Umbilical cord hernia in a child with autosomal recessive chondrodysplasia punctata

O CHANDAVASU AND F DESPOSITO

Neonatal Services, Jersey City Medical Center; and Departments of Pediatrics and Human Genetics, University of Medicine and Dentistry of New Jersey, Jersey City, New Jersey, USA.

SUMMARY An infant with congenital chondrodysplasia punctata with a secondary deformation of umbilical cord hernia is reported. The paper discusses deformation syndromes as anomalies due to unusual mechanical or intrinsic factors.

We report a case of an unusual umbilical cord hernia arising as a secondary deformation in a malformed infant with congenital chondrodysplasia punctata.

Case report

The male proband, the product of an uncomplicated term pregnancy, was delivered by caesarean section because of cephalopelvic disproportion and clinical evidence of fetal distress to a 25 year old primigravida with blood group AB, Rh positive, and VDRL negative. The pregnancy was uneventful except for one episode of maternal urinary tract infection which was successfully treated with ampicillin. An ultrasound at 30 weeks' gestation was normal, and the fetus was said to be compatible in size for the date. The parents are first cousins.

At birth, the baby was deeply cyanotic and in shock and was noted to have multiple congenital anomalies including a large umbilical cord hernia (fig 1). He required immediate resuscitation; Apgar score was 9 at 5 minutes. After stabilisation, the patient was transferred to the Intensive Care Nursery (ICN) at the Jersey City Medical Center. Physical examination in the ICN revealed a term male newborn, weighing 3050 g, length 42 cm, and head circumference 34.5 cm. Multiple anomalies were noted including small skin tags on the lips, short thorax, and widely spaced nipples. Scrotal folds were normal but the testes bilaterally were

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References
undescended. There was a large umbilical cord hernia measuring 12\times 12 \text{ cm} with intestines and liver visible in the sac. A peripheral cataract was present in the left eye. Bone x-rays revealed epiphyseal stippling of the long bones (femora, humeri), abnormal calcification of the sternum, spine, ribs and scapula (fig 2), and periarticular calcification of the left foot (fig 3). After stabilisation, the patient underwent surgical correction of the umbilical cord hernia at 3 hours of age. Postoperatively, the patient went into shock, became cyanotic, and died at 35 hours of age. The mother’s perineal area and the infant’s superficial skin culture grew group B Streptococcus.

Discussion
Chondrodysplasia punctata is a rare genetic disorder involving the long bones, spine, and pelvis with soft and connective tissue defects with characteristic punctate epiphyseal mineralisation. Microcephaly, mental deficiency, cataracts, and an ichthyosiform skin dysplasia are also associated with this disease. It is invariably lethal.
The presence of a large umbilical cord hernia containing intestinal viscera and liver with excessive abdominal skin folds has not been previously noted in this disease. The findings in this case suggest that the umbilical cord hernia arose as a secondary 'deformation' resulting from a combination of extrinsic forces from maternal factors (primigravida uterus and small pelvis) and a malformed fetus (bone dysgenesis and short thorax). The deformative process presumably occurred during the later stages of the pregnancy. The extrinsic forces acted on the malformed fetus and the abdominal contents of the fetus under increased pressure 'herniated' through the area of least resistance, the umbilical cord (fig 4). Adequate abdominal skin folds attest to the fact that there was no primary abdominal wall deformity. As the abdominal contents extruded into the hernial sac the testes were withdrawn from their well-formed scrotal sacs (fig 5). It is also possible that the weakening of the connective tissue of the abdominal wall, because of the basic genetic defect, made it easier for the herniation to occur, given the external pressure.

A careful analysis of congenital malformation syndromes may help to distinguish those abnormalities which represent pleiotrophic manifestations of the abnormal gene from those arising as secondary deformative processes as a result of maternal and intrinsic fetal factors.

References

Correspondence and requests for reprints to Dr O Chandavasu, Neonatal Services, Jersey City Medical Center, Baldwin Avenue, Jersey City, New Jersey 07304, USA.

Trisomy 14 mosaicism in a 2 year old girl

MICHAEL B PETERSEN*, LARS O VEJERSLEV*, AND BENTE BECK†

*Department of Medical Genetics, The John F Kennedy Institute, Glostrup; and †Department of Pediatrics, Kolding Hospital, Kolding, Denmark.

SUMMARY Trisomy 14 mosaicism with 6% trisomic cells in blood and 16% in skin fibroblasts was found in a 2 year 2 month old girl with mild psychomotor retardation, craniofacial dysmorphism, pectus carinatum, curved fifth fingers, retarded bone age, and signs of an