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Sonographic Appearance of Callosal Agenesis: Correlation with Radiologic and Pathologic Findings

Marta Hernanz-Schulman¹
F. C. Dohan, Jr.²
T. Jones³
P. Cayea³
J. Wallman¹
Rita L. Teele¹

Sonographic features are described in six infants in whom total or partial agenesis of the corpus callosum was confirmed by either computed tomographic or pathologic examination. The six patients demonstrated a range of abnormalities involving the neuraxis as well as other systems, notably cardiovascular, gastrointestinal, and genitourinary. Chromosomal abnormalities were present in two patients. The sonographic features of callosal agenesis seen in these patients included: lack of characteristic acoustic interfaces to define the corpus callosum on both coronal and sagittal sonograms; increased separation and parallelism of the bodies of the lateral ventricles; loss of the characteristic convexity of the medial border of the anterior horns of the lateral ventricles; variable prominence of the occipital horns of the lateral ventricles; variable degree of superior extension of the third ventricle; alteration or absence of the cavum septi pellucidi; and radial arrangement of cerebral sulci about the prominent third ventricle. Cases of partial agenesis may show the dysplastic features found in complete agenesis. However, only some of the callosal echoes are present. The sonographic features of partial agenesis in one infant had not been described before.

Pneumoencephalographic and computed tomographic (CT) findings of agenesis of the corpus callosum are well known, but its sonographic features have not been described as well or as often in the English literature. We report six cases of agenesis of the corpus callosum together with radiologic and pathologic correlation. The pathogenesis and implications of this lesion are reviewed briefly.

Materials and Methods

Six patients with callosal agenesis were recently evaluated at our institutions. In four, the initial diagnosis was made sonographically and later confirmed by either CT or pathologic examination. Two patients were examined with sonography after the diagnosis had been made by CT.

Sonography was performed in the coronal and sagittal planes using real-time equipment fitted with either a 6.0 or 7.5 MHz transducer. CT was performed with a General Electric 8800 scanner. The clinical records of all infants were reviewed and the clinical and genetic findings were recorded.

For reference, the usual sonographic appearance of the corpus callosum in the coronal and sagittal plane is illustrated in figure 1. Note that the corpus callosum is easily recognized as the relatively hypoechoic structure between two highly reflective acoustic interfaces, that is, the sulcus of the corpus callosum superiorly and the bodies of the lateral ventricles inferiorly. Note the similarity to the pathologic section illustrated in figure 1C (different patient).

Case Reports

Case 1

A male infant was the 3.95 kg product of a 34 week gestation. His size and weight were appropriate for gestational age. Head circumference was 39 cm (greater than 90th percentile).

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¹ Department of Radiology, Children's Hospital, 300 Longwood Ave., Boston, MA 02115. Address reprint requests to R. L. Teele.

² Department of Neuropathology, Children's Hospital, Boston, MA 02115.

³ Department of Radiology, Brigham and Women's Hospital, Boston, MA 02115.

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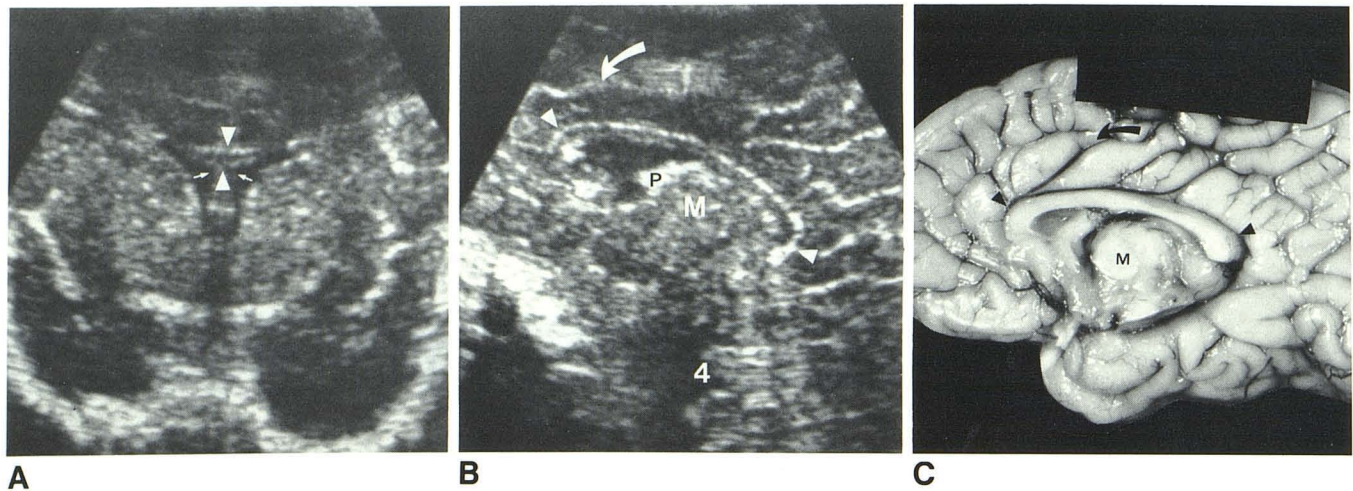


Fig. 1.—**A**, Coronal sonogram anterior to foramen of Monro in normal infant, 34 weeks gestation. Medial convexity of anterior horns of lateral ventricles (arrows), septum pellucidum between them, and reflective interfaces defining corpus callosum (arrowheads). **B**, Midline sagittal sonogram, same infant. Characteristic interfaces define corpus callosum (arrowheads), cingulate gyrus

(curved arrow), massa intermedia (M), choroid plexus (P), and fourth ventricle (4). **C**, Midline pathologic specimen in normal infant. Corpus callosum (arrowheads), cingulate gyrus (curved arrow), massa intermedia. Posterior fossa structures not included.

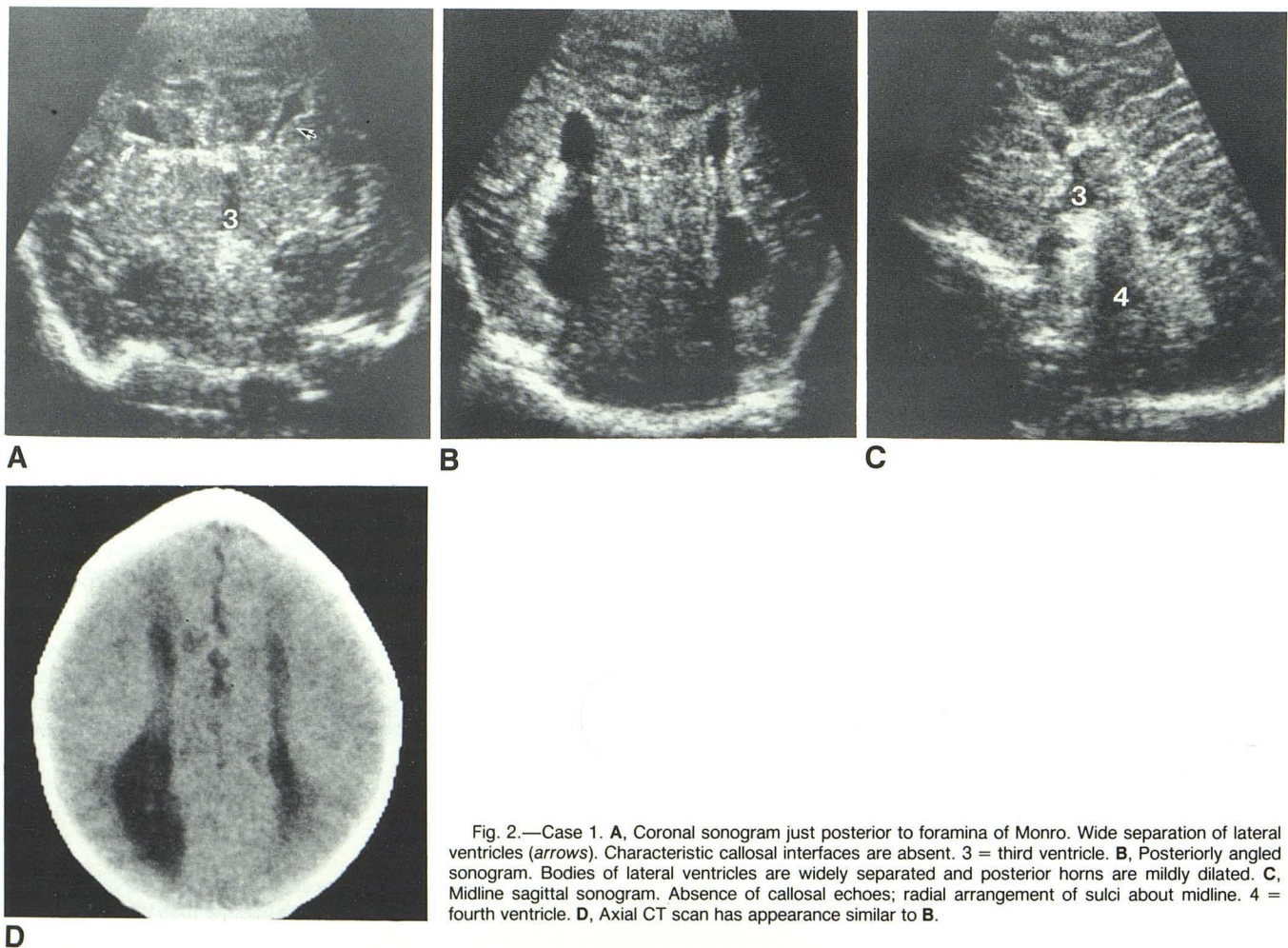


Fig. 2.—Case 1. **A**, Coronal sonogram just posterior to foramina of Monro. Wide separation of lateral ventricles (arrows). Characteristic callosal interfaces are absent. 3 = third ventricle. **B**, Posteriorly angled sonogram. Bodies of lateral ventricles are widely separated and posterior horns are mildly dilated. **C**, Midline sagittal sonogram. Absence of callosal echoes; radial arrangement of sulci about midline. 4 = fourth ventricle. **D**, Axial CT scan has appearance similar to **B**.

At birth he was noted to have dysmorphic features characterized by trigonocephaly, flat nasal bridge, hypertelorism, and abnormal segmentation of the sternum and omphalocele. Many of his dysmorphic characteristics have been associated with deletion of the short arm of chromosome 9 (46, 9p-). Genetic examination, however, showed a reciprocal translocation between 3p and 9p. Sonographic and CT findings are shown in figure 2.

Case 2

A male infant was the 3.24 kg product of a 39 week gestation. Prenatal sonography had indicated an abnormally large head for the calculated gestational age. At birth his head circumference was at the 98th percentile. Hypertelorism was present. Electroencephalo-

graphic findings were consistent with agenesis of the corpus callosum. Sonographic and CT findings are shown in figure 3.

Case 3

A 10-month-old boy from Colombia was adopted and his perinatal history was not known. He had a large frontal encephalocele and extremely dysmorphic features with marked hypertelorism. His sonographic and CT findings are shown in figure 4.

Case 4

A male infant was the 2 kg product of a 33 week gestation. At birth he was noted to have an omphalocele, 13 ribs, and a butterfly

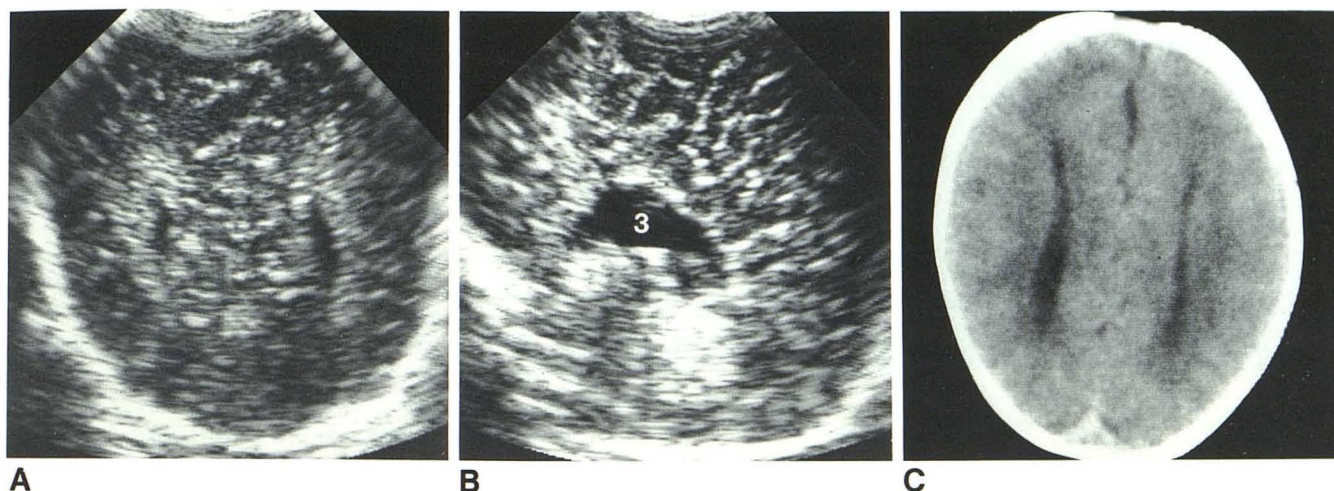


Fig. 3.—Case 2. **A**, Steeply angled coronal sonogram. Parallelism and wide separation of posterior horns of lateral ventricles. **B**, Midline sagittal sonogram. Callosal echoes are absent. Third ventricle (3) is mildly dilated, with gyri

arranged radially around its superior border. **C**, CT scan is similar in orientation to **A** and shows similar features.

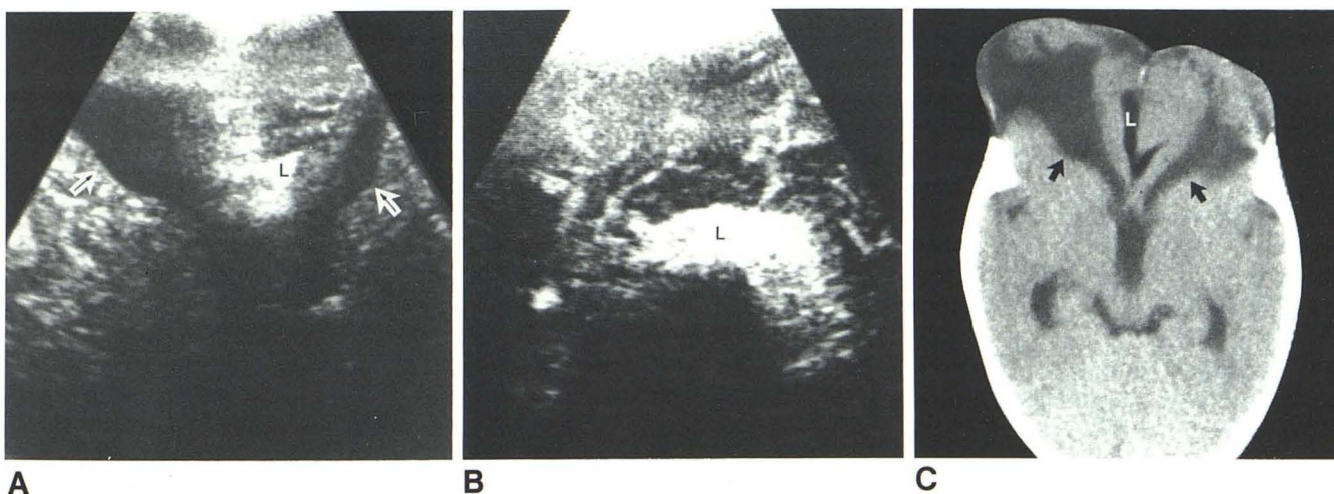


Fig. 4.—Case 3. **A**, Coronal sonogram through anterior encephalocele. Inferolateral border of dilated anterior horns of lateral ventricles (arrows). Echogenic midline mass represents lipoma (L). Right frontal horn is more ectatic than left. **B**, Midline sagittal sonogram. No corpus callosum is identified.

Echogenic midline lipoma prevents imaging of deeper structures. **C**, CT scan. Inferolateral border of lateral ventricles (arrows) corresponds to **A**. Concavity of medial walls of ventricles.

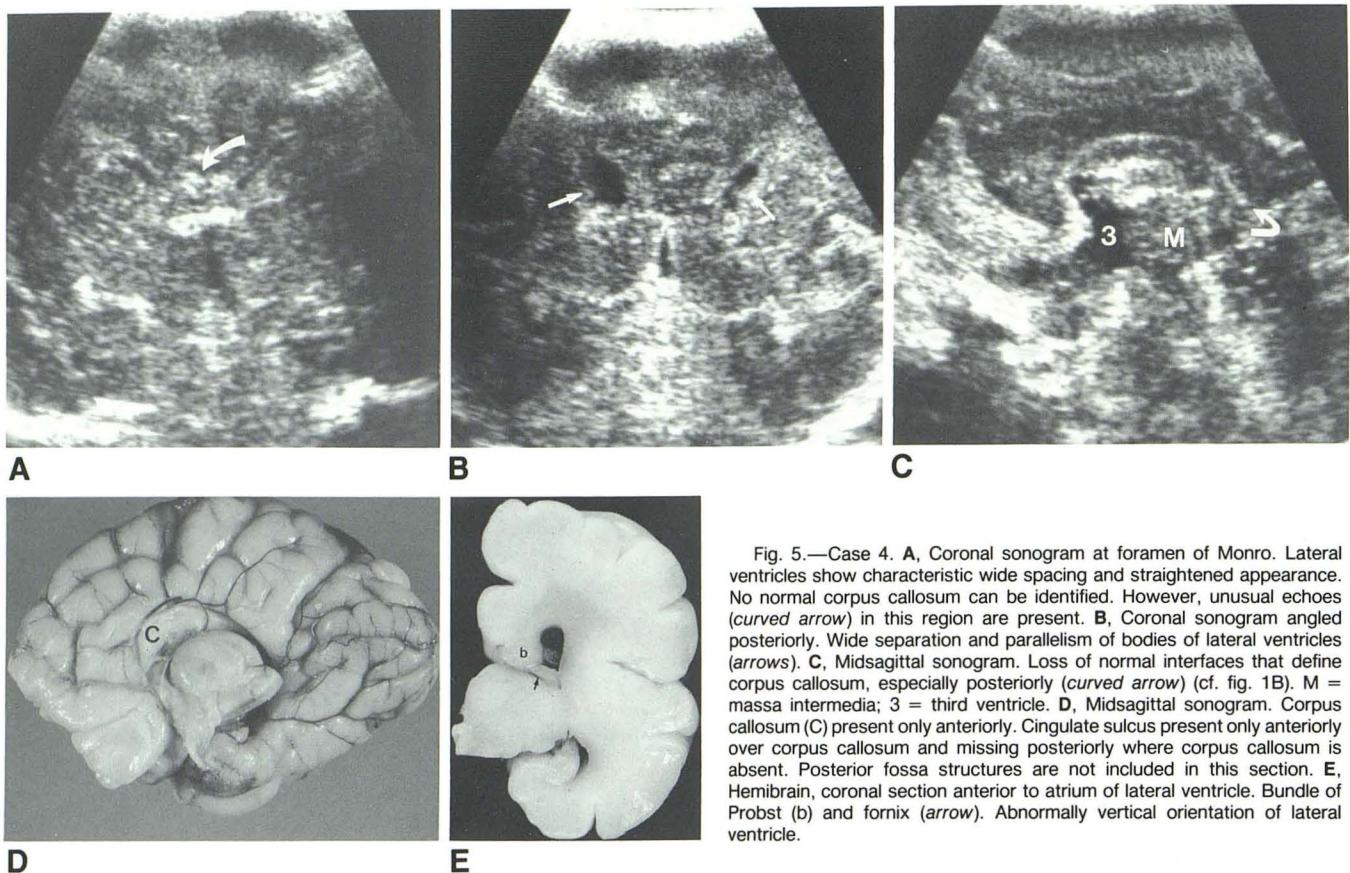


Fig. 5.—Case 4. **A**, Coronal sonogram at foramen of Monro. Lateral ventricles show characteristic wide spacing and straightened appearance. No normal corpus callosum can be identified. However, unusual echoes (curved arrow) in this region are present. **B**, Coronal sonogram angled posteriorly. Wide separation and parallelism of bodies of lateral ventricles (arrows). **C**, Midsagittal sonogram. Loss of normal interfaces that define corpus callosum, especially posteriorly (curved arrow) (cf. fig. 1B). M = massa intermedia; 3 = third ventricle. **D**, Midsagittal sonogram. Corpus callosum (C) present only anteriorly. Cingulate sulcus present only anteriorly over corpus callosum and missing posteriorly where corpus callosum is absent. Posterior fossa structures are not included in this section. **E**, Hemibrain, coronal section anterior to atrium of lateral ventricle. Bundle of Probst (b) and fornix (arrow). Abnormally vertical orientation of lateral ventricle.

vertebra at T5. His facies was described as dysmorphic with hypertelorism. Radiographic studies revealed dysplastic kidneys with vesicoureteral reflux; there were complex cardiac defects as well. Death followed oliguric renal failure at 6 days of age. Sonographic findings are shown in figures 5A–5C.

Postmortem examination demonstrated brachycephaly, microphthalmia, absent philtrum, congenital mitral stenosis, two ventricular septal defects, multiple atrial septal defects, hypoplastic lungs, dysplastic kidneys, and absent right radius. Neuropathologic examination of the brain revealed partial agenesis of the corpus callosum with absence of the posterior third. A bundle of Probst was identified posteriorly on each side, beginning at the caudal margin of the corpus callosum. The lateral ventricles were normal in shape anteriorly, but were widened and shortened posteriorly, with an abnormally vertical orientation. The fornices were normal in size although slightly malformed, and, in the region of the absent corpus callosum, they were situated more laterally than usual. The genu and posterior limb of the internal capsules were malformed and both the cerebral peduncles and the corticospinal tracts were slightly reduced in size. Multiple areas of gray-matter heterotopia were present in the cerebellar white matter. Figures 5D and 5E illustrate the pathologic sections obtained in this infant, which correlate well with the sonograms.

Case 5

A female infant was the 2.1 kg product of a 38 week gestation that was remarkable for the presence of hydramnios. At birth the

infant was noted to have an omphalocele and other dysmorphic features consistent with those found in trisomy 18. These included growth retardation (less than 10th percentile for height, weight, and head circumference), microphthalmia, micrognathia, hypertelorism, low-set ears, esophageal atresia with distal tracheoesophageal fistula, and camptodactyly involving several fingers. Genetic studies were diagnostic for trisomy 18 47XX. Postmortem examination in addition demonstrated choanal stenosis, renal cortical polymicrocystic changes, and tetralogy of Fallot with pulmonary infundibular and valvar atresia. Neuropathologic examination of the brain demonstrated complete agenesis of the corpus callosum as well as a somewhat simplified gyral pattern, small superior temporal gyri, malformed hippocampi, mild hypoplasia of the cerebellum, and a small basis pontis. A bundle of Probst was present on either side. The fornices were normal in size. Anteriorly, the lateral ventricles had an abnormally vertical orientation and were somewhat narrowed and shortened. There was moderate widening of the atria and the temporal and occipital horns. Figure 6 shows the sonographic and pathologic appearance of the brain in this case.

Case 6

A female infant was the 3 kg product of a 37 week gestation. At birth she was noted to have an omphalocele and macrocephaly with a head circumference (40.7 cm) greater than 2 SD above the norm. Additional diagnostic workup revealed bilateral inguinal hernias, an atrial septal defect secundum, normal 46XX chromosomal complement, and retinal discolored areas, which raised the possibility of

Aicardi syndrome. A sonographic (fig. 7) and CT study of the head revealed absence of the corpus callosum with a large interhemispheric paramedian cyst and ventricular dilatation.

Results

We identified nine sonographic signs of callosal agenesis in our six cases: (1) lack of the characteristic acoustic interfaces that define the corpus callosum; (2) wide separation of the bodies of the lateral ventricles; (3) parallel orientation of the lateral ventricles; (4) loss of the characteristic convexity with straightened appearance of the medial walls of the lateral ventricles (in some cases, this may actually produce a concave appearance of the medial borders of the anterior horns); (5) variable dilatation of the occipital horns of the lateral ventricles; (6) variable superior extension of the third ventricle; (7) alteration or absence of the septum pellucidum; (8) loss of the C-shaped conformation of the cingulate gyrus; and (9) radial arrangement of the sulci around the roof of the third ventricle. We found that the degree of superior extension of the third ventricle was quite variable and sometimes not prominent. Only one of our patients (case 3) showed the medial hornlike concavity of the anterior lateral ventricles as seen on pneumoencephalography and coronal CT. However, there was loss of the normal medial convexity in all of our cases.

Findings such as angular dorsal margins of the lateral ventricles and elongation of the foramina of Monro, which are seen on pneumoencephalography, cannot be appreciated sonographically. However, sonography affords direct demonstration of the entire corpus callosum on a single sagittal section. When it is absent, the morphologic alteration of the cingulate gyrus and the cerebral sulci is best appreciated sonographically. In instances of partial agenesis, illustrated by case 4, the absence of only the posterior part of the

structure may be appreciated along with many of the morphologic alterations seen when agenesis is total.

Discussion

Embryologic Overview

At about 7 weeks of gestation [1], the primitive midline lamina terminalis is invaded by cells from the right and left septal area [2]. This cellular, thickened ridge in the lamina terminalis becomes the commissural plate, which provides a bridge through which nerve fibers can cross the midline from one hemisphere into the other (fig. 8). The fibers group

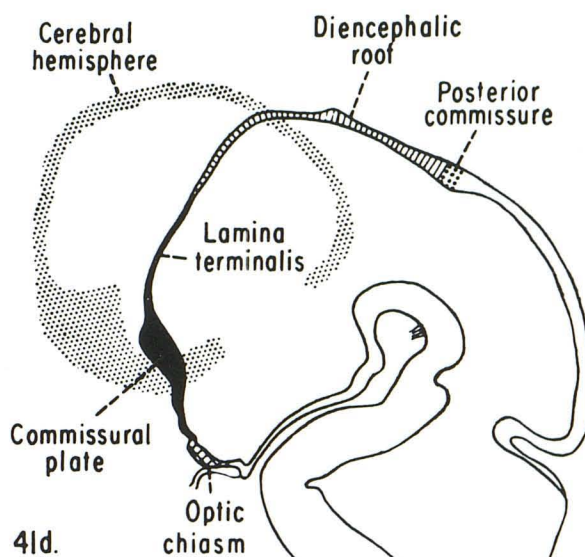
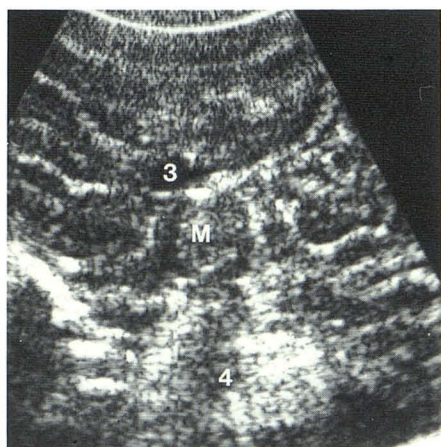
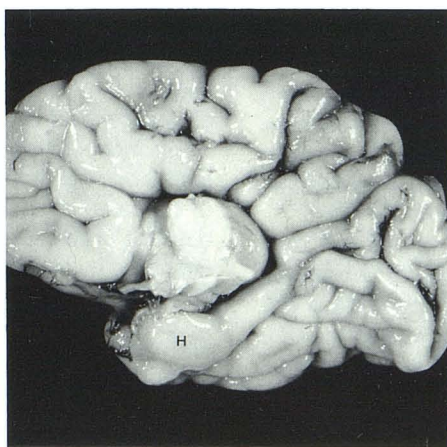


Fig. 8.—Sagittal section in 41-day-old embryo. (Reprinted from [3].)



A



B

Fig. 6.—Case 5. **A**, Midsagittal sonogram. Complete lack of callosal echoes, high position of third ventricle, and lack of definition of cingulate gyrus. M = massa intermedia; 4 = fourth ventricle. **B**, Midsagittal pathologic specimen similar to **A**. Posterior fossa structures are not included, enabling demonstration of malformed hippocampus (H).



Fig. 7.—Case 6. Coronal sonogram. Lack of callosal echoes, wide separation of anterior horns (arrows), and large midline cyst (C). Shunt catheter (arrowhead).

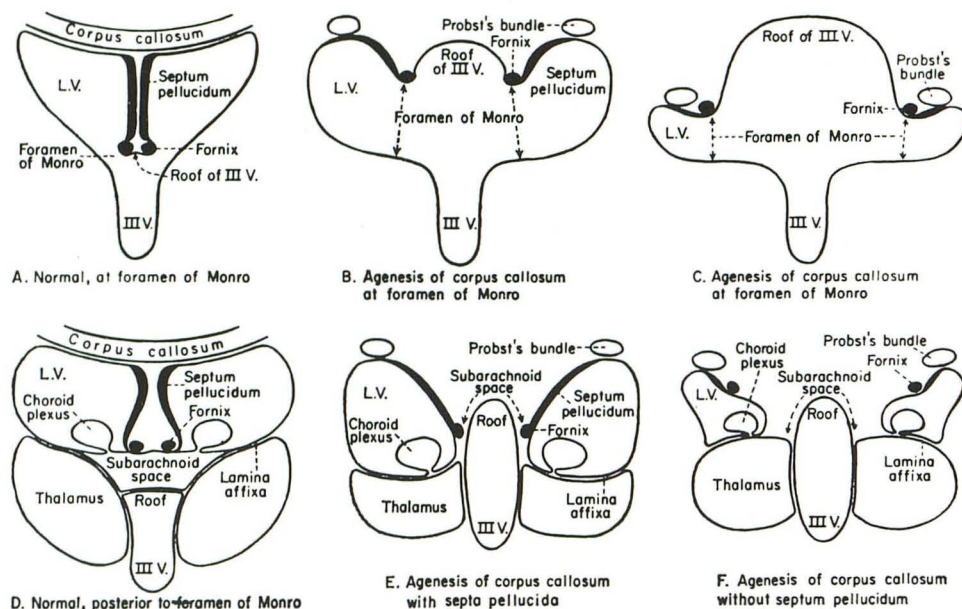


Fig. 9.—Coronal sections of normal and acallosal brains. L.V. = left ventricle; III V. = third ventricle. (Reprinted from [3].)

themselves into three divisions: the anterior commissure, the hippocampal commissure, and the corpus callosum itself.

The corpus callosum begins its formation at about 12 weeks of gestation and has achieved its adult morphology by 18–20 weeks [1]. The earliest callosal fibers lie anterior and inferior to the foramen of Monro. As more fibers join in, the corpus callosum grows posteriorly in a C shape over the diencephalon.

Pathologic-Sonographic Correlation

In most instances of callosal agenesis there is an aberrant bundle of uncrossed fibers named the bundle of Probst, which was first observed by Onufrowicz in 1887 and was described in detail by Probst in 1901 [4]. It takes origin in the frontal white matter and continues along the medial walls of the roofs of the lateral ventricles to the posterior white matter (fig. 9). As described by DeLange [5] in 1925, when callosal agenesis is partial, this bundle is continuous anteriorly with the crossed fibers, its posterior course being unaltered from that in complete agenesis. This was true in our case 4. When examined microscopically, the fibers of this bundle seem to have a random termination and the total number of fibers is less than in the corpus callosum [6].

The anterior separation of the lateral ventricles and their characteristic anterior concave medial borders in callosal agenesis arises from hypertrophy of the cingulate gyrus [6]. The bundle of Probst may contribute to this separation [1].

With no corpus callosum above, the third ventricle extends superiorly to a variable degree. It is postulated that this is the embryonic anatomy and that genesis of the corpus callosum normally "pushes" the third ventricle inferiorly. When the former is absent, the latter remains in its embryonic high position [6]. This third ventricular extension serves to separate further the bodies of the lateral ventricles posterior to the foramina of Monro.

The characteristic dilatation of the posterior horns of the lateral ventricles is postulated to occur secondary to the absence of the splenium [3].

According to Loeser and Alvord [3], cystic degeneration of the central part of the commissural plate gives rise to the cavum septi pellucidi. The medial walls of the lateral ventricles are therefore a remnant of the commissural plate. Carried one step further, the septum pellucidum is the tissue that connects the fornix either to the corpus callosum or, in its absence, to Probst bundle [3, 6]. In agenesis, the septum is not maintained in its midline position by the corpus callosum. Instead, it is allowed to migrate laterally and becomes greatly attenuated or obliterated [3].

The radial arrangement of the sulci is postulated to occur secondary to the abnormal mechanical stresses at work when the corpus callosum is absent [1, 3], rather than to the persistence of an embryonic pattern, as suggested by some authors [7], since the radial arrangement of the sulci does not appear to be a fetal pattern [1]. This arrangement has been compared by Kirshbaum [8] with the drawing-in of gyri around a porencephalic cyst.

The inconstant pathologic finding of nonintersection of the parietooccipital and calcarine sulci along the medial surface of the cerebrum has been postulated to be secondary to the absence of the splenium [3]. This abnormality was not demonstrated by sonography in any of our patients.

Spectrum

First described by Reil in 1812 [7] and reviewed by Bruce in 1899 [6, 9], agenesis of the corpus callosum may be partial or complete. When partial, it nearly always involves absence of the posterior part of the structure (as in case 4), since embryologically the corpus callosum grows in a rostrocaudal direction. If only the splenium is absent, the leaves of the septum pellucidum may be present but deviated laterally and

there may be superior extension of the pineal recess of the third ventricle [1]. When callosal agenesis is complete, the anterior and hippocampal commissures may be absent as well.

Callosal agenesis occurs hereditarily in the mouse, *Mus musculus* [10]. In humans, agenesis of the corpus callosum in any of its forms usually occurs sporadically with a cited frequency of about 2.3% of mentally retarded persons [1]. Jellinger et al. [11] reported a 0.7% incidence found incidentally at pneumoencephalography performed for other reasons. Familial cases have been reported and autosomal recessive inheritance has been suggested [12]. Menkes et al. [13] reported callosal agenesis in one family, where it followed a gender-linked recessive pattern. Callosal agenesis is also a component of other syndromes inherited in diverse fashions, for example, leprechaunism, which appears to follow an autosomal recessive mode of inheritance [1]. In addition, it is a common component of the phenotypic manifestations of certain chromosomal abnormalities, notably trisomy 13, 18, and 8 [1]. Our case 5 had a trisomy 18, and case 1 was considered to have a translocation involving the C group of chromosomes.

Agenesis of the corpus callosum is considered to be a malformation distinct from the holoprosencephalies [14], although in the more severe forms of the latter, the corpus callosum is necessarily absent [1, 4]. Both malformations have been reported in the fetal alcohol syndrome [15]. Trigonocephaly and midline facial abnormalities may be found in both groups of patients with a lower frequency in cases of callosal agenesis (37.8%) than in the holoprosencephalies (92.8%) [11]. The craniofacial abnormalities found in the latter group tend to be more severe as well and include such conditions as cebocephaly and cyclopia.

The literature to date suggests that most cases of callosal agenesis are associated with other intracranial abnormalities. Jellinger et al. [11], in a review of 50 autopsied cases of callosal agenesis, cited an overall 88% incidence of various cerebral malformations in these patients. Thirty-eight percent of the cases exhibited gyral dysplasias (agyria, polymicrogyria) and failure of migration (heterotopia). Our case 4 was found to have multiple areas of gray-matter heterotopia in the cerebellar white matter at autopsy. Encephaloceles are also found in cases of both partial and complete callosal agenesis. This malformation was present in our case 3. Interhemispheric cysts also occur, and our case 6 demonstrates the sonographic appearance in this context. A high incidence of non-nervous-system malformations was also reported by Jellinger et al., most often involving the cardiovascular, gastrointestinal, and genitourinary systems. Such malformations were present in our cases 1, 4, 5, and 6.

Case 3 illustrates the occurrence of midline mesenchymal tumors, notably lipomas, in callosal agenesis. These tumors are rare intracranially. When they occur, they usually arise in the corpus callosum, or its anatomic location if it is absent [16, 17]. The sonographic appearance of midline lipoma was reported recently [18].

Although patients with agenesis of the corpus callosum often exhibit a variety of neurologic symptoms related to the

various associated cerebral anomalies, uncomplicated callosal agenesis implies no functional impairment except that detected by designed tests of interhemispheric communication [19, 20].

In conclusion, when performing sonography in a neonate or infant suspected of callosal agenesis, it is essential to obtain good sagittal and coronal sonograms. The midline sagittal sonogram will show the absence of callosal interfaces and the altered contour and superior extension of the third ventricle with the characteristic radial arrangement of the sulci around its roof in a full-term or older infant in whom sulci are ordinarily visible. Careful search might demonstrate lack of intersection of the parietooccipital and calcarine sulci.

Coronal sonograms will again show absence of callosal echoes and separation of the lateral ventricles by the hypertrophied limbic system and bundle of Probst with consequent loss of convexity of the anterior horns. The usual appearance of the septum pellucidum will not be seen. The posteriorly angled axial scan will show the characteristic, though variable, dilatation of the posterior horns and the parallel orientation of the lateral ventricles.

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