

Case Report

BILATERAL SYMMETRICAL LISSENCEPHALY WITH PACHYGYRIA: A CASE REPORT

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ABSTRACT

Background: Lissencephaly is a rare developmental disorder characterized by absence of cerebral convolutions. Pachygyria (broad gyri) or agyria (no gyri) are terms used to describe appearance of cerebral surface. Together these associated conditions are a part of congenital cortical malformations and may result due to arrest of brain development before third or fourth month of gestation. Patients suffering from these conditions presents with significant developmental delays which further depends on the degree of malformation. Results: We report a case of bilateral symmetrical extensive lissencephaly with pachygyria. The major MRI findings during evaluation of our case were smooth gyral pattern with thickened cortex, thinning of periventricular white matter and prominent VR (Ventricular) spaces.

Conclusion: These defects can be idiopathic, associated with chromosomal abnormalities LIS 1 (chromosome 17) or can be to environmental factors (prenatal drugs or intrauterine perfusion failures). In our case Chromosome 17 defect was suspected as the parieto-occipital regions were more involved.

KEY WORDS: Lissencephaly, Pachygyria, Smooth gyral pattern, LIS 1.

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INTRODUCTION

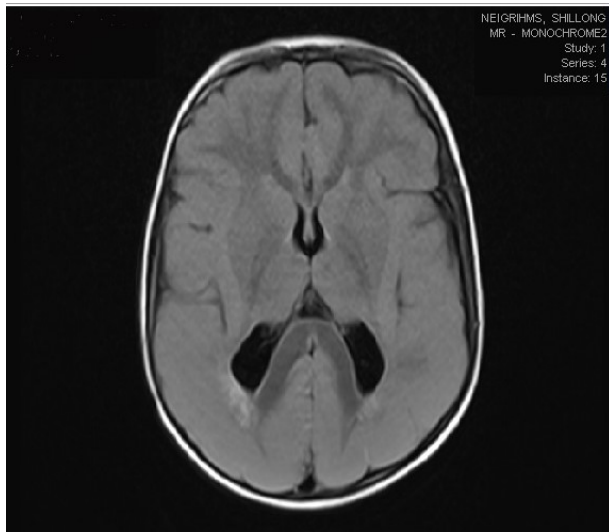
Lissencephaly or smooth brain is a rare congenital disorder known to be caused by defective neuronal migration during early third or fourth month of gestation resulting in lack of development of sulci and gyri. This condition is usually associated with pachygyria (broad gyri) or agyria [1] (no gyri). These spectrums of disorders may be idiopathic, may be seen associated with insult in early pregnancy or may be due to

chromosomal disorders (LIS 1). The malformation starts late in gestation at 12-24 weeks, because of neuroblastic migration not proceeding completely to the superficial layers of cortex [2].

Defective neuronal migration in these groups of disorders may be seen on MRI as smooth brain surface, decreased white matter with thick brain cortex, broad or absent gyri with shallow sulci and ventriculomegaly. We are hereby, reporting

a case of bilateral symmetrical extensive lissencephaly with pachygyria which was evident on MRI.

CASE REPORT



A three year old female child with complain of delayed motor milestones reported to the outpatient department, Department of Pediatrics, NEIGRIHMS, Shillong. The newborn with a history of home delivery at term presented with delayed speech and visual impairment. The patient had a history of delayed cry after birth. The breast feeding was normal. At 1 month of age the baby developed high degree fever and convulsions for which she was admitted in hospital for 1 month and also admitted several times for fever and convulsions thereafter. The baby was born of a non-consanguineous marriage, the mother being a 34 year's old 5th gravida. During antenatal period the pregnancy was monitored regularly and was uneventful with no history of any maternal illness. There was no history of any drug intake during pregnancy except for iron-folic acid supplementation, 2 doses of TT injections. There was no history of any neonatal death in the family. On examination the baby could not recognize her mother; could not hold head properly and was not able to sit. Speech and language evaluation revealed that even though vocalization was present the baby could not speak words. The baby could recognize familiar voices and could indicate her needs through differential cry. Ophthalmic examination revealed a normal fundus, not reacting to light and impaired eye movements revealing signs of cortical blindness. Bilateral Spasticity was observed which was

more prominent in the lower limbs. Chromosomal analysis to rule out LIS-1 syndrome could not be done. MRI done revealed thickening of cortex, more prominent in parieto-occipital and temporal regions with thinning of periventricular white matter and prominent ventricular spaces. A diagnosis of bilateral symmetrical extensive lissencephaly with Pachygyria was made.

DISCUSSION

Pachygyria (incomplete lissencephaly) is probably a form of lissencephaly in its lesser form, with the same cortical pattern. The term "lissencephaly" was introduced by Owen to describe the flat, smooth malformed brain in humans to distinguish it from the normal smooth brain found in lower animals [3]. It is characterized by short, broad, fat gyri caused by abnormal sulcation and gyration of the cortical mantle [2]. The exact distinction between the two entities is ambiguous [3, 4]. At its extreme, pachygyria may present with no sulcation at all creating a smoothly oriented hourglass configuration to the brain [2].

These forms of cortical dysplasia's, such as agyria, lissencephaly, pachygyria and neuronal heterotopias (disorganized brain tissue), are common neuro-pathological findings in newborns with intractable epilepsy and mental retardation. Cortical dysplasia's may be a result of environmental factors such as cytomegalovirus infections or may be seen associated with certain genetic abnormalities. Association of X- chromosome is seen more commonly with patterns effecting frontal lobe whereas chromosome 17 associated patterns affects parietal lobe more [2]. In our study even though the genetic study could not be done but Chromosome 17 defect was suspected as the parieto-occipital regions were more involved (fig.1). Cortical dysplasias may again be seen associated with a number of specific syndromes [5]. Two clinicopathological types have been identified [6]. Type I lissencephaly is characterized by a thick cortex with four rather than six layers of neurons, and it can be seen associated with phenotypes such as the Miller–Dieker syndrome (17p13.3monosomy) or the Norman–Roberts syndrome. Type II lissencephaly is pathologically characterized by a disorganized,

unlayered cortex, hydrocephalus is common feature, and clinically it presents as Walker–Warburg syndrome [7]. In both cases, areas of thick cortex (pachygyria), enlarged ventricles with decreased myelination are present. Details of the pathogenesis still remain provisional. However, cortical and sub-cortical laminar necrosis in the fourth month of fetal life was suspected to be causing the defective neuronal migration which was again linked to intra uterine hypoxia or perfusion failure [8].

CONCLUSION

Dysplasia like Lissencephaly and Pachygyria are rare disorders. MRI is one of the most preferred diagnostic tool in post-natal life with a potential to investigate these congenital disorders [9] particularly when a newborn presents with mental retardation or epilepsy. Although there is no specific treatment or cure for it prevention during early pregnancy, using trans-vaginal ultrasound in high risk cases can circumvent the existing disease burden in long term. Recurrence risk further should be addressed in confirmed cases by detailed cytogenetic analysis and genetic counseling.

Conflicts of Interests: None

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