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SIRENOMELIA IN AN INFANT OF A DIABETIC MOTHER*

A Case Report

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SUMMARY: Gürakan B, Karaaslan E, Balcı S. (Social Security Maternity Hospital, Ankara, Turkey). Sirenomelia in an infant of a diabetic mother: a case report. Turk J Pediatr 1986; 38: 393-397.

Sirenomelia is a rare fatal condition characterized by fusion of the lower limbs. Its etiology is unknown. It is believed that there is a connection between sirenomelia and maternal diabetes but this association has not been firmly established. Here a case of a sirenomeliform infant born to a diabetic mother is reported. *Key words: diabetic mother, sirenomelia.*

The characteristic feature of sirenomelia is fusion of the lower extremities. The most common associated anomalies are the absence of the sacrum and other defects of the vertabrae; imperforate anus and absence of the rectum; absence of external and internal genitalia; and renal agenesis and absence of the bladder[1].

Sirenomelia is a rare congenital malformation with an estimated incidence of one in 60,000 births[2]. This defect is also accepted to be the most severe form of caudal regression syndrome (CRS), which is known to be frequently associated with maternal diabetes. As a result, the association between maternal diabetes and sirenomelia may be expected; however, there are only a few reported cases of sirenomelia as the product of a diabetic pregnancy. Here an additional case is detailed to emphasize the connection between sirenomelia and maternal diabetes.

Case Report

This investigation focused on the seventh pregnancy of a 31-year-old woman who had an eight-year history of diabetes mellitus (para 3, abortus 4). Her diabetes had reportedly been controlled satisfactorily by oral antidiabetics. During her pregnancies, however, she had been treated with insulin. Her father was also diabetic, but there was no family history of neonatal deaths or anomalies of the limbs.

Her first child, who was apparently normal, died because of gastroenteritis when he was three months old. Among the remaining pregnancies, the only surviving child, born from her fourth pregnancy, was seven years old as of this writing and had club foot deformity.

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In the fifth month of her last pregnancy, the mother was informed by her doctor that oligohydramnios was present and the fetus was likely to be anomalous. Termination of pregnancy was recommended, but the patient refused. Three months later she was admitted to our hospital, and in the 38th week of gestation a sirenomeliform infant was delivered by cesarean section because of fetal distress. Oligohydramnios was confirmed.

The infant was 2400 g in weight and 46 cm in length. The lower extremities were fused to the level of the heels (Fig. 1). The feet were separated and rotated laterally. Each foot had five toes. There were no external genitalia or urinary tract orifice, and the anus was imperforate. The upper limbs, chest and abdomen showed no evidence of malformation. Low-set ears, a flat nose and prominent epicanthal folds compatible with "Potter's facies" were present.

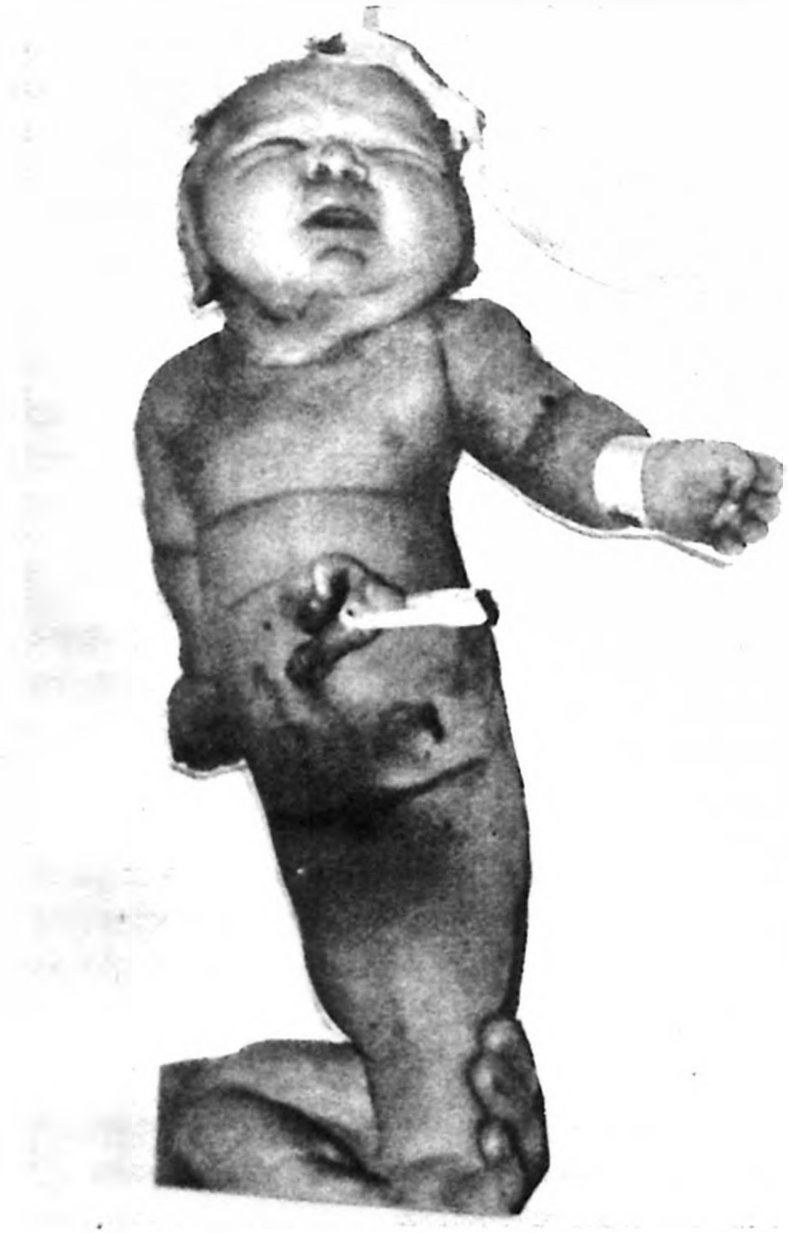

Fig. 1: Anterior aspect of the sirenomelic infant. No external genitalia are present.

X-ray studies demonstrated no abnormalities in the upper limbs, head or trunk. In the caudal region, the sacrum was absent and the pubis was represented by a fused midline bone. Both femurs and tibias were present, but there was only one fibula in the midline between the tibias (Fig. 2).

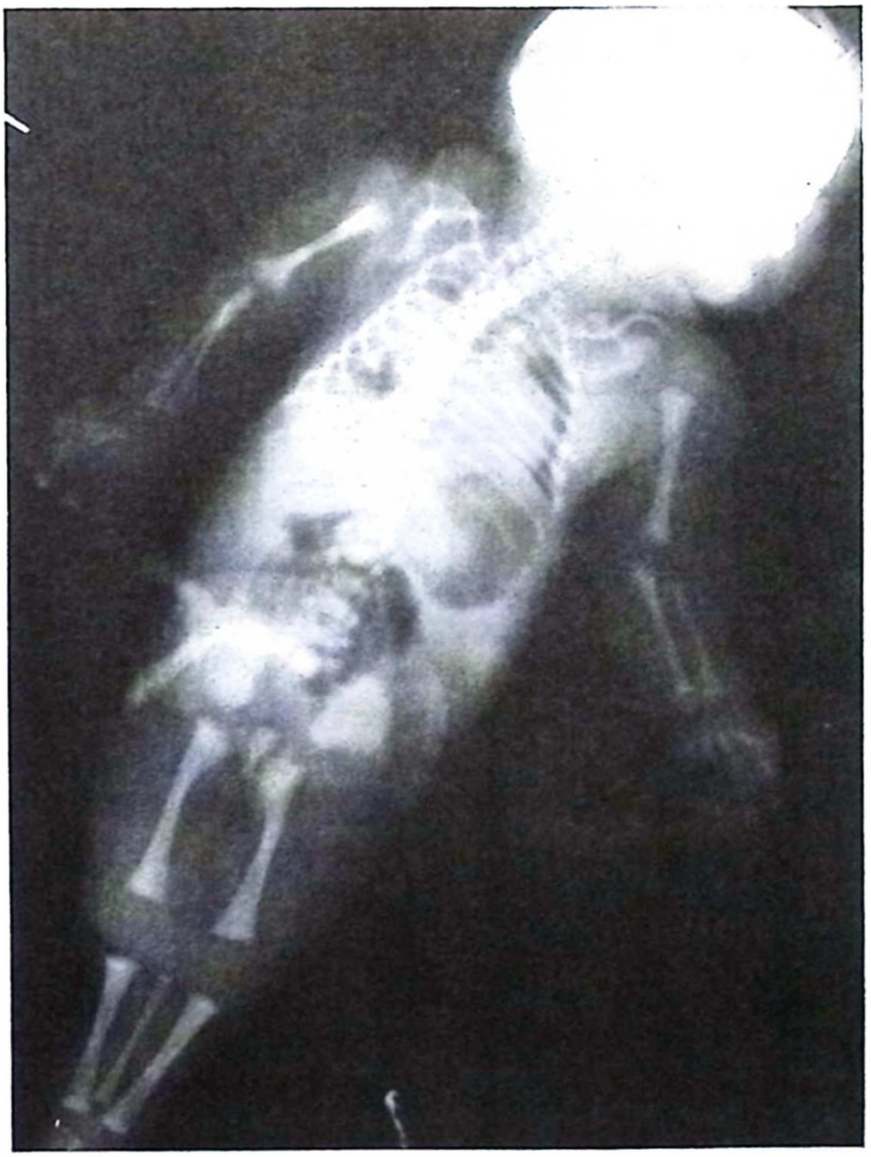

Fig. 2: Antero-posterior radiograph of the infant.

The baby was in respiratory distress and survived only six hours. Unfortunately, the parents refused postmortem examination.

Discussion

The precise cause of sirenomelia is still unclear. Associated conditions have led to speculation regarding its etiology. Intrauterine forces causing pressure on the tail end of the embryo, primary failure of development of the caudal somites, and nutritional deficiency of the lower part of the body have been suggested as pathogenetic mechanisms.

The nutritional deficiency theory, supported by Kampmeimer[3] and later by Stevenson, et al.[4] has been most widely accepted. According to Stevenson, et al.[4] defects found in sirenomelia can be explained by a vascular steal produced by a single, anomalous umbilical artery, Whether this vascular morphology is the consequence or the casuse of the caudal maldevelopment remains unproven[5].

Various combinations of cranial abnormalities and sirenomelia have been reported. Russel et al.[6] tried to explain the association between cephalic defects and caudal regression anomalad, by a generalized disturbance in cell migration from the primitive streak with secondary damage to the cranial and caudal mesodermal masses. Chondebois and Brunet[7] hypothesized that the speeding-up of autonomous differentiation in the notochord anlage caused malformations in both regions.

An additional and significant factor providing insight into the etiology has been the increased incidence of sirenomelia in twins. In eight to 15 percent of cases, sirenomelic births have been reported as the products of monozygotic twin pregnancies[8]. This association led to the suggestion that the process of monozygotic twinning can cause sirenomelia by interfering with the determination or differentiation of caudal embryonic structures[8, 9].

Sirenomelia is generally accepted as constituting the most severe end of the spectrum of CRS[2, 10]. The frequent observation of CRS among the offspring of diabetic mothers strongly suggests that maternal diabetes plays an important role in the pathogenesis of these malformations[11]. This relationship, however, has not been established in sirenomeliform infants. maternal diabetes was associated with only two of the 80 cases of sirenomelia reviewed in a 1987 study[12]. The low incidence of sirenomelia among pregnancies of diabetics may be explained by the effects of separate nondiabetogenic genes, as hypothesized for CRS. From another point of view, the teratogenic effect of diabetes differs from that of other known teratogens, in that it has no specific phenotypic expression. Many of the defects that are common in the general population occur at increased rates in infants of diabetic mothers. Although anomalies such as sacral dysgenesis carry a high risk in diabetic pregnancies, their actual incidence may be very low as they are extremely rare in the general population[13]. Indeed, among more than 50.000 births that took place in this maternity hospital during the last four years, sirenomelia was observed only once, and this was in the infant of a diabetic mother.

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