The role of 3D/4D ultrasound in the diagnosis of fetal facial abnormalities

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Abstract

Fetal facial anomalies are quite frequent and constitute grounds for major stress for parents because they are very visible and have a high emotional impact. Examining fetal face and facial abnormalities recognition is very important because allows to suspect the presence of possible chromosomal anomalies or genetic syndromes. Chromosomal anomalies are present in a proportion of 32-77% of the fetuses with facial congenital malformations, depending on the type of the malformation. Therefore, over 250 genetic syndromes are associated with facial anomalies. Ultrasonography 3-dimensional (D) or 4D provides improved viewing of fetal anatomy in relation to facial examination in 2D mode. The 3D ultrasound examination either multiplanar or surface rendering reconstruction mode is one of the ways to obtain an excellent initially target volume of organ investigated. The view in 3D or 4D make it easier understanding anatomical location and the consequences of fault being studied.
Keywords: *facial malformations, 3D/4D ultrasound, fetal face, cleft lip and palate, prenatal diagnosis*

Introduction

The examination of the fetal face using 3-dimensional (D) ultrasound enables the evaluation of structures using surface rendering reconstruction and the multiplanar or maximum mode. Sometimes are used static volume contrast imaging mode or tomographic ultrasound imaging (TUI)[(1)]. The 3D or 4D sonography offers many benefits because fetal face can be turned into a standard symmetrical orientation and examined millimeters by millimeters by scrolling through the volumes acquired[(2)]. This type of ultrasound can highlight in addition different activities and emotional statuses of the fetus.

Sonographyc diagnosis

Sonographyc diagnosis of the fetal facial abnormalities raises two major problems: low sensitivity detection and the difficulty of establishing a precise anatomical diagnosis. Fetal abnormalities diagnosis incidence is 20-30%[(3)]. We review the literature in order to standardize a protocol for the examination of fetal face and to increase the rate of detection of fetal abnormalities.

Fetal facial malformations

These include: 1. ear abnormalities; 2. absence/hypoplasia of nasal bone; 3. anomalies of orbit; 4. anomalies of the mandible; 5. tongue protrusion; 6. facial tumors; 7. cleft lip and palate. When we diagnose a facial malformation we must always answer the question: it is an isolated fault or is it about a genetic syndrome?

1. Ears assessment should be carried out especially in case of the existence of family history with congenital malformations of the ear. Ear malformations (i.e. microtia, large ears, abnormal positioning) is teaming up with some genetic syndromes, such as trisomy 13, trisomy 18 and 21. Treacher Collins syndrome (i.e. autosomal dominant congenital disorder characterized by craniofacial deformities, micrognatia, underdeveloped zygoma, orbits anomalies, cleft lip and/or palate and malformed or absent ears), Fraser syndrome (i.e. microcephaly, cryptophthalmos, syndactyly, abnormal genitalia, urinary tract abnormalities), VACTERL association (i.e. vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula and/or esophageal atresia, renal anomalies and limb defects) have been associated with congenital malformations of ear. The 3D examination of the ear is now evaluated in various studies. Shih[(4)] did made 3D reconstruction of one or both ears in 80% of fetuses from the study showing a more accurate picture and clarity of detail in comparison with 2D images.

2. Absence or hypoplasia of nasal bone is associated with an increased risk of Down syndrome. Nasal bone was initially measured on 2D images of facial profile, technically quite difficult for that involves obtaining a perfect midsagittal image. Using 3D multiplanar imaging Lee identified 45% of fetuses with Down syndrome in the second or third trimester of pregnancy using absence of the nasal bone as a sonographyc marker[(5)].

3. Orbits abnormalities include: hypotelorism, hypertelorism, anophthalmia, microphthalmia, orbital and periorbital tumors, cataract and retinian coloboma. Hypotelorism can be associated with holoprosencephaly, facial defects and trisomy 13. Also, hypertelorism is encountered in the following syndromes: Noonan syndrome (i.e. nucal fold, coronary disease, low set ears, pectus excavatum, cryptorchidism), Gorlin syndrome (i.e. odontogenic keratocysts of the jaw, macrocephaly, ventriculomegaly, vertebral anomalies, scoliosis or other skeletal abnormalities, cleft lip or palate), Neu-Laxova syndrome (i.e. microcephaly, micrognathia, lissencephaly, Dandy-Walker anomaly or agenesis of the corpus callosum, hypoplasia of cerebellum). Orbital and periorbital tumors (i.e. dacro-

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cystocele, hemangioma, glioma and teratoma) can be valued more exactly in the 3D or 4D ultrasound examination.

4. Mandible anomalies are associated with over 100 genetic syndromes (Treacher-Collins syndrome, Pierre Robin syndrome, orofaciodigital syndrome, Nager syndrome - acrofacial dysostosis, *cri du chat*, trisomy 13 or 18, Turner syndrome - monosomy X). Micrognathia and retrognathia are two different entities but they appear frequently together. Retrognathia can be diagnosed by measuring the inferior facial angle, when the value is <49.2 degrees - as shown in Figure 1[6]. The mandible width/maxxila width ration <0.785 was used by Rotten in diagnosing of micrognathia[6]. Also jaw index (the ratio between the anteroposterior diameter of the mandible and biparietal diameter, APD/BPD X100) was used to diagnose micrognathia. Hypoplastic maxilla appears in Down syndrome, Apert syndrome (i.e. hypertelorism, syndactyly, agenesis of the corpus callosum), Crouzon syndrome (i.e. craniosynostosis, ear canal malformation, partial syndactyly, hypertelorism), in the consumption of alcohol and drugs.

5. Tongue protrusion is frequently associated with Down syndrome but it is possible to appear in other situations also, sometimes is difficult to establish an exactly antenatal diagnosis. Tongue protrusion must be differentiated from macroglossia (occurs frequently in Beckwith-Wiedemann syndrome), small mouth (in DiGeorge syndrome, sometimes in Pierre Robin sequence), tongue hypotonia (i.e. Prader Wili syndrome, Rett syndrome), tumor of ductus tireoglosus[7].

6. Ultrasonography 3D/4D has proved a real help in facial tumors diagnostic (i.e. hemangioma, teratoma, tumors of the maxxila) as well as in evaluating growth of the tongue volume.

7. Cleft lip (i.e. cheiloschisis) and cleft palate (i.e. palatoschisis) are the most frequent congenital malformations of the face. Approximately 1 in 700 children born have a cleft lip or a cleft palate or both. Palate separates the oral and the nasal cavities and is composed of hard palate (bony, immobile, anterior located) and soft palate (muscular, mobile, posterior located). Hard palate is formed (Figure 2) by the primary palate located anterior of the incisive foramen and secondary palate composed of palatine process of the maxxila and horizontal plates of the palatine bone.

Cleft palate (palatoschisis) is an opening gap or fissure at the hard palate and soft palate level. Cleft palate may vary from a simple opening of the rear of the palate up to a complete separation of the roof of the mouth (i.e. hard palate and soft). In general it is considered that there are several genes concerned. There have been described over 300 syndromes that include this type of malformation. It has also been demonstrated involvement of environmental factors acting in the course of embryonic development (i.e. viral or parasitological infections, diabetes, epilepsy, low levels of vitamin A, D, E, alcohol, smoking, radiation).

Cleft lip and/or palate has an incidence of 1.4 to 1000 live births and represents 30-33% of all fetal abnormalities[8]. Incidence of the ultrasound diagnosis is variable, various studies suggesting that this abnormality is recognized with a frequency between 27%[9] and 59%[10]. Incidence of the isolated cleft palate and/or lip is 25-80% and the detection rate is 0-22%[11]. Tessier described fifteen lines of cleft.

Classification of cleft lip and/or palate:

- cleft lip; cleft palate; cleft lip and palate;
- cleft of the primary palate - unilateral, bilateral, median;
- cleft of the secondary palate - total, partial, submucosal;
- cleft of the primary and secondary palate - unilateral, bilateral and median.

Median cleft is rare and appears embryologically from incomplete fusion of the medial nasal prominences. It is teaming up frequently with chromosomal anomalies and holoprosencephaly. Cheilognathopalatoschisis may appear as constituent part of some syndromes: trisomy 13 and 18, holoprosencephaly, Van Der Woude syndrome (i.e. autosomal dominant congenital disorder 1q32-41, characterized by: cleft lip and/or palate, hypodontia, syndactyly, congenital heart disease, and cerebral abnormalities), hemifacial microsomia or Goldenhar syndrome (i.e. incomplete development of the ear, nose, soft palate or lip, and mandible on usually one side of the body), Majewsky syndrome (characterized by hyperostosis, craniodiaphyseal dysplasia, dwarfism, syndactyly, bra-

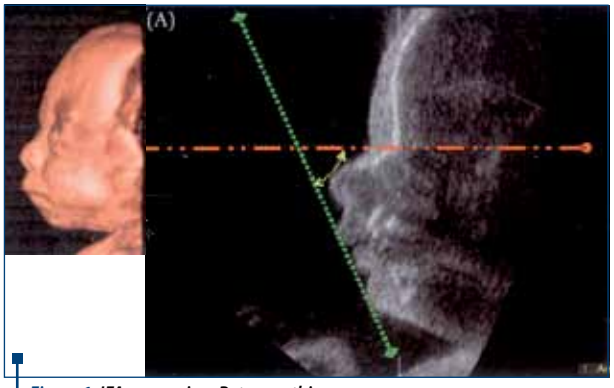

Figure 1. IFA measuring, Retrognathia

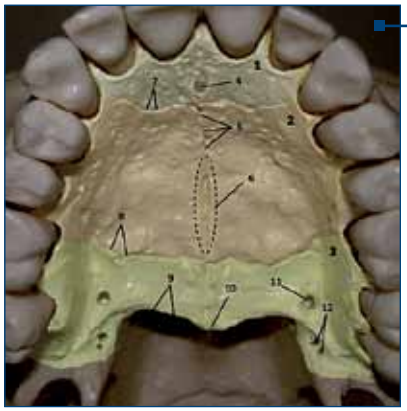

Figure 2. Hard palate: 1. incisive bone; 2. palatine process of the maxxila; 3. horizontal plates of the palatine bone; 4. incisive foramen; 5. median palatine suture; 6. palatine bone; 7. incisive suture; 8. transverse palatine suture; 9. posterior border of the horizontal plate; 10. posterior nasal spine;11. greater palatine foramen; 12. lesser palatine foramina

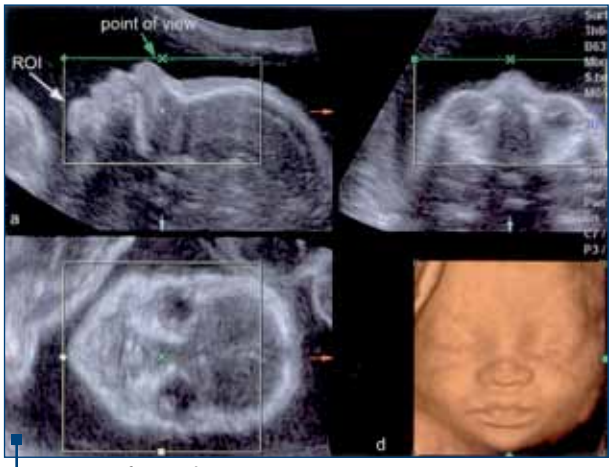

Figure 3. Surface rendering - 3D view

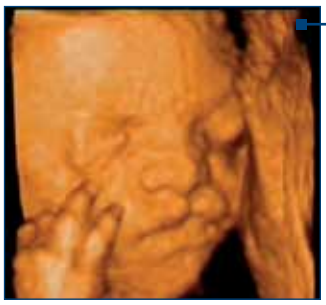

Figure 4. Cleft lip - 3D view

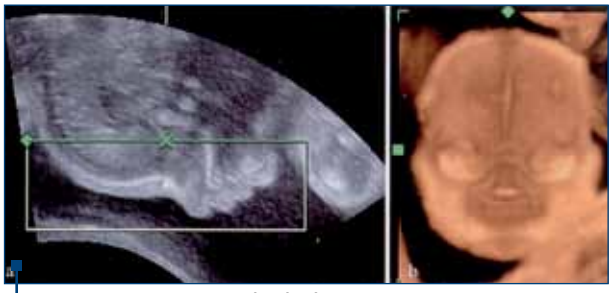

Figure 5. Reverse Face View - hard palate viewing

chydactyly, and mental retardation), Roberts syndrome (i.e. hypertelorism, microbrachycephaly, micrognathia, and hypomelia). Cleft of the secondary palate is hard to detect in the absence of others anomalies associated, easier to diagnosis such as retrognathia, micrognathia or anterior cleft.

Clinically cheilognathopalatoschisis may cause severe distortions of facial skeletal system or soft tissues which begins in the embryonic period and continue childbirth. Children with maxillo-facial clefts presents difficulties of feeding, speech problems, hearing impairment, dental abnormalities, disorders of facial integrity. Adolescents with clefts are at an elevated risk for developing psycho-social problems.These are dependent on the location and the severity of clefts.

Facial profile investigation

Investigating facial profile is a fundamental part of the sonographic examination. It can be done in 2D but also using 3D or 4D ultrasound. Using 3D multiplanar reconstruction in coronal, axial or sagittal planes has allowed for visualization of hard palate, otherwise difficult to evaluate. Gestational age recommended for fetal abnormalities screening remains, after most authors, at 21 -22 weeks.

The protocol for the facial abnormalities screening has several stages:

Step1: 2D ultrasound screening in the midsagittal and axial planes, then in anterior coronal plane.

Step 2: Examining using 3D/4D multiplanar reconstruction in the midsagittal plane and axial plane, then in the coronal plane by surface rendering mode (Figures 3 and 4) to highlight soft tissues of the face or in maximum mode or transparent mode for better visualization of bony structures.

The three reference planes allow a good anatomical view of the elements of the fetal face.

- in sagittal plane is highlighted: the nose, upper lip, partially alveolar ridge, maxilla, bottom lip, the chin/mandible;
- in the axial plane: upper lip, maxilla, the bony alveolar ridge, orbits;

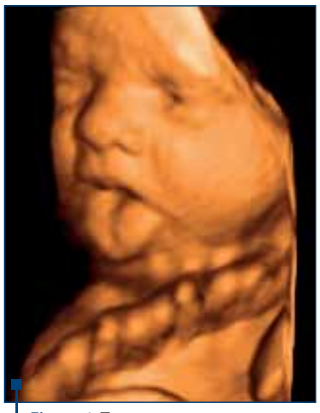

Figure 6. Tongue movement

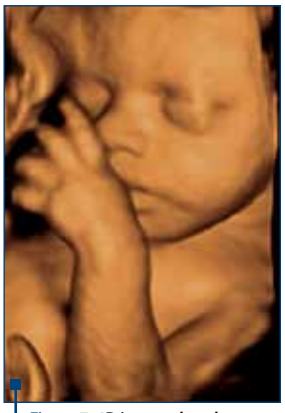

Figure 7. 4D image - hand movement

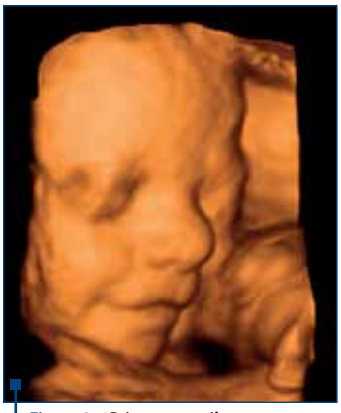

Figure 8. 4D image - smile

- in coronal plane: the fetal face, orbits, the nose, lips, the alveolar ridge and the hard palate integrity.

If it is suspected or it is observed a facial malformation then we may pass to the next step of examination.

Step 3: detailed analysis using 3D/4D multiplanar reconstruction concentrated on the area of interest. Secondary palate analysis can be more efficient in the posterior coronal plane or in the axial plane. Soft palate is difficult to view. At this stage you can use TUI which enables simultaneous analysis of multislice images (similar to tomographic imaging) representing contiguous sections in the area of interest.

By obtaining a volume whith 3D or 4D reconstruction it can be studied in detail secondary palate and soft palate, in the posterior coronal plane, in oblique sagittal plane[12] or using Reverse Face View technique described by Campbell in 2003[13,14] which allows you to view from the back to the front secondary palate and avoiding shadowing from the maxilla (Figure 5).You can also use Flipped Face View technique. This allows by roll-over the acquired volume analysis of anatomical elements in the following order: the chin, lower lip, the tongue, alveolar ridge, maxilla, hard palate, the upper lip, nose, forehead and in posterior plane, secondary palate and soft palate are assessed.

Another technique, described by Faure[15], which starting on the 2D image of the maxilla in the axial plane, then by 3D multiplanar reconstruction and rotating from the axial plane into frontal plane, unable to analyze accurately the primary palate but also secondary palate[16-18].

Ultrasonography 4D show the different movements and facial expressions in dynamic that can be signs of fetal consciousness (Figures 6, 7 and 8). It can be studied in this way, in terms of quality, certain patterns of fetal facial movements. The most common facial activities surprised by 4D ultrasound examination between 30 and 33 weeks of gestation are simultaneous movements of eyelids and mouth[19].

Ultrasonography 4D is a powerful tool to assess fetal behavior and several studies show a certain continuity between fetal and neonatal behavior, in particular as regards isolated flashing movements, opening mouth and eyelids, tongue expulsion, frown, smile or hand movements toward to the face[20-22].

Conclusions

The 3D or 4D ultrasound examination may reveal a detailed facial anatomy. The oligohydramnios, fetal position (i.e. pin of the fetal face with the placenta or uterine wall, overlapping limbs, anterior position of the fetal spine) and maternal obesity limits detailed view of the fetal face. False positive or false negative results may be given by the acoustic shadow of the maxilla, the tongue confused with the intact hard palate. Advantages of using 3D/4D ultrasound are clear. Fetal face can be viewed in a standard orientation. Ultrasonography 3D/4D allows you to view fetal face for a long time without care of fetal movements and by obtaining multiplanar or multislice images can be achieved the exact location of the malformation.

Ultrasound examination 3D/4D has an important role in assessing fetal face because it allows a better specification of fetal facial anatomy and increase facial abnormalities diagnosis incidence. At the same time comes to help parents through the establishment of early emotional connections to the child by easier understanding of possible malformations which may affect the fetus, as well as the therapeutic options. Recognition of clefts during 3D or 4D ultrasound examination allows psychological preparation of the couple as well the decision about of a possible reconstruction surgery. ■

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