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Case Report

Lissencephaly: Pachygyria Type

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Abstract

Lissencephaly, also known as "smooth brain," is a severe and rare malformation of the cerebral cortex. It is defined as a gyration anomaly, either complete (total absence of cortical sulci) known as agyria, or pachygyria if there are still some sulci present. This condition is most often secondary to an anomaly in neuronal migration during embryogenesis. It is frequently observed in developing countries and its management is complex and challenging. We report the case of a one-and-a-half-year-old female patient with no significant medical history, admitted to the emergency department for repeated convulsive seizures. Her primary care physician requested a brain MRI.

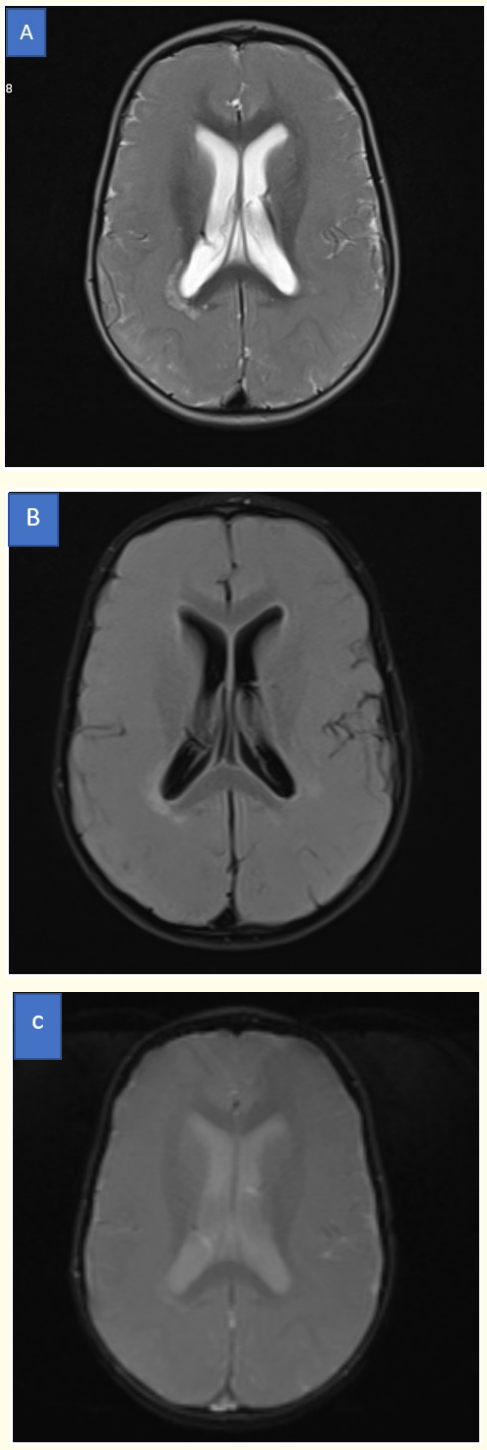
Keywords: *Lissencephaly; Gyration Anomaly; Pachygyria; MRI*

Introduction and Case Report

This is a one-and-a-half-year-old female patient who consulted her pediatrician for repeated convulsive seizures. Her primary care physician requested a brain MRI, which revealed punctate and clumped periventricular subcortical signal anomalies, more pronounced near the occipital horn of the right lateral ventricle, with iso T1, hyper T2, and FLAIR signals without diffusion sequence abnormalities, and no enhancement after GADO injection. There is also dedifferentiation of the white-gray matter with a reduction of white matter and a gyration anomaly characterized as pachygyria. These findings are consistent with pseudo-leukodystrophy with laminar heterotopia associated with type 6a lissencephaly.

Discussion

Lissencephaly is a rare and severe malformation of the cerebral cortex, defined as a gyration anomaly characterized by either a complete absence of cortical sulci (agyria) or the presence of a few cortical sulci (pachygyria). This malformation is frequently observed in developing countries, and its management is challenging and costly.


A
B
C

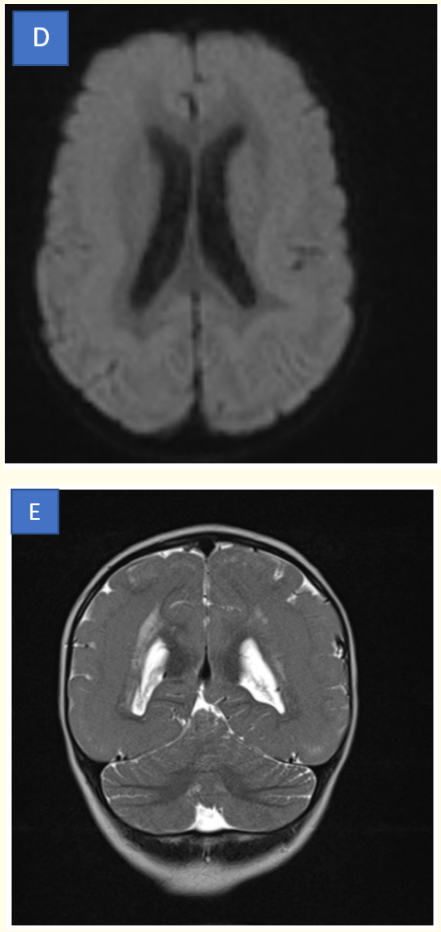


Figure 1: *Objectifying brain MRI: Punctate and clumped periventricular subcortical signal anomalies, more pronounced near the occipital horn of the right lateral ventricle, with iso T1, hyper T2 (A), and flair (B) signals without diffusion sequence abnormalities (C), and no enhancement after GADO injection. There is also dedifferentiation of the white-gray matter with a reduction of white matter and a gyration anomaly characterized as pachygyria (E). A: Axial section: T2, B: Axial section: T2 Flair, C: Axial section: Diffusion, D: Axial section: Echo grad, E: Coronal section: T2.*

Clinically, lissencephaly manifests in the first few weeks of life with neurological deficits, particularly hypotonia. Later, during the first year of life, it presents with convulsive seizures, severe psychomotor delay, feeding difficulties, growth retardation, and muscle spasms.

Lissencephaly can be secondary to genetic or non-genetic causes. Genetic causes mainly include abnormalities in the LIS1 gene (isolated lissencephaly and Miller-Dieker syndrome (MDS)), mutations in the TUBA3 and DCX genes, and mutations in the ARX gene (XLAG syndrome, X-linked lissencephaly with agenesis of the corpus callosum). Cobblestone lissencephaly (also known as type II lissencephaly) presents in three forms: Walker-Warburg syndrome, Fukuyama congenital muscular dystrophy, and muscle-eye-brain disease (MEB) [1]. Lissencephaly with facial dysmorphisms is observed in MDS, Baraitser-Winter syndrome (BWS), and Norman-Roberts syndrome [2,4].

Non-genetic causes include mainly viral infections during pregnancy and decreased cerebral vascularization. The prognosis of lissencephaly is severe, affecting both life expectancy and functionality.

The diagnosis of lissencephaly primarily relies on imaging, particularly brain MRI, which allows for the detailed examination of the cortex and the periventricular region to identify signal anomalies consistent with grey matter heterotopia (as seen in our patient) and the hippocampus. Brain MRI also helps study the morphology of the cortex and sulci, the white-grey matter differentiation, and the cortex periphery for thickening. If the sulci are completely absent, it is termed agyria; if not entirely absent, it is termed pachygyria. A key MRI finding is the focal widening of the subarachnoid spaces, which should always be systematically searched for [3].

The established classification scheme for lissencephaly is based on the severity (grades 1 - 6) and the gradient [5]:

- Grade 1: Generalized agyria
- Grade 2: Variable degree of agyria
- Grade 3: Variable degree of pachygyria
- Grade 4: Generalized pachygyria
- Grade 5: Mixed pachygyria and subcortical band heterotopia
- Grade 6: Subcortical band heterotopia alone
- Gradient 'a': From posterior to anterior gradient
- Gradient 'b': From anterior to posterior gradient.

Grade 1 and grade 4 are very rare. Grade 2 is observed in children with Miller-Dieker syndrome (a combination of lissencephaly with dysmorphic facial features, visceral abnormalities, and polydactyly). The most common lissencephaly observed, consisting of frontotemporal pachygyria and posterior agyria, is Grade 3. Another malformation worth mentioning because of its connections to pachygyria is polymicrogyria. Polymicrogyria is characterized by many small gyri separated by shallow sulci, slightly thin cortex, neuronal heterotopia and enlarged ventricle and is often superimposed on pachygyria [5].

Conclusion

In summary, this case of pachygyria-type lissencephaly highlights the importance of early diagnosis and comprehensive imaging in the management of cortical malformations. Given the severe neurological implications and the complex genetic associations, a multidisciplinary approach involving radiologists, geneticists, and pediatric neurologists is essential for optimal patient care and counseling. Further research into the genetic mechanisms underlying lissencephaly may provide insights into potential therapeutic strategies in the future.

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