

CASE REPORT

X-linked Lissencephaly: a Smooth Brain and a Rough Journey for a 15-Year-Old-Patient

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Abstract

Lissencephaly („smooth brain”) is a malformation of cortical development associated with deficient neuronal migration and abnormal formation of cerebral convolutions or gyri. Although once thought to be rare, malformations of the cerebral cortex are increasingly involved as a major cause of recurrent seizures in children and adults. We described the case of 15-year-old female patient diagnosed with lissencephaly, mental retardation and focal seizures. At 6 months old, the patient developed focal motor seizures. Despite the fact that a CT was performed short after the occurrence of the first seizures, its quality did not allow the identification of cerebral anomalies like subcortical band heterotopia. Genetic testing through multiplex ligation-dependent probe amplification (MLPA) did not reveal any deletion or duplication in genes relevant to the subcortical band heterotopia diagnostic. Thus, in these cases, MRI assessment represents a useful tool to establish the diagnosis.

Keywords: lissencephaly, X-linked, seizures, MRI

Rezumat

Lisencefalia („creierul neted”) este o malformație a dezvoltării corticale asociată cu migrația neuronală deficitară și formarea anormală a circumvoluțiilor cerebrale sau a girilor. Deși sunt considerate patologii rare, malformațiile cortexului cerebral reprezintă din ce în ce mai frecvent o cauză majoră de producere a crizelor epileptice recurente la copii și adulți. Vă prezentăm cazul unei paciente în vârstă de 15 ani diagnosticată cu lisencefalie, retard mental și crize focale. La 6 luni, pacienta a prezentat crize motorii focale. Deși examenul CT a fost realizat la scurt timp după apariția primelor crize epileptice, calitatea acestei investigații nu a fost suficientă pentru identificarea heterotopiei subcorticale. Testarea genetică prin metoda MLPA nu a identificat nicio deleție sau duplicație în genele relevante pentru diagnosticul de heterotopie subcorticală în bandă. În astfel de cazuri, evaluarea RMN constituie un instrument foarte util în stabilirea diagnosticului.

Cuvinte cheie: lisencefalie, X-linkată, crize epileptice, RMN

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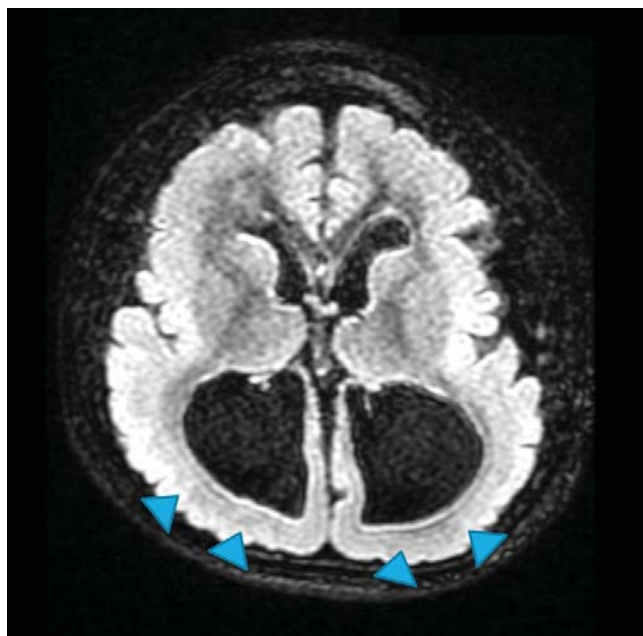


Figure 2. Cerebral MRI T1 Axial View. Ventriculomegaly (arrows).

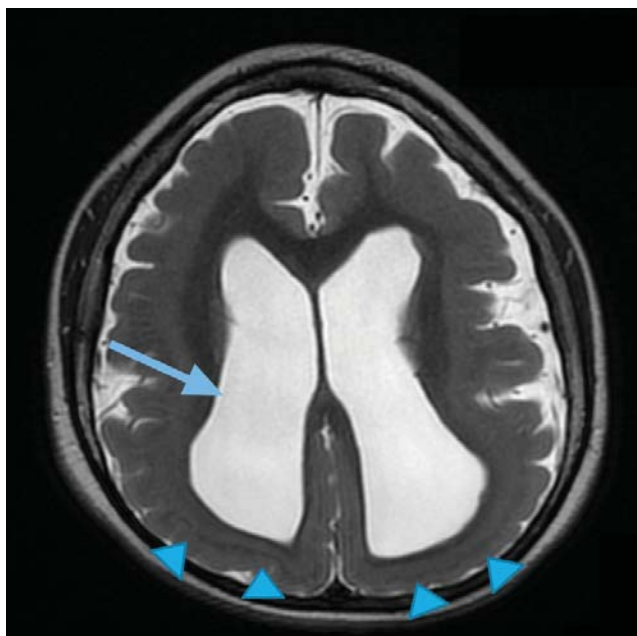


Figure 3. Cerebral MRI T2 Axial View. Ventriculomegaly (arrows). Disorganized gray matter located just beneath the cortex and separated from it by a thin band of white matter (triangles).

an efficient long term management, thus increasing quality of life. Despite the fact that a CT was performed right after the first seizures occurred, its limitations regarding resolution did not allow the detection of anomalies like subcortical band heterotopia. Also, the importance of the MRI is greater in certain cases such as this one, considering the fact that genetic testing did not detect any alteration in the genes associated with subcortical band heterotopia.

Compliance with ethics requirements:

The authors declare no conflict of interest regarding this article.

The authors declare that all the procedures and experiments of this study respect the ethical standards in the Helsinki Declaration of 1975, as revised in 2008(5), as well as the national law. Informed consent was obtained from all the patients included in the study.

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