MORPHOLOGICAL AND VOLUMETRIC ULTRASONOGRAPHIC EXAMINATION IN THE DIAGNOSTIC OF RARE FACIAL ANOMALIES. A CASE PRESENTATION

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MORPHOLOGICAL AND VOLUMETRIC ULTRASONOGRAPHIC EXAMINATION IN THE DIAGNOSTIC OF RARE FACIAL ANOMALIES. A CASE PRESENTATION (Abstract): Proboscis is a rare craniofacial anomaly, characterized by a rudimental tubular structure in the middle of the face. It is classified in 4 general types: holoprosencephalic, lateral nasal, supranumerary, and disruptive. Its presence could be associated to other anomalies, such as: trispomy 13, holoprosencephaly, etmocephaly, cyclopia. The ultrasound examination of the face should follow the nose-mouth coronal plane, the strictly sagittal plane for profile, and step by step axial sections for eyes, maxillary, tongue and mandibula. The volumetric 3D ultrasound scan can confirm the proboscis diagnostic and differentiate the anomaly from other facial abnormalities. Case report: We present a congenital abnormalities foetus with proboscis, alobar holoprosencephaly, hypotelorism, polyhydramnios, which was diagnosed prenatally using late morphological and volumetric scan at 32 weeks of gestation. A male foetus is extracted through cesarean section at 38 weeks, with a weight of 2850 grams, and Pagar score at 1 min 2-3. After birth, at 4 hours life, the foetus dies. The external anatomical evaluation confirmed the ultrasound anomalies as described. Further genetic analysis are under way to confirm a probable trisomy 13. In conclusion, the face examination is important in the diagnostic of congenital abnormalities. This requires a systematic and sometimes interdisciplinary approach. The profile analysis (forehead, nose, upper lip, chin) is a fast examination, that could warn the obstetrician regarding the possibility of an anomaly, and the use of standardized exam for sagittal section is essential. **Keywords:** PROBOSCIS, HOLOPROSENCEPHALY, 3D ULTRASOUND TRISOMY 13

INTRODUCTION

Craniofacial malformations include a wide spectrum of fetal anomalies.

Sonographic diagnosis of fetal craniofacial malformations is possible and has been described in many publications.

However, a meticulous scanning technique is required and this is reflected in two main problems.

Firstly, the sensitivity of identifying these anomalies is low, with a general tendency to recognize facial malformations associated with other anomalies and to miss the isolated ones;

Secondly, the specific diagnosis of a craniofacial malformation may prove challenging even to an experienced sonographer.

Most guidelines for the standard examination of fetal anatomy include views of the fetal face (1). Prenatal diagnosis of craniofacial anomalies is indeed possible.

However, meticulous scanning is required, and the detection rate remains low in routine sonographic studies, usually below 20%.

Furthermore, most of these malformations are identified following the recognition of extrafacial anomalies (2,3).

Fig. 1. Proboscis – rare craniofacial anomaly

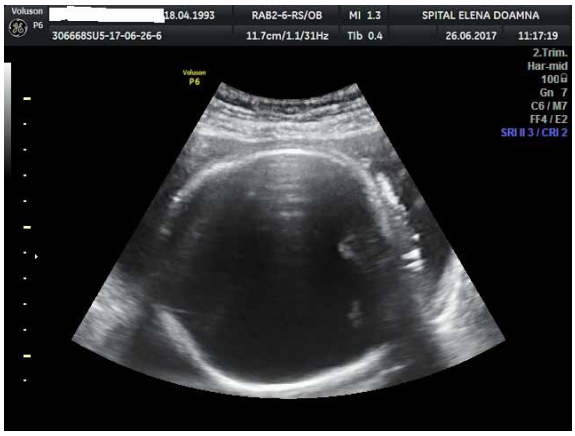

Fig. 2. Alobar holoprosencephalia

Proboscis (fig. 1) is a rare craniofacial anomaly, characterized by a rudimental tubular structure in the middle of the face. It is classified in 4 general types : holoprosencephalic, lateral nasal, supranumerary, and disruptive. Its presence could be associated to other anomalies, such as : trispomy 13, holoprosencephaly, etmocephaly, cyclopia (4,5).

The ultrasound examination of the face should follow the nose-mouth coronal plane, the strictly sagittal plane for profile, and step by step axial sections for eyes, maxillary, tongue and mandibula.

The volumetric 3D ultrasound scan can confirm the proboscis diagnostic and differentiate the anomaly from other facial abnormalities (6,7).

The etiology of the disease is still unclear. Environmental and genetic factors may be at work. Among environmental factors there are maternal diabetes mellitus, alcoholism, cytomegalovirus, rubella or toxoplasma infections, and some drugs (retinoic acid, cholesterol synthesis inhibitors, fenitoin, salicylates) (8,9).

The diagnosis of the disease can be made with abdominal ultrasound examination and MRI during prenatal period.

Trisomy 13 and trisomy 18 are the most frequently encountered chromosomal anomalies (10,11).

The etiopathogenesis of the disease is still unclear. Holoprosencephaly without a recognizable gene or chromosome defect has been already described occasionally.

The ethmoid complex, rooted in the prechordal mesoderm, plays an important role in the development of the midline and symmetry of the fetal face. Flaws in the development of the ethmoid complex lead to severe malformations of the whole, middle or upper parts of the face. In situations without ethmoid process a structure, called the proboscis, develops above the eyes. This structure is a hollow tube made of cartilage coated with respiratory epithelium (12, 13).

Histologic findings show the proboscis is a similar structure to the nose, which includes respiratory epithelium. This indicates that it results from a problem during development of the frontal region of the nose (14).

In English-language literature 257 cyclopian cases have been reported until now and 81 of these cases had chromosomal abnormalities.

The combination of true cyclopia, proboscis and holoprosencephalia (HPE) (fig. 2) is an extremely rare condition.

All cases of this combination were reported with the absence of a nose (15).

If the facial defects are not easily detected when the fetal position is suboptimal during the 2-D sonographic imaging, 3-D (fig. 3) or 4-D scan may play an important role in confirming such defects. Additionally 3-D and 4-D sonography of the facial features can provide more convincing evidence for prenatal diagnosis and counseling (16).

CASE REPORT

We present a congenital abnormalities foetus with proboscis, alobar holoprosencephaly, hypotelorism, polyhydramnios, which was diagnosed prenatally using late morphological and volumetric scan at 32 weeks of gestation.

A male foetus is extracted through cesarean

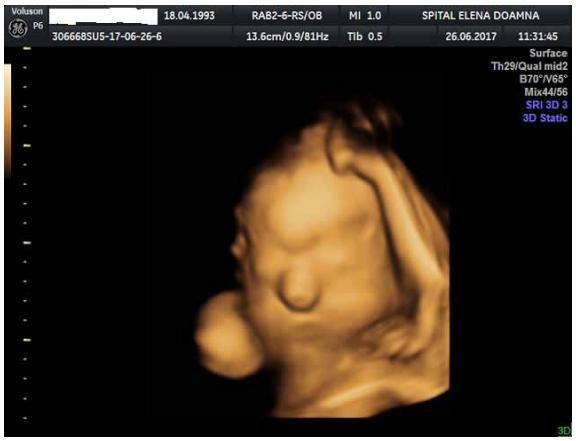

Fig. 3. Proboscis – 3D ultrasound

Fig. 4. A male fetus with proboscis

section at 38 weeks, with a weight of 2850 grams, and Apgar score at 1 min 2-3 (fig. 4).

After birth, at 4 hours life, the foetus dies.

The external anatomical evaluation confirmed the ultrasound anomalies as described.

Genetic analysis confirmed the presence of trisomy 13 (fig. 5).

Dysmorphism in the case of trisomy 13 is often very spectacular, covering a wide range of possibilities from a simple median incisor ti cyclopia, with a full range of intermediary aspects possible including more or less hypotelorism, single nostrils.

Clefting is found in 60-80 % of cases, and is both unilateral and bilateral. Median clefts are characteristic.

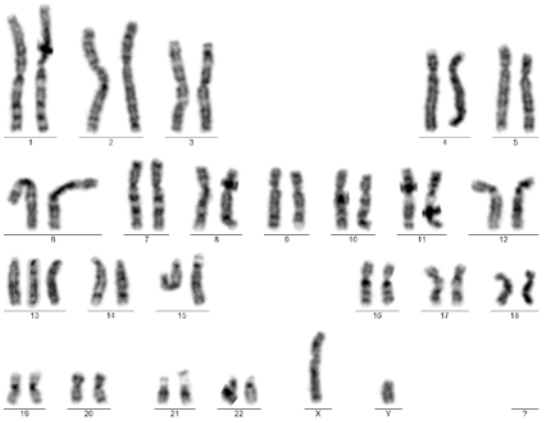

Fig. 5. Trisomy 13 – genetic analysis

CONCLUSSIONS

The face examination is important in the diagnostic of congenital abnormalities.

This requires a systematic and sometimes interdisciplinary approach.

The profile analysis (forehead, nose, upper lip, chin) is a fast examination, that could warn the obstetrician regarding the possibility of an anomaly, and the use of standardized exam for sagittal section is essential.

The study of fetal face anomalies using precise, reproductible, anthropometric criteria helps avoid the too-frequent subjectivity found in an approach that is not truly quantified. The discovery of dysmorphism and its characterization is one way that is frequently used to uncover various pathological fetal syndromes (17).

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