Case Report

SUBCORTICAL BAND HETEROTOPIA AND PACHYGYRIA WITH COGNITIVE DETERIORATION IN AN ELDERLY PATIENT

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ABSTRACT

Subcortical band heterotopia and pachygyria are rare conditions characterized by ectopic neuronal migration, leading to the appearance of "continuous double cortex appearance" and loss of normal convolutions of the cortex, respectively on neuroimaging. They usually present in childhood with disorders of muscle tone, craniofacial dysmorphism, refractory seizures, intellectual disability and feeding disturbances. Patients with such presentations have lowered life expectancy, but many mild cases survive till adulthood. Here we report subcortical band heterotopia and pachygyria – an incidental finding – in the case of an elderly lady with intellectual disability and seizure disorder, who presented with recent onset of cognitive decline and behavioural symptoms.

Keywords: Subcortical band heterotopia, pachygyria, seizure disorder, intellectual disability, cognitive decline

INTRODUCTION

Disorders of cortical formation are characterized by abnormal cortical structure occurring due to interruption in normal development during the stages of proliferation, migration or organization of the cortex. Ectopic neuronal migration leads to heterotopia and pachygyria.[1] Pachygyria refers to the thickening of the convolutions in cerebral hemispheres, with few gyri. Clinically, they present with intellectual disability, seizures, craniofacial dysmorphisms and often have reduced life expectancy.[2] Here, we present a case of an elderly female with pachygyria and heterotopia who presented with intellectual disability, seizure disorder and recent onset of cognitive deterioration.

CASE REPORT

Miss P, a 63-year-old, unmarried, illiterate woman with intellectual disability and seizure disorder, presented with recent worsening of her cognitive functions and behavioural problems. As a child, she had poor social skills and difficulty in learning new skills; but used to maintain self-care without supervision and help in various household chores. From her childhood, she had recurrent attacks of generalized tonic-clonic seizures. She was on Tab. Carbamazepine from the age of 30 years, which was continued irregularly, and she used to get one or two seizure per month. There were no pervasive mood symptoms, suspicions, fearfulness, hallucinatory behaviour or other behavioural problems.

Over the past 1½ years, she had sleep impairment and inability to do the usual chores. There was a history suggestive of agnosia—she was unable to identify objects which she used to do earlier. This led to impairment in her functioning; she could not help in the chores she used to earlier. There were no behavioural issues at that time. Five months back, there was a sudden onset of behavioural symptoms characterized by impaired vegetative and social functioning. She had fatigue, showed odd movements but had no drowsiness,

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confusion, misidentification of relatives, picking movements or agitation. For this, she was treated at a local hospital, identified to have hyponatremia and managed with IV fluids. After this, her sleep and appetite improved, but cognitive dysfunction persisted.

One month back, she developed decreased sleep, poor social interaction and communication and was refusing feeds. She was treated elsewhere, had some improvement, then medications were stopped. Two weeks later, she had worsening of her symptoms and difficulty in walking. She had a seizure one week later, following which she was brought to our institution for further management. On examination, she was fully conscious but agitated, irritable, aggressive and not cooperative for detailed examination. Physical examination revealed bilateral hypertonia, hyperreflexia and bilateral flexor plantar reflex. Laboratory investigations were normal (See Table 1).

Table 1. Blood investigation results of the patient

investigations	values
Total WBC count	5200 Cells/cu.mm
Hemoglobin	12.7 g/dl
Platelet count	6,05,000/cu.mm
Random blood sugar	95mg%
Blood Urea	13 mg%
Serum Creatinine	0.7 mg %
Serum Sodium	140 mmol/L
Serum Potassium	3.2 mmol/L
Serum Magnesium	1.8 mg/dl
Serum Calcium	8.4 mg %
Serum Phosphorus	3.1 mg %
Serum TSH	0.77 microIU/ml
Serum T3	3.23 pg/ml
Serum T4	0.89 ng/dl

CT Scan Brain showed pachygyria, malfolded sylvian fissure, subcortical band heterotopia (SBH) and deep white matter ischemia (See Figures 1 and 2). Considering her poor financial background, MRI Brain was deferred as it would not have contributed to the patient's diagnosis or management.

She was managed with parenteral Haloperidol initially and then Tab. Quetiapine, the dose of which was titrated slowly. Tab. Carbamazepine 200 mg HS was continued as per the advice of the Neurologist. Supportive care was given. Adequate nutrition and hydration were ensured by Ryle's tube feeds and multivitamin tablets. Gradually her irritability and aggressive behaviour reduced, and she began taking oral feeds. But her communication was poor. She

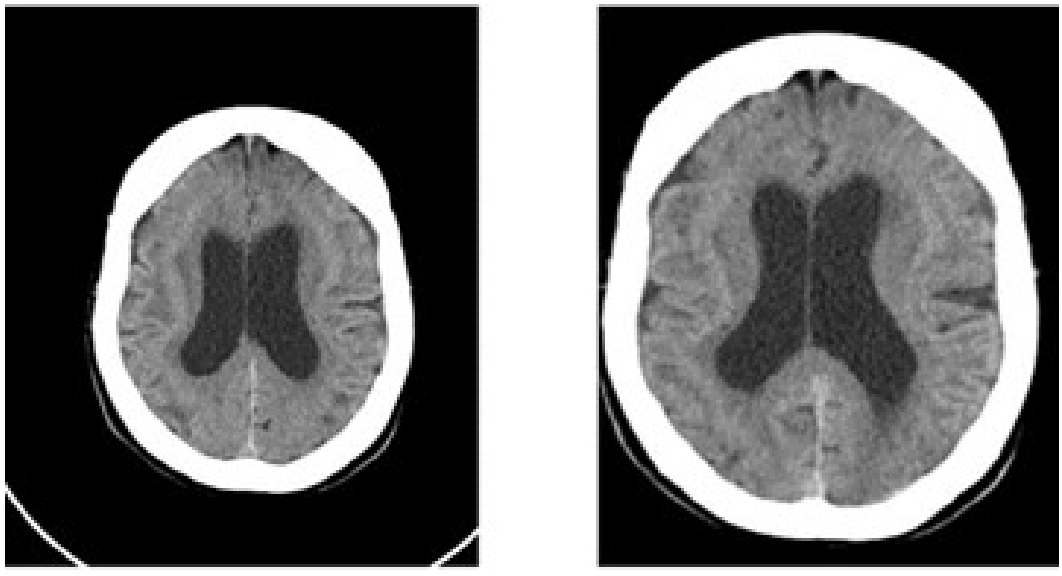

Figure 1. CT Scan Brain showing pachygyria and subcortical band heterotopia

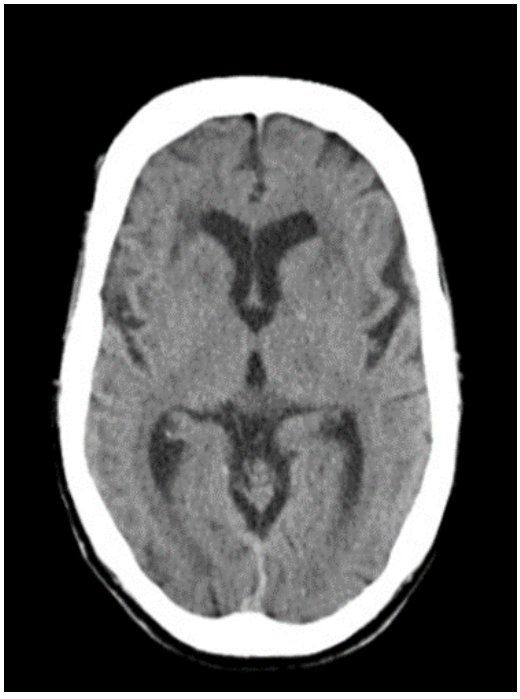

Figure 2. CT Scan Brain showing malfolded sylvian fissures and deep white matter ischemic changes

conscious and alert but was not cooperative for a detailed cognitive examination. She was mostly bedridden and had urinary incontinence. Her everyday activities were completely supervised. She was discharged after two weeks of inpatient care on Tab. Carbamazepine 200 mg HS, Tab. Quetiapine 25 mg 1-0-1½ with ½ SOS if agitated and multivitamin tablets. After one week, the patient was taking oral feeds, her sleep improved, and she was manageable at home.

Considering the past history, recent worsening of cognitive functions over the past eighteen months and impairment in activities of daily living, with an acute deterioration five months back along with behavioural disturbances, clinical diagnoses of Moderate Intellectual Disability, Seizure disorder and Major Neurocognitive Disorder (MNCD) due to vascular cause, with behavioural disturbances, were made.

DISCUSSION

SBH and pachygyria are generally associated with low life expectancy. Cases of psychosis, schizo-affective disorder, mania and seizures associated with neuronal migration disorder have been reported.[3,4] Here, we report a case that presented with seizure disorder and intellectual disability who survived to old age and developed further cognitive deterioration, impairment in functioning and behavioural disturbances suggestive of MNCD, possibly due to vascular cause, in an elderly female.

Among the disorders of cortical formation, lissencephaly—including agyria and pachygyria—and SBH belong to a single malformation spectrum. There is an arrest of neuronal migration in both these conditions. SBH is a smooth layer of grey matter seen subcortically, separated from the overlying cortex's curvature by a thin band of white matter, giving it a characteristic appearance of "continuous double cortex." Usually, the cortex is normal or pachygyric (with few gyri) in these cases.[1] Although mutations of *LIS1* and *DCX* genes were initially implicated in the aetiology, with advancements in molecular genetics, almost 19 LIS-associated genes have been identified.[5] DCX gene mutations cause lissencephaly in males and SBH in females. Classical lissencephaly manifests with epilepsy and psychomotor retardation, while SBH presents with a milder course, with mild to moderate intellectual disability and late-onset epilepsy usually.[6] This patient had presented with a history suggestive of mild to moderate intellectual disability and seizure disorder. Due to the milder form of presentation, she could survive to old age, when she developed further deterioration of cognitive function and behavioural disturbances.

In a review of clinical, imaging and molecular data, 188 cases with lissencephaly and SBH, aged one day to 40 years, were studied. The case we present is aged 63 years—the oldest reported as per review of literature. Generally, such patients present with mild to moderate clinical severity—borderline to moderate intellectual disability and seizures of variable severity, often poorly controlled. Although life expectancy is reduced, many of these patients survive to adulthood.[5] This patient survived to old age and developed significant cognitive impairment, including agnosia and impairment in language function, associated with impairment in everyday activities and behavioural disturbances. CT Scan of the brain revealed white matter ischemic changes along with pachygyria and SBH. National Institute of Neurological Disorders and Stroke and the Association Internationale pour la Recherche et l'Enseignement en Neurosciences (NINDS-AIREN) criteria states that a diagnosis of possible vascular dementia can be made even in the absence of a clear

temporal relationship between dementia and stroke or in patients with a subtle onset and variable course of cognitive deficits and evidence of cerebrovascular disease.[7] In this patient, the abrupt onset of behavioural symptoms, gait difficulty and worsening of cognitive symptoms could be due to vascular insults as evidenced by the deep white matter ischemic changes. Hence, a clinical diagnosis of MNCD due to vascular causes was made along with intellectual disability. The history, physical examination and investigations were not suggestive of any other causes of dementia.

CONCLUSION

Detection of pachygyria and SBH was an incidental finding in this patient, who presented with a history of intellectual disability and seizure disorder. The genetic mutation was not probably severe enough, so that she could survive to old age and present with further cognitive impairment due to vascular causes. This is the oldest case of pachygyria and SBH that has been reported to the best of our knowledge.

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