Assessment of fetal malformations in the first trimester of pregnancy by three-dimensional ultrasonography in the rendering mode. Pictorial essay.

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Abstract

We present our experience in the contribution of three-dimensional ultrasonography, using the rendering mode, to the prenatal diagnosis of congenital anomalies including neurological defects (acrania/anencephaly, encephalocele, holoprosencephaly), facial anomalies (cyclopia and facial clefts), abdominal wall defects (omphalocele and gastroschisis) and defects of extremities (fetal muscle-skeletal dysplasias). Three-dimensional ultrasonography may contribute to improve the prenatal diagnosis with further revision of the fetal images, allowing a better prenatal counsel to the parents.

Keywords: prenatal diagnosis, congenital anomaly, three-dimensional ultrasound

Introduction

With advancing knowledge and technology, the application of ultrasound during the first trimester of pregnancy has been gaining widespread interest and increasing utility. Indeed, these advances have allowed for more accurate pregnancy dating, earlier determination and assessment of multifetal gestations, and vastly improved screening and diagnosis of embryonic and fetal malformations [1].

Perhaps, one of the most exciting advancements, and one with the greatest potential, has been the advent of three-dimensional ultrasonography (3DUS). This technology, combined with traditional two-dimensional imaging, has afforded sonologists the possibility of evaluating fetal structural morphology through the render mode, the patients the ability to better conceptualize and understand the problems facing their fetus and geneticists the ability to see the phenotype as a tool in making their assessment [2].

Consequently, the aim of this pictorial essay is to illustrate examples of several fetal anomalies, which can be detected in early pregnancy by 3DUS in the rendering mode.

Acrania/Anencephaly

The incidence of acrania/anencephaly is approximately 1:1,000 births [3]. Genetic and environmental factors have been implicated in its etiology. The single most important environmental factor is diet, namely maternal peri-conceptual folic acid intake [4]. Anencephaly involves an absent upper cranial vault accompanied by an absence of the cerebral hemispheres above the level of the orbits. Acrania includes an absence of the cranial vault with preserved brain parenchyma and normal facial structures. It is believed that first trimester cases of acrania, will ultimately progress to anencephaly by the second trimester [3,4].

In the first trimester, the defect is best appreciated in coronal imaging of the face (fig 1A), demonstrating ab-
ence of the calvaria above the bony orbits, which may appear prominent. As demonstrated in fig 1B, the defect can also be evaluated in the sagittal view. Polyhydramnios, and other co-existing major congenital anomalies are best identified in second trimester [3,4].

Encephalocele

Encephalocele, whose incidence is reported to vary between 1-3:10,000 and 1:5,000 live birth [3], is a rare skull defect with resultant herniation of intracranial contents including the meninges, neural tissue, and meningoceles [4].

Depending on defect location, the encephalocele may be subdivided in occipital (defect between lambda suture and foramen magnum), or frontal (defect between bregma and anterior margin of ethmoid bone). The importance of the location of the herniation is because to different prognosis [4].

Various conditions have been associated with encephaloceles including congenital rubella, diabetes, genetic syndromes (e.g. Meckel-Gruber Syndrome), and amniotic-bands; however, a true causative factor remains unknown. The insult is hypothesized to occur from failure of closure of the rostral neural pore, versus disruption of fetal-skull formation (amnion rupture sequence), especially in the midline, which may be associated with other malformations [3,4].

As demonstrated in fig 2, sonographic findings in the first trimester include a calvarial defect with herniation of a mass, which may be cystic, solid (with giral pattern), or combined cystic and solid mass. The figures 2 and 3 illustrate an occipital encephalocele.

Holoprosencephaly

The incidence of the holoprosencephaly in the first trimester of pregnancy is unknown. However, in one series, this defect was reported in 27% of 181 fetuses with trisomy 13 at 11–13 weeks of gestation [5].

Holoprosencephaly includes a large spectrum of cerebral and facial malformations that result from absent or incomplete division of the embryonic forebrain (prosencephalon) [4]. There are three types of holoprosencephaly: alobar, semilobar and lobar. The sonographic findings are the best illustrated in coronal imaging especially the most severe form, the alobar holoprosencephaly, where there is absent midline structures, no division of the cerebral hemispheres, single common ventricle, and fused thalami (fig 4). In semilobar holoprosencephaly there is partial separation of the hemispheres. Only in lobar holoprosencephaly is normal cortical division with the presence of two thalami, albeit accompanied by abnormalities of the corpus callosum, septum pelucidum, and/or the olfactory tract [4]. The facial anomalies accompanying holoprosencephaly include a proboscis (cylindrical protuberance), ethmocephaly (the eyes are separated but closely placed), cyclopia (most severe form of hypotelorism), and a palatine defect [4,5] (fig 5).
Omphalocele (otherwise known as exomphalos) is a congenital abnormality with herniation of abdominal contents through a ventral wall defect, and thought to occur during the process of body infolding. The incidence this anomaly ranges in 4,000-7,000 live births [7]. The herniation it is generally covered by a thin membrane consisting of peritoneum and amnion. The defect can vary in size from a few centimeters to as much as the entirety of the ventral abdominal wall. The umbilical cord inserts into the covering membrane at a location distant from the abdominal wall. Physiologic herniation of the fetal bowel into the base of the umbilical cord occurs normally between 8 and 12 weeks gestation. Failure of the intestinal loops to return to the abdominal cavity results in the formation of an omphalocele [7]. This anomaly is characterized by a high association with others malformations and chromosome abnormalities (trisomies 18 and 13), especially with smaller lesions [6]. The diagnosis can be made as early as 10 to 12 weeks when no herniation should be visible once, especially when the crown rump length is 45 mm or more. As illustrated in figures 6 and 7, the sonographic finding this malformation is an echogenic mass anterior to the fetal abdomen with the umbilical cord insertion into the membrane covering the abdominal wall defect [6,7].

Gastroschisis

The worldwide incidence of gastroschisis has seen an increase in recent decades [8]. Gastroschisis occurs because a defect of the abdominal wall secondary to incomplete closure of the lateral folds during the initial gestation. Contrary to an omphalocele (unruptured), the eviscerated bowel characteristically does not have a covering membrane. Characteristically, the lesion is to the right of the intact umbilical cord [7]. The differentiation between gastroschisis and omphalocele is important, as gastroschisis is not associated with aneuploidy [6].
In first trimester vaginal ultrasound may help in distinguishing the contents of the herniation, especially as early as 12 weeks when the diagnosis of gastroschisis may be possible. The figure 8 demonstrates a fetus with gastroschisis.

**Facial anomalies**

Facial clefts are one of the most frequent congenital anomalies, with an incidence between 1:625 and 1:1,000 newborns in the USA [9]. The cleft palate stems from a failure in the fusion of maxillary lateral prominences. Most cleft palates affect the hard palate, causing interruption of the alveolar arch of the maxilla. The classification of clefts is determined by localization (unilateral, bilateral, or median) and anatomic involvement (only the lip is engaged; lip and palate are engaged; and only the palate is engaged) [10].

3D ultrasound (rendering mode) in the first trimester of pregnancy can serve as an invaluable adjunct to 2D ultrasound in cases of cleft lip and palate. As illustrated in figure 9, the rendered visualization from 3DUS may aid parents in conceptualizing the defect and help with the counseling [11].

Cyclopia is a facial midline anomaly frequently associated with alobar holoprosencephaly (fig 10). To date, few reports have examined prenatal diagnosis of cyclopia in the first trimester [12,13]. Namely, Blass et al [12] described a case of cyclopia associated with alobar holoprosencephaly diagnosed at 9 weeks, whereas similarly Dane et al [13] described a case of semilobar holoprosencephaly with cyclopia and radial aplasia at 12 weeks.
Extremities defects – fetal muscle-skeletal dysplasias

Skeletal dysplasias are a heterogeneous group of bone disorders characterized by short long-bone length. More than 500 types of skeletal dysplasias have been described with 50% of believed to be lethal. Usually, the prenatal diagnosis may be made in the second trimester of pregnancy with an accuracy of 30 to 50% [14]. In the first trimester, lethal skeletal dysplasia has been reported to be associated with an increased nuchal translucency in 85% of cases [15]. Figures 11 show two case of fetal immobilism by 3DUS in the rendering mode with upper limbs contractures and marked skin edema. Figure 12 shows a case of tanatophoric dysplasia with markedly shortened limbs and a great disproportion between the fetal head and body.

The figure 13 shows a case of neural tube defect at 14 weeks of gestation. The 3DUS in the rendering mode with maximum transparency demonstrates the bony defect in the lumbar spine. The figure 14 shows a case of Femoral Hypoplasia Unusual Facies Syndrome at 14 weeks of gestation. The 3DUS in the rendering mode demonstrates the large metopic suture and the micoglnathia.

Conclusions

The 3DUS is gaining widespread use and utility in the assessment of the growing embryo and fetus in the first trimester of pregnancy [16]. This technologic advancement and the future improvements have and will lead to increased prenatal diagnosis of fetal anomalies in the first trimester [17]. First trimester diagnosis will permit earlier counseling of parents, and in some countries interruption of the pregnancy. The 3DUS images are more similar to the reality, which seems to be a useful tool during prenatal consultations with the parents. In addition, 3DUS allows registration of the fetal images and further revision and discussion in a distant referral institute. These images can be faster acquired and stored in the hard disk for further analysis. In this pictorial essay, we attempt to show examples of the feasibility of first trimester prenatal diagnosis of some fetal anomalies using 3DUS.

Conflict of interest: none

References

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