

A Case of a Newborn with Agenesis of the Corpus Callosum Complicated with Ocular Albinism

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Keywords

Ocular albinism · Agenesis of the corpus callosum · Hypopigmentation · Vici syndrome

Abstract

Purpose: To report a case of ocular albinism found in a newborn infant in whom agenesis of the corpus callosum (ACC) was indicated in utero. **Case Report:** This study involved a female newborn who was delivered after a gestational period of 41 weeks. The patient was referred to the Obstetrics Department at Takatsuki Hospital, Takatsuki City, Japan, after the indication of ACC by magnetic resonance imaging (MRI) at a nearby clinic during the fetal period. At birth, the baby's weight was 2,590 g, and ACC and ventricular enlargement were found by cranial sonography and cranial MRI. While initial ophthalmic findings noted partial loss of pigmentation of the iris and hypopigmentation of broad areas of the fundus in both eyes, nystagmus was not observed. The patient's hair pigment was slightly diluted, and the color of her skin was slightly off-white. At 2 years after birth, obvious mental retardation was observed. With regard to other systemic findings, no apparent heart, kidney, or immune system abnormalities were found. **Conclusion:** Although the patient in question is presently growing without any major systemic problems, it will be necessary in the future to pay attention to any changes in systemic and ophthalmic findings.

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Introduction

Albinism is a congenital disorder of hypopigmentation that, due to an abnormality in the melanin pigment formation process, results in a decrease or absence of melanin in the eyes, hair, and skin. The reported incidence of albinism is estimated to be 1 in every 5,000–10,000 individuals worldwide. From the site of onset, the condition is broadly classified into generalized albinism or oculocutaneous albinism and ocular albinism [1]. Generalized symptoms include whitening of the skin over the entire body, hair, eyebrows, and eyelashes, while ocular findings include a decrease or absence of pigment in the iris, retinal pigment epithelium, or choroidea, as well as other symptoms such as photophobia, nystagmus, and macular hypoplasia [2]. While some cases of albinism combined with agenesis of the corpus callosum (ACC) have been reported, they are extremely rare [3]. In this study, we report a case of ocular albinism in a newborn in whom ACC was indicated in utero, including the manner in which the original disease was diagnosed.

Case Report

This study involved a female newborn born after a gestational period of 41 weeks and who was the couple's first child. Magnetic resonance imaging (MRI) performed at a nearby clinic during the fetal period revealed the indication of ACC, so the mother was referred to the Obstetrics Department at Takatsuki Hospital, Takatsuki City, Japan, where she subsequently delivered the baby by natural spontaneous birth. The baby was admitted to the neonatal intensive care unit for ACC examination and then referred for consultation to the Department of Ophthalmology at Takatsuki Hospital for review of congenital eye anomalies.

No past history of consanguineous marriage, albinism, or other systemic disease was recorded.

Findings at Birth

The weight of the baby at birth was 2,590 g, with a cranial circumference of 31 cm and an Apgar score of 8/8. There was no coiling of the umbilical cord or amniotic fluid turbidity at the time of birth. Cranial sonography showed ventricular enlargement, and ACC and cerebellar hypoplasia were observed. In addition, cranial MRI revealed atrophy of the occipital lobe and abnormalities in the development of the cerebral gyrus and brain sulcus (fig. 1). Echocardiography showed good heart contractility. With regard to other systemic findings, no apparent heart, kidney, or immune system abnormalities were found. The color of the baby's hair was brown, while her eyebrows and skin were white.

Initial Ophthalmic Findings

The patient showed no obvious nystagmus, but light reactions were somewhat sluggish. The iris showed a ring-shaped area of hypopigmentation around the edge of the pupil, and the pupils exhibited a slightly irregular round shape (fig. 2). No obvious cataracts were found. The fundus showed hypopigmentation in both eyes from the posterior pole to the equator and exhibited a slightly irregular and albinism-like character (fig. 3). The patient's macular pigment was comparatively well preserved. Although optical coherence tomography was not performed, we theorize that macular hypoplasia might have been present since the foveal reflex of the fundus oculi was not observed. Although the optic discs were slightly reduced in size, they showed good color, and there was no evidence of optic nerve atrophy

or congenital anomalies in the optic nerve head. Moreover, no noticeable abnormalities were observed in the color, diameter, or flow of the retinal blood vessels. At 2 years after birth, the patient's eye tracking and fixation skills are evident, yet weak, and the foveal reflex was still not observed.

Discussion

While there have been scattered reports of ACC complicated with albinism in the past, the majority of these have been combined with abnormalities in the central nervous system [3]. In terms of the original underlying disease, the most frequently reported is Vici syndrome [4–10], with occasional reports of microcephalic osteodysplastic primordial dwarfism type I [11], hypomelanosis of Ito [12, 13], Lowe syndrome [14], oculocerebrocutaneous syndrome [15], and other diseases.

Vici syndrome was first reported in 1988 by Dionisi Vici et al. [4] in the case of two brothers presenting with albinism, ACC, cataracts, cardiomyopathy, mental retardation, convulsions, and immunodeficiency, and was subsequently named 'Vici syndrome' by del Campo et al. [9] in 1999. The findings of previously reported cases show that the prognosis has usually been poor due to cardiomyopathy or infection, with frequent reports of death during infancy. Vici syndrome has been observed in both males and females, and a number of related mutations have been identified in the EPG5 gene [16]. There have been numerous reported cases of Vici syndrome, and although this syndrome was initially considered for the case in this present study, which appeared to be oculocutaneous albinism from the findings of the fundus and the anterior segment of the eye, as well as the coloring of the hair and eyebrows, there was no obvious cardiomyopathy or immunodeficiency, and the systemic findings were inconsistent with many aspects of the syndrome. However, since no diagnostic criteria have been established for Vici syndrome, and because mild cases have been reported in the past [17], this possibility was also considered for our case. Although no gene search was carried out on this current patient, it is expected that it will be considered a necessity for diagnosis in the future.

Since microcephalic osteodysplastic primordial dwarfism type I presents in combination with microcephaly, it is not consistent with the findings in this present case. Hypomelanosis of Ito also frequently presents in combination with facial dysplasia, yet no obvious facial abnormalities were observed in our patient [11]. Oculocerebrorenal syndrome (also known as Lowe syndrome) was also rejected as the possible underlying disorder, since the main symptoms, as its name suggests, are mental retardation as a cerebral symptom and renal tubular acidosis as a renal symptom, and, also, because it inevitably presents with congenital cataracts in both eyes [14]. Similarly, oculocerebrocutaneous syndrome was rejected as the possible underlying disorder, as it is known to present in combination with localized underdevelopment of the skin and congenital anomalies of the optic disc such as morning glory syndrome [15].

Although the patient in question is presently growing without any major systemic problems, it is considered quite possible that various complications may arise in the future, including those related to the central nervous system, and it will be necessary to proceed with careful follow-up observation.

Although even ophthalmic scrutiny will be difficult as a result of the presence of mental retardation, we believe that it will be necessary, in the future, to pay attention to any changes in ophthalmic findings, including cataracts.

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Statement of Ethics

This case study has been approved by the Ethics Committee of the Osaka Medical College.

Disclosure Statement

The authors have no conflicts of interest to report.

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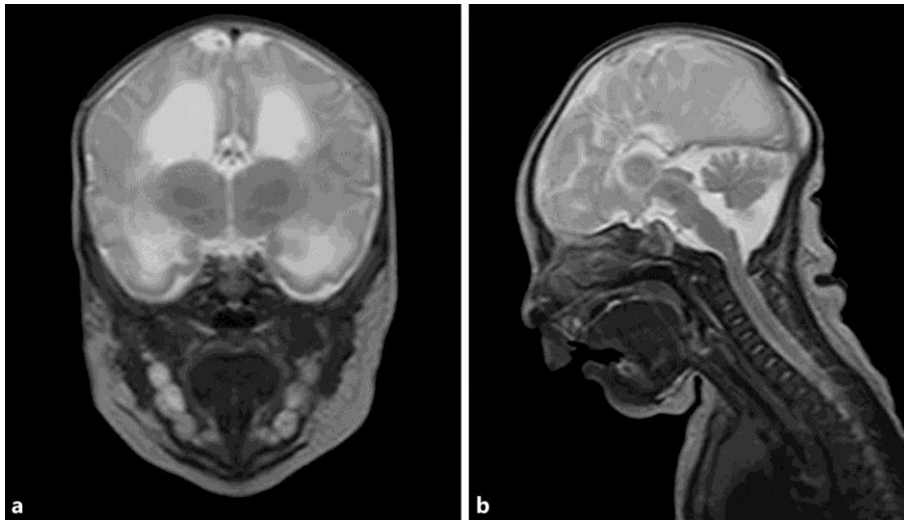


Fig. 1. Cranial MRI findings of the patient (coronal section, **a**; sagittal section, **b**). Atrophy of the occipital lobe and abnormalities in the development of the cerebral gyrus and brain sulcus were observed.

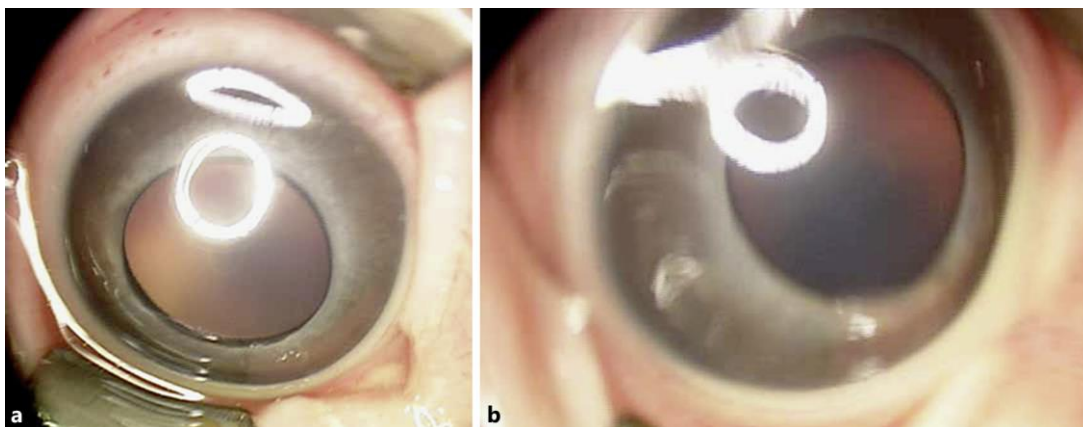


Fig. 2. Anterior segment photographs of the patient (right eye, **a**; left eye, **b**). The iris showed a ring-shaped area of hypopigmentation around the edge of the pupil, and the pupils exhibited a slightly irregular round shape in both eyes.

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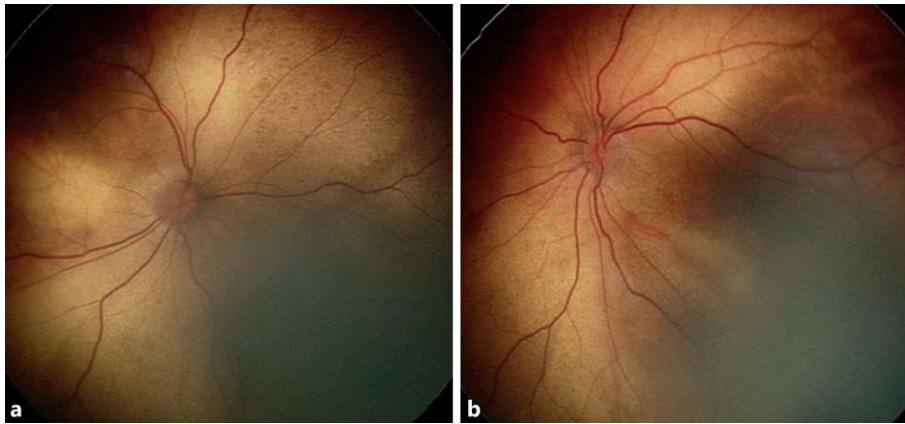


Fig. 3. Fundus photographs of the patient (right eye, **a**; left eye, **b**). Fundus photographs showed hypopigmentation in both eyes from the posterior pole to the equator that exhibited a slightly irregular and albinism-like character.