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CASE REPORT

Callosal agenesis followed postnatally after prenatal diagnosis

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ABSTRACT **Callosal agenesis is a congenital brain anomaly caused by embryonal hypogenesis of the corpus callosum. Concerning the neurological prognosis, epilepsy and motor disturbance are noted in some cases, while many cases are asymptomatic and the prognosis is good. We report a fetus tentatively diagnosed with hydrocephaly on prenatal echoencephalography, which was performed without adequate explanation to and understanding of the parents. The parents had not expected an abnormality before the screening, and were subsequently not psychologically prepared for the discovery of the congenital brain anomaly on imaging. Moreover, they received no guidance on how to deal with any possible abnormalities. The pregnant mother was referred to our hospital. Prenatal MRI was performed after informed consent was obtained, and the fetus was diagnosed with callosal agenesis. The patient was followed for 5 years, and neurological development was normal. However, the parents have remained anxious while raising the child. Thus, the prenatal diagnosis of callosal agenesis in this case caused unnecessary mental burden to the parents. Here, we report the course of the case, and discuss the way prenatal ultrasonography should be used as a prenatal screening method, and the importance of counseling before the test.**

Key Words: callosal agenesis, fetal magnetic resonance imaging, genetic counseling, prenatal echogram, screening

INTRODUCTION

Accuracy in the diagnosis of callosal agenesis and hydrocephaly remains unclear. Many cases are asymptomatic, and diagnosis may not be made without radiological imaging in many cases. The disorder is incidentally detected on radiography of the brain performed for other purposes, suggesting that it is not rare. Thus, it should be kept in mind that prenatal ultrasonic screening may detect callosal agenesis and hydrocephaly even though the fetal development is normal. We encountered a case prenatally diagnosed with callosal agenesis by fetal MRI performed upon discovery of an abnormality on ultrasonography. Prenatal ultrasonic screening had been performed with inadequate informed consent. The parents had received a serious psychological impact from the echogram result, which had a negative impact on raising the child even though her neurological development was normal. Here, we report the clinical course of the case, and discuss prenatal echographic screening.

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CLINICAL REPORT

The patient was the mother of a 5-year-old girl who has been under counseling from before the delivery of the child because of anxiety concerning the development and raising of her child with a congenital brain anomaly. There was no problematic past or familial medical history. The mother had undergone ultrasonic screening at 31 weeks of gestation, and fetal hydrocephaly was tentatively diagnosed. Fetal development was normal and consistent with the gestational age on ultrasonography before 31 weeks, and the mother had not expected this abnormality. The mother was shown the echogram during the examination, and was shocked by the finding of congenital brain anomaly. She was subsequently referred to our hospital, and underwent ultrasonography. The lateral ventricle of the fetus was enlarged in a teardrop shape, and the corpus callosum could not be confirmed. After obtaining informed consent from the parents, prenatal MRI was performed at 34 weeks of gestation. Colpocephaly was noted in the fetal skull, the callosum was not imaged in the cerebrum, the bilateral ventricles were parallel, and the posterior horn was enlarged. Since the medial cerebral sulci radiated toward the 3rd ventricle in the sagittal view, callosal agenesis was diagnosed (Fig. 1A,B).

The child was born by natural delivery at 38 weeks of gestation with a birth weight of 2480 g, and the 1- and 5-minute Apgar Scores were 8 and 9, respectively. No anomaly was found on physical examination. Feeding was normal, and there was no problem in the development. However, the parents were very anxious about their child with a congenital abnormality of the brain. Thus, we periodically counseled the parents, and explained that the prognosis of callosal agenesis varies, and many cases are asymptomatic, although epilepsy and motor disturbance occur in some cases. The mother and infant were discharged three weeks after birth. At 3.5 months after birth, spindles were noted in electroencephalogram of the bilateral central regions, but no epileptic spikes were detected. The motor development of the infant was normal and consistent with the age: the neck stabilized at 3.5 months of age, tossing and turning appeared at 5.5 months, being able to maintain a sitting position at 8 months, and walking at one year and 1 month of age. Speech of single words appeared at one year of age. On MRI at two years and 2 months of age, callosal defect was noted, but myelination corresponded to the age (Fig. 1C,D). The child is now five years old; psychomotor and speech developments are normal. But the mother is still anxious about the child's future. We follow under periodic counseling with examination of the child.

DISCUSSION

The corpus callosum appears at approximately 10 weeks of gestation, and the development is almost complete at 17 weeks. Callosal agenesis occurs during this period (Kendall 1983). Thus, although

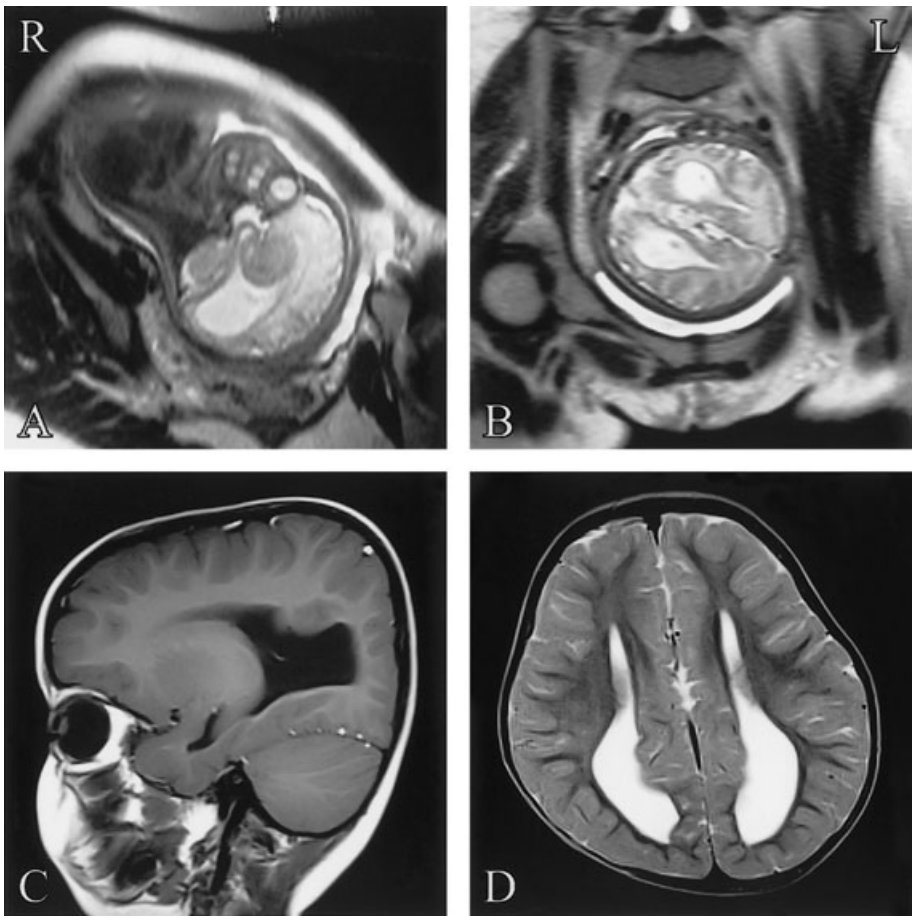

Fig. 1 Fetal magnetic resonance imaging (HASTE: half-Fourier acquisition single-shot trubo spin echo method: effective TE = 64.0/1 ms, TR = 4.4 ms, TA = 00:15, matrix 128 × 256, FOV 300–400 mm) showed agenesis of the corpus callosum (A) and colpocephaly (B). Magnetic resonance imaging. Sagittal T1 (right: spin echo: TE = 15, TR = 400 ms) and axial T2 (left: spin echo: TR = 3800 TE = 110) weighted-images. Myelination was consistent with her age (C, D).

ultrasonography is normal before 10 weeks, callosal agenesis may be diagnosed thereafter. Drugs, infection, cerebrovascular disorder, and trauma have been speculated to be the causes of callosal agenesis. Its prevention is not possible, and no therapy is available. Early diagnosis of callosal agenesis is important for the investigation of complications. A defect of the callosum alone may be noted, but it may also be complicated by congenital hydrocephaly, Dandy–Wakwer syndrome, Chari deformity, myelomeningocele, total frontal cerebral vesicle, agyria, encephalodysplasia, Aicardi syndrome (Jellinger *et al*. 1981). The degrees of symptoms and developmental disorders vary depending on the type of complicating anomalies and other cerebral disorders which may be present. Clinically, there is no callosal defect-specific symptom. Convulsive attacks, motor paralysis, and psychomotor developmental retardation may occur from the neonatal period, or diagnosis can be made based on epilepsy and various neurological abnormalities, whereas some cases present no clinical symptoms throughout their life, and the defect is incidentally diagnosed after adolescence in others. In this patient, motor and mental development was normal. However, the parents are still very anxious about raising their child, and under periodic counseling. If the prenatal ultrasonography had not been performed, the parents would not have such anxieties about raising their child. The early diagnosis resulted in a long-term negative effect on raising their child associated with their anxiety in this case.

Advances in prenatal MRI and ultrasonography have allowed early prenatal diagnosis of various disorders, which is of great medical benefit, enabling us to initiate fetal treatment from an early stage (Gupita & Liford 1995; Sonigo *et al*. 1998). However, parents under perinatal management may see an abnormal image of the fetus and become anxious. Prenatal imaging diagnosis is very problematic in that it may provide non-beneficial medical information and cause anxiety, and it may detect hereditary diseases. However, in actual practice, ultrasonography is performed as a screening tool to examine the gestation sac and fetal heart beats to confirm pregnancy in the early phase without adequate explanation of various disorders that may be incidentally discovered. The possibility of such findings should be explained before prenatal imaging, such as ultrasonography and fetal MRI, is performed, and the family should be counseled concerning possible unexpected findings including obtaining informed consent on whether or not the findings are to be disclosed.

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