Late-onset obstructive hydrocephalus associated with occipital encephalocele with large skull defect successfully treated by endoscopic third ventriculostomy: A case report

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Case Report

Keywords: aplasia cutis congenita, aqueductal stenosis, encephalocele, hydrocephalus, ventriculostomy

Posted Date: June 21st, 2022

DOI: https://doi.org/10.21203/rs.3.rs-1732147/v1

Abstract

Background: Hydrocephalus is one of the most common presentations of occipital encephaloceles and usually develops within the first year of life. This case report presents a rare case of late-onset obstructive hydrocephalus associated with occipital encephalocele with an extraordinarily large occipital skull defect.

Case Report: At birth, a newborn girl presented with an absence of a vast amount of occipital cranium and skin and was diagnosed with occipital encephalocele. Under meticulous sterile management, the affected area was successfully epithelialized, and the patient was discharged without infectious complication. Despite an obstructed cerebral aqueduct, she grew without any signs of hydrocephalus until the age of 7 years. Her gait gradually worsened, and imaging tests at the age of 8 years revealed markedly enlarged lateral and third ventricles but not the fourth ventricle. Endoscopic third ventriculostomy successfully relieved her symptoms with improvement of hydrocephalus.

Conclusion: This is the first case of late-onset obstructive hydrocephalus associated with an occipital encephalocele characterized by large-scale cranial bony defects. Although further investigation is required to elucidate the mechanism of hydrocephalus, this rare phenomenon should be noted during neurological and radiological follow-up.

Introduction

Encephalocele is a type of neural tube defect characterized by an abnormal protrusion or herniation of the intracranial contents through a calvarial bone defect [1], with an estimated incidence of 0.8–5.6 per 10,000 live births [2]. Despite recent developments in diagnostic neuroimaging, surgical techniques, and post-operative care, the mortality and morbidity of encephalocele are still high [1, 3–5], and the neurological prognosis is relevant for several factors, such as associated anomalies, the amount and type of dysfunctional brain elements, and the presence of hydrocephalus [1, 2, 6].

Hydrocephalus is an important factor associated with prognosis, and as many as 50–90% of occipital encephalocele patients develop hydrocephalus [1, 2, 6]. Hydrocephalus mostly develops within the first year of life [1, 6–9], and the literature reports the development of hydrocephalus after excision and repair of the encephalocele [2, 6, 10].

In this report, a rare case of late-onset obstructive hydrocephalus secondary to an occipital encephalocele with an extremely large infra-and supra-torcular cranial bone defect is described. Endoscopic third ventriculostomy (ETV) successfully improved the hydrocephalus.

Case Report

Prenatal screening revealed a marked cerebral abnormality and absent occipital cranium, indicating cranial dysraphism. The patient was delivered via Caesarean section at 39 weeks of gestation. At birth, an extraordinarily large amount of the occipital cranium and skin was absent, and the parieto-occipital

lobe and cerebellum were covered only by the subarachnoid membrane, leading to the diagnosis of encephalocele and aplasia cutis congenita (Fig. 1A, B). She also had hypoplasia of the corpus callosum, ectopic grey matter, enlarged massa intermedia, and aqueduct obstruction (Fig. 1C). Cerebrospinal fluid (CSF) leakage was closed with polyglycolic acid felt and fibrin glue, and an artificial dermis was used to cover the skin defect. Under meticulous sterile management in the neonatal care unit, the affected area was epithelialized without any sign of infection by 8 months of age, and the patient was discharged with the use of a custom-made headgear apparatus, while having mild motor deficit and severe mental retardation (Fig. 1D). Starting at 5 years of age, she experienced intractable epileptic spasms requiring treatment with multiple anti-seizure medications (phenobarbital, levetiracetam, lamotrigine, and vigabatrin), while she grew without the sign of infection and hydrocephalus in follow-up magnetic resonance imaging (MRI) until the age of 7 years and 4 months (Fig. 1E). Since then her unstable gait gradually became obvious. She was subsequently referred to our neurosurgical department at 8 years of age.

Neurological examination on admission revealed that the patient was alert, and there was no noticeable paresis in the extremities. However, her gait disturbance was significant; therefore, she was unable to walk without support. MRI revealed markedly enlarged lateral ventricles. The third ventricle was also dilated, the floor of which projected downwards. In contrast, the fourth ventricle was not dilated (Fig. 2).

The patient underwent ETV under general anesthesia. A burr hole was placed in the right frontal region near the cranial edge (Fig. 3A). A plastic sheath was inserted into the anterior horn of the right lateral ventricle through which a flexible neuroendoscopic system (EVIS LUCERA SPECTRUM video imaging system; Olympus, Tokyo, Japan) was introduced. The third ventricle was reached via the abnormally dilated foramen of Monro. Anatomical structures, such as bilateral mammillary bodies and infundibular recess, were easily identified, and the ETV was completed using endoscopic forceps and a balloon catheter without complication (Fig. 3B, C). The postoperative course was uneventful, and her gait improved after surgery. Postoperative head MRI revealed that the lateral and third ventricles were markedly reduced in size (Fig. 4). In addition, the images showed an improvement in the downward extension of the third ventricular floor. The patient’s clinical status remained stable over the subsequent 5-month period.

Discussion

This report describes a rare case of delayed-onset obstructive hydrocephalus associated with occipital encephalocele that was successfully managed with ETV. This case was also characterized by cranial bony defects on an enormous scale. Although hydrocephalus is one of the most common presentations of occipital encephalocele, most patients develop hydrocephalus before 1 year of age [1, 6, 7]. Our patient showed clinical signs of hydrocephalus at 8 years of age, which was not detected at 7 years of age.

Thus, two hypotheses for the poorly-understood pathophysiology of this condition are proposed. The first theory is that the slowly progressing cerebellar herniation in the caudal direction might have distorted the

aqueduct, leading to obstructive hydrocephalus. However, in this case, aqueduct obstruction was already evident in the MRI by 4 months of age (Fig. 1C). Another hypothesis is that the resorptive capability of the CSF in the lateral and third ventricles or supratentorial cerebrum impaired as time progressed, leading to dilation of only the lateral and third ventricles. As this patient had no episode of CSF infection, frequent compression of the venous system through the site of a large bony defect in daily life might have induced hydrocephalus.

Regarding the treatment of hydrocephalus, ETV is perceived as a suitable alternative to extracranial shunting, particularly for obstructive hydrocephalus, as it offers a more physiological solution and a chance for a shunt-free life for patients with hydrocephalus [11]. Although intracranial developmental anomalies were considered relative contraindications for ETV because of the perceived technical difficulties with navigation in the past [12], ETV has now gained widespread acceptance as an effective alternative to ventricular shunt placement in patients with hydrocephalus in many congenital abnormalities, such as hydrocephalus in infants with encephalocele [7, 13, 14]. Regarding the ETV in this case, a slightly modified location of the burr hole was needed, considering the patient's specific abnormalities. Concerning this modification, we used a flexible neuroendoscope with a higher degree of mobility to access the targeted ventricle in a non-linear manner [14–17].

Conclusion

This is the first case of delayed-onset obstructive hydrocephalus associated with severe occipital encephalocele, which is characterized by an extraordinarily large skull defect. Although further investigation is needed to elucidate hydrocephalus' true mechanism, this rare phenomenon should be noted during the clinical follow-up.

Abbreviations

ETV; endoscopic third ventriculostomy, CSF; cerebrospinal fluid, MRI; magnetic resonance imaging

Declarations

Ethics approval and consent to participate: This is an observational study. The institutional review board of Hokkaido University Hospital has confirmed that no ethical approval is required.

Consent for publication: Written informed consent has been obtained from the parents of the patient included in the study.

Availability of data and material: The data and material in this study are available from the corresponding author, Taku Sugiyama, upon reasonable request.

Competing interests: The authors declare that they have no conflict of interest.

Funding: This study does not receive funding.

Authors' contributions: Taku Sugiyama and Miki Fujimura contributed to the conceptualization. Yuki Munekata, Taku Sugiyama, and Miki Fujimura drafted the manuscript. Taku Sugiyama, Yuki Ueda, and Miki Fujimura critically revised the manuscript. All authors contributed to the data acquisition and interpretation of the data. All authors approved the version to be published and agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Acknowledgements: The authors would like to thank Editage (www.editage.com) for English language editing.

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Figures

Figure 1

Photograph of the back of the patient's head (**a**) and computed tomography (**b**) at birth show extremely large defects of the skin and bony skull in the occipitoparietal area. A sagittal view of constructive interference in steady-state magnetic resonance imaging (MRI) at 4 months (**c**) shows aqueduct obstruction with no observable dilation of the third ventricle floor. Photograph (**d**) and T2-weighted MRI (**e**) at the age of 7 years and 4 months show a well-epithelialized occipital area of the skin defect and no sign of hydrocephalus.

Figure 2

T2-weighted magnetic resonance imaging (MRI) at the age of 8 years and 0 months (**a**) shows the significant dilation of the lateral and third ventricles. A sagittal view of steady-state MRI (**b**) shows the downward projection of the third ventricle floor and the shrinkage of the fourth ventricle.

Figure 3

Preoperative computed tomography angiography (**a**) indicates the anomaly of the cerebral venous system; thus, a burr hole was made in the right frontal region near the cranial edge (circle) to avoid venous injury. Endoscopic image depicting the third ventricular floor before perforation (**b**) and after perforation and ballooning (**c**).

Figure 4

Postoperative axial T2-weighted magnetic resonance imaging (MRI) (**a**) confirms the marked reduction in size of the lateral and third ventricles. A sagittal steady-state MRI (**b**) illustrates reduced extension of the third ventricular floor.