Research Paper Medical Science

Corpus Callosum Agenesis with Interhemispheric Cyst Associated Foot and Face Anomalies: Radiological Study

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ABSTRACT

In case of interhemispheric cyst the characteristic imaging triad of asymmetric ventriculomegaly, a large interhemispheric cyst, and partial or complete agenesis of the corpus callosum (ACC) is seen. Most cases were initially referred as aqueduct stenosis and hydrocephalus or focal porencephaly. We describe the imaging findings that identify an absent corpus callosum associated with interhemispheric cyst in newborn initially thought to have hydrocephalus attributable to aqueductal stenosis. The presented case showed mildly symmetric ventriculomegaly on sonography. Computed tomography (CT) imaging was performed. Associated foot and facial abnormalities are also reported. Technological improvements in sonography, CT and magnetic resonance imaging (MRI) allow improved characterization of associated intracranial anomalies in the setting of hydrocephalus. Accurate diagnosis can aid parental counseling, especially because isolated aqueductal stenosis suggests a better prognosis than hydrocephalus with anomalies. Markedly asymmetric ventriculomegaly in most of cases was the key to excluding isolated aqueductal stenosis and was associated with callosal malformation with a type 1a interhemispheric cyst. The presented case is associated with mildly symmetrically ventriculomegaly with complete agenesis of the corpus callosum and midline large interhemispheric cyst instead of markedly asymmetric ventriculomegaly. We also highlighted the associations of foot and facial anomalies.

KEYWORDS Aqueductal stenosis, Corpus callosum, Hydrocephalus, Interhemispheric cyst, Ventriculomegaly

INTRODUCTION

The origin of the interhemispheric cyst in agenesis of the corpus callosum (ACC) is controversial. Neurenteric, arachnoid, and ependymal cysts have all been suggested as possible causes. The cyst developed in concert with increasing ventricular size, which suggests that the cyst is a form of communicating hydrocephalus and is an ependymal-lined cyst.[1]

In agenesis of corpus callosum involvement of organs and apparatus other than the central nervous system was present in 65% of the cases. Cranio-facial abnormalities (65%), and skeletal abnormalities (31%), were present to various degrees. Cranio-facial abnormalities involved: the skull conformation (macrocephaly, trigonocephaly); the ocular region (hypertelorism, broad and depressed nasal bridge); the palate (high arched palate and cleft palate); the oral region (cleft lip, macrostomia). Skeletal abnormalities comprised: hand malformations brachydactyly, absent proximal and distal phalanges, clinodactyly of the fifth fingers); cervical spine anomalies (scoliosis, hyperkyphosis); feet abnormalities (bilateral club foot, valgus foot, bilateral syndactyly of the second/third toes).[2]

In the presented case of corpus callosum agenesis with association of large interhemispheric cyst and mild symmetrical ventriculomegaly associated with right valgus foot and prominent nose.

STUDY:

A newborn female baby came in our radio-diagnosis department with history of premature delivery. Baby weight was 900 grams. On inspection face was abnormal with prominent nose [Figure 1a and 1b] and lower limbs also showed right valgus foot [Figure 2]. On ultrasound examination, we found large midline interhemispheric cyst with absence of corpus callosum and symmetrically mild enlarged both lateral ventricles. Third ventricle was directly communicated with large midline interhemispheric cyst [Figure 3a, 3b, and 3c] Cerebellum, brainstem and fourth ventricle were normal. No evidence of macrocephaly and depressed nasal bridge seen. Abdominal ultrasound was normal.

CT scan of brain of newborn was also obtained and the above said findings were corroborated by the images [Figure 4a, 4b and 4c].

DISCUSSION

Malformation of the corpus callosum is known to be associated with interhemispheric cysts and an absent cavum septi pellucidi.[3,4,5,6,7] Barkovich et al[3] classified interhemispheric cysts as type 1 or 2, with subcategories in both. The classification of Barkovich et al was based on pediatric imaging.

Embryologically, communicating cysts are expansions of the ventricular tela choroidea, which is detached from the thalamus on one side (or both if the findings are bilateral).[8] The underlying cause is not certain but could be related to a meningeal disorder. We hypothesize that the presence of the ventricular diverticulum/cyst is the initial abnormality that disrupts the normal cellular cues for directional migration of the commissural fibers in the developing brain. This process hypothetically leads to incomplete or failed formation of the corpus callosum. Given the male predominance, there may be an underlying, as yet unknown, genetic abnormality.[9] But our case was female child.

Markedly asymmetric ventriculomegaly was the initial and key sonographic finding; careful additional imaging led to recognition of an absent cavum septi pellucidi, callosal malformation, and an interhemispheric cyst. In all cases, macrocephaly increased steadily throughout pregnancy, and in few cases, skin-covered fontanelle meningoceles developed. These meningoceles protruded through the sutures in the region of the anterior or posterior fontanelles, likely from increased intracranial pressure. Features of Chiari I malformation can be present, presumably due to downward pressure on the tentorium and posterior fossa from the enlarging ventricles.[9] In our presented case ventriculomegaly is mild and symmetrically. Features of Chiari I malformation absent and head size is within normal limit.

Bedeschi M F *et al.*[2] performed clinical and genetic study in 63 young patients in agenesis of corpus callosum. They evaluated the agenesis of the corpus callosum with other central nervous

system (CNS) malformations and other than CNS malformations. They observed 53% partial genesis and 47% complete agenesis with slightly male prevalence. Other associated CNS anomalies pachygyric or heterotopic areas, septo-optic dysplasia, frontal operculi hypoplasia, schizencephaly, frontal lissencephaly, hydrocephalus, interhemispheric cysts, holoprosencephaly, cerebellar vermis and hemisphere hypoplasia were observed. An involvement of organs and apparatus other than the CNS was present in 65% of the cases. Cranio-facial abnormalities involved: the skull conformation (macrocephaly, trigonocephaly); the ocular region (hypertelorism, broad and depressed nasal bridge); the palate (high arched palate and cleft palate); the oral region (cleft lip, macrostomia). Skeletal abnormalities comprised: hand malformations (brachydactyly, absent proximal and distal phalanges, clinodactyly of the fifth fingers); cervical spine anomalies (scoliosis, hyperkyphosis); feet abnormalities bilateral club foot, valgus foot, bilateral syndactyly of the second/third toes).

In our presented case is female, no macrocephaly, no hypertelorism, and no broad and depressed nasal bridge, no other CNS malformations showed. We observed agenesis corpus callosum with large midline interhemispheric cyst and mild symmetrical ventriculomegaly associated with right valgus foot and prominent nose, instead of markedly asymmetrical ventriculomegaly with lateral/ midline large interhemispheric cyst and depressed nasal bridge, which was were commonly associated with agenesis of corpus callosum.

CONCLUSION

Although agenesis corpus callosum with large midline interhemispheric cyst and markedly asymmetrical ventriculomegaly are not rare malformation, but agenesis corpus callosum with large midline interhemispheric cyst, mild symmetrical ventriculomegaly, feet abnormalities (right valgus foot) are rare malformation. The presented study also highlightened the association prominent nose malformations in female newborn, which was proven to be a rare study.

Legends

1a

1b

Figure 1a and 1b: Photograph newborn face showing prominent nose.

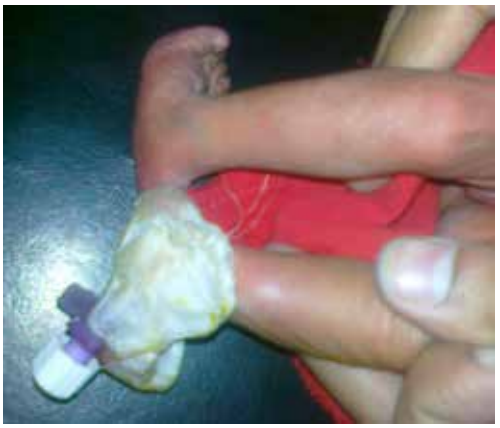
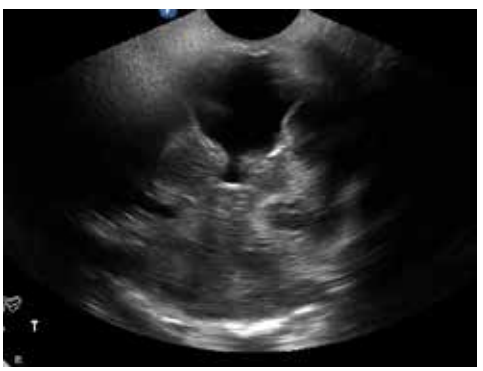
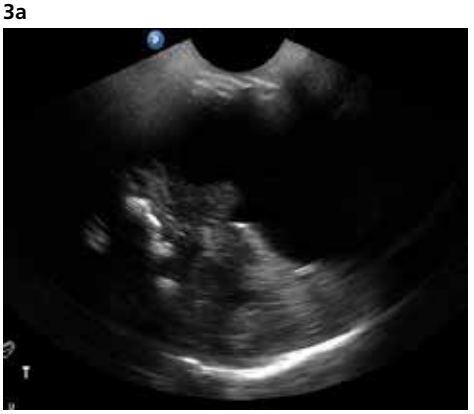


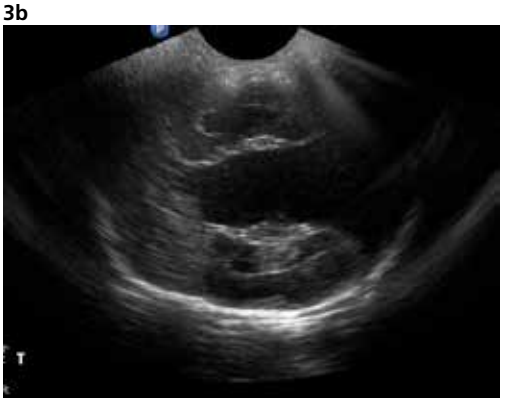
Figure 2: Photograph newborn lower limb showing right club foot.



3a

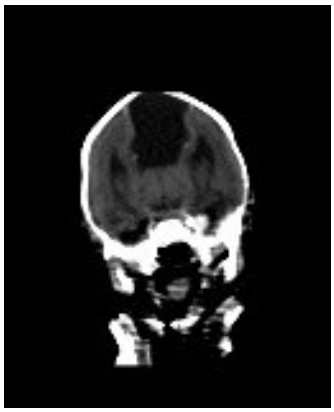


3b

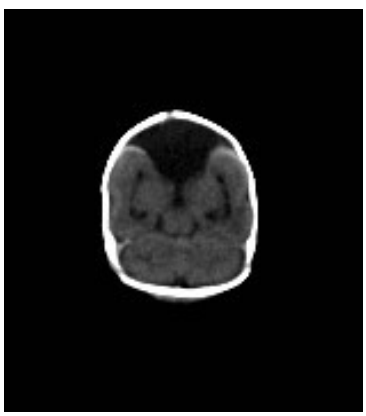


3c

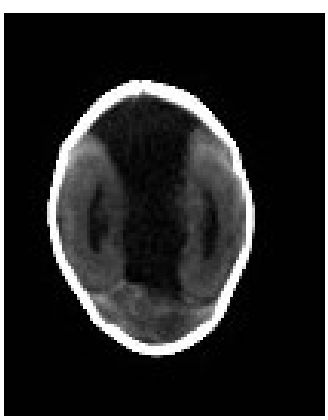
Figure 3a, 3b and 3c: Coronal, sagittal and axial ultrasound images showing large midline interhemispheric cyst with absence of corpus callosum and symmetrically mild enlarged both lateral ventricles.



4a



4b



4c

Figure 4a, 4b and 4c: Coronal and axial CT images showing large midline interhemispheric cyst with absence of corpus callosum and symmetrically mild enlarged both lateral ventricles.

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