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A CASE OF INCIDENTALLY-DIAGNOSED AGENESIS OF THE CORPUS CALLOSUM: CASE PRESENTATION

1.Atiye Fedakâr. MD. Pediatrician[1] 2. Zakir Sakcı MD, Radiology[2]

[1]The Departments of Pediatrics. Afiyet Hospital. Ümraniye, İstanbul, Turkey.

[2] The Departments of Radiology. Sağlık Bilimleri Üniversitesi Ümraniye Eğitim Araştırma Hastanesi , Ümraniye, İstanbul, Turkey.

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Corresponding author: Dr Atiye Fedakâr

Address: Armağan evler mah. Akdeniz cd. Sandra evleri c blok d-4. 34762 Ümraniye, İstanbul, Turkey

Phone:+905327439762

ABSTRACT

Corpus callosum is an anatomic structure that ensures the motor, sensory and cognitive performance of the brain by connecting stimuli originating from the cortex to the contralateral hemisphere. Agenesis of the corpus callosum is a commonly observed cerebral malformation mainly characterized by the absence of interhemispheric commissures. It can occur either in isolation or together with various syndromes. It is generally identified by tomography or cranial MR scans taken during the evaluation of epilepsy or other neuropsychiatric conditions.

This study presents a newborn infant case who was admitted to the newborn intensive care unit of our institution due to respiratory distress, and was then diagnosed with an agenesis of the corpus callosum following a cranial ultrasonography and cranial MR performed because the newborn infant was born with a cord around the neck and had a low birth weight.

Key Words: Corpus callosum, agenesis, newborn

Introduction

Corpus callosum (CC), the largest commissural structure of the brain located at the center of the brain, is responsible from providing inter-hemispheric sensorial connections, and it is essential for motor and high cognitive information (1,2). It develops during the 8th to 20th

gestational weeks and several factors such as radiation, chemical agents, infectious causes, nutritional disorders, hypoxia, chromosomal abnormalities, maternal hormones, and storage diseases may result in complete or partial defects in the CC (1.3.4.5). Corpus callosum agenesis (ACC) is a brain abnormality which, by virtue of the widespread use of imaging modalities such as magnetic resonance imaging (MRI), has become more frequently detected. It is more common among men and may occur as an isolated entity or accompany other syndromes (6). In the clinical presentation, it may manifest with findings such as growth retardation, learning difficulties, epilepsy, and mental retardation (7). Herein, we present a case of ACC which was detected by cranial ultrasonography (USG) alone, as the patient was born with nuchal cord and low birth weight in the lack of any clinical symptoms, and was later confirmed by cranial MRI.

Case Report

Our case was an interm infant born with nuchal cord by normal spontaneous delivery. Her mother was 26 years old and had her second pregnancy. The birth weight was 2,420 g, height 48 cm, head circumference 33 cm, and physical examination did not reveal any finding other than tachypnea. The Apgar score was 7 in the first and 9 in the fifth minute. Family history included influenza infection of mother and kinship. Complete blood count, C-reactive protein (CRP), and complete urinalysis results were normal. Biochemistry analysis showed elevated aspartate aminotransferase (AST) (66 IU/L). Echocardiography findings were normal. No other malformation was detected. The karyotype was normal. She was internalized in the neonatal intensive care unit due to tachypnea and received oxygen support by hood and parenteral nutrition. Enteral nutrition was initiated in the postnatal second day and the patient was discharged in the sixth day. In our clinics, we perform routine cranial and abdominal USG in cases of premature birth, babies of diabetic mothers, babies born with low birth weight or babies born large for gestatational age, babies born with a nuchal cord, in cases with a history of birth asphyxia, convulsion and in babies with hypernatremic dehydration. In our case, we performed cranial and abdominal USG due to low birth weight and the presence of a nuchal cord. Cranial USG indicated corpus callosum agenesis (CCA). The diagnosis was later confirmed by cranial MRI. The patient is currently six month-old and being followed in the outpatient setting.

Discussion

The name CC was first given by Galenus of Bergama during the second century. Later, it was described as the center of imagination, the place where the soul exists. Although it has been known for many years, there are still some unknown facts about CC and it is one of the brain tissues which are the most frequently associated with structural abnormalities (1). In CC, myelin tissue is 40% decreased, compared to other central nervous system and peripheral nervous system tissues; therefore, the absence of a protective myelin sheath exposes CC to a higher risk of damage by external factors (1). In particular, any harmful effect during embryogenesis period at the third month of pregnancy can result in ACC. Main factors causing agenesia include metabolic and toxic causes, inheritance, chromosomal abnormalities, asphyxia, chronic hypoxia, subdural hematoma, hydrocephalus, and storage disorders (1,3,4,5). According to current literature data, its frequency is estimated to vary between 0.3 and 0.7 (8). It is more common

among men (6). Literature shows that 50% of the cases occur as an isolated entity, while it may also accompany 65 hereditary or non-hereditary syndromes (1). Our case was a girl, who had an isolated disease without any accompanying malformations. Her karyotype was normal. The cause of CCA was associated with elevated AST levels and in-utero hypoxia due to nuchal cord.

Furthermore, CC plays a significant role in brain functions, including formation and development of inter-hemispheric sensorial connection, memory functions, regulation of attention span, restitution events in brain damage, and providing visibility in unilateral writing difficulties (1,9,10). Review of the literature shows that the clinical findings vary according to accompanying abnormalities (6). Epilepsy occurs in 50% of the cases, while the disease may also represent with mental retardation, growth retardation, learning difficulties, eating disorders, schizophrenia, and microcephaly (2,11,12). Mental retardation is often identified at school term (2). In a study performed by Alkan et al. (13) in 160 cases with callosal dysgenesis, agenesis was noted in 22, while the most common clinical findings were convulsion (40.9%), microcephaly (27.2%), and growth retardation (22.7%). In another study, Taylor et al. (14) reported that convulsion was the most frequent clinical finding in 50% of CCA cases. Our case, who is currently six-month-old, did not develop any pathological condition during follow-up. In addition, diagnosis of CCA is based on brain imaging (ultrasound transfontanellar, computed tomography, MRI). Brain imaging rules out other abnormalities. Today, antenatal diagnosis of the malformation is commonly performed by antenatal USG and MRI. It is, however, very difficult to give a prenatal counseling (15,16). Similarly, diagnosis was made by USG and MRI in our case.

Treatment of CCA is symptomatic and the treatment is tailored individually based on the clinical status of the patient. Anti-epileptic treatment should be preferred in case of seizures, while psychotherapy should be given in the presence of psychomotor disorder (17). As our case had an isolated disease, she is being monitored clinically.

In conclusion, ACC is a common brain abnormality. As in our case, non-invasive and practical cranial USG in high-risk infants is of utmost importance for early diagnosis, monitoring, and treatment of such abnormalities.

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Figure 1:MRI of the patient agenesis of the corpus callosum. sagittal image

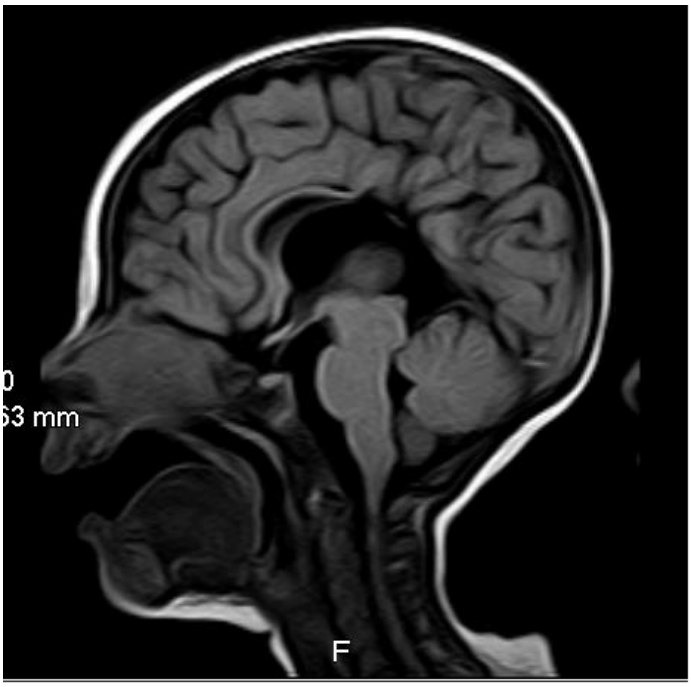

Figure 2 :MRI of the patient agenesis of the corpus callosum. axial image