IMPORTANCE OF ANTENATAL KARYOTYPING IN THE MANAGEMENT OF A CASE WITH VESTIGIAL RADII

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ABSTRACT

An uncommon presentation of vestigial radii in a Trisomy E fetus was detected prenatally with ultrasound scanning. Appropriate counselling and obstetric management were thus carried out avoiding an unnecessary caesarean section. The importance of identifying high risk pregnancy and prenatal diagnosis by a qualified personnel cannot be overstressed.

Key words: Antenatal, Karyotyping, Vestigial, Radii.

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CASE REPORT

Mrs T L G, a 40 year old Chinese G2 P1 lady, was referred by the general practitioner to have specialist care at 29 weeks of amenorrhoea (7.8.86). She had no amniocentesis in early pregnancy but was happy because an ultrasound examination done at 28 weeks in the private sector showed a normal male baby. She was healthy, the uterine size corresponded to 30 week gestation with clinically excessive liquor. The fetal parts and heart sounds were detected with difficulty. The fetus was a singleton in longitudinal lie presenting by the breech.

Ultrasound scanning for hydramnios in our Antenatal Diagnostic Centre by one of the co-authors (Y C W) confirmed increased amount of liquor amnii, and asymmetrical growth retardation of the fetus. This led to search for fetal anomalies which revealed: vestigial radius on the right, and trace of radius on the left associated with bilateral shortening of ulna. The interocular distance was well below 2 standard deviations. Screening of the renal, cardiovascular and axial skeletal system did not reveal major abnormality.

Though normal babies are born with absent or vestigial radii, the patient's age, polyhydramnios, growth retardation and the presence of hypotelorism, which is known to be associated with chromosomal aberrations (1, 2) prompted to do an amniocentesis for karyotyping. The aim of the procedure was to enable a better decision regarding continuation of pregnancy and for avoidance of abdominal delivery in case of fetal distress.

She was well until 32 weeks of amenorrhoea (27.8.86) when she was admitted to the hospital with rupture of membrane at 0200 hrs and mild infrequent uterine contractions (1 every 6 min). Vaginal speculum examination confirmed the membrane ruptured and drainage of clear liquor. The baby had a cephalic presentation. Cardiotocography showed some variable decelerations.

The amniocentesis result was still pending. It was decided, after discussing the situation with the couple, that no active intervention on fetal grounds would be taken.

The fetal heart sounds were not detected the next morning. Intrauterine death was confirmed by an ultrasound examination. The couple appeared to be psychologically relieved despite the grief. The labour was induced by 1 intramuscular injection of a prostaglandin analogue and labour started 1 hour later. The cervix was fully dilated in 30 minutes. A fresh stillbirth with multiple congenital abnormality was delivered spontaneously together with the placenta and cord. Karyotyping of the amniotic cell culture which was available at this stage showed 47, XY + 18 (Trisomy E).

Postmortem examination showed a fresh stillbirth with rudimentary external ears, partial syndactyly between 2nd and 3rd digit of the right lower limb, both hands showed fixed flexion deformity with 4 digits seen on right hand and 2 digits on left hand. X-rays confirmed bilateral absent radii. Patent ductus arteriosus, patent foramen ovale and high ventricular spetal defect were present. Both right and left lungs showed three lobes. All other systems were normal.

DISCUSSION

This abnormality would have been diagnosed earlier and pregnancy terminated in second trimester if she had her amniocentesis in second trimester in view of her age. The psychological trauma and the difficulty in decision to operatively intervene would not arise. This emphasizes the need for early referral of those patients who need prenatal diagnosis.

This case reports the unusual phenotype of Trisomy E, and also illustrates some important points in prenatal diagnosis. In the classical syndrome (3), the baby may have microcephaly, micrognathia, abnormal position of the fingers, etc. None of these occurred in this case. The association of absent radii has been described as an unusual feature (2). Other associated conditions with vestigial radius include Aase Syndrome, Holt-Oram Syndrome, Radial Aplasia-Thrombocytopenia Syndrome, Roberts Syndrome, VATER Association, deLange Syndrome, Fanconi Pancytopenia Syndrome, Seckel Syndrome and Trisomy 13 Syndrome.

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This case illustrates the importance of complete and systematic assessment of fetus which should be exercised in screening for fetal anomalies. The findings of polyhydramnios or growth retardation should prompt a search for congenital malformation or chromosomal aberrations. Chromosomal aberration should be considered if features of hypotelorism or an absent radii is observed on ultrasonography. In case the karyotype report is needed urgently, the use of a short culture technique using foetal cord blood or placental biopsy will be advantageous. Such analytical work-out is required of all those involved in antenatal ultrasonic diagnosis. Meire (4) has pointed out the increasing responsibility in ultrasound antenatal diagnosis shouldered by radiographer (or ultrasonographer) of variable standard. We agree with Meire's (4) suggestion that some form of qualification (eg a diploma in ultrasound) has to be attained before a radiographer (or ultrasonographer) is allowed to carry out an ultrasound examination for fetal anomaly to minimise the chance of misdiagnosis and help him/her to defend himself in case of medicolegal disputes.

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