A Case of Seizure Disorder - Pachygyria A Rare Presentation

A.K. Badrinath[1], K. Suresh[2], R. Raghunathan[2], M. Balachandran[2], Suresh Babu S.[3]

ABSTRACT

Introduction: Pachygyria is neuronal migration disorder with thick convolutions on cerebral cortex. It leads to mental retardation and intractable seizures. It is also called as Macrogyria. It is a rare disease and diagnosis is increasing with advances in the imaging techniques. The main stay of treatment is supportive.

Case report: Here we present a 14 year old patient who presented with right focal seizures and mental retardation was diagnosed as a case of pachygyria.

Conclusion: We wanted to present this case report as one the rarer presentation and highlight their response to antiepileptic therapy.

Keywords: Pachygyria, lissencephaly, seizures, mutations

INTRODUCTION

Pachygyria is a congenital disorder with defect in the neuronal migration. As a result of abnormal migration of the neurons there is thickening of the convolutions in the cerebral hemispheres. The cause of this rare neuronal migration disorder may be genetic or viral infections during pregnancy. Children with pachygyria have varying degree of mental retardation and seizure disorder. The other disorder synonym with pachygyria is lissencephaly which is characterized by a smooth surface of the cerebral cortex due to loss of the gyri. Pachygyria is milder form of lissencephaly. The diagnosis is by imaging studies, Magnetic resonance imaging is better compared to Computed tomography. There is no specific treatment for this disorder and treatment is only supportive.[1,2]

CASE REPORT

A 14 year old male patient presented to the Department of General Medicine, Sri Manakula Vinayagar Medical college and hospital, Puducherry, with complaints of seizures 3 episodes involving the right upper and lower limb (right focal seizures) (figure-1). Patient was a known case of seizure disorder on treatment but still patient had repeated episodes of seizures. Patient is also having mental retardation. Patient is born to third degree consanguineous marriage and the antenatal history was uneventful and delivered by caesarean section due to a macrocephy. Patient mother had recurrent abortions following his birth. Patient is conscious and hypertelorism present. Patient vitals stable and cardiac and respiratory system examination was normal. Examination of the central nervous system - patient is conscious, abnormal behaviour with speech disturbances were present. Patient had hypertonia, muscle power was normal and deep tendon reflexes were exaggerated and plantar extensor on the right side. Patient also had persistent myoclonic jerks involving the right upper and lower limb.

Patient was further evaluated and the complete haemogram, renal function tests and serum electrolytes of the patient was normal. MRI Brain was done to find the cause for intractable seizures and diagnosed to be a case of pachygyria with apparent left hemimeglanecaphaly (figure-2).

DISCUSSION

Pachygyria a greek word (pachy - thick and gyri - fat) is a neuronal migration disorder which leads to thickened and few gyri and in some patients the brain substance may be increased. Lissencephaly is severe form of the condition associated with a smooth cortical surface. The abnormal migration results in a four layered cortex instead of a six layered cortex.[1,2]

The possible causes of pachygyri are genetic mutations, viral infections during pregnancy and hypoxic insults during pregnancy. The genetic mutations involved is autosomal and X linked mutations. The L1S1 gene is responsible for autosomal lissencephaly. It is located in the chromosome 17p13.3. L1S1 gene encodes a protein for proper neuronal migration. Most cases are due to deletions of mutation in L1S1 gene. Patients with misense mutations have pachygyria. Doublecortin (DCX) is responsible for X linked disorders. Mutations causes anterior malformations and is linked to lissencephaly in males and subcortical band heterotopias in females. It is located in chromosome Xq22-3q23.[2,3]

The clinical presentation of patients with pachygyria is varying degree of mental retardation depending upon the extent of cortex involved. Patients often have seizure (persisting spasms, focal seizures, tonic seizures atypical seizures and atonic seizures) which is recurrent and resistance to treatment. The other symptoms include hypotonia, hypertelorism and estropia. Patients with pachygyria often have reduced life expectancy.[3,4]

With the advances in imaging techniques the incidence is increasing and is diagnosed by computed tomography (CT) of the Brain and MRI (magnetic resonance imaging) and MRI Brain is better than CT in diagnosis.[4,5]

Grading of Lissencephaly

Grade 1: generalised agyria
Grade 2: variable degree of agyria
Grade 3: variable degree of pachygyria
Grade 4: generalised pachygyria
Grade 5: mixed pachygyria with SBH
Grade 6: sub cortical band heterotopia

Treatment is usually symptomatic and rehabilitation therapy. Seizures are controlled with one or more anticonvulsants but patients have frequent episodes. The therapies include assistive technology speech therapy, behavioural therapy, occupational

[1]Professor, [2]Associate Professor, [3]Resident, Department of General Medicine, Sri Manakula Vinayagar Medical College and Hospital, Puducherry, India

Corresponding author: Dr. A.K. Badrinath, No: 4, shree apartments, II floor, 29 #, Avvainagar, Lawspet, Puducherry- 605008

How to cite this article: A.K. Badrinath, K. Suresh, R. Raghunathan, M. Balachandran, Suresh Babu S. A case of seizure disorder - pachygyria a rare presentation. International Journal of Contemporary Medical Research 2016;3(11):3243-3244.

Figure-1: Showing macrocephaly with hypertelorism (increased inter-pupillary distance) in the patient.

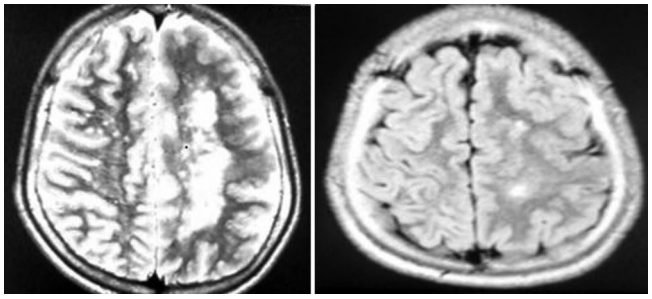

Figure-2: MRI Brain of the patient; the first figure showing thickened convulsions with few gyri (pachygyria) of the left cerebral hemisphere and the next figure showing apparent left hemimegalencephaly.

therapy, counselling, symptomatic drug therapy. But still there is no drug available to improve motor functions and higher cortical functions in these patients. Pachygyria is basically a structural defect; specific treatment of this pachygyria is still not available especially for associated seizures.[4,5]

CONCLUSION

Pachygyria is an inherited disorder with abnormal neuronal cell migration with mental retardation and refractory seizures with treatment being symptomatic. We wanted to highlight one of the rarer neuronal disorder and role of imaging sciences to diagnose these disorders to prevent it early by prenatal diagnostic modalities to reduce the burden of these disorders.

REFERENCES

1. Ramirez D, Lammer EJ, Johnson CB, Peterson CD. Autosomal recessive frontotemporal pachygyria. Am J Med Gemet A. 2004;124:231-8.
2. Kurul S, Cakmakci H, Dirik E. Agyria-pachygyria complex. MR findings and correlation with clinical featurez. Pediatr Neurol. 2004;30:16-23.
3. Francis F, Meyer G, Fallet-Bianco C, Moreno S, Kappeler C, et al. Human disorders of cortical development: from past to present. Eur. J. Neurosci. 2006;23:877–893.
4. Kuzinecky R. Familial diffuse cortical dysplasia. Arch Neurol. 1994;51:307-10.
5. Aronica E, Becker AJ, Spreafico R. Malformations of Cortical Development. Brain Pathol. 2012; 22:380-401.

Source of Support: Nil; **Conflict of Interest:** None

Submitted: 13-10-2016; **Published online**: 25-11-2016