Porencephaly: A Possible Complication of Chorion Villus Sampling?

A.K. Sharma
S. Phadke

Chorion villus sampling (CVS) has been a major advance in the field of prenatal diagnosis. Many studies have shown that the technique is safe with minimal immediate risks (1-3) but the long term effects on the fetus are not well documented. It is, therefore, necessary to take cognisance of all adverse outcomes to establish the long term safety of this procedure.

Case Report

The patient a 38-year-old G3 P2 Muslim lady with a non-consanguineous marriage underwent CVS at 9 weeks gestation because of her age. The procedure was carried out transcervically at another centre and therefore the details of number of insertions and weight of tissue obtained were not available. She had spotting for two days following the procedure but the pregnancy remained uneventful. Cyto genetic study of the aspirated tissue revealed a normal karyotype.

The patient had not taken any drugs during pregnancy other than vitamins and hematines, her previous pregnancies and deliveries were normal and there was no family history of congenital malformations. At 28 weeks of gestation a GTT was performed which indicated gestational diabetes (F-110 mg/dl, 1 h- 146 mg/dl, 2 h-180 mg/dl). She was controlled on diet and crystalline insulin 5 I.U. twice a day.

An ultrasound performed at 37 weeks revealed an intracranial, unilateral cystic area occupying most of one hemisphere. There was a marked mid line shift (Fig.). Biparietal diameter was 11 cm and head circumference was 38 cm. A provisional diagnosis of porencephaly was made. The patient had an elective cesarean section. The neonate weighed 3.7 kg, had an Apgar score of 8 and had no other apparent congenital malformation. The head circumference was 38.5 cm. CT scan confirmed the diagnosis of a porencephalic cyst.

Discussion

Porencephaly is an extremely rare
condition having an incidence of 1 in 9000. True porencephaly is a developmental anomaly caused by a failure in the migration of cells destined to form the cerebral cortex, and is characterised by cystic cavities usually bilateral and symmetrical. Pseudoporencephaly arises as a consequence of a vascular, infectious or traumatic cause and is usually unilateral.

The following etiological factors have been implicated: trauma, CO exposure, alcohol abuse, antiepileptic drugs, and isoretinoin intake. There has been one case associated with amniocentesis(4). On the other hand there are reports whereby CVS has been associated with congenital malformations, particularly limb reduction defects(5).

In this case it is not possible to conclude whether CVS has been a causative factor since our patient had other predisposing factors in the form of advanced maternal age and gestational diabetes, nonetheless the association is rare enough to deserve mention.

REFERENCES


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Short Rib-Polydactyly Syndrome Type I
(Saldino-Noonan Syndrome)

O.P. Mishra
C. Mohanty
Manoj Kumar
B.D. Bhatia
M. Singh
V. Bhargava

Short rib-polydactyly syndrome type I is an uncommon skeletal dysplasia transmitted by autosomal recessive pattern. It is characterized by respiratory distress due to pulmonary hypoplasia within a narrow dysplastic thorax and extremely short ribs. Abnormal osseous development and multiple visceral abnormalities are common.

Case Report

A 2000 g baby was born of a non-consanguinous marriage at 34 weeks gestation. There was no history of exposure to drugs or irradiation in the antenatal period. The baby suffered severe birth asphyxia (Apgar score 2/10 at 1 min and 3/10 at 5 min). The patient continued to have severe respiratory distress and developed repeated apneic spells at 2 h. He expired 4 h after birth.

The baby's weight was appropriate for gestational age(1); other measurements revealed crown heel length of 37 cm, head circumference 32.5 cm, mid arm circumference 8.5 cm, chest circumference 20.5 cm, abdominal girth 29.5 cm, upper segment (US) 26 cm, lower segment (LS) 11 cm, US/LS ratio 2.36, arm span 32 cm, upper limb length 8 cm and lower limb length 11.5 cm. The anterior (2.5 × 2.5 cm) and posterior (1.5 × 1.5 cm) fontanelle were open and all the sutures were separated. Head appeared relatively large, neck was short and the thorax was hypoplastic with markedly diminished expansion. Abdomen was protuberant with hepato-splenomegaly (3 cm each). No other organs were palpable and there was no ascites. There were loose folds of skin over the chest and extremities. The extremities were small with short hands and feet with postaxial polydactyly (Fig. 1). The scrotum was bifid and the testes were undescended. The phallus was absent while the urethral opening was seen at the base of the scrotum.

On autopsy examination, the lungs were hypoplastic and the right and the left lungs weighted 6.3 and 5.1 g, respectively.

From the Departments of Pediatrics and Anatomy, Institute of Medical Sciences, Banaras Hindu University, Varanasi 221 005, U.P.

Reprint requests: Dr. O.P. Mishra, 6, Rashmi Nagar Colony, B.H.U. Post Office, Varanasi 221 005.

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