevelopmental milestones until 2 months of age.

In her initial physical examination, she was lethargic and had a large head, and multiple pigmented, hairy nevi were observed over her whole body (Fig. 1). Computerized tomography (CT) showed vermian hypoplasia and bi-compartmental hydrocephalus. The fourth ventricle was enlarged and seemed to be continuous with the cisterna magna, suggesting a Dandy-Walker malformation (Fig. 2A). Magnetic resonance imaging (MRI) was also compatible with Dandy-Walker malformation and high-pressure hydrocephalus. The images demonstrated a large posterior fossa and communication with the enlarged fourth ventricle through the remaining cerebrospinal fluid (CSF) pathway, and aplasia of the cerebellar vermis, hypoplasia of the cerebellar hemisphere, and a high position of the tentorium (Fig. 2B). The patient developed generalized seizures, and a repeated radiological study suggested a possible tonsillar herniation. She underwent a ventriculoperitoneal shunting procedure (program-

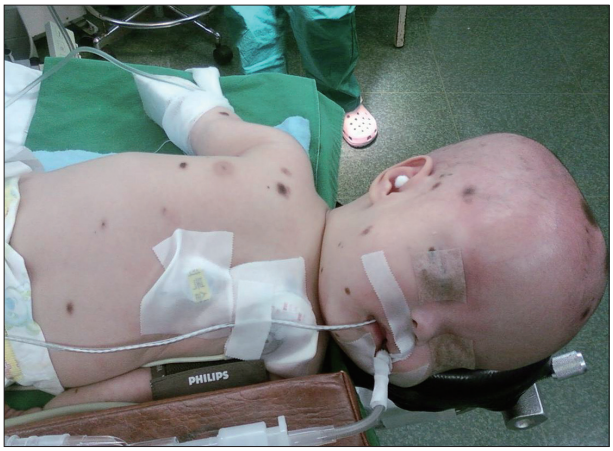

Fig. 1. Photograph demonstrating multiple small and medium sized plaque of melanosis in face and chest.

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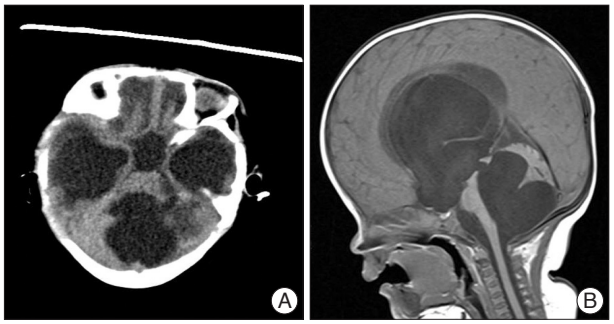

Fig. 2. A : Computed tomography axial image. B : T1-weighted sagittal magnetic resonance image. It demonstrates the hypoplasia of cerebellum and enlarged fourth ventricle, and hydrocephalus, findings of characteristics of Dandy-Walker malformation.

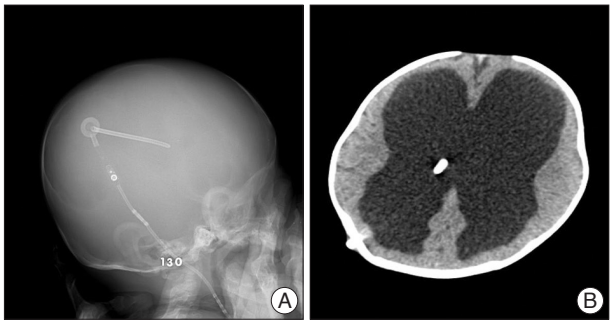

Fig. 3. Posterative operation image (ventriculoperitoneal shunt). A : Plain skull image. B : Computed tomography image.

mable valve) (Fig. 3) in order to prevent subsequent hydrocephalus. Examination of the CSF cytology was unremarkable. She did well after surgery and was discharged after 3 weeks. Three months after the operation, routine follow-up CT scans demonstrated reduction of the hydrocephalus. She had no more problems during the following periods, except for neurodevelopmental retardation.

DISCUSSION

Neurocutaneous melanosis is a rare, congenital nonheritable neuroectodermal dysplasia, which was first described by Rokitansky[17]. A few more than 100 cases have been published so far [1-5,10,11,13-16,18,19]. This disease is characterized by one or multiple large congenital melanocytic nevi and diffuse or nodular leptomeningeal involvement of melanocytes. First, Fox[6] in 1972 and, later, Kadonaga and Frieden[9] in 1991 proposed criteria for the diagnosis of this lesion.

The criteria for this lesion were as follows; 1) large nevus (greater than 20 cm in adults and lesions that approximately 9 cm of diameter on the head or 6 cm on the body in infants), 2) multiple nevi (greater than or equal to 3 lesions), 3) no evidence of cutaneous melanoma, except in cases where meningeal lesions are histological benign, 4) no evidence of meningeal melanoma except in cases where the cutaneous lesion are benign. According to above criteria, our case was compatible to diagnosis for neurocutaneous melanosis (large nevus greater than 6 cm in body, multiple nevi, no evidence of cutaneous melanoma and meningeal melanoma).

Although the pathogenesis of neurocutaneous melanosis has not been elucidated, theories focus on defective morphogenesis of the neural ectoderm in the developing embryo[1-5,8,10-15]. Melanocytes of the skin and pia mater are believed to be derived from multipotential precursor cells of the neural crest. The neural crest is responsible for the formation of skin melanocytes and basal leptomeninges. In addition, Dandy-Walker malformation has been shown to have an association with neurocutaneous melanosis. Dandy-Walker consists of a spectrum of midline anomalies of the hindbrain. The main features of Dandy-Walker malformation are cystic enlargement of the fourth ventricle, cerebellar dysgenesis, and an enlarged posterior fossa resulting from maldevelopment of the rostral embryonic roof of the rhombencephalon, with or without hydrocephalus. Although the cause of Dandy-Walker malformation is not known, it has been theorized that it is caused by an induction failure of the opposing cerebellar plates, with persistence of the membranous area of the fourth ventricle at or before the seventh week of embryonic development. A secondary component may involve atresia of the foramina of Magendie and Luschka, which produces non-communicating hydrocephalus and cyst dilation. The physiologically increased intravascular pressure after birth may explain the development of hydrocephalus after birth instead of during intrauterine life. Whether the ventricular outlet is obstructed or not, the dysgenesis of the cerebellum and fourth ventricle may result in the gradual accumulation of CSF in the posterior fossa, but still allow absorption in a critical equilibrium because of the relatively high-pressure gradient between the CSF pathway and the venous compartment. The physiological increase in systemic blood pressure after birth results in a decreased pressure gradient and a disruption of the absorption of CSF, leading to hydrocephalus[1-5,7,10-14,18,19].

At least 100 cases of neurocutaneous melanosis have been reported; in 15 cases, there were associations between Dandy-Walker malformation and neurocutaneous melanosis[1-4,10-16,18,19]. This fact indicates that these developmental abnormalities may share a common origin. The common neurological manifestations of these two abnormalities are irritability, seizures, lethargy, nausea and vomiting, neck stiffness, bulged fontanels, papilledema, and cranial nerve palsies, which are the signs and symptoms of increased intracranial pressure. Once the neurological symptoms manifest, the course is usually progressive and fatal due to primary CNS melanosis[1,3,5,10,12,14-16]. The association of Dandy-Walker malformation with neurocutaneous melanosis seems to have an extremely poor prognosis. In all reported cases, the patients showed rapid neurological deterioration and death by four years of age. The presence of these two abnormalities represents a phenotypic marker for more profound melanotic infiltration of the leptomeninges, which increases the risk of malignant transformation[4,12].

The characteristic MRI findings are severe cerebellar vermian

and hemispheric dysgenesis, compression and rotation of the cerebellum and brain stem, retrocerebellar cysts, and extra-axial anomalies of the posterior fossa. In addition, the T1 shortening of the involved structures (hyperintensity on T1-weighted images) is due to the presence of pathologic involvement of the brain and leptomeninges by melanin pigment deposition[2,4,5,11,14-16]. Calcium, blood, and fat are the other causes of T1 shortening on MRI, but these can be eliminated by CT[2]. Elevated serum levels of CA-125 and homovanillic acid in a metastatic melanoma case have also been recorded in the literature[3].

The characteristics of neuroimaging studies such as CT or MRI for our case of neurocutaneous melanosis with Dandy-Walker malformation are not different from previous cases[14]. The interesting feature of our case is that no leptomeningeal infiltration of the melanosis or melanoma was seen, but they were found in the previous six cases in the literatures[14]. Large congenital melanocytic nevi in trunk were found only 4 cases (25%), these lesions are also found in our case. Other characteristics such as hydrocephalus, development delay, and seizure were not different from previous cases.

CONCLUSION

The findings in our case support the possibility that neurocutaneous melanosis not only interferes with the normal inductive effects of the primitive meningeal tissue, but also mesenchymal development, which may cause concurrent Dandy-Walker malformation. This case provides considerable information suggesting one pathogenesis of both Dandy-Walker malformation and neurocutaneous melanosis.

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