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Imaging congenital brain malformations: A cause of drug-resistant epilepsy in children

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Abstract

Congenital brain defects are abnormalities in brain development that occur during foetal life. These anomalies can have a variety of causes, including genetic factors, intrauterine infections, maternal exposure to toxins and disturbances in the intrauterine environment. Cerebral malformations can lead to drug-resistant epilepsy, one of a variety of neurological problems. The imaging of drug-resistant epilepsy is a critically important topic, as seizures that are refractory to drug treatment pose a significant challenge for clinicians. In children with congenital brain malformations, drug resistance is particularly common. Advances in brain imaging have led to significant improvements in diagnosis, localisation of epileptogenic foci and surgical planning. The main imaging techniques used, each of which has a role to play in the management of drug-resistant epilepsy, are magnetic resonance imaging (MRI), computed tomography (CT) and transfontaneous ultrasound (FUS). Five cases were collated to illustrate the different forms of cerebral malformations in children presenting with drug-resistant epileptic seizures at the Abderrahim Harouchi Mother and Child Hospital in Casablanca.

Keywords: Epilepsy; Cerebral; Malformations; Drug-Resistant

1. Introduction

Congenital cerebral malformations are structural abnormalities of the brain that are present from birth. These malformations can result from a variety of factors, including genetic mutations, maternal infections, exposure to toxic substances during pregnancy or disturbances in foetal development [1]. Medical imaging plays a crucial role in the diagnosis, classification and management of these malformations.

The different types of congenital cerebral malformations are classified as follows:

- Neuronal migration malformations: These are disorders in the movement of neurons to their final position during embryonic development, including: lissencephaly, polymicrogyria, heterotopia.
- Cell division malformations: Abnormalities affecting the division and growth of brain cells, such as: microcephaly, megalencephaly.
- Malformations of the cerebral structure: Affect the structure and organisation of the brain, such as: holoprosencephaly, agenesis of the corpus callosum [2].

The aim of this work is to demonstrate the role of medical imaging in the diagnosis and management of the above-mentioned malformations, responsible for epilepsy refractory to the usual drug treatments.

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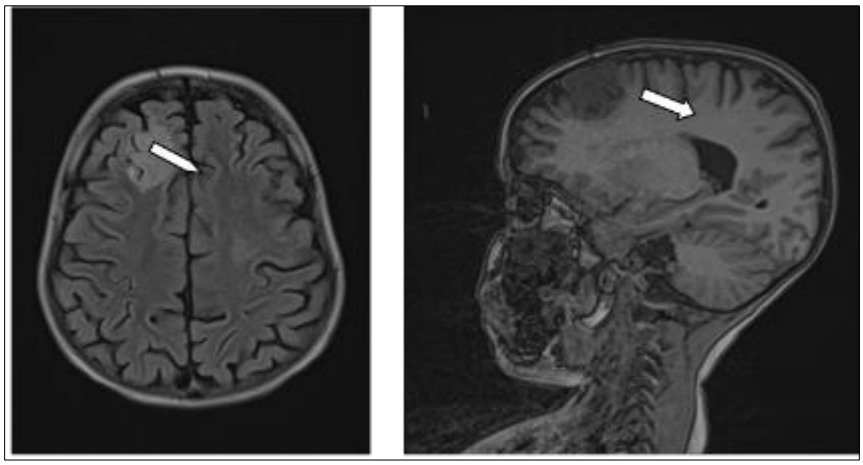
2. Methods

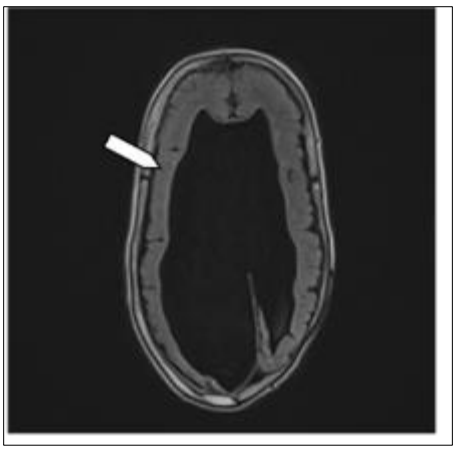
Our work is a retrospective study of seven (5) archived cases of children presenting with convulsive seizures who underwent brain imaging at the Abderrahim Harouchi Mother and Child Hospital. A complete clinical examination was carried out to look for neurological deficits, abnormal psychomotor development and speech disorders. Four (4) patients underwent combined CT-MRI and (1) patient underwent CT. The CT scan was performed in helical acquisition without injection of contrast medium, in double windowing, from C2 to the vertex. The 1.5 Tesla MRI was performed according to the protocol: 3D T1, axial T2 / T2*, coronal TIR on the hippocampi, FLAIR for patients over 2 years old, Diffusion; only one patient received a gadolinium injection.

3. Results

The extremes of age were 8 days and 12 years, with 4 girls and 1 boy. 4 patients had a history of prematurity and 3 of neonatal distress. Seizures were present in all patients. Each patient included in this illustrative study presented with at least one of the cerebral malformations and 3 of them presented with a lesion association. These were focal cortical dysplasia (figs. 1 and 2), gyration anomalies: polymicrogyria, schizencephaly, lissencephaly and malrotation of the hippocampi (figs. 3-6), grey matter heterotopias (figs. 7 and 8) and haemimegalencephaly (fig. 9).



Figures 1 and 2 Focal cortical dysplasia; axial FLAIR and sagittal T1 sections showing focal thickening of the right fronto-parietal cerebral cortex (arrow), cortico-subcortical FLAIR hypersignal of the dysplastic area (arrowhead), in a 5-year-old girl presenting with partial convulsive seizures since the age of 3.



Figures 3 Polymicrogyria; FLAIR axial section showing triventricular dilatation and numerous shallow right parietal supratentorial cortical sulci (arrowhead) in an 8-year-old girl with seizures and congenital hydrocephalus

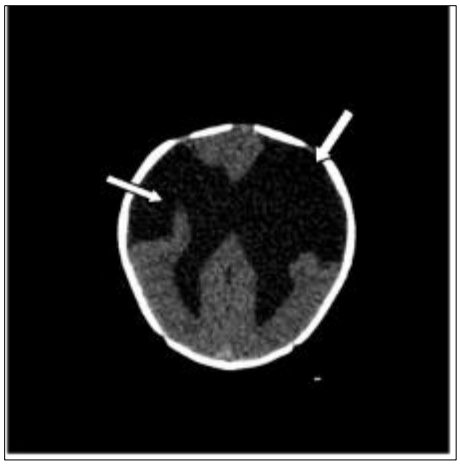


Figure 4 Bilateral schizencephaly with open cleft; axial CT sections at 8 days of age showing biventricular hydrocephalus communicating bilaterally with the subarachnoid spaces via a cleft (arrows)

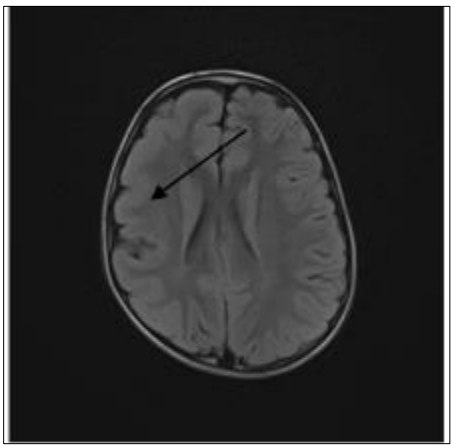


Figure 5 Lissenencephaly; FLAIR axial section showing a smooth right hemispheric cortical appearance, with rarefaction of the cortical sulci (arrow), in a 7-year-old boy with delayed height

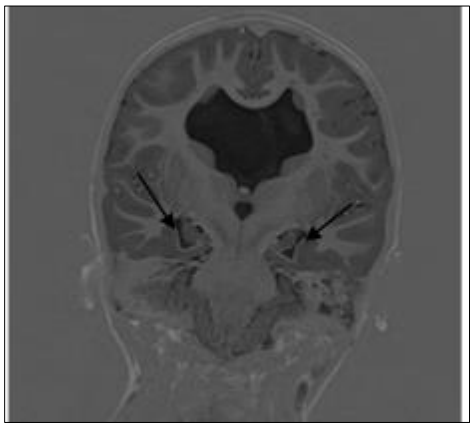
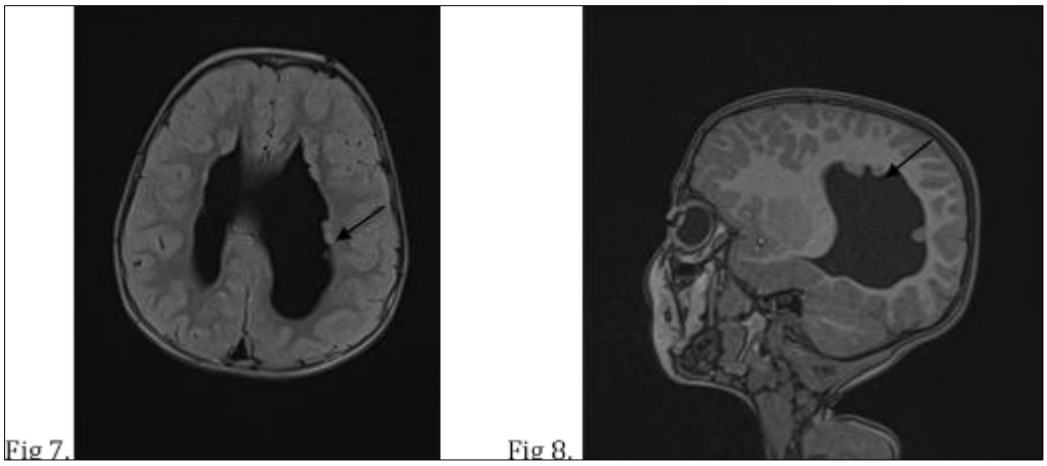


Figure 6 Malrotation of the hippocampi; coronal TIR sections showing bilateral inversion of the hippocampi in relation to the collateral sulci, with no signal anomaly (arrows), in a 2-year-old girl treated for callosal agenesis and seizures



Figures 7 and 8 Nodular heterotopias of the grey matter; FLAIR axial and T1 sagittal sections showing triventricular dilatation and hypoplasia of the corpus callosum associated with ectopic SG nodules in the subependymal region (arrows), in a 3-year-old girl being treated for hydrocephalus

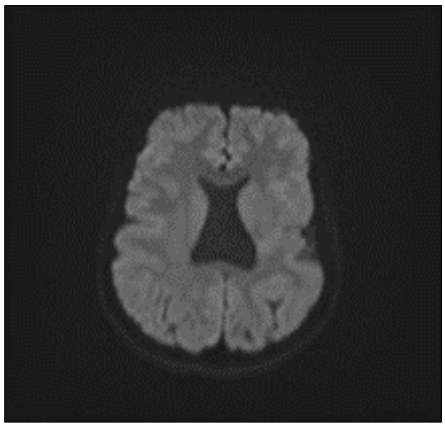


Figure 9 Right hemimegalencephaly; Diffusion sequence showing hypertrophy of the right cerebral hemisphere compared with the left, associated with contralateral pachygyria, in a 12-year-old girl with right hemiparesis being treated for epilepsy

4. Discussion

Focal cortical dysplasia (Figures 1 and 2) is due to cytomegaly of neurons and glial cells. Initially, CT scans reveal a discretely hyperdense area in the cortex extending into the white matter. In advanced stages, this area becomes hyperdense due to microcalcifications. There is no mass effect or contrast after injection. On MRI, this localised thickening of the cortex is reflected by an area of hypersignal extending to the white matter/grey matter (WM/GM) junction [3,4].

Polymicrogyria is characterised by the presence of multiple microgyri separated by shallow grooves (Figure 3). The cortex is discretely thickened with a sawtooth-like appearance of the SG/SB junction [5, 6].

Schizencephaly is defined by the presence of a trans-cerebral cleft with edges separated or fused by an ependymopial suture, known as type I or closed cleft schizencephaly or type II or open cleft schizencephaly (Figure 4). The cortex bordering the cleft is polymicrogiric. This is associated with agenesis of the septum, which may be found, depending on its topography, most often frontal, rolandic or insular [5].

Lissencephaly (agyria or pachygyria) is a severe cerebral malformation manifested by a smooth surface of the cortex abnormally composed of four (4) abnormal layers. Agyria refers to a thick cortex with no detectable furrows (Figure 5), whereas in pachygyria a few cortical furrows are visible [6].

Grey matter heterotopia is the presence of ectopic grey matter at a variable distance from the ventricles. The diagnosis may be suspected on CT, but is evident on MRI (Figures 7 and 8). Neurological symptoms include school delay, epilepsy and language difficulties. Three types are defined: nodular, subependymal and laminar heterotopias [6, 7 and 8].

Haemimegalencephaly is a hyperplasia of one hemisphere, with a poor prognosis. It is easily diagnosed on MRI by the presence of a large lateral ventricle with stretched horns and a thick cortex with short gyri and broad cortices [9, 10].

Tuberous sclerosis of Bourneville (TSB) is also known as tuberous sclerosis complex (TSC). It is characterised by the formation of benign tumours in various organs, principally the brain. The most common radiographic manifestations are cortical or subependymal tubercles and white matter abnormalities, 50% of which are in the frontal lobe; T2 hypersignal and T1 hyposignal on MRI with only 10% of tubercles showing enhancement with gadolinium injection; frequently calcifying after two years [11]. We did not report a similar case in this study.

Hippocampal malrotation or inversion of the incomplete left hippocampus is most common. The three main forms illustrated on MRI are verticalization of the collateral sulcus, pyramidal or rounded shape and inferior displacement of the fornix [12]. In our study, the malrotation was of the verticalization of the collateral sulcus bilaterally.

5. Conclusion

Cortical malformations are a major cause of epilepsy and delayed psychomotor development. Continuing advances in imaging promise to transform clinical approaches and improve outcomes for patients with refractory epilepsy, from diagnosis through to post-surgical planning and assessment. The various types of malformation are clearly identified and classified according to the stages of cortical development. Antenatal diagnosis has become possible thanks to the development of ultrasound and MRI, enabling a more precise and exhaustive study of gyration and the foetal cortex.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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