Subependymal Grey Matter Heterotopia with Seizure in a 6-Month-Old Child

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Title Page

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Statement of Contribution

Bibek Shrestha played a central role in the research, contributing to conceptualization, data curation, formal analysis, and methodology. He was responsible for project administration and took the lead in writing the original draft, as well as in the review, editing, and visualization processes. Priyesh Shrestha, Vivek Karn and Shivaram Ale Magar were involved in the supervision, validation, and investigation phases of the project, ensuring the study's accuracy and credibility.

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Disclosure

None

Data availability statement

None

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Conflict of interest

None

Patient consent statement

Written informed consent was obtained from the parents of the patient for publication of this case image and accompanying images, complying with the requirements as mentioned in Wiley's CCR Consent Form.

Ethical Approval

The institutional review board (IRB) of Institute of Medicine, Maharajgunj Medical Campus does not mandate ethical approval for Case Image.

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Key Clinical Message

A 6-month-old male presented with seizures. Non-contrast CT revealed subependymal grey matter heterotopia, corpus callosum dysgenesis, colpocephaly, and suspicious closed-lip bilateral parieto-occipital schizencephaly. Benign posterior fossa and subarachnoid space enlargement were noted. This case highlights the diagnostic value of imaging in rare neuronal migration disorders and associated structural abnormalities.

Introduction

Gray matter heterotopia (GMH) is a rare cause of seizures resulting from abnormal neuronal migration during fetal development. GMH can be classified into three types: subependymal (periventricular), subcortical, and band (laminar) heterotopia. Radiological investigation such as Computed Tomography (CT) and Magnetic resonance imaging (MRI) is crucial for diagnosing GMH, revealing ectopic gray matter in various locations [1].

Case

A 6-month-old male infant presented with a primary complaint of seizures persisting for one week. A non-contrast CT scan of the head revealed several significant findings. There was a marked thinning of the corpus callosum accompanied by colpocephaly, characterized by disproportionate dilation of the occipital horns of the lateral ventricles. Along the bilateral lateral ventricular walls, nodular tissue with attenuation patterns consistent with grey matter was observed, suggestive of subependymal grey matter heterotopia. Additionally, features suspicious for closed-lip bilateral parieto-occipital schizencephaly were identified. Further CT scan dilated cerebrospinal fluid spaces within the left posterior fossa and benign enlargement of the subarachnoid spaces. (Figure 1, 2, 3, 4, 5) However, the calvarium, cerebral hemispheres, basal ganglia, and brainstem were structurally normal, with no evidence of midline shift or mass effect. These imaging findings collectively suggest a diagnosis of subependymal grey matter heterotopia associated with corpus callosum dysgenesis, colpocephaly, and potential schizencephaly. The dilated posterior fossa and subarachnoid spaces appear to be incidental findings, likely of benign origin.

Discussion

Subependymal grey matter heterotopia (SGMH) is a rare neurodevelopmental disorder resulting from defective neuronal migration during embryogenesis. This condition leads to ectopic grey matter along the ventricular walls and has been implicated in various neurological manifestations, particularly epilepsy [1]. The case of this 6-month-old male with seizures highlights the clinical significance and diagnostic challenges of SGMH in the context of associated neuroanatomical abnormalities, such as corpus callosum dysgenesis and colpocephaly. GMW is genetically heterogeneous, with 146 genes and chromosomal loci identified as potential causes and is associated with epilepsy, cognitive deficits, and other neurodevelopmental disorders [2]. CT is an important modality for diagnosis which reveals ectopic gray matter in various locations. Interestingly, this case also showed features suspicious for closed-lip bilateral parieto-occipital schizencephaly, a structural anomaly characterized by abnormal clefts in the cerebral hemispheres. While schizencephaly and SGMH are distinct conditions, their coexistence has been reported in the literature, likely reflecting a shared disruption in neuronal migration and cortical organization during fetal development. The dilated cerebrospinal fluid spaces in the left posterior fossa and benign enlargement of the subarachnoid spaces are incidental findings that are not uncommon in infants. These findings do not appear to contribute to the primary neurological presentation but may warrant monitoring to rule out progressive hydrocephalus or increased intracranial pressure. The clinical presentation of seizures in this case is consistent with the known association between SGMH and epilepsy, attributed to the abnormal cortical organization and ectopic grey matter's role in generating epileptiform activity [2]. Management of such cases typically involves a multidisciplinary approach, including seizure control with antiepileptic medications, developmental assessments, and genetic counseling.

List of Figures

References

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