Case Report

A rare case: agenesis of cerebellum

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Abstract

The absence of a cerebellum is extremely rare condition and the clinical presentation is variable. We aimed to present a case with isolated cerebellar agenesis in a pregnant woman at 23 weeks of gestation. Twenty seven years old primigravida patient was referred to our obstetrics outpatient clinic with a suspicion of cerebellar in her fetus. Cerebellum agenesis has been identified by ultrasound evaluation and magnetic resonance imaging. The family was informed about cerebellar agenesis. After counseling and taking written informed consent, pregnancy was terminated. In conclusion, a termination choice may be presented to a family with the early diagnosis of isolated cerebellar agenesis.

Key words: Cerebellar agenesis, magnetic resonance imaging, prenatal diagnosis, ultrasonography

Introduction

The cerebellum is located at the bottom of the brain. It is divided into three sections: an anterior lobe (above the primary fissure), posterior lobe (below the primary fissure) and flocculonodular lobe (below the posterior fissure). A narrow strip of protruding tissue along the midline is called the cerebellar vermis [1]. The cerebellum is a part of the brain that plays an important role in motor control. It may also be contained in some cognitive functions such as attention and language [2]. The cerebellum does not initiate movement, but it contributes to coordination. Cerebellar damage does not cause paralysis, but instead produces disorders in fine movement, equilibrium, posture, and motor learning. Posterior fossa disorders include classic Dandy-Walker malformation, Blake's pouch cyst, mega cisterna magna, and posterior fossa arachnoid cyst [3]. Agenesis of cerebellum is a rare disorder [4]. Cerebellar agenesis can be diagnosed prenatally with ultrasonography and magnetic resonance imaging (MRI) used in modern medicine. Cerebellar agenesis often is associated with meningomyelocele. Here, we have presented a case of isolated cerebellar agenesis.

Case presentation

A 27-year-old otherwise healthy primigravida at 23-weeks of gestation was referred to our hospital with suspicion of cerebellar pathology for a prenatal diagnosis. While ultrasound examination at 13 weeks had been considered as normal, cerebellum agenesis has been identified in the ultrasound in our clinic. Extracranial morphological examination and amniotic fluid volume were normal. Fetal MRI showed cerebellar agenesis (Figure 1). Parents were informed about the disease. After counseling and taking written informed consent, pregnancy was terminated using vaginal misoprostol (400 μg). A male fetus weighting 420 gram was aborted. After termination, genetic screening was performed from collected cord blood and karyotype was normal. The family did not want an autopsy.

Discussion

Cerebellar agenesis is the complete absence of the cerebellum regardless of the underlying cause. There is no known incidence because of its rarity. Isolated cerebellar agenesis is more seldom. Here is presentation of an isolated cerebellar agenesis.
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Posterior fossa disorders include classic Dandy-Walker malformation, Blake’s pouch cyst, mega cisterna magna, and posterior fossa arachnoid cyst [3]. Agenesis of cerebellum is a rare disorder [4]. Cerebellar agenesis can be diagnosed prenatally with ultrasonography and magnetic resonance imaging (MRI) used in modern medicine. Cerebellar agenesis often is associated with meningomyelocele. Here, we have presented a case of isolated cerebellar agenesis.

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Agenesis of the cerebellar is most often associated with motor movement disorder. Coordination of voluntary movements is impaired in affected individuals (ataxia). Walking may be delayed up to 4-7 years. Speech disorders can be seen due to the discoordination of the muscles associated with speech and nystagmus that can be seen due to the discoordination of eye muscle. Memory and mental disorders may be seen [5,6,7,8]. Cerebellar agenesis was found in men and women in equivalent proportions. Our patient was male fetus. Diagnosis of cerebellar agenesis is confirmed by neuroimaging while ultrasound and MRI is helpful in diagnosis in the midtrimester. In our case, cerebellar agenesis was first seen in the ultrasonographic evaluation in the 23 weeks of gestation, and then confirmed with fetal MRI study. As a result, a termination choice may be presented to a family with the early diagnosis of isolated cerebellar agenesis.

Conflict of interest statement

The authors declare no conflict of interest.

References