Lissencephaly-pachygyria spectrum: Computed tomography diagnosed
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Imaging sequences
Imaging Technique: CT
Special Focus: Congenital Case Type: Anatomy and Functional Imaging
Authors: Dr Asma Jatoi, Dr Ghazala Shahzad, Dr Adnan Ahmed
Patient: 15 months, female

Clinical History:
A 15-month-old female baby was brought to us by her mother with complaints of frequent seizures and muscular hypotonia since birth. She was taken to multiple nearby medical clinics. She was then referred to a tertiary hospital where she was advised to undergo plain CT scan of the brain.

Imaging Findings:
Plain CT of the brain revealed smooth thickened cortex (agyria) along with scanty white matter involving temporo-parietal and occipital lobes, associated with band heterotopia (Fig. 1 and 2). There are widened thickened gyri (pachygyria) noted in frontal lobes with presence of intact white matter (Fig. 2). These are forming a "hour glass"-shaped cerebral contour (Fig.3). Ventricles appear dilated and dystrophic (Fig. 3). There is further evidence of hypoplasia of splenium of corpus callosum and pons (Fig 1).

Discussion:

DISCUSSION

The lissencephaly-pachygryria spectrum is a rare brain disorders where the whole or parts of the surface of the brain appear smooth, characterised by absent or minimal sulcation [1].

Lissencephaly is caused by defective neuronal migration during embryonic development [4].

Lissencephaly-pachygryria is characterise as:
1. Type I (classic) lissencephaly
2. Type II (cobblestone) lissencephaly

The pattern of inheritance depends on the gene abnormality. The severity gradient in lissencephaly type I predicts whether the affected gene is LIS1 (posterior greater than anterior) or DCX (anterior greater than posterior). [5]

- LIS1 (acquired) accounting for 50% of all cases, with a posterior to anterior gradient of most typical pattern being
agyria in the occipital regions transitioning to pachygyria in the frontal regions. [5]

-DCX (X-linked) accounting for 10% of all cases, with an anterior to posterior gradient of agyria in the frontal or midfrontal regions, transitioning to pachygyria in the occipital lobes.

CLINICAL FEATURES

Presentation will vary from patient to patient. [1, 4]
In type I lissencephaly, there is marked hypotonia, seizures and difficulty feeding.

In type II lissencephaly, there is developmental delay, hypotonia and ocular abnormalities.

IMAGING FEATURES

Lissencephaly can be identified on all modalities (antenatal and neonatal ultrasound, CT and MRI). MRI is the modality of choice. [3].

CT demonstrated the main features [3]
(1) a cerebral surface that is agyric or agyric with pachygyric areas.
(2) a cerebral contour that appears as "hour glass" due to lack of or incomplete opercularisation of the brain.
(3) an abnormal gray-white-matter distribution in the cerebral hemispheres (hypomyelination).

Type I (classic) lissencephaly can appear as the classic hour glass or figure-8 appearance or with a few poorly formed gyri (pachygyria) and a smooth outer surface usually associated with band heterotopia.

Type II lissencephaly has a microlobulated surface referred to as a cobblestone complex. Band heterotopia is not evident and the cortex is thinner than in type I. [1]

OUTCOME

Lissencephaly can’t be reversed. Treatment aims to support and comfort affected children. The prognosis depends on the degree of brain malformation. Many will die before the age of 10 years. [4]

TAKE HOME MASSAGE

Lissencephaly is a rare congenital malformation of the brain that has characteristic radiographic features. MRI is the modality of choice while CT scan establishes the diagnosis [2]

Differential Diagnosis List: Type I classical lissencephaly with posterior to anterior gradient., Polymicrogyria, Lissencephaly Type II

Final Diagnosis: Type I classical lissencephaly with posterior to anterior gradient.

References:

A.Prof Frank Gaillard Lissencephaly-pachygyria spectrum. Radiopedia
Rachel Nall, RN, BSN Lissencephaly.
Description: There is agyria of temporo-parietal and occipital lobes forming smooth cerebral contour with scanty white matter.
Splenium of corpus collasum appears hypoplastic with distorted ventricles. Origin: Liaquat Medical Hospital, hyderabad, Pakistan
Description: There are sulcations in frontal lobes with intact white matter. Origin: Liaquat Medical Hospital, Hyderabad, Pakistan
**Description:** The contour of cerebrum shows hour glass cerebrum. Ventricles appear dilated. **Origin:** Liaquat Medical Hospital, hyderabad, Pakistan