

Case Report

Pachygyria Presented as Mania

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ABSTRACT

Psychiatric manifestation of pachygyria, a neuronal migration disorder is rare in literature; rarer if it is bipolar disorder specifically. Here, we report a case of mania and seizure who subsequently diagnosed as pachygyria. Proper literature about pathophysiology is discussed and recently discovered putative genetic role in bipolar disorder explained. This case also emphasis the importance of detailed history taking and imaging investigation even in a pure psychiatric presentation.

Key words: *Bipolar disorder, magnetic resonance imaging, pachygyria*

INTRODUCTION

Pachygyria is a congenital malformation of the cerebral hemisphere due to abnormal cell migration resulting in unusually thick convolution of the cerebral cortex. Usually presented with mental retardation and epilepsy, pachygyria may rarely be associated with mania, though the literature from India is scarce. Here we present a female presented with mania who subsequently diagnosed as pachygyria. To the best of our knowledge, it is the first reported report of pachygyria presented mania from Eastern India and also emphasis the importance of imaging and detailed investigation even in a patient presented with pure psychiatric presentation.

CASE REPORT

History proper

A 21-year-old unmarried female patient presented with symptoms of excessive talkativeness, irritability,

decreased sleep, overdressing, and hyper sexuality for last 20 days.

Patient was born out of a nonconsanguineous marriage and is the only child with no significant personal or family history. Her motor and developmental milestones were normal.

Patient was found to be a known case of seizure disorder (generalized tonic-clonic seizures) for last 5 year for which she had been on phenytoin 300 mg with occasional breakthrough seizures.

Mental status examination revealed psychomotor hyperactivity, over-familiarity; speech was increased in volume with decreased reaction time; expansive, labile affect. In thought there were grandiose delusion and delusion of reference.

Physical examination

Physical examination revealed obesity with moderate anemia and increased pulse rate.

Neurological examination showed decreased power in upper and lower limb, increased tone in right lower limb, right plantar reflex extensor, mild ataxia, ocular nystagmus (fast component in left side) in both eye.

Investigation

Laboratory analysis, including complete blood count,

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urine analysis biochemical and thyroid profile and liver function test normal.

At first we did a computed tomography scan, which showed possibilities of pachygyria [Figure 1] for which magnetic resonance imaging (MRI) ordered.

Magnetic resonance imaging revealed thickening of cortical gray matter, left frontotemporal lobes with an indistinct gray-white differentiation and pachygyria [Figures 2-5]. Left sylvian fissure was deformed [Figure 6].

Electroencephalogram demonstrated nonspecific slow-wave activity in left frontal and temporal cortices and some epileptiform discharges.

Psychometry

- On Wechsler Adult Intelligence Scale (WAIS) IQ was 75.
- Young Mania Rating Scale (YMRS) score was 33.

Course and follow-up

Patient was diagnosed as a diagnosis of bipolar affective disorder, current episode manic with psychotic

symptoms [F31.2] and put on olanzapine 15 mg and Na valproate 200 mg/day. Phenytoin was optimized to 350 mg after neurology consultation. Her mood symptoms subsided within 2 weeks with a repeat YMRS score of 10. On discharge, the dose of olanzapine was decreased to 10 mg with other medication remaining the same. She was maintaining well during follow-up at 3 months.

DISCUSSION

Definition

Normally the brain cells begin to develop from periventricular region and then migrate from medial to lateral to form the cerebral cortex.

Defective neuronal migration results in the formation of a disorganized cerebral cortex in which neurons are not normally connected with one another.^[1] Neurons that failed to reach their destination at the cortex remain at subcortical positions and differentiate composing islands of mature nerve cells, resembling cortical

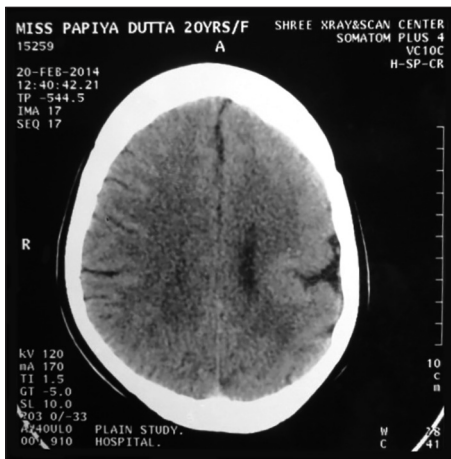


Figure 1: CT scan showing cortical malformation

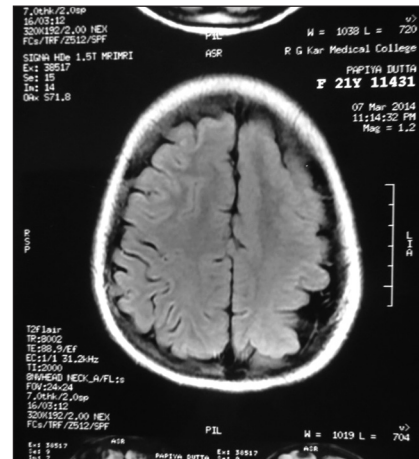


Figure 2: Loss of gyri

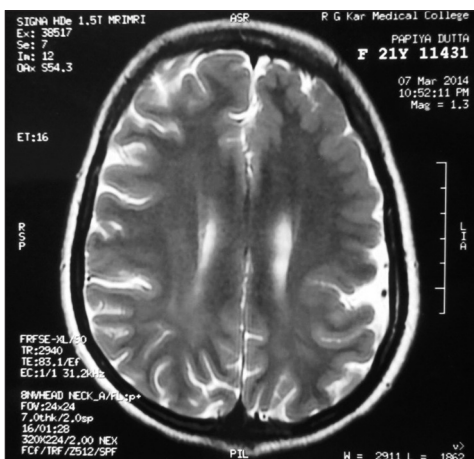


Figure 3: T2-weighted magnetic resonance imaging showing thickened gyri

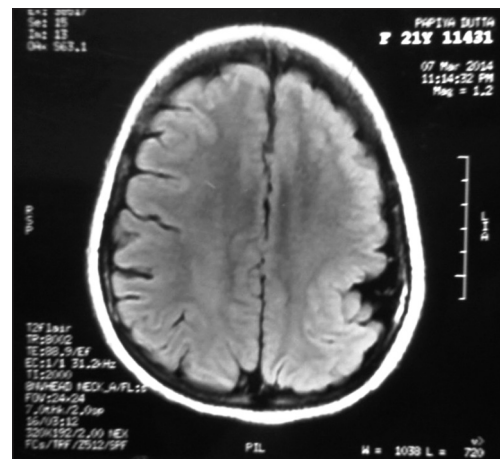


Figure 4: T1 weighted FLAIR, superior cut section showing cortical malformation



Figure 5: T1-weighted magnetic resonance imaging showing cortical malformation

neurons (subcortical band heterotopia) separated from the overlying cortex by an intervening band of white matter. The gyral pattern is also abnormal and is the basis for the morphologic classification of neuronal migration defects into lissencephaly, pachygyria, and polymicrogyria. Pachygyria, in which there are focal thickening and convolution of gyri, is a type of neuronal migration disorder (NMD) closely resembles lissencephaly type 1, others in this entity being pachygyria, agyria, porencephaly, lissencephaly or schizencephaly, the most severe form.^[2]

The lissencephaly-pachygyria spectrum is useful in describing the spectrum of diseases that cause relative smoothness of the brain surface and includes agyria (no gyri), pachygyria (broad gyri) and lissencephaly (smooth brain surface). It is a basket term for a number of congenital cortical malformations characterised by absent or minimal sulcation.^[3]

Clinical presentation

Neuronal migration disorder generally presented as seizure disorder, developmental delay and mental retardation and mental dysfunction. It may be asymptomatic and being symptomatic depends on the size and severity of dysfunction.

Mental retardation is not always present and depends on the size and severity of the abnormality.^[4]

Seizure may be focal or generalized and may be the first presenting symptoms.^[5]

To discuss psychiatric manifestation, NMD is previously reported to be associated with psychosis,^[6] schizoaffective disorder-depressive type,^[7] bipolar psychotic depression,^[8] bipolar 1 mania with psychotic symptoms,^[9] bipolar 2 disorder.^[5]



Figure 6: Deformed sylvian fissure

Until date, there was only one case report of pure pachygyria associated mania.^[5,10]

Structures that have been implicated in the pathophysiology of bipolar disorder include the temporal cortex, the entorhinal cortex.^[11] Furthermore, temporo-occipital cortical dysplasia has been reported to be associated with rapid-cycling bipolar disorder and learning disability.^[12] In bipolar 2 disorder patients, abnormalities in the right superior and middle temporal gyri, cingulate gyrus, precuneus and adjacent frontal and parietal white matter abnormalities were reported in high-resolution MRI studies, which correlated with the change in IQ.^[13]

Genetics

Various genes have found to be associated with both NMD and psychiatric disease and can help to elucidate specific genetic role in psychiatric disease.

RELIN gene is associated with NMD and also with schizophrenia, bipolar disorder and autism. DISC1, Ndel1^[1] and an SNP rs31745^[14] are found to be key regulators of neuronal migration. Also, both epilepsy and BMD can be explained by abnormal kindling, which was shown to be disturbed in NMD.^[15]

In our case, all the findings corroborated with the above findings still it is the first reported such case from Eastern India. In our case, defect involved frontoparietal region. It is worth mentioning that recently it is found that frontotemporal pachygyria is a distinct genetic entity inherited as an autosomal recessive trait.^[16]

CONCLUSION

Although uncommon, pachygyria might be considered as one of the differentials in cases of bipolar disorder

along with mental retardation and/or seizures. Neuroimaging, preferably MRI, should be done to confirm the diagnosis. Functional neuroimaging and genetic studies in such cases might shed further light on the pathophysiology of bipolar disorder.

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