THE ULTRASONIC DIAGNOSIS AND DIFFERENTIAL DIAGNOSIS OF FETAL HYDRANENCEPHALY MALFORMATION

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ABSTRACT

To discuss the features of ultrasound images and the differential diagnosis of fetal hydranencephaly malformation. First, analyze the images of twelve cases of fetal hydranencephaly malformations diagnosed by prenatal ultrasound examination, then, summarize the key points of ultrasonic diagnosis of the fetal hydranencephaly malformations. Combining the misdiagnoses, put forward the differential diagnosis of fetal hydranencephaly malformation and severe hydrocephalus, alobar holoprosencephaly and porencephalia. Among the twelve cases of fetal hydranencephaly malformations, the ultrasound images all show that the cranial cavities are filled with liquid, large anechoic areas, no echo from cerebral hemisphere, thalamus and brainstem can be seen. Eight cases are cerebral falx strip-shaped high echo, four cases are no cerebral falx echo. Six cases can see few cerebral cortex in occipital region. Three cases are hydramnios. Among the twelve cases of fetal hydranencephaly malformations, nine cases are correctly diagnosed, two cases are misdiagnosed as holoprosencephaly, and one case is misdiagnosed as severe hydrocephalus. All three misdiagnosed cases are all diagnosed as hydranencephaly malformations after the ultrasound reexaminations. The key points of examining the fetal hydranencephaly malformations are observing whether there are cerebral hemisphere structures after finding the large anechoic areas in cranial cavity. If only pay attention on the image features and do differential diagnosis with similar diseases like severe hydrocephalus, alobar holoprosencephaly and porencephalia, it is not difficult to get the right diagnosis.

Key words: Ultrasound diagnosis; Fetus; Hydranencephaly malformation; Identification; Hydrocephalus; Holoprosencephaly; Porencephalia.

INTRODUCTION

Fetal hydranencephaly malformation is known as hydrocephalus. The hydrops is in the cranial cavity, not the ventricle. It is a kind of fatal brain abnormality that lack of the both side brain hemisphere, but has the thalamus, brainstem and the ependymal. Cruveilhier found this disease and reported it in 1829, then Spielmeyer named it hydranencephaly malformation.

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Since then, the cases of hydranencephaly malformation became more and more (Jia Guangyi et al., 2003; Liu Mingyu et al., 1995; Lu Jingguang et al., 2002; Mori K, 1985; Shi Zhong xian et al., 1997; Sarwar M, 1985; Su yongneng et al., 1999; Wang Shicheng et al., 2004; Yang Fangyu et al., 2007; Yu Xuesu et al., 2001). Through most documents and reports, the occurrence rate of hydranencephaly malformation is 0.05~0.2% (Mori K, 1985; Sarwar M, 1985. In this article, there are twelve cases of hydranencephaly malformation. The key point is to analyze the features of the ultrasound images and do the differential diagnosis with related malformations.

MATERIALS AND METHODS

Objects of study
Between January 2010 and December 2014, there are 35670 cases of prenatal ultrasound diagnosis on middle and terminal pregnancy in our hospital. Among these cases, there are twelve cases of hydranencephaly malformation. The ages of the pregnant women are among 22 to 35, the average is 26. Nine cases of them are unipara, three are multipara. The earliest gestational week is 11+4 w, the latest is 24w, and the average is 16w.

**Instruments and methods**

Using Color Doppler diasonograph, including GE Logiq E9, Voluson E8, Voluson 730 expert, Philips iu 22, the frequency of the probe is 3.0 ~ 5.0 HMz. Regularly observe the fetus, placenta, amniotic fluid and umbilical cord while the pregnant woman is in supine position. Acquire the growth and development situation of the fetus, measure the biparietal diameter, head circumference, femur length, max anteroposterior diameter of the amniotic fluid amount (midtrimester pregnancy), AFI (late pregnancy), and measure the S/D value of umbilical arterial blood, drag index and pulsatility index. During the brain examination of the fetus, we use the transaction, sagittal section and coronal section. After finding the abnormal in the fetal brain, we keep the ultrasound images of every section.

**RESULTS**

The ultrasound examinations show that there are twelve cases of hydranencephaly malformation, and further confirmed by CT, MR and dissection after induced labour. Through the ultrasound examination, twelve cases all show that the cranial cavity is full of liquid, with large anechoic areas. There is no echo from cerebral hemisphere outside the anechoic areas and the thalamus and brainstem can be seen. The thalamus and brainstem inburst into anechoic areas (See Picture One). Eight cases show cerebral falx strip-shaped high echo. Four cases show no cerebral falx echo. Six cases can see few cerebral cortex in occipital region (See Picture Two). Three cases are hydramnios. One case is thickened nuchal translucency. Among the twelve cases of fetal hydranencephaly malformations, nine cases are correctly diagnosed by the first ultrasound examination, two cases are misdiagnosed as alobar holoprosencephaly, and one case is misdiagnosed as severe hydrocephalus. All three misdiagnosed cases are all diagnosed as hydranencephaly malformations after the ultrasound reexaminations before induced labour.

**DISCUSSION**

Hydranencephaly malformation is the result of brain damaging. The earliest occurrence is 6 weeks in gestation, and the general occurrence is after 11 weeks gestation. This disease is sporadic and occasionally shows the feature of familial inheritance. But the aetiological agent is still not clear now. It is commonly believed that it is related with the obliteration of cervical internal carotid artery or bilateral arteries in brain. The obliteration of artery causes progressive cerebral infarct, then liquefying and absorbing. Frontal lobe, temporal lobe, parietal lobe and partial occipital lobe are filled with cerebrospinal fluid. It happens in the process of the fetal development. The malformation of the internal carotid causes the obstacle of the blood circulation. Or the blood-supply of internal carotid is influenced by some unknown reasons. For example, cord around neck or the formation of thrombus in internal carotid.

The above viewpoint is also supported by the pathological change of hydranencephaly malformation. Because frontal lobe, temporal lobe and parietal lobe (all depend on the blood-supply by internal carotid) are missing, while posterior fossa structures, occipital lobe, thalamus and brainstem (all depend on the blood-supply by verteobasilar artery) are basically normal. Depending on the feature of blood-supply, cerebral falx can be normal or hypogenetic(Bei Xia et al., 2001; Li Shengli, 2004; Tu Changyu, 2014; Michael Entezami, 2003). There are also some reports believe that other reasons cause hydranencephaly malformation are diffuse infection leading to cerebral necrosis, severe hydrocephalus leading to cerebral atrophy, or cerebral defects during the development of the embryo(Li Shengli, 2004). Hydranencephaly malformation is normally bilateral and unilateral hydranencephaly malformation is very rare.

According to the remaining situation of the brain tissue, the hydranencephaly malformation can be divided into severe and slight. According to the position and scope of the brain damage, the hydranencephaly malformation can be divided into Supratentorial type, hemisphere type, brain hypoplasia type and posterior cranial fossa type(Yang Fangyu et al., 2007). Also, according to the pathogenesis combing the CT and MRI analysis, the hydranencephaly malformation can be divided into supratentorial type, hemisphere type and brain hypoplasia type (Su Yongneng et al., 1999).

Hydranencephaly malformation is a kind of severe malformation that can not be treated. Because of the bad prognosis and most of them are stillbirth, most of the live birth will die in three months. It is very rare that the live birth can last for one year. There are reports that the live birth can live for three years, it is very scarce. So, once it is diagnosed positively, it should immediately terminate the gestation. Hence, the ultrasound diagnosis of hydranencephaly malformation is of great importance.

The ultrasound findings of hydranencephaly malformation is of noticeable features: skull hyperechoic structure is normal, there is no images of frontal lobe, temporal lobe and parietal lobe brain tissue. These brain tissue areas are replaced by anechoic area from
cerebrospinal fluid. Thalamus, brainstem and incomplete occipital lobe can be seen from ultrasound. Cerebral falk can be seen completely or partially, or vanished. It is mainly because the blood supply of endocranium and brain are separated, the former one can have multiple sources. Endocranium anterior cerebral artery is a branch of arteria ophthalmica, supplying the blood for the front part of endocranium. Arteria meninge media, a branch of maxillary artery, goes into cranial cavity through spinous foramen. The blood vessels spread over meninx which covers frontal lobe and parietal lobe.

When do ultrasound diagnosis of hydranencephaly malformation, people should tell the difference from severe hydrocephalus, alobar holoprosencephaly and porencephalia. In this group of cases, there are two cases misdiagnosed as alobar holoprosencephaly; one case is misdiagnosed as severe hydrocephalus. When reviewing the reasons for misdiagnoses, it mainly because the image observation is not carefully enough. At the same time, doctors ignore the differential diagnosis of several fetal malformation. In addition, the early gestational weeks might be another reason in two cases. Among the three misdiagnoses, the pseudomorphism between anechoic areas and skull is regarded as compressed brain tissue. The cerebral falk can only be seen the first half in two cases and are misdiagnosed as alobar holoprosencephaly. In another case, the cerebral falk is complete and is misdiagnosed as severe hydrocephalus.

Although the hydranencephaly malformation shows the feature of large anechoic areas in cranial cavity, the key point is that the hydrops place is in the extensive ventricle. The thinned substantive echo of the brain can be seen around the ventricle. In the hydrops anechoic areas, choroid plexus high echo can be seen, especially in the ventricle which is far away from the probe.

The feature of alobar holoprosencephaly is that the forebrain can not normally divide into two cerebral hemispheres. It leads to the paracele merges with ventriculus tertius and the facial and brain tissue functional defect. The large anechoic areas of the cranial cavity are in the paracele, showing the feature of connecting the two paraceles. The large anechoic areas show the shape like horseshoe. Substantive brain tissue can be seen in the periphery of anechoic area. The brain hemisphere is not separated, the thalamus is can be seen but is blended. Cerebral falk can not be seen, septum pellucidum, diacele and callosum disappear. The fourth ventricle and postfovea are broadened. In addition, because the brain hemisphere is not separated, it causes a series of facial central line structure malformation. For example, eye span is too short; cyclopia. Also some nasal deformity: single nostril; no nostril; long nose deformity.

Long nose deformity normally places above the cyclopia. The central harelip, bilateral harelip and microstomia happen occasionally. All these characteristic features can tell the difference between alobar holoprosencephaly and hydranencephaly malformation.

Porencephalia is a pseudocystic lesion caused by cerebral ischemia, bleeding, infection or brain tissue development abnormity. The cystic anechoic area is inside the brain substance with irregular form. The cystic area is connected with paracele or arachnoid lower cavity. If the range of porencephalia is large, it is easy to confuse with hydranencephaly malformation. The ultrasound feature of porencephalaias is the substantive echo around the cystic area. Doctors can identify the hydranencephaly malformation and porencephalia using this point. Hydranencephaly malformation, like hydrocephalus, alobar holoprosencephaly and porencephalia, there are large anechoic area in the cranial cavity, but the anechoic area is not in the ventricle and the brain hemisphere tissue can not be seen around the periphery of the anechoic area. When there is cerebral falk structure in the ultrasound image, doctors should tell the differences between hydranencephaly malformation and severe hydrocephalus. The key point is that the periphery of the anechoic area of hydranencephaly malformation has no brain tissue. When there is no cerebral falk structure in the ultrasound image, doctors should tell the differences between hydranencephaly malformation and alobar holoprosencephaly. The key point is that the thalamus of hydranencephaly malformation is not blended. In addition, in cases of severe hydrocephalus and alobar holoprosencephaly, it is very easy to see the choroid plexus high echo inside the anechoic area. This is another key point for differential diagnosis of fetal hydranencephaly malformation.

It is easy to diagnose the hydranencephaly malformation if grasping the features of the ultrasound images. Also, pay attention on the differential diagnoses with severe hydrocephalus, alobar holoprosencephaly and porencephalia. But there are several points should be noticed: when checking the fetal brain, doctors should observe every section, including cross section, sagittal section and coronal section. Because in different sections, there are different findings. When observing the brain, the hemispheres, thalamus, brainstem and ventricular system, no details should be neglected. For the cases of small gestational weeks, it is difficult to ensure whether there is brain tissue in the periphery of anechoic area in the cranial cavity and where the thalamus is blended. In the examination, doctors should make fully use of the functions of ultrasonic instruments, like magnify the images or tone-up, for better observation. All in all, the key point of ultrasound examination of hydranencephaly malformation is that observe whether there is brain hemisphere tissue after finding the large anechoic area in the cranial cavity.
CONCLUSION

Fetal hydranencephaly malformation is a kind of untreatable malformations, with unfavourable prognosis. So, it is vital to discover it early. Ultrasound examination is a very effective way to diagnose fetal hydranencephaly malformation.

The images of ultrasound have the following features: there are a large piece of anechoic area in the skull, there is no images of frontal lobe, temporal lobe and parietal lobe brain tissue, or there are only thalamus and brainstem, or partial occipital lobe. Cerebral falx can be seen completely or partially, or maybe disappeared.

It's worth noting that people should do differential diagnosis between fetal hydranencephaly malformation with severe hydrocephalus, alobar holoprosencephaly and porencephaly. The key point of the differential diagnosis is that the anechoic area of fetal hydranencephaly malformation lays neither in the ventricle nor in the brain parenchyma, while the anechoic area of severe hydrocephalus and alobar holoprosencephaly lays in the ventricle and the anechoic area of porencephaly lays in the brain parenchyma.

If doctors grasp this key point, it is not hard to do the ultrasound diagnosis and differential diagnosis of fetal hydranencephaly malformation.

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