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Beyond the Smooth Brain: MRI Findings of Lissencephaly in Two Pediatric Cases

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Abstract **Case Report**

Background: Lissencephaly is a rare malformation of cortical development caused by abnormal neuronal migration, leading to severe neurological impairment from the neonatal period or early childhood. ***Objective:*** To report two pediatric cases of lissencephaly and to highlight their clinical and MRI characteristics in light of recent literature. ***Case Presentation:*** The first case involved a 3-year-old boy with type I lissencephaly associated with parieto-occipital band heterotopia (double cortex). The second case concerned a female neonate presenting with lissencephaly with pachygyria associated with diffuse leukoencephalopathy, suggesting a complex genetic and metabolic etiology. ***Conclusion:*** MRI remains the cornerstone for the diagnosis and characterization of lissencephaly. The association with white matter abnormalities or band heterotopia suggests syndromic and genetically heterogeneous forms, requiring multidisciplinary management.

Keywords: Lissencephaly; Magnetic resonance imaging; Pachygyria; Band heterotopia; Cortical development malformations; Pediatric epilepsy.

INTRODUCTION

Lissencephaly is a rare disorder of cortical development resulting from defective neuronal migration between the 12th and 24th weeks of gestation [1]. It is characterized by absent or simplified cortical gyration associated with abnormal cortical thickening. Clinically, lissencephaly presents with early-onset epilepsy, severe developmental delay, and motor impairment [2].

Recent advances in molecular genetics have significantly expanded the spectrum of genes implicated in lissencephaly and related disorders, allowing improved genotype–phenotype correlations [3,4]. Brain MRI remains the reference imaging modality for diagnosis, classification, and etiological orientation of malformations of cortical development [5]. We report two pediatric cases illustrating the radiological and clinical heterogeneity of lissencephaly.

CASES PRESENTATION

Case 1

A 3-year-old male patient was followed for cerebral palsy. He presented with generalized tonic–clonic seizures since the first month of life. There was no documented history of perinatal asphyxia or neonatal infection. Neurodevelopment was severely delayed.

Case 2

A female neonate, aged 4 days, was admitted for generalized tonic–clonic neonatal seizures present since birth. Initial evaluation revealed no evidence of metabolic disorders or neuro-meningeal infection. Brain MRI was performed in the context of persistent neonatal seizures.

Imaging Findings

Case 1

Brain MRI demonstrated a diffusely smooth cortical surface at the supratentorial level, related to abnormal cortical gyration of the pachygyria type (figure 1 (a, b, c)), with a characteristic anteroposterior gradient (figure 1 c), consistent with a disorder of neuronal migration.

There was diffuse cortical thickening, best appreciated on coronal TIR sequences, associated with a thin parieto-occipital subcortical band showing signal intensity similar to gray matter, resulting in a “double cortex” appearance (figure 2 (a, b)), highly suggestive of band heterotopia.

In addition, enlargement of the supratentorial and infratentorial subarachnoid spaces was noted, associated with moderate ventriculomegaly, without signs of transependymal cerebrospinal fluid resorption (figure 1 d). An enlarged posterior cranial fossa communicating with the fourth ventricle was also observed (figure 1 c).

Overall, these findings were consistent with type I lissencephaly associated with parieto-occipital band heterotopia.

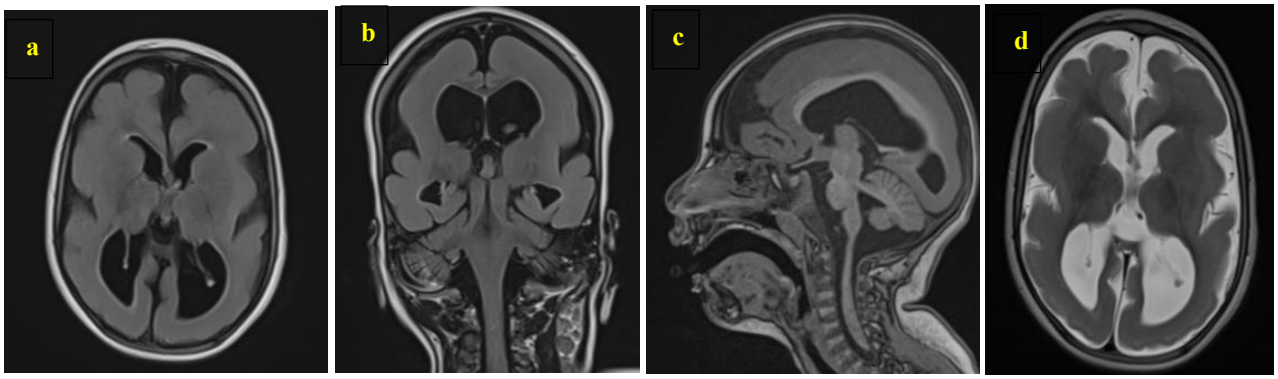

Figure 1: Brain MRI of the first case (FLAIR axial (a) and coronal (b); 3D T1 sagittal (c) and T2 axial (d)) demonstrated a diffusely smooth cortical surface at the supratentorial level (a, b, c), with a characteristic anteroposterior gradient (c). Enlargement of the supratentorial and infratentorial subarachnoid spaces with **moderate ventriculomegaly**, **without signs of transependymal cerebrospinal fluid resorption (d). An enlarged posterior cranial fossa communicating with the fourth ventricle was also observed (c)**

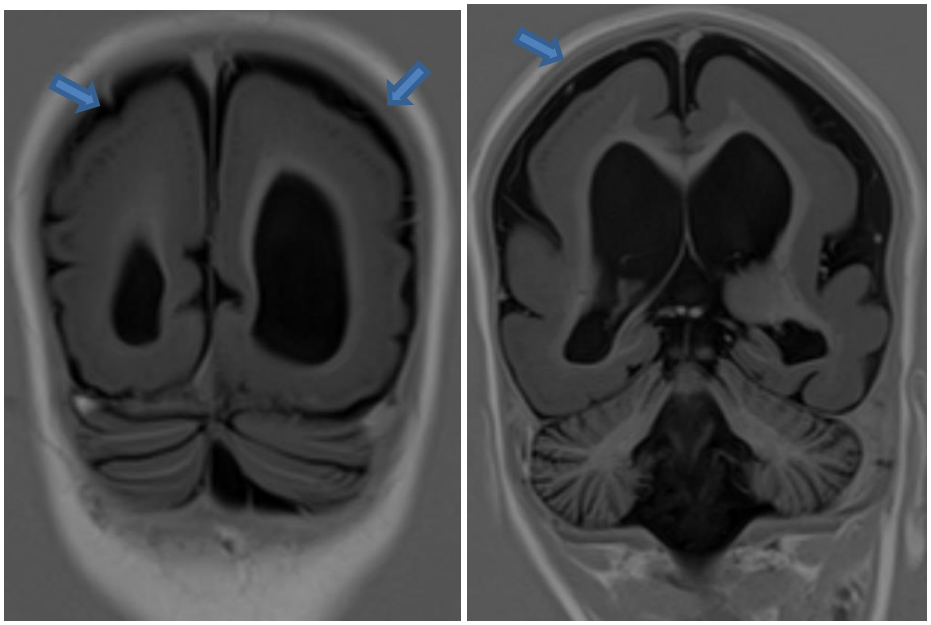

Figure 2: Diffuse cortical thickening appreciated on coronal TIR sequences (a, b) of the first case, associated with a thin parieto-occipital subcortical band showing signal intensity similar to gray matter, resulting in a "double cortex" appearance (arrow), highly suggestive of band heterotopia

Case 2

Brain MRI revealed diffuse, bilateral signal abnormalities of the periventricular, deep, and subcortical white matter, appearing hyperintense on T2-weighted images (figure 3 a) and hypointense on T1-weighted images (figure 3 b), consistent with diffuse leukoencephalopathy [7].

Cortical gyration was abnormal, with rarefaction of cortical sulci and broad gyri, consistent with pachygyria, associated with areas of smooth cortex and mild focal cortical thickening (figure 3). No evidence of neuronal heterotopia was identified.

There was enlargement of the subarachnoid spaces and ventricular system, related to cortico-subcortical atrophy (figure 3 a). Additionally, an abnormal signal involving the tentorium cerebelli and falx cerebri, hyperintense on T1-weighted images (figure 4 a) and showing signal void on T2* sequences (figure 4 b), suggested a hemorrhagic component.

The overall MRI findings were consistent with lissencephaly with pachygyria associated with diffuse leukoencephalopathy, suggesting a genetic etiology with possible associated metabolic involvement.

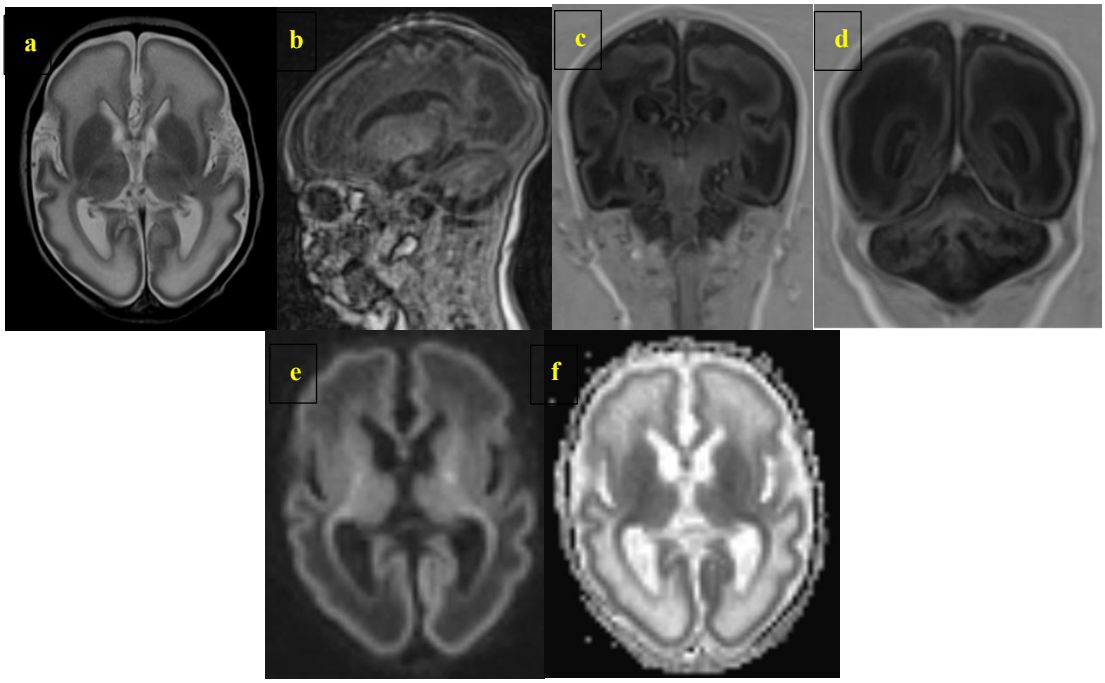

Figure 3 : Brain MRI of the second case (T2 axial (a), 3D T1 sagittal (b) and TIR coronal (c, d), DWI (e, f)) revealed diffuse signal abnormalities of the periventricular white matter, appearing hyperintense on T2-weighted images (a) and hypointense on T1-weighted images (b), with no diffusion restriction (e, f).
Cortical gyration was abnormal, with rarefaction of cortical sulci and broad gyri, associated with areas of smooth cortex and mild focal cortical thickening (a, b, c, d).
Enlargement of the subarachnoid spaces and ventricular system, related to cortico-subcortical atrophy (a)

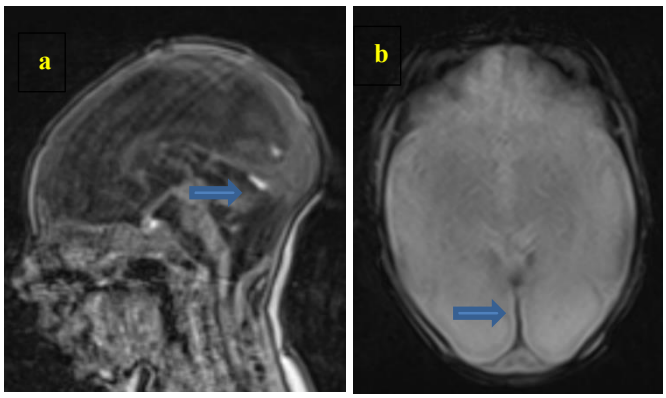

Figure 4: seconde case : Abnormal signal involving the tentorium cerebelli and falx cerebri, hyperintense on T1-weighted images (a) and showing signal void on T2* sequences (b), suggested a hemorrhagic component

DISCUSSION

Lissencephaly is a rare malformation of cortical development caused by abnormal neuronal migration during early gestation, leading to simplified gyration and abnormal cortical thickness on MRI [1,5]. Clinically, it is typically associated with early-onset epilepsy, severe developmental delay, and motor impairment, as observed in both of our patients [2].

The first case represents type I (classic) lissencephaly, characterized by pachygyria with a clear anteroposterior gradient and diffuse cortical thickening. The associated parieto-occipital band heterotopia (double cortex) reflects partial arrest of neuronal migration and places this patient within the lissencephaly–subcortical band heterotopia spectrum [6,8]. This imaging pattern is classically linked to mutations in *DCX* and other microtubule-related genes, emphasizing the importance of genetic testing for diagnostic confirmation and counseling [4,10].

The second case illustrates a more complex phenotype, with lissencephaly associated with pachygyria and diffuse white matter abnormalities, suggestive of a syndromic form involving both neuronal migration and myelination processes [3,7]. Recent

studies report that combined cortical malformations and leukoencephalopathy are frequently associated with genetic or metabolic disorders, particularly in neonates presenting with early seizures [9]. The absence of neuronal heterotopia and the presence of diffuse white matter involvement support this hypothesis.

MRI remains the cornerstone for the diagnosis and classification of lissencephaly. Key imaging features include cortical smoothness, gyral pattern, cortical thickness, distribution gradients, and associated anomalies such as ventriculomegaly or band heterotopia [5,12]. In both cases, ventricular and subarachnoid space enlargement likely reflects secondary cerebral volume loss rather than primary hydrocephalus, which has important clinical implications.

L'échographie transfontanellaire (ETF) constitue un outil de première ligne accessible pour le dépistage de la lissencéphalie chez le nouveau-né, permettant d'objectiver une simplification des sillons et une anomalie de la gyration, mais reste limitée par sa faible résolution spatiale et sa dépendance opérateur, rendant souvent nécessaire le recours à l'IRM pour une caractérisation précise [13].

In conclusion, these cases highlight the radiological heterogeneity of the lissencephaly spectrum and the critical role of MRI in guiding etiological evaluation. Early recognition of specific imaging patterns allows appropriate genetic investigations, prognostic assessment, and multidisciplinary management.

CONCLUSION

Lissencephaly is a rare but severe malformation of cortical development, best characterized by MRI. The reported cases emphasize the diversity of imaging patterns, ranging from classic type I lissencephaly associated with band heterotopia to complex forms with diffuse white matter involvement. A multidisciplinary approach integrating neuroimaging, genetics, and clinical follow-up is essential to optimize diagnosis, management, and prognostic evaluation.

Conflict of Interest: The authors declare no conflict of interest.

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