

Dysgenesis of the Corpus Callosum Presenting as First-onset Seizures in an Apparently Normal 32-year Old Female

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Abstract

Introduction: Dysgenesis of the corpus callosum is a brain abnormality involving the large nerve fibers connecting the two hemispheres of the brain. The corpus callosum connects the left and right cerebral hemispheres and facilitates interhemispheric communication. When it is malformed, these functions might be affected.

Clinical Presentation: This case report documents a patient with a malformed corpus callosum. She came in for first-onset generalized tonic clonic seizures. As part of a routine workup for patients with first-onset seizures, a computed tomography (CT) scan of the brain was done. It revealed

dysgenesis of the corpus callosum. She was started on valproic acid and was discharged improved.

Conclusion: Callosal disorders usually present with some degree of neurologic impairment. The index case however has no detectable neurologic deficits and is apparently normal. The rarity of a dysgenetic corpus callosum mandates more epidemiological studies to further elucidate this disease.

Keywords: dysgenesis; corpus callosum; seizure; normal

Introduction

Dysgenesis of corpus callosum is a rare disorder that is present at birth. It is characterized by a malformed corpus callosum which is an area of the brain that connects the two cerebral hemispheres. This part of the brain is normally composed of transverse fibers. The cause of dysgenesis of corpus callosum is usually not known, but it can be inherited as either an autosomal recessive trait or an X-linked dominant trait. It can also be caused by an infection or injury during the twelfth to the twenty-second week of pregnancy leading to developmental disturbance of the fetal brain.¹ In some cases, mental retardation may result but intelligence may be only mildly impaired and subtle psychosocial symptoms may be present.

It is unclear how often disorders of the corpus callosum actually occur since no one knows how many individuals have callosal conditions which are undiagnosed. Although it is rare, it is the most common brain abnormality observed in human beings.² Current research suggests callosal conditions may occur in as many as four persons per 1,000 in the general population. They occur more frequently among persons with developmental disabilities (22-24 per 1,000).³

In one case series involving 41 patients with callosal disorders,⁶ all of the patients with callosal disorders had severe neurologic impairments ranging from feeding and visual problems to abnormal muscle tone and paralysis. Interestingly, a majority of the patients had seizures just like the index case.

Callosal disorders are lifelong conditions. Individuals can benefit from early intervention services, supportive therapies, special education, and adult support services based on their individual needs. Evaluations and therapies should begin early in life and continue throughout childhood and into adult life. As researchers continue to learn more about the causes and impact of callosal conditions, families and professionals may gain additional knowledge to improve interventions and supports.

Objectives

1. To discuss briefly the anatomy and physiology of the corpus callosum
2. To discuss the causes of a dysgenetic corpus callosum
3. To present past studies of callosal disorders
4. To present the course in the ward of the index case
5. To discuss possible reasons why the index case is apparently normal

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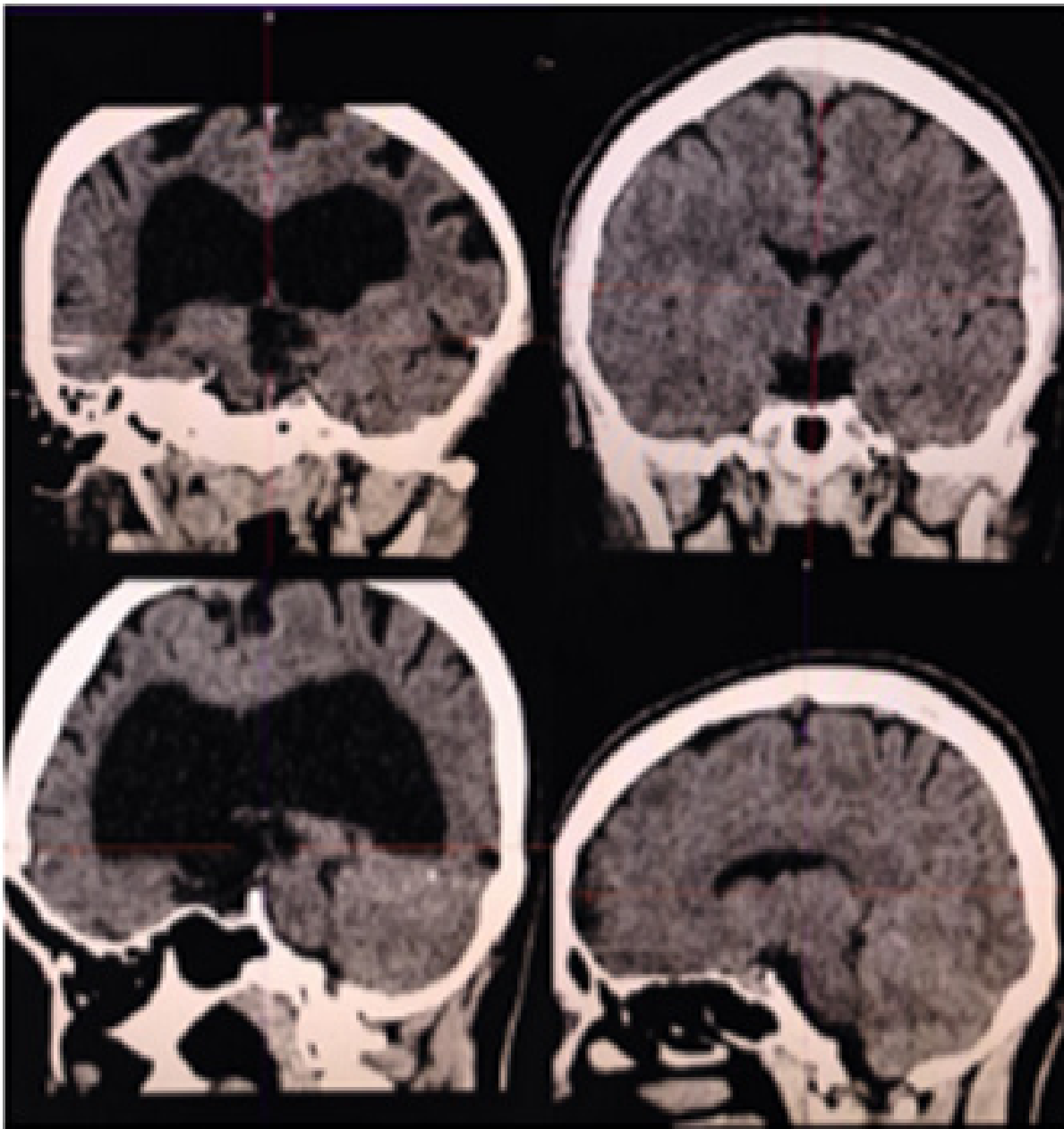


Figure 1. Shown on the right is patient's CT images compared with a normal CT scan from someone of the same age as the patient.

Clinical Presentation

The index case is a 32-year-old single female. She has a Bachelor's degree in Nursing. She worked as a nurse for three years in a private hospital but she currently works as a retail banker in one of the biggest call centers in the country. She grew up apparently with no neurological abnormalities. She presented at the emergency room for first-onset generalized tonic-clonic seizures. Neurologic examination and mental status

examination were unremarkable. She was admitted for observation and six hours after, patient's seizures recurred. Complete blood count, blood sugar and electrolytes (sodium, potassium, magnesium and total calcium) were all within normal limits. Lumbar tap was initially done to rule out infection and cerebrospinal fluid (CSF) studies which were all negative. Computed Tomographic (CT) scan of the brain with contrast was subsequently done. The CT scan revealed dysgenesis of the corpus callosum. There is widening of the cortical sulci and interhemispheric fissures.



Racing car sign

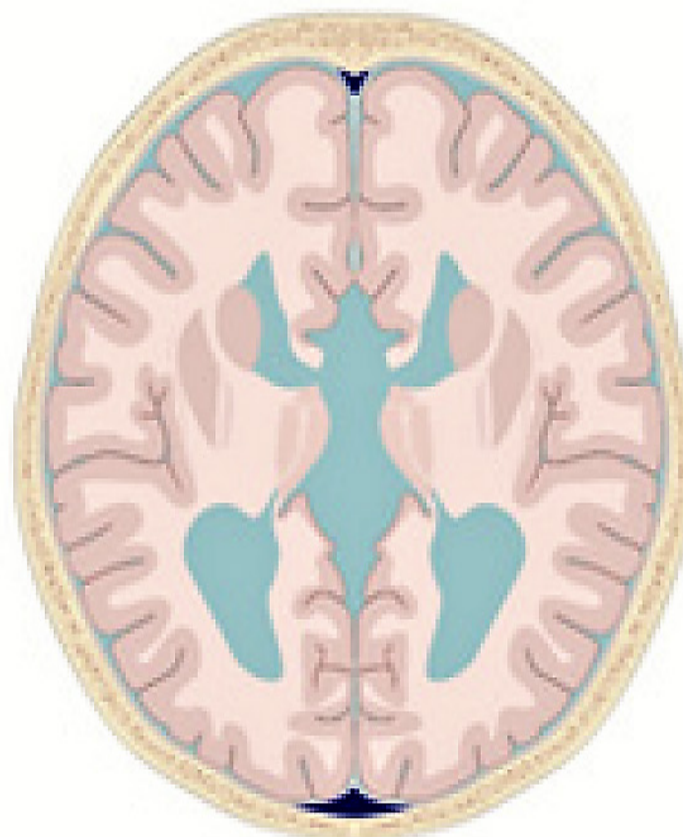


Figure 2. The Racing Car Sign. The patient's CT image compared with a drawing of the racing car sign for clarity.

The lateral and third ventricles are dilated. The fourth ventricle is normal in configuration. The parenchymal density is normal with no focal lesions evident. The gray white matter interface is maintained. Patient was started on valproic acid. Seizures did not recur and patient was discharged improved.

Discussion

As previously described, patients with dysgenesis of the corpus callosum usually have some sort of neurologic

deficits. It can range from mild cognitive deficits to severe mental retardation and intractable seizures. In our patient, she was completely asymptomatic until this recent admission. She even finished her education with distinction. Callosal disorders can be part of a syndrome or an isolated condition. If it occurs as an isolated condition, patients can be physically healthy and completely asymptomatic. If it occurs as part of a syndrome, it is associated with other physical abnormalities and it is usually apparent at birth. In the patient, it is more likely that it is an isolated condition.

The patient's case is unusual in three aspects. First, it was an incidental finding as part of the work-up of an adult patient who presented with first-onset generalized tonic-clonic seizures. Second, her age of presentation is at 32 years old. There has been no recorded data of callosal dysgenesis that presented in the adult age group. More than 90% of cases are discovered during infancy and the rest within the first two years of life. As previously discussed, the true incidence of dysgenesis of the corpus callosum is unknown.

The incidence might rise because of the frequency and availability of brain imaging studies such as CT scan and MRI. And lastly, the patient has no cognitive or mental impairments. She has in fact lived a normal life. Accounts from her friends and family reveal no social impairments. There is no family history of seizures. Patient's mother claim that prenatal history is unremarkable.

The concept of neuroplasticity could possibly explain the reason why the index case is apparently normal. The corpus callosum is formed early during gestation. A dysgenetic corpus callosum enables the fetal brain to make new inter-hemispheric connections that can overcome this structural abnormality. Two known abnormal tracts develop in patients with dysgenetic corpus callosum: the Probst bundle and the Sigmoid bundle that connects the frontal lobe to the contralateral occipital lobe. It is hypothesized that the severity of neurologic impairments depends on how well these tracts develop.

Conclusion

Callosal conditions are rare disorders with an unknown incidence. More than 95% of these conditions present in the first two years of life. It commonly present as seizures and neurologic impairments. Treatment is supportive. Depending on the severity of the neurologic impairments, these patients can live a long and normal life. Callosal conditions are rare disorders with an unknown incidence. More than 95% of these conditions present in the first two years of life. It commonly present as seizures and neurologic impairments. Treatment is supportive. Depending on the severity of the neurologic impairments, these patients can live a long and normal life.

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