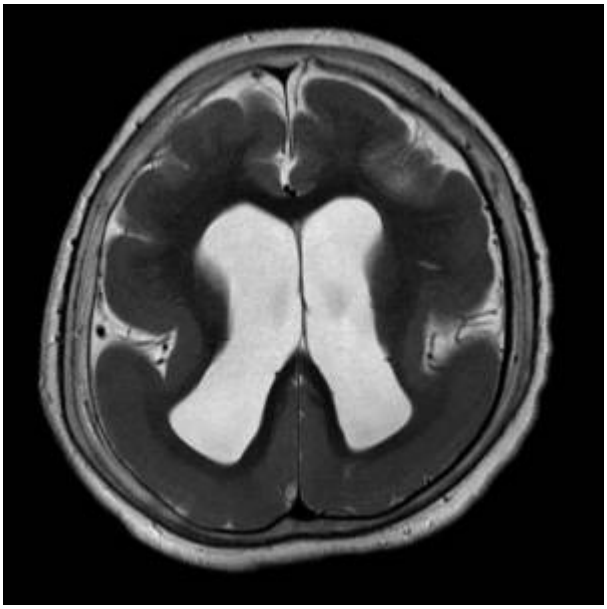
Pachygyria



Pachygyria is a developmental condition due to abnormal migration of nerve cells (neurons) in the developing brain and nervous system [1)].

The lissencephaly-pachygyria spectrum is a useful way to describe the spectrum of diseases that cause relative smoothness of the brain surface and includes:

agyria: no gyri

pachygyria: broad gyri

lissencephaly: smooth brain surface

Polymicrogyria: small gyri with shallow sulci. May be difficult to diagnose by CT/MRI, and may be confused with pachygyria.

With pachygyria, there are few gyri (the ridges between the wrinkles in the brain), and they are usually broad and flat.

The condition is also known as “incomplete lissencephaly.

Pachygyria may occur alone (isolated) or as part of various underlying syndromes.

Clinical features

Symptoms vary among affected people and may include moderate to severe developmental delay, seizures, poor muscle tone and control, feeding or swallowing difficulties, and small head size (microcephaly).

In most cases it is not inherited, but various inheritance patterns have been reported.

Diagnosis

Few broad & flat gyri with shallow sulci

Treatment

Treatment is symptomatic and supportive.

Case series

A retrospective study of 40 children with some form of pachygyria was performed at the Children's Memorial Hospital in Chicago. All 40 children had MR brain scans. We analyzed the MR findings, and correlated these findings with the clinical symptoms and course in all the children. We have autopsy findings in 15% these children. Based on our clinical, MR and autopsy findings, in conjunction with the medical literature, we found the following: (1) Pachygyria can occur as an isolated entity without an association with lissencephaly. The MR findings in these children consisted of a brain that demonstrated normal opercularization with either focal or diffuse areas of pachygyria without areas of agyria. These children live longer and have less severity of symptoms than the children with lissencephaly. (2) The MR findings in children with lissencephaly consisted of a brain that demonstrated abnormal opercularization with areas of total agyria or areas of agyria with pachygyria. (3) The MR findings in 25% of our children with polymicrogyria simulated pachygyria. The MR findings of the brain in these children consisted of a 'nubby' appearance to the outer surface of these abnormal gyri which resembled pachygyria but on histologic exam was polymicrogyria [2].

[1]
NINDS Neuronal Migration Disorders Information Page. National Institute of Neurological Disorders and Stroke (NINDS) Web site. https://www.ninds.nih.gov/Disorders/All-Disorders/Neuronal-Migration-Disorders-Information-Page. Accessed 9/30/2017.

[2]
Byrd SE, Osborn RE, Radkowski MA. The MR evaluation of pachygyria and associated syndromes. Eur J Radiol. 1991 Jan-Feb;12(1):53-9. PubMed PMID: 1999213.