Macrocephaly is defined as an unusually large head circumference, measuring more than two standard deviations (SDs) above the mean. While benign familial macrocephaly is the most common diagnosis, various neuropathological disorders—including megalencephaly, subdural fluid collection, hydrocephalus, intracranial masses, and conditions involving abnormal skull growth—are also occasionally found in patients exhibiting neurological symptoms [1-3]. However, there are only a few documented instances where individuals with macrocephaly have been diagnosed with neuropathological conditions without any accompanying neurological risk factors.

A 2-year-old patient visited the outpatient clinic due to macrocephaly, with a head circumference of 51.5 cm (99.2 percentile, 2.409 SD), detected during a routine clinical checkup. He was born at full term, weighing 3,100 g, with no dysmorphic features and a head circumference of 32.5 cm (6.1 percentile, –1.546 SD). There was no family history of macrocephaly, and no abnormal neonatal history, including perinatal asphyxia or neonatal seizures, was reported. Additionally, the child achieved normal developmental milestones and exhibited no focal neurological deficits during the neurological examination. Other nonspecific symptoms such as lethargy, irritability, or vomiting were absent, and the fontanelle was closed, although the caregiver could not recall the exact timing. Further review of the regular checkup records revealed that the patient's height (89.7 cm, 61.0 percentile) and weight (12.7 kg, 54.4 percentile) remained within the interquartile range, while his head circumference increased from 46.1 cm (42.7 percentile, –0.184 SD) at 14 months to 51.5 cm (99.2 percentile, 2.409 SD) at 24 months. Although the patient only experienced mild headaches and no severe neurological symptoms, brain magnetic resonance imaging (MRI) was conducted due to the development of severe macrocephaly. MRI revealed a large cystic lesion (10.9×8.6×10.2 cm) occupying the left cerebral hemisphere, causing rightward midline shifting and cerebral compression of the frontal lobe, occipital lobe, and midbrain (Fig. 1A and B). The patient was subsequently transferred to the Department of Pediatric Neurosurgery, where he underwent endoscopic fenestration of the cyst. This procedure successfully reduced the size of the cyst and alleviated the midline shift (Fig. 1C and D). Pathological analysis of the biopsy revealed tiny strips of squeezed tissue with simple columnar cells, supporting a diagnosis of an ependymal cyst. The patient was monitored in the hospital for 5 days and continued outpatient follow-up for 1 month to check for acute complications, with none detected. Further outpatient follow-up with MRI is scheduled after 1 year.

Macrocephaly is a relatively common clinical feature, affecting up to 5% of the normally developed pediatric population [4]. However, severe
Macrocephaly, defined as a head circumference exceeding +3 SD, is sometimes linked to neuropathological conditions, including neurogenetic or structural brain disorders. This association underscores the importance of neuroimaging. Furthermore, a recent study highlighted the need for neuroimaging when risk factors such as neurological deficits or developmental delays are present, while recommending regular observation for patients without these risk factors [5].

In newborns with an open anterior fontanelle, transcranial ultrasound can quickly differentiate pathological conditions. However, after the age of 1 year, performing ultrasound in children becomes more challenging due to the closure of the anterior fontanelle. Additionally, brain MRI is the most precise method for diagnosing brain abnormalities, but it requires sedation in pediatric patients. This sedation can lead to several complications, necessitating a precise indication for its use [6]. Currently, there are no major guidelines regarding the use of brain MRI in pediatric patients diagnosed with macrocephaly.

Based on the present case, healthcare providers should consider neuroimaging in the management of severe macrocephaly, even when there are no neurological risk factors present. It is hoped that this case will contribute to the development of comprehensive guidelines for conducting neuroimaging in instances of macrocephaly.

This study was approved by the Institutional Review Board of the Gangnam Severance Hospital, Yonsei University College of Medicine (3-2023-0254). The review board waived the need for informed consent for this retrospective study.

Conflicts of interest

Young-Mock Lee is an editorial board member of the journal, but he was not involved in the peer reviewer selection, evaluation, or decision process of this article. No other potential conflicts of interest relevant to this article were reported.

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