

Prenatal diagnosis of otocephaly using two-dimensional and three-dimensional ultrasonography

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ABSTRACT

We report a case of the prenatal diagnosis of a very rare anomaly, namely isolated otocephaly, using two-dimensional and three-dimensional ultrasound at 24 weeks of gestation. Two-dimensional ultrasound revealed polyhydramnios, absence of a stomach shadow, hypotelorism and a proboscis-like mass. Three-dimensional ultrasound gave a precise demonstration of the striking craniofacial features of otocephaly including agnathia, synotia, microstomia and protuberance of the nose–mouth fusion. The use of three-dimensional ultrasound provided a whole view of the very rare lethal malformation in utero and contributed significantly to prenatal diagnosis.

INTRODUCTION

Otocephaly is a very rare congenital anomaly characterized by extreme hypoplasia or absence of the mandible (agnathia), synotia, microstomia and aglossia^{1,2}. A variety of associated malformations, including defects of the central nervous, cardiovascular, genitourinary and axial skeletal systems have been reported^{1–3}. Prenatal diagnosis of this lethal anomaly is extremely infrequent, usually late and always obscure^{4–8}. Precise and direct prenatal evidence of the striking craniofacial features has not previously been reported, because of the limitations of conventional two-dimensional ultrasound. With the advent of three-dimensional ultrasound, digital documentation of whole volumes has enabled us to demonstrate the striking features of otocephaly at 24 weeks of gestation, without any loss of information.

CASE REPORT

A 30-year-old woman, gravida 2, para 1, was referred at 24 weeks' gestation to our outpatient clinic owing to the suspicion of a fetal anomaly and the presence of poly-

hydramnios. The patient was healthy, her past obstetric and family histories were unremarkable and neither parent had been exposed to hazardous environmental factors. A level II two-dimensional ultrasound examination revealed a live female fetus with a biparietal diameter of 6.0 cm, an abdominal circumference of 22.0 cm and a femur length of 4.2 cm (appropriate for 24 weeks of gestation). The following anomalies were also seen: polyhydramnios with an amniotic fluid index of 28.4 cm, absence of a stomach shadow and facial anomalies including hypotelorism and a proboscis-like mass. There were no defects noted in the central nervous system, cardiovascular system, genitourinary system or axial skeletal system.

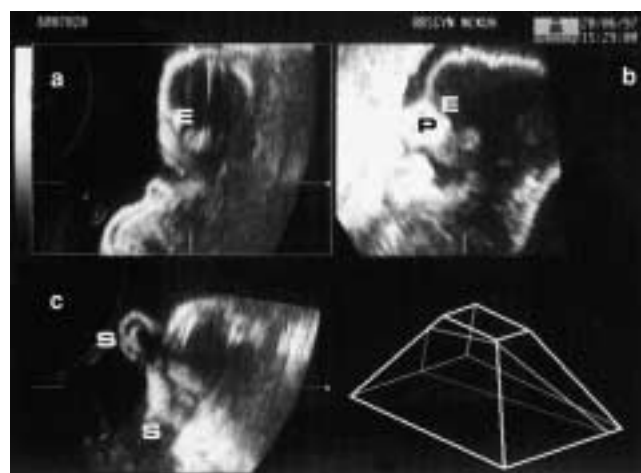


Figure 1 Images of three perpendicular planes in three-dimensional ultrasound: (a) coronal view; (b) sagittal view; (c) horizontal view. Note the proboscis-like mass near the eye in panel b, and the contiguous ears below the absent mandible on panel c. E, eyeball; P, proboscis-like mass; S, synotia – the contiguous low-set ears



Figure 2 The transparent mode (sagittal view) of three-dimensional ultrasound, demonstrating protuberance of the nose-mouth complex (see the corresponding gross picture in Figure 3b). E, eyeball; P, protuberance of the nose-mouth complex; M, maxillary process; S, low-set ears

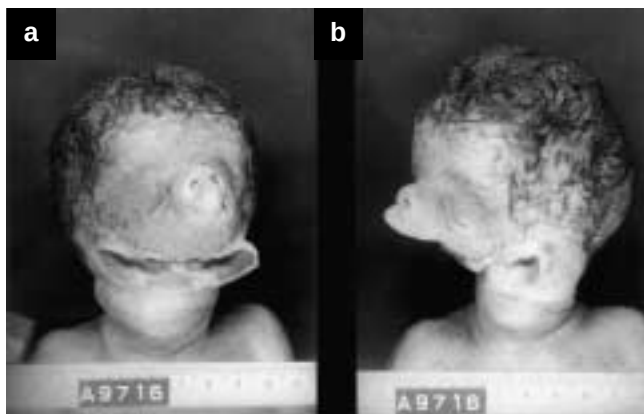


Figure 3 (a) Frontal view of the infant showing synotia, nose-mouth fusion, microstomia and hypotelorism; (b) lateral view of the infant showing agnathia, synotia and protuberance of the nose-mouth fusion

Further investigation by three-dimensional ultrasound, using a Combison 530D machine (Kretztechnik, Zipf, Austria), demonstrated facial anomalies (Figures 1 and 2) which included synotia, i.e. adjacent low-set ears in the anterior cervical region in the absence of the mandible; microstomia, presenting as nose-mouth fusion; protuberance of the nose-mouth complex; and agnathia, due to the absence of bony tissue between the maxillary process and the ears.

Isolated otocephaly was diagnosed and, following counselling, the couple opted for therapeutic termination. A stillborn female fetus with otocephaly (Figure 3) was delivered 5 days later. Informed consent was obtained for an autopsy examination to be performed. The pathological findings correlated with the prenatal sonographic images. Tissue culture from skin and muscle cells showed a normal female karyotype (46,XX).

DISCUSSION

Otocephaly is a very rare, lethal, developmental complex, characterized by extreme hypoplasia or absence of the mandible, synotia, microstomia and aglossia. In humans, otocephaly is often found in association with other anomalies including holoprosencephaly, encephalocele, situs inversus totalis, renal defects and axial skeletal abnormalities¹⁻³. The ultrasound images and pathological findings in our case revealed an isolated otocephaly, which is a defect of the ventral portion of the first branchial arch derived from defective neural cell migration and formation³.

Prenatal diagnosis of otocephaly is extremely rare and usually very difficult. The majority of the previous cases have been incidentally found following identification of other associated anomalies such as holoprosencephaly, encephalocele, situs inversus totalis or renal defects⁴⁻⁸. To date, there have been no convincing prenatal images of the characteristic features of otocephaly. In this case, we were able to distinguish, using three-dimensional ultrasound, the craniofacial features at 24 weeks of gestation.

In our experience^{9,10}, three-dimensional ultrasound offers several advantages over two-dimensional ultrasound. The complete defect can be captured in the ultrasound volume in a few seconds and the anatomic features can be displayed step by step from the stored volumes without keeping a highly anxious woman on the examination table. In this case, the three perpendicular planes displayed simultaneously showed the relative anatomic relationships of synotia and the protuberance of the nose-mouth fusion. The transparent mode also clearly illustrated the microstomia, proboscis and agnathia. A combination of these modes provided convincing evidence of the prenatal diagnosis of otocephaly and gave credence to our belief that three-dimensional ultrasound may be the method of choice in the early prenatal diagnosis of congenital anomalies.

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