Sirenomelia in uneventful pregnancy

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Summary

A rare case of sirenomelia at 38 weeks of gestation is reported. Fusion of the lower extremities and incomplete development of the bony pelvis was associated with agenesis of the urinary and genital systems, anorectal atresia and a single large umbilical artery. There was complete situs inversus of the single lower limb. This was composed of two partially fused femurs, a common tibia without fibula, and a rudimentary foot having three metatarsal bones and their corresponding toes. The present case was also interesting for its association with hypoplasia of the lungs. The pathogenesis of sirenomelia is discussed.

Key words: Sirenomelia; Sympodia; Pulmonary hypoplasia; Oligohydramnios.

Introduction

Sirenomelia, also known as the mermaid syndrome or sympodia, is a rare congenital anomaly characterised by complete or partial fusion of the lower extremities; it occurs in one of 60,000 births [1, 2]. The disorder is invariably associated with severe malformations of the urinary, genital and the lower gastrointestinal organs. Not infrequently, neural tube defects are present. The severity of these malformations are incompatible with life. Prenatal diagnosis is not always possible because of oligohydramnios [2, 3].

The cause of sirenomelia remains obscure, but a genetic mechanism has been refuted because there are no familial cases on record. This conditions occurs with an excess frequency in monozygotic twins and, to a lesser extent, in infants with a maternal diabetic background. There is, indeed, a 100-150 fold increase in the incidence of sirenomelia in identical twins, and a relative risk of eight among infants of women with insulin-dependent diabetes [4].

We report a case of sirenomelia associated with pulmonary hypoplasia in a pregnancy complicated with oligohydramnios. Our case was connected with a large single umbilical artery. The pathogenesis of this sporadic defect is discussed.

Case report

The infant was stillborn at 38 weeks' gestation with a body weight of 1210 g. It was the child of a 33-year-old married women, gravida 4 para 0. Both parents were healthy. There was no connection with identical twins, maternal diabetes mellitus, family history of congenital malformations or infectious disease during the gestational period. The mother denied any exposure to known teratogens, including irradiation, drugs, alcohol and tobacco. Her pregnancy was uneventful except for oligohydramnios that was noted during the third trimester. At 34 weeks' gestation, ultrasonography indicated intrauterine growth retardation (IUGR). A repeat ultrasonographic examination at 38 weeks' gestation indicated intrauterine fetal death.

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Postmortem examination

Postmortem examination of the stillbirth showed profound disturbances in the caudal part of the infant (Figures 1 and 2), both skeletal and visceral. The skeletal abnormalities included incomplete development of the pelvis with absence of both ischial bones and failure of acetabulum formation. The iliac bones and the pubis were unaffected. Both hip joints lied underneath the lateral edges of the pubic symphysis. The femurs were fused at the level of the upper metaphysis. The right femur ended in deformed condyles that articulated with a laterally lying patella, while the left femur ended in a normal knee joint with an intact extensor mechanism, normal ligaments and

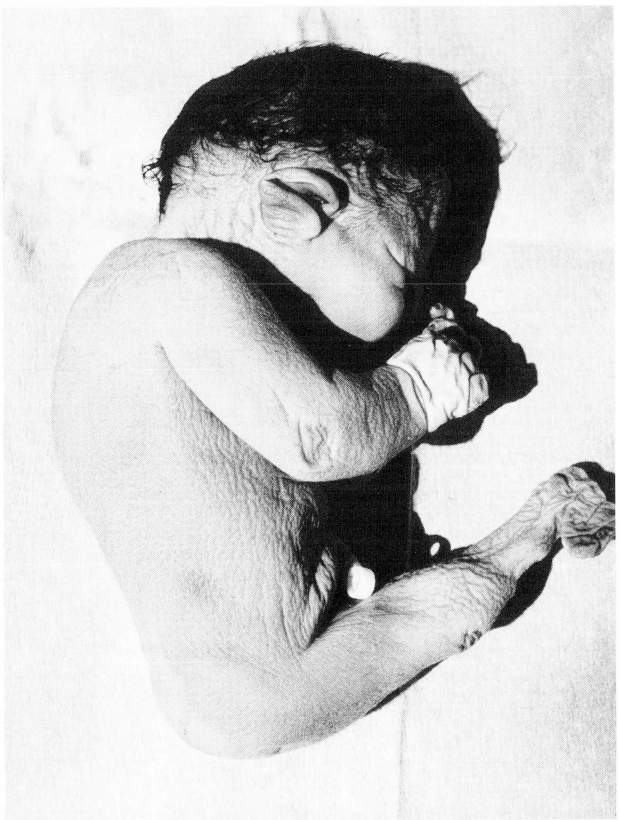

Figure 1. – Sirenomelia showing fusion of the lower extremeties.

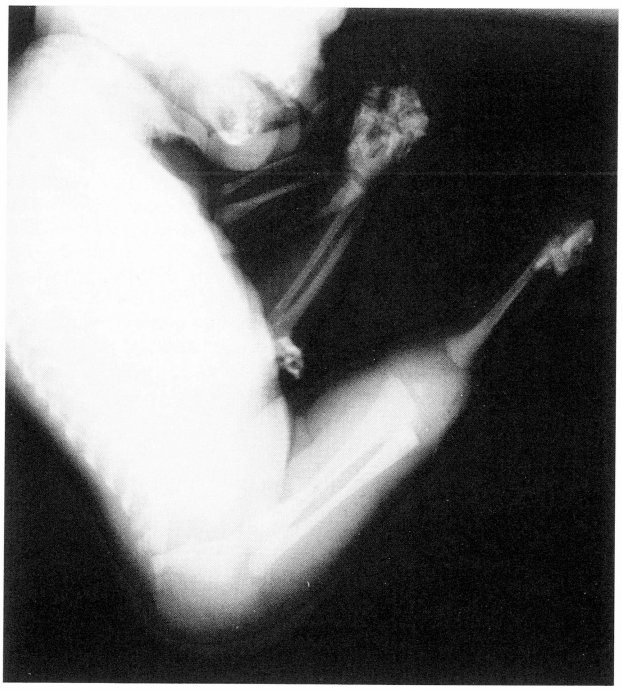

Figure 2. – Radiograph of the sirenomelus infant.

menisci. The lower end of the femur was rotated 180° posteriorily (complete situs inversus). The single tibia was not accompanied by a fibula. There was absence of the fourth and fifth metatarsal bones together with the corresponding phalanges of the toes.

The associated visceral abnormalities comprised agenesis of the kidneys, ureters, bladder, urethra, and also agenesis of the internal and external genitalia. The anus was imperforated. A large single umbilical artery in direct communication with the abdominal aorta was detected immediately below the diaphragm. The abdominal aorta distal to the origin of this vessel gave off no branches before its bifurcation into the common iliac arteries.

The lungs were smaller than normal and had a combined weight of 10 g. The lung: body weight ratio was 0.008. The heart, brain, liver, spleen and thymus were unremarkable. The placenta weighed 200 g and measured 12 x 12 x 2 cm. The umbilical cord was 35 cm in length.

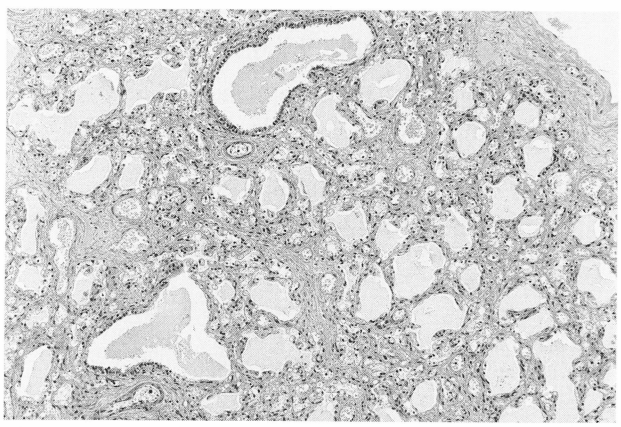

Figure 3. – Hypoplasia of the lungs. Note multiple outpouchings of the terminal bronchioles, paucity of terminal alveolar units, and much connective tissue.

Histological examination

Histological examination of the right and left lung revealed paucity of terminal alveolar units and replacement by connective tissue (Figure 3). The terminal bronchioles showed multiple outpouchings forming terminal alveolar sacs, lined by cuboidal epithelium. The picture was that of severe pulmonary hypoplasia. The placenta showed extensive fibrosis of chorionic villi and fetal artery stenosis with focal areas of infarction and calcification. The umbilical cord contained only an artery and a vein. Histological examination of the remaining tissues was within normal limits.

Discussion

Although the defining feature of sirenomelia is fusion of the lower limbs, the disorder is invariably associated with severe visceral malformations, such as agenesis of the kidneys and pelvic organs, leading to intrauterine death. An equally important characteristic in the sirenomelia complex is the presence of a single large anomalous umbilical artery, usually designated persistent vitteline artery [1], which was thought to be the cause rather than consequence of this syndrome (vascular disruption hypothesis). Other common abnormalities include deformities of the bony pelvis and neural tube defects, while cardiovascular, respiratory and upper gastrointestinal tract malformations are less frequently seen [1]. Despite the plethora of fetal abnormalities, the intrauterine pregnancy progressed uneventfully by to term and resulted in the birth of a stillborn infant. Ultrasound scan revealed intrauterine growth retardation (IUGR), but failed to diagnose sirenomelia, apparently because of oligohydramnios. It is indeed difficult to appreciate the condition in the absence of an adequate amount of amniotic fluid because of the poor quality of imaging [2, 3]. Sirenomelia is most commonly encountered in identical twins and infants of diabetic mothers, but the latter is controversial and, certainly, did not mark our case. An increased incidence of sirenomelia was also noted among mothers with previous abortions [5], and this conforms with our report.

Sirenomelia originates early in embryonic life [6], but its aetio-pathogenesis remains obscure. A genetic mechanism has been refuted, since chromosomal abnormalities and familial inheritance are lacking. It is possible to represent a developmental field defect of the posterior axis caudal blastema, resulting in complete or partial fusion of the lower limb buds [5]. Stevenson and coworkers introduced a fascinating hypothesis: the incomplete development of the lower half of the embryo is the result of "vascular steal" through a single large umbilical artery arising directly from the abdominal aorta, instead of the usual paired umbilical arteries originating from the internal iliac arteries [6]. The abdominal aorta distal to the origin of this major vessel is always subordinate and gives off no branches before its bifurcation into the common iliac arteries. This deprives the caudal part of the embryo of nutrients [2].

The phenomenon of a single large umbilical artery, combined with the absence of distal branching of the abdominal aorta, not only substantiates a pathogenetic

mechanism for sirenomelia, but also forms an invaluable criterion for the prenatal diagnosis of this rare anomaly by Doppler ultrasound [7].

Our case of sirenomelia is also interesting for its association with hypoplasia of the lungs. Pulmonary hypoplasia signifies a reduced number or size of alveoli, but is best defined as the ratio of lung weight to body weight. This ratio is less than 0.012 in term babies [8]. Pulmonary hypoplasia is not infrequently associated with other congenital malformations, often having a causal relationship. It is most probably that in our case pulmonary hypoplasia was secondary to renal agenesis.

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