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Diagnosis of fetal transverse facial cleft by magnetic resonance imaging negative interval scanning sequence: a case report

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► ***Case report***

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ABSTRACT

The Transverse facial cleft is a rare congenital malformation of the fetal face. Color Doppler ultrasound is one the common methods of screening for this condition, but its diagnostic accuracy can be affected by the position of the placenta, the shape of the pregnant woman's body, and whether the fetus is oriented in an open or closed position. Magnetic resonance imaging has the advantages of high soft tissue resolution and wide field of vision, which facilitate objective and comprehensive evaluation of fetal development. It can be an important supplementary means of prenatal screening. We here report a case of fetal bilateral lip angle asymmetry revealed by color Doppler ultrasound screening at 26 weeks' gestation. Magnetic resonance image negative interval scanning was used to diagnose transverse facial cleft, which was confirmed after induced labor. We also retrospectively analyzed the process of embryonic development, imaging manifestations, and treatment methods of transverse facial cleft.

INTRODUCTION

Cranio-facial cleft is a congenital facial malformation that can develop during the fetal stage. Tessier classification divides craniofacial cleft into 30 types based on the combination of disease description and anatomy [(1)]. The cranial fissure develops above the orbit, while the facial fissure develops below the orbit. Two common types of surface cracks are transverse surface cracks and oblique surface cracks. Transverse facial fissure is a Tessier7 fissure, and it can be unilateral or bilateral. The main manifestations are enlargement of the corners of the mouth and cracks extending outward from the corners of the mouth. Mild cases may have simple soft tissue defects, while severe cases may involve multiple facial clefts that involve the middle ear and other structures. Patient prognosis is related to the severity of cracks, whether or not they are accompanied by other deformities. People with mild cases can benefit from plastic surgery in both appearance and function. Color Doppler ultrasound is the primary method of prenatal screening, but its diagnostic accuracy is affected by fetal position and the pregnant woman's body shape. Magnetic resonance imaging (MRI) has the advantages of high soft tissue resolution, objectivity, and no radiation. It is an important supplementary means of prenatal screening. Here, we discuss the value of MRI negative interval scanning technology in screening for fetal facial diseases by reporting one fetus with transverse facial fissures. We used both conventional MRI scanning technology and negative interval scanning technology.

Case presentation

At the time of examination, the pregnant woman was 30 years old and 26 weeks +5 pregnant with her first pregnancy, a singleton. There were no relevant abnormalities in her or her family's medical history. Ultrasound was performed during her prenatal examination using a GE E8 color Doppler ultrasonic diagnostic instrument. Results showed the bilateral lip angles to be asymmetrical (figure 1). We then recommended a fetal MRI examination, which we later performed using a Siemens 1.5T magnetic resonance instrument, Aera model.

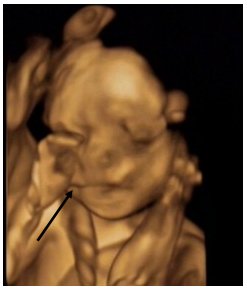

Figure 1. 3D color Doppler image of fetal face: The fetus is in a closed position, with asymmetric squabble on both sides and deep corners of the mouth on the right side (arrow indicates the right squabble).

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This study was approved by the ethics committee of Tangshan Maternal and Child Health Hospital under registration number (2023-040-01), date of registration: March 24, 2023.

We used the following techniques for fetal MRI examination: ① Haste-T2WI, TR 1100 ms, TE 97 ms, slice thickness 4.5 mm, slice spacing 20%, FOV 310 mm×310 mm; ② Trufi-T2WI, TR 684.79 ms, TE 2.62 ms, slice thickness 4.5 mm, slice spacing 0%, FOV 310 mm×310 mm. Negative interval scanning technology: thin slice Trufi-T2WI, TR 582.20 ms, TE 2.39 ms, slice thickness 4.5 mm, slice spacing -50%, FOV 340 mm×340 mm with the patient in supine position, breathing freely.

Fetal MRI findings: On conventional T2WI, only the axial and coronal images of the right maxillofacial region of the fetus were visible with high signal intensity, and they were faint (figure 2a–b). A deep cleft could be seen in the sagittal position (figure 2c), bilateral external ears were visible and structurally symmetrical (figure 2d), and the mandibular appearance was basically normal (figure 2e). Thin slice Trufi-T2WI showed the local soft tissue of the right cheek to be invaginated and the skin incomplete. The right corner of the mouth was horizontal, and there was a fracture with an up-and-down diameter of about 0.2 cm, which was horizontal and extended to the maxilla. The length of the fracture was about 1.2 cm (figure 3a–c). We found these findings consistent with the light imaging findings of fetal transverse facial cleft.

After consulting with the patient, we repeated the fetal color Doppler ultrasound examination. It showed that the fetal right facial isthmus was depressed, and the continuity between the facial isthmus and the outer edge of mandible was interrupted. After the boundary of the lip vermilion, the cleft continued to extend outward about 0.49 cm, and the right lateral maxilla seemed to be interrupted by a gap about 0.18 cm in width (figure 4). We diagnosed the fetus with right lateral cleft with maxillary cleft.

The pregnant woman and her family carefully considered the situation, and she agreed to induce labor at the 27th week of pregnancy. Upon examination of the gross specimen, we found that the bilateral spat of the fetus was asymmetrical, the facial cleft extended horizontally from the right spat, the soft tissue near the cheek was slightly collapsed, and the external ear and mandible had developed well (figure 5). There was no autopsy, but gene and chromosome examinations were performed.

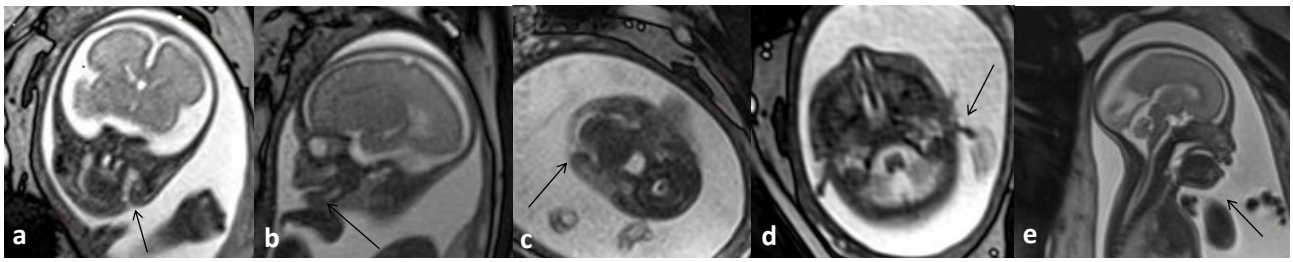

Figure 2. Conventional MR examination image: **a)** MRI Trufi-T2 weighted image coronal position, a strip of high signal was faintly visible on the right maxillofacial region (arrow indicates the lesion); **b)** MRI Trufi-T2 weighted image sagittal position, a deep fissure can be seen (arrow indicates the lesion); **c)** MRI Trufi-T2 weighted image axial position, a strip-shaped high signal was faintly visible on the right maxillofacial region (arrow indicates the lesion); **d)** MRI Trufi-T2 weighted image axial position, bilateral symmetrical external auricle structures can be seen (arrow indicates the ear); **e)** MRI Haste-T2 weighted image sagittal position, the mandibular contour was normal and no adduction sign was found (arrow indicates the mandible).

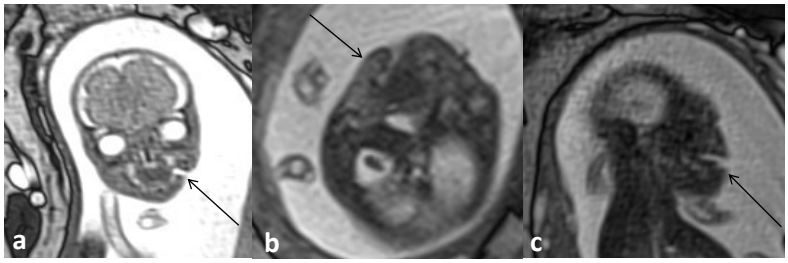

Figure 3. Negative interval MR images: **a)** MRI Trufi-T2 weighted image coronal position; **b)** MRI Trufi-T2 weighted image sagittal position; and **c)** MRI Trufi-T2 weighted image axial position. All show high signal cracks on the horizontal side of fetal squabble (arrows).

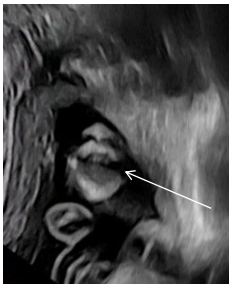

Figure 4. Re-examination of the 3D color Doppler image: The fetus is in the open position, the right corner of the mouth is enlarged, and a crack can be seen (arrow).

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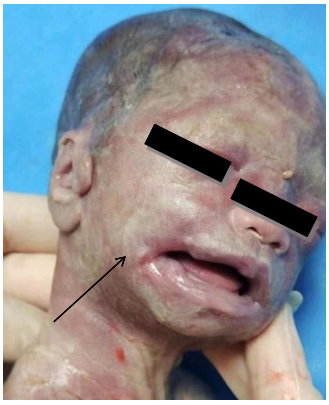

Figure 5. Gross specimen after induced labor: The right corner of the mouth is enlarged; that is, the corner of the mouth is cracked (arrow), and the shape of the outer auricle is normal.

DISCUSSION

Between 7 and 12 weeks of pregnancy, the lip and palate form bilateral maxillary processes, which grow toward the midline, while the medial nasal processes grow downward. The two processes fuse with each other to form the upper lip. Both mandibular processes grow toward the midline and fuse at the midline to form the lower lip, mandible, teeth, and mandibular soft tissue, and the maxillary process and mandibular process combine to form the corner of the mouth. When some or all of them fail to fuse, any of several maxillofacial deformities can result: when the maxillary process and the nasal frontal process fail to completely fuse, some degree of facial oblique cleft form. When the mandibular process and maxillary process fail to completely fuse on one or both sides, the fetus develops different degrees of angular cleft on one or both sides after birth. Severe cases are called transverse facial cleft, also known as big mouth deformity, or mouth cleft. The incidence of severe cases is low, accounting for about 0.3%-1.0% of all facial deformities [2]. This type of cleft can occur alone, or it can be accompanied by other deformities and manifest as other syndromes, such as the first and second branchial arch syndrome and the otospinal cord syndrome [3-5]. The pathogenic factors may be teratogenic agents, heredity, gene mutations, or other factors.

Color Doppler ultrasound is an important method of prenatal screening, but because transverse facial cleft usually occurs as a unilateral cleft extending from the mouth to the external ear [6], it is readily visible only when the fetus is in the mouth-open position [7]. Conventional ultrasound examination can easily lead to missed diagnosis, and some reports have shown that three-dimensional ultrasound can fill gaps in the screening of fetal phenotypic abnormalities [8]. MRI has the advantages of high soft tissue resolution and the ability to scan in any direction, and it has seen increasing use in the screening of fetal prenatal diseases [9, 10]. We here used parameters of 4.5 mm slice thickness and 0% slice spacing for scanning, and the lesions were faintly visible on the right maxillofacial region, but they were not sufficiently clear. The thin scan of the fetal face performed with a negative interval clearly showed the depth, length, shape, direction, and position of transverse cleft of fetal face, which manifested as an increase of the right corner of mouth and a gap filled with amniotic fluid due to T2 high signal and has nothing to do with whether the fetal mouth is open or closed. This is because the facial cleft is caused by abnormal skin and the abnormal development of orbicularis oris muscle, which leads to incomplete closure of the mouth. Thin-layer MRI can objectively and symmetrically display the structure of bilateral squabble, so the asymmetrically widened corners of the mouth can indicate the deformity of the facial transverse cleft. In this case, in addition to showing the fetal face, the large field of vision inherent in this technique can allow observation of the development of fetal external ear, nose, mandible, and other parts. Sagittal position can facilitate the evaluation of mandibular size [11]. The results of this study showed that MR negative interval scanning is suitable for the diagnosis of facial fissures and can objectively display the course and range of facial fissures. Because it may be a part of multi-system malformation, axial position and coronal position can present the symmetrical external auricle in a manner beneficial to the comprehensive evaluation of fetal prenatal development. These orientations are also helpful to the judgment of simple transverse facial cleft and some other syndromes. At present, it is clear that SWI facilitates the evaluation of the fetal vertebra and has a strong ability to show bone structure. In the future, we may attempt to use SWI sequencing to scan fetal craniofacial bones and assess SWI in the evaluation of craniofacial cleft bone defects.

Simple transverse facial cleft affects the aesthetics and feeding ability of newborns. It is treated surgically. The treatment scheme mainly includes reduction of orbicularis oris muscle, repair of the skin flap, bone grafting, osteotomy, and distraction osteogenesis [12]. The operation is carried out in stages. Surgeons and other clinicians are continually improving surgical methods with an eye to reducing complications and improving the postoperative visual effect [13].

In conclusion, negative interval MRI scanning sequencing facilitates the screening of fetal facial deformities and is not dependent on the position of the fetus. In practical work, we should use it reasonably, considering the types of diseases that may be present in each case. Fetal MRI examination may serve as an important part of prenatal screening alongside fetal color Doppler ultrasound.

Ethical considerations: The ethics committee of Tangshan Maternal and Child Health Hospital under registration number (2023-040-01), date of registration, approved this study: March 24, 2023.

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The patient signed an informed consent form.

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***Author contribution*:** L.S. recruited the patient and performed the MRI imaging analysis. L.C. reviewed the literature, drafted, and revised the manuscript.

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