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SHORT COMMUNICATION

THE ULTRASONIC DETECTION OF AN ISOLATED CRANIOSYNOSTOSIS

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SUMMARY

The prenatal detection of scaphocephaly, an isolated form of craniosynostosis, is presented. The diagnosis was made at 34 weeks of gestation in a woman with polyhydramnios. The ultrasound appearance and postnatal follow-up are presented.

KEY WORDS: isolated craniosynostosis; scaphocephaly; prenatal diagnosis

INTRODUCTION

The use of real-time ultrasound equipment makes prenatal diagnosis of fetal skull deformities possible. Commonly encountered deformities include the lemon shape associated with spina bifida and the strawberry shape associated with trisomy 18 (Nicolaides *et al.*, 1986, 1992). Isolated or simple craniosynostosis, however, is a developmental abnormality that is rarely found at routine ultrasound examination, although estimations based on the data of Hunter and Rudd (1977) and Tessier (1971) reveal a frequency between 0·4 and 1 in 1000.

The only cases of craniosynostosis that have been reported concern fetuses with complex and marked craniosynostosis syndromes such as cloverleaf skulls, involving multiple sutures (Brahman *et al.*, 1979; Banna *et al.*, 1980; Salvo, 1981); Apert syndrome, characterized by craniosynostosis, midfacial malformations, and symmetric syndactyly of the hands and feet (Hill *et al.*, 1987); Crouzon syndrome, characterized by craniosynostosis, maxillary hypoplasia, and ocular proptosis (Menashe *et al.*, 1989); and Pfeiffer syndrome, consisting of craniosynostosis, broad thumbs, broad big toes, and partial soft tissue syndactyly of the hands (Hill and Grzybek, 1994). In general, the craniosynostosis syndromes represent less than 1 per cent of all cases with craniosynostosis (Cohen, 1986).

To our knowledge, this is the first report on the prenatal detection of an isolated form of craniosynostosis.

CASE REPORT

A 31-year-old woman, gravida 2, para 1, with a normal previous pregnancy and delivery was referred to our Division of Prenatal Diagnosis. A detailed ultrasound scan was undertaken at 33$\frac{1}{7}$ weeks because of suspected polyhydramnios. The present pregnancy had so far been uncomplicated. There was no consanguinity. The sonographic evaluation showed a single fetus in cephalic

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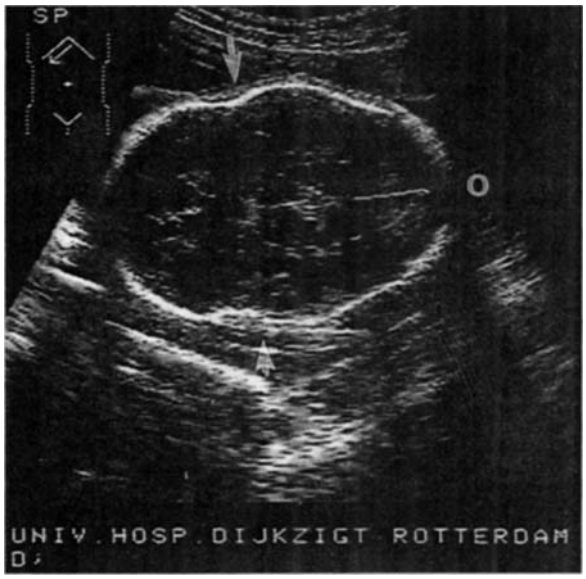

Fig. 1—Axial scan of the fetal head showing sharp circumscribed dilatation on both sides of the cranium (arrows) and a pointed occiput (o)

presentation. The mild polyhydramnios was confirmed. No clinical explanation for the polyhydramnios could be determined. The fetal biparietal diameter (BPD) was 8·0 cm, which is just above the tenth percentile for gestational age. The head circumference (30·5 cm) and femur length (6·6 cm) were both on the 60th percentile (Snijders and Nicolaides, 1994). At the level of the BPD a relatively sharp circumscribed dilatation on both sides of the cranium was seen, giving the appearance of a cloverleaf in the transverse section (Fig. 1). The occiput appeared pointed. The intracranial anatomy was normal and no other anomalies were detected. In brief, the fetal skull showed dolichocephaly with a cloverleaf aspect, suspicious of craniosynostosis. An amniocentesis was performed, which revealed a normal male karyotype. The alpha-fetoprotein (AFP) level was normal.

At a gestational age of $38\frac{6}{7}$ weeks, a 3685 g male newborn was delivered spontaneously. The skull showed frontal bossing, a non-palpable sagittal suture, and a projection of the back of the head. No other congenital deformities were diagnosed. The postnatal skull X-rays confirmed a partially closed sagittal suture and patent lambdoid and coronal sutures (Fig. 2).

At the age of 5 months, a healthy infant was seen with a head circumference of 46 cm, which is above the 90th percentile. An X-ray and CT-scan of the skull showed partial closure of the sagittal suture. The remaining sutures were all open. An EEG showed no abnormalities.

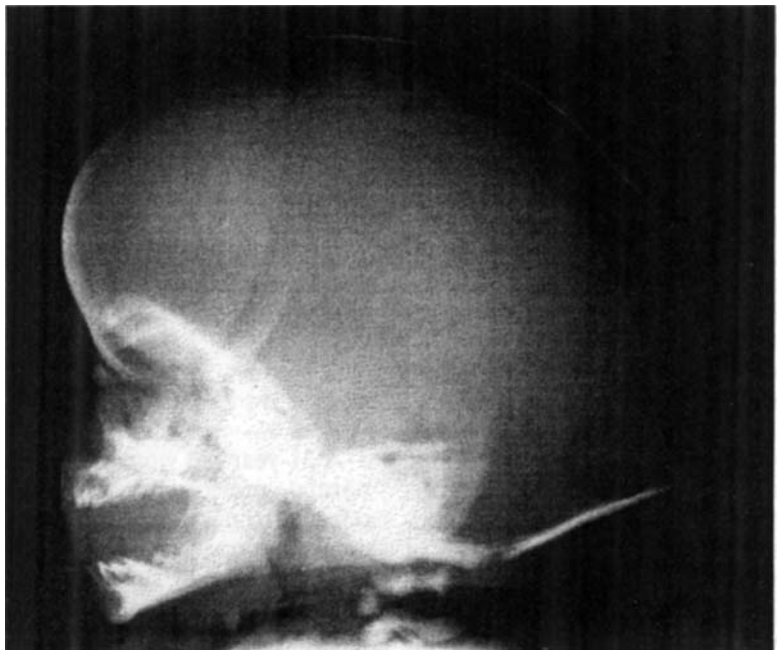

Fig. 2—Postnatal lateral skull X-ray showing the scaphocephalic deformation

The skull shape was surgically remodelled by remoulage of the parieto-temporo-occipital part of the skull. The postoperative period was without complications. At the age of 4 years, the child's physical and mental development corresponds to his age.

DISCUSSION

In general, a cloverleaf skull is regarded as a severe and complex malformation, as in most instances it is combined with other congenital abnormalities and results in early death (Cohen, 1986). In this case, the malformation, which was diagnosed prenatally as a cloverleaf-like deformity, appeared to be a scaphocephaly. Patients with a scaphocephalic malformation have a better prognosis than patients with a cloverleaf skull, in the absence of other abnormalities and the less severe character of the skull deformity.

The term 'scaphocephaly' was introduced in 1860 by Von Bauer. It describes the elongated and narrow shape of the skull (Van der Meulen *et al.*, 1990). Besides the abnormally formed calvaria, a decreased cephalic index and an increase in head circumference are frequently found. Patients with a scaphocephalic skull most often show a premature closure of the sagittal suture (craniosynostosis). Scaphocephaly is seen as an isolated entity or as part of a complex of abnormalities. Sagittal involvement is the most common type of isolated craniosynostosis (Cohen, 1986). Hunter and Rudd (1977) noted an incidence at birth of sagittal synostosis of 1 in 4200, with a marked male predominance (7:3). In spite of its relatively high incidence, scaphocephaly, like other forms of isolated craniosynostosis, has not been reported at routine ultrasound examination up to the present case.

In contrast, all the reported syndromic craniosynostoses were diagnosed during the third trimester of pregnancy, in particular between 31 and 36 weeks, either on the skull deformities and/or on the associated anomalies. In one of these cases, however, abnormal skull measurements had already been found at 18 weeks' gestation (Brahman *et al.*, 1979). This finding coincides with expectations based on normal and abnormal skull development. During normal development, the majority of the sutures are formed between bone centres of different skull bones at 16 weeks' gestation (Vermeij-Keers, 1990). In synostotic skulls, it has been postulated (Vermeij-Keers, 1990) and subsequently proven (Mathijssen *et al.*, 1995) that instead of normal suture formation, direct fusion of dislocated bone centres takes place at this period of gestation or even earlier. Therefore, more attention should be paid during ultrasound examination to whether the abnormal fusion of bone centres does lead to a detectable skull deformity between 18 and 22 weeks of gestation.

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