Case 16266

Mixed pachygyria and subcortical band heterotopia (grade 5 lissencephaly).
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Diagnostic procedure
Procedure: Staging
Imaging Technique: MR
Imaging Technique: CT
Imaging Technique: Image manipulation / Reconstruction
Special Focus: Congenital Case Type: Clinical Cases
Authors: Carlos Fernández Cabrera, Laura Koren Fernández, Irene Navas Fernández-Silgado, Patricia Martín Medina, Amaya Hilario Barrio, Ana Ramos González
Patient: 24 years, female

Clinical History:

A 24-year-old female patient with known symptomatic epilepsy and global developmental delay presented in the emergency department due to a recent episode of generalized tonic-clonic seizure with a secondary head trauma. A CT scan was requested.

Imaging Findings:

Non-contrast-enhanced CT of the brain (Fig 1) showed a frontal extracranial haematoma. Incidentally, a marked hypodensity of periventricular white matter with a very straight white-grey matter junction was noted.

Brain MRI (Fig 2 and 3) revealed a continuous ribbon of grey matter seen just deep to the cortex, separated from it with a thin layer of white matter, giving the appearance of a double cortex. Incidental cavum septum pellucidum et vergae was noted. The overlying brain demonstrated wide gyri and mild shallowness of sulci (pachygyria).

Discussion:

Heterotopias belong to the malformations of cortical development caused by abnormal neuronal migration during embryogenesis. They are divided into three groups depending on the location of the ectopic formations [1]. Subependymal heterotopia is characterized by nodules of grey matter located immediately beneath the ependyma of the lateral ventricles. Focal subcortical heterotopia appears as nodules that extend from the ventricular surface outward into the white matter without continuity with the cerebral cortex. Subcortical band heterotopia is a form of diffuse heterotopia, resulting in grey matter deep to the cortex symmetrically distributed between the hemispheres.

Subcortical band heterotopia is a malformation of cortical development characterized by bands of gray matter in the white matter parallel to the surface of the cortex. More than 90% of the affected individuals are females due to mutations of the DCX gene (located on the long arm of chromosome X), which is essential for neuronal migration during cerebral embryogenesis. Most cases are sporadic, although a familial form has also been reported with X-
linked dominant inheritance. Affected patients typically present with refractory epilepsy and a variable degree of mental retardation [2].

Due to its contrast resolution, the mainstay of diagnosis is the MRI, which shows the characteristic “double cortex appearance”, formed by smooth ribbons of grey matter interposed in the white matter between the cerebral cortex and the ventricular surface. The signal intensity of the heterotopic grey matter is the same as normal cortex on all sequences [3].

Subcortical band heterotopia is included in the group of cortical malformations caused by abnormal neuronal migration together with lissencephaly [4]. The term lissencephaly refers to a smooth brain with absent (agyria) or abnormally broad gyri (pachygyria). These entities represent by themselves a category called “lissencephaly - subcortical band heterotopia spectrum” which includes apart from the abnormal gyri and the heterotopia, other associated findings such as enlarged lateral ventricles, mild hypoplasia of the corpus callosum, cerebellar hypoplasia and cavum septi pellucidi et vergae [4, 5].

The neuroradiological appearance of this lissencephaly – subcortical band heterotopia spectrum disorders is graded using a 6 point grading system based on the appearance of sulci and the extent and distribution of the heterotopia on MRI, where lower grade signifies more severe disease, as is shown in Fig 4. [6]. The patient of our case presented band heterotopia and broad and flat gyri and therefore was classified as a grade 5 lissencephaly.

Written informed patient consent for publication has been obtained.

**Differential Diagnosis List:** Mixed pachygyria and subcortical band heterotopia (grade 5 lissencephaly), Subcortical band heterotopia (grade 6 lissencephaly), Cortical dysplasia

**Final Diagnosis:** Mixed pachygyria and subcortical band heterotopia (grade 5 lissencephaly).

**References:**


William B Dobyns, MD and Soma Das. (2009) PAFAH1B1-Associated Lissencephaly/Subcortical Band Heterotopia. GeneReviews® [Internet]

**Figure 1**

*Description:* CT examination shows a frontal extracranial haematoma (white arrow). Incidentally, homogeneous bands of gray matter located between normal white matter and the cerebral cortex are noted (blue arrows). *Origin:* Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
Description: CT examination shows a frontal extracranial haematoma (white arrow). Incidentally, homogeneous bands of gray matter located between normal white matter and the cerebral cortex are noted (blue arrows). Origin: Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
**Description:** Axial T2 demonstrates symmetric bands of abnormal hyperintense signal intensity deep to the cerebral cortex (white arrows), giving rise to a “double cortex” appearance. **Origin:** Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
Description: Coronal T2 demonstrates symmetric bands of abnormal hyperintense signal intensity deep to the cerebral cortex (white arrows), giving rise to a "double cortex" appearance. Origin: Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
**Description:** The same complete band of subcortical tissue with signal intensity similar to gray matter is shown (white arrows). Note the shallowness of sulci and broad gyri. Cavum septi pellucidi with cavum vergae is also demonstrated. **Origin:** Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
Description: The same complete band of subcortical tissue with signal intensity similar to gray matter is shown (white arrows). Note the shallowness of sulci and broad gyri (pachygyria). Origin: Department of Radiology. Neuroradiology Section. Hospital 12 de Octubre. Madrid. Spain.
**Figure 4 a**

<table>
<thead>
<tr>
<th>GRADE 1</th>
<th>GRADE 2</th>
<th>GRADE 3</th>
<th>GRADE 4</th>
<th>GRADE 5</th>
<th>GRADE 6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Complete agyria</td>
<td>Diffuse agyria with a few undulations at the frontal or occipital poles</td>
<td>Mixed agyria and pachygyria</td>
<td>Diffuse pachygyria or mixed pachygyria and normal or simplified gyri</td>
<td>Mixed pachygyria and subcortical band heterotopia</td>
<td>Subcortical band heterotopia only</td>
</tr>
</tbody>
</table>

**Description:** Grading System for Classic Lissencephaly and Subcortical band heterotopia. **Origin:** Carlos Fernández Cabrera. Department of Radiology, Hospital Universitario 12 de Octubre, Madrid, Spain.