Impact of fetal magnetic resonance imaging on prenatal diagnosis of fetal craniofacial anomalies

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Craniofacial malformations include a wide spectrum of fetal anomalies. Facial clefts are by far the most frequent among these malformations with an estimated incidence between 1 and 2.2/1000, depending on the geographical region[1].

The prenatal ultrasound diagnosis of fetal craniofacial malformations by conventional two-dimensional (2D) ultrasound is possible, however the reported sensitivity is low, in particular in isolated cases. Moreover, the specific diagnosis of a craniofacial malformation by 2D ultrasound may be very difficult even for an experienced sonographer [2-4]. The incorporation of 3-dimensional (3D) ultrasound into clinical practice has resulted in remarkable progress in visualization and anatomic examination of the fetal face. Several authors have reported improvement in the visualization of the fetal face and neck in high-risk pregnancies. Cleft lip, micrognathia, malformed ears, and frontal bossing have all been reported to be better displayed and analyzed by 3D ultrasound[3-6].

Magnetic resonance imaging (MRI) is a valuable complement to sonography, adding useful information about the maxillofacial anatomy and allowing precise evaluation of the primary and secondary palate[7-9]. Apart from facial clefts, the use of magnetic resonance has been described in diagnosis of several other craniofacial anomalies (retrognathia, micrognathia, craniosynostosis, cephalocele, vascular anomalies, craniofacial tumors, dacryocystoceles, microphthalmia and other ocular and orbital abnormalities)[10]. Although the use of magnetic resonance in prenatal diagnosis of many craniofacial anomalies, its real impact needs to be proven in other studies. The reported numbers of cases were generally low[9], gestation age at the time of magnetic resonance high[8] and studied population limited to only high-risk cases[7].

The aim of our study was to evaluate the impact of fetal magnetic resonance on diagnosis of fetal craniofacial abnormalities.
Methods and Materials

Material

This was a prospective study of 15 pregnancies referred to our clinic for suspected craniofacial anomaly. The gestational age ranged from 18. until 38. weeks. Only pregnancies with complete and detailed outcome were included.

Methods

Imaging methods

In all cases, both 2D/3D ultrasound examination and fetal magnetic resonance were performed on the same day and the images were stored on digital device for complex retrospective analysis. All examinations and the complex evaluation of ultrasound and MRI images were performed by the same operator specialized in fetal medicine. Diagnosis based on MRI examination was each time confirmed by specialized paediatric radiologists.

Ultrasound examination included 2D evaluation of fetal face in all three planes with consequent volume acquisition with high-resolution 3D ultrasound using a multi-frequency transabdominal volume transducer (Voluson 730 Expert, GE Medical Systems, Zipf, Austria). In all cases we obtained several static volumes of the fetal face, while care was taken to to avoid artefacts caused by fetal movements or the limbs, umbilical cord or placenta above the fetal face. Both 2D images and 3D volumes were stored on digital device for retrospective analysis on computer using 4D View software (GE Medical Systems). 3D volumes were manipulated and evaluated in multiplanar mode with consequent 3D rendering and reconstruction.

Magnetic resonance straight-line followed 2D/3D ultrasound images and was performed on Philips Gyroscan Intera 1.5T with additional Sense Cardiac Coil on maternal abdomen. Imaging included sB-TFE and SS-T2/TSE in all cases with additional FLAIR, SSh-FFE-Bone, DIF and T1-TFE in selected cases in all three orthogonal planes. The mother was in supine position unless she was rotated oblique due discomfort. No drugs for fetal sedation were administrated.

Outcomes

Primary information about outcomes were obtained from parents or referral centres, the detailed diagnosis was retrieved from co-operating paediatric specialists or pathologists.
Fig. 0: Figure 1: Ultrasound and magnetic resonance examination of fetal face in a normal case. a-c/ 2D ultrasound examination of fetal face in three planes: a/coronal (arrow pointing to lip), b/transversal (arrow pointing to alveolar ridge of palate), c/sagittal (arrow pointing to nasal bone); d-f/ 3D ultrasound examination of fetal face in d/multiplanar mode and surface 3D rendering of e/face and f/lip and palate; g-k/ magnetic resonance examination of fetal face in three planes: g/sagittal (arrow pointing to palate), h/transversal (arrow pointing to alveolar ridge of palate), i-k/coronal (arrow pointing to i/lip and j-k/palate).

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Results

16 consecutive pregnancies referred for suspected craniofacial anomalies were examined.

Our 2D/3D US examination revealed 1 normal and 15 abnormal pregnancies affected by craniofacial abnormalities (Figure 1).

The abnormal cases included: 1 case of encephalocele associated with anophtalmia and bilateral cleft lip and palate (Figure 2), 10 facial clefts, (Figure 3) 1 epidermal scalp cyst (Figure 4) and 3 tumors affecting fetal head suspected to be most likely lymphangiomas.

Fetal MR confirmed our 2D/3D US findings, but it provided additional information about location, size and borders of craniofacial tumors and epidermal scalp cyst. Moreover, it improved our confidence into the correct diagnosis in these cases.
Images for this section:

Fig. 0: Figure 1: Chart showing the spectrum of 15 detected craniofacial abnormalities.

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Fig. 0: Figure 2: Encephalocele & unilateral anophthalmia & bilateral cleft lip/palate: 2D/3D ultrasound & magnetic resonance. a-d/ 2D ultrasound images presenting: a/ encephalocele, b/anophtalmia, c/bilateral cleft lip & palate, d/bilateral cleft lip; e-g/ rendered 3D (surface mode) ultrasound images presenting: e/anophtalmia & bilateral cleft lip, f/encephalocele, g/bilateral cleft lip & palate; h-k/ magnetic resonance images presenting: h/encephalocele, i/encephalocele & anophtalmia, j/ bilateral cleft palate (with deviated nasal septum), k/cleft palate.
Fig. 0: Figure 3: Unilateral cleft lip & cleft palate: 2D/3D ultrasound & magnetic resonance. a-b/2D ultrasound images presenting: a/unilateral cleft lip, b/unilateral cleft lip & cleft palate; c-d/rendered 3D ultrasound images presenting: c/unilateral cleft lip, d/unilateral cleft lip & cleft palate; e-f/magnetic resonance images presenting cleft palate.

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**Fig. 0:** Figure 4: Epidermal squamous cyst: 2D/3D ultrasound & magnetic resonance. a-b/2D ultrasound images in transversal plane presenting the cyst; c-e/3D ultrasound images presenting: c/the cyst in transversal plane, d-e/3D surface rendering presenting the cyst as a prominence on fetal face; f-g/magnetic resonance images presenting the cyst: f/T2-weighted images (SS-T2/TSE), g/T1-weighted images (T1-TFE).

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Conclusion

Ultrasound remains to be the primary prenatal imaging method, however magnetic resonance can add clinically relevant information in selected group of pregnancies affected by craniofacial abnormality.

The additional knowledge provided by MRI improves the certainty and accuracy of prenatal diagnosis, thus allowing better prenatal counselling of parents and postnatal therapeutic planning by the oral and craniofacial surgical team[11]. Anatomical extension of oropharyngeal and neck masses can be accurately assessed relative to the fetal airway with MRI, and can assist in managing delivery[10]. Our study was too small and larger studies need to confirm our data. We keep continuing the study and hope to further improve effectiveness of our diagnosis by improvement of image quality and experience.

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References


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