SIRENOMELIA (MERMAID SYNDROME) IN AN INFANT OF A DIABETIC MOTHER

F. Davari Tanha, N. Googol and M. Kaveh

Department of Obstetrics and Gynecology, Mirza Koochak Khan Hospital, School of Medicine, Tehran University of Medical Sciences, Tehran, Iran

Abstract- Caudal regression syndrome (caudal dysplasia sequence) is a rare congenital malformation. It has a spectrum ranging from simple anal atresia to the absence of sacral, lumbar and possibly lower thoracic vertebrae and the most severe form called sirenomelia (Mermaid syndrome). Sirenomelia has a sole characteristic which is the lower limbs fusion, with multiple internal structural abnormalities particularly in the renal tract (bilateral renal agenesis). This is a rare condition with a relative risk of 200-250 in diabetic pregnancies. The etiology of this syndrome is not well-known. Maternal diabetes is considered to be the most important causative factor. Genetic predisposition and vascular hypoperfusion have been suggested as the other possible factors.We present birth of an infant with great congenital defect which was categorized as the most intense form of caudal regression syndrome (sirenomelia). The baby was born from an uncontrolled diabetic mother who was ignorant of her diabetes. She had a sonographic report at early third-trimester of pregnancy, which had shown severe oligohydramnios and according to this reason the anomaly of the fetus was not detected at that time (antenatal). Since sirenomelia is a lethal abnormality, the infant died a few hours after birth. As noted above caudal regression syndrome is strongly associated with maternal diabetes; due to metabolic derangement in uncontrolled serum glucose.

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Key Words: Sirenomelia, Mermaid syndrome, caudal regression syndrome, caudal dysplasia sequence, sacral agenesis

INTRODUCTION

Caudal regression is a rare syndrome which represents a spectrum of congenital malformations ranging from lumbosacral spine agenesis to the most severe cases of sirenomelia with lower extremities fusion characteristics (1). We have encountered a case of this defect, born from an uncontrolled diabetic mother. In Sirenomelia, the lower limbs are fused together, sometimes with a single femur. Associated malformations include absent external genitalia, imperforate anus, lumbosacral vertebral and pelvic abnormalities and renal agenesis. The condition has been thought to be part of the caudal regression spectrum. The prevalence of this syndrome is 0.1-0.25: 10,000 in normal pregnancies and the same prevalence has been reported in our country (Iran). As there is strong association between this syndrome and maternal diabetes, it has been estimated that this syndrome has a relative risk of 200 - 250 in diabetes. Up to 22% of fetuses with this anomaly will have diabetic mothers.

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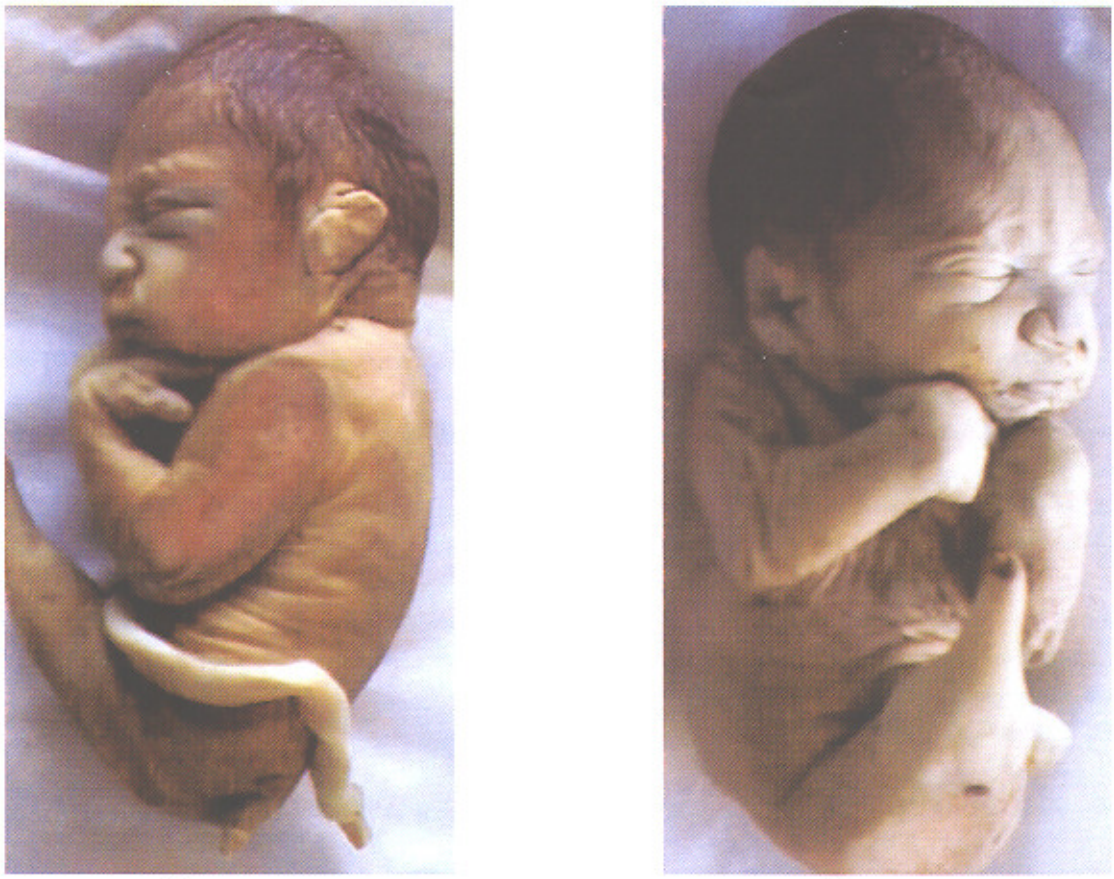
Correspondence:
F. Davari Tanha, Department of Obstetrics and Gynecology, Mirza Koochak Khan Hospital, School of Medicine, Tehran University of Medica Sciences, Tehran, Iran
Tel: +98 21 7484428 Fax: +98 21 8904172
E-mail: Fatedavari@yahoo.com

CASE REPORT

A 33-years-old pregnant woman (3-0-1-3) presented to our hospital with the chief complaint of labor pain. Her gestational age (GA) was estimated 26 weeks by LMP (Last menstrual period) and sonography. She had a sonographic report at early third-trimester of pregnancy which had shown severe oligohydramnios, with no evidence of skeletal or other organ anomaly. Since diminished or lack of amniotic fluid volume (oligohydramnios) disturbs sonographic image resolution, anomalies had not been diagnosed by the radioloist.

Speculum examination showed cervical dilatation of about 3 cm. Ferning and nitrazine-paper tests were negative (the tests done for rupture of membrane diagnosis). Initial lab tests revealed plasma glucose, 210 mg/dl and 333 mg/dl at two consecutive times. Unfortunately the mother was not aware of her diabetes, although she gave a past obstetrical history of diabetes in her previous pregnancy.

Several hours after her admission in labor room, she delivered an infant with cephalic presentation and Apgar Score of 3/0 at 1 and 5 minutes. The newborn baby had gross anomaly known as sirenomelia (Fig. 1 and 2), characterized as fusion of lower limbs. Absence of external genitalia and imperforate anus were also apparent.



A B

Fig. 1A, 1B. Post-mortem photograph of infant with sirenomelia which shows its characteristic feature

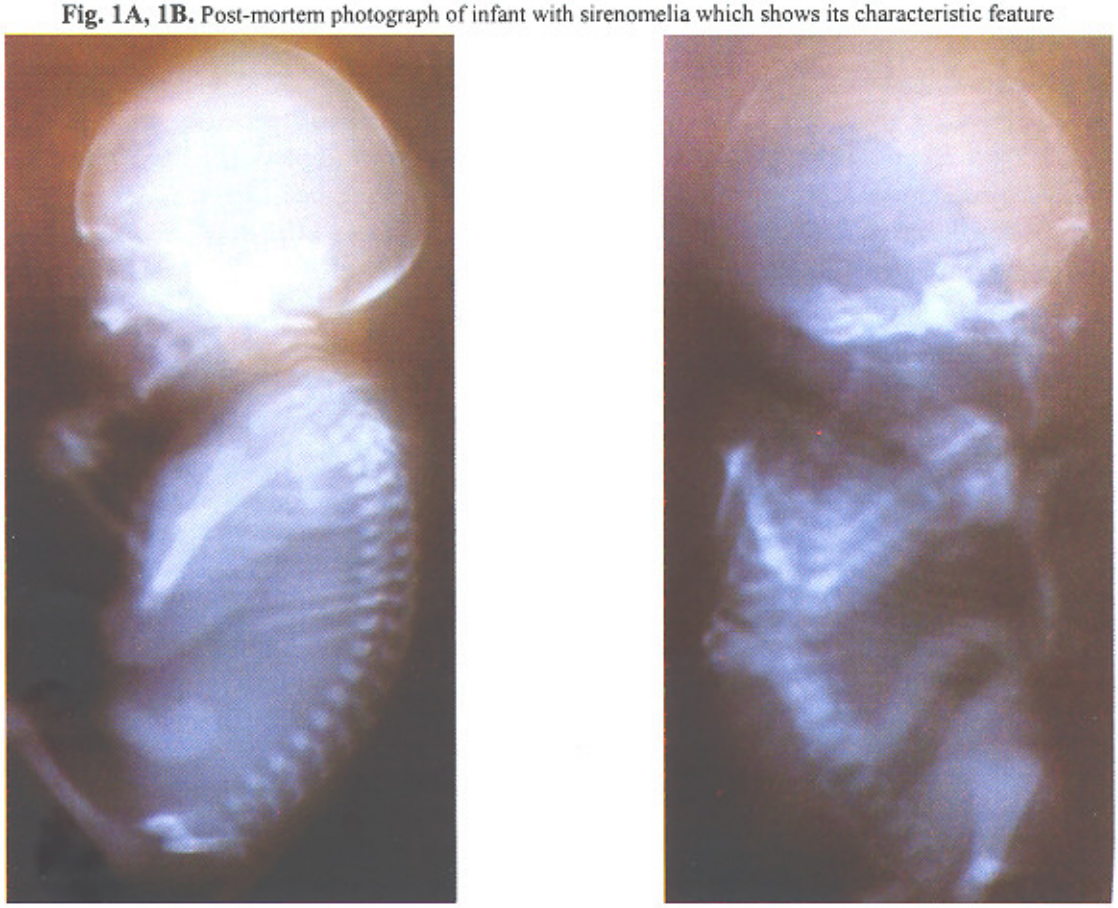


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Fig. 2A, 2B. Radiographs of the same infant, with subtotal sacral agenesis and one femur. Lateral and oblique views

X-rays of the neonate allowed categorization on the basis of skeletal deformity, subtotal sacrococcygeal agenesis and a single dysmorphic lower limb was present (Fig. 3 and 4). Internal organ studies revealed bilateral renal agenesis but other organs such as gastrointestinal tract, cardiovascular system and etc. were normal.

DISCUSSION

The sacrum is a bone at the base of the spinal column that is formed following the fusion of 5 vertebrae. It lies in the region of the buttocks and just below it there is another small series of bones called the coccyx. The presence of the sacrum gives rise to the normal shape of the pelvis. The nerves from the spinal cord pass through a bony canal within the sacrum and exit the sacrum in a number of places to provide nerve supply to the bowel anal sphincters, the bladder and the bladder sphincter and also to the muscle and sensory organs in the lower limbs. Sacral agenesis is a condition that exists when either part or all of the sacrum is absent (2). Total agenesis of the sacrum is manifested by a narrowed pelvis with the medial portion of the iliac ala almost in contact and is included in the group of malformations known as caudal regression syndrome (1, 3).

Caudal regression syndrome is a rare congenital defect in the general population, with a prevalence of 0.1-0.2: 10,000 in normal pregnancies but a relative risk of 200-250 in diabetics (3,4). It represents a broad spectrum of lumbosacral agenesis in the most severe cases of sirenomelia (5). Both sacral agenesis and caudal regression syndrome are related in that the latter syndrome has variable spinal anomalies varying from partial sacral agenesis to complete absence of the lumbosacral spine (5).

Sirenomelia is the most relentless condition of this syndrome. The term comes from "siren" or "mermaid" resulting from characteristic fusion of the lower extremities. The specific anomaly of sirenomelia is based on the presence of lower limbs fusion, associated with other skeletal and lumbar deformities (sacral agenesis). Besides there may be genitouninary, gastrointestinal, cardiovascular and central nervous system abnormalities, and in this aspect there is some overlap with the VATER constellation of anomalies. Sirenomelia is a lethal condition because of bilateral renal agenesis, which leads to severe oligohydramnios and lung hypoplasia (6,7). Both sacral agenesis and caudal regression are strongly associated with maternal diabetes. The aspect of anomalies is probably due to a generalized alteration of mesodermal cell migration in primitive streak period. Maternal metabolic derangement in diabetes may act through a diminished turnover of phosphoinositide or arachidonic acid. These seem to have their effect in the first few weeks of pregnancy so diabetic control should be strict in this period, starting before conception (8,9).

Classification of sirenomelia from caudal regression syndrome is still debated (8) and there are different theories about its pathogenesis. Sirenomelia as a part of caudal regression syndrome has its own pathogenesis which is maternal metabolic derangement in diabetes (as explained above), but in evidences by Twicker et al. sirenomelia and caudal regression are two different entities. In this aspect the pathogenesis of sirenomelia has been proposed to be a vascular steal phenomenon with the single, aberrant, umbilical artery stealing blood supply from the lower torso and limbs (l0,11). Ultrasound is the only reliable way of diagnosing these conditions antenatally as there is no open defect to raise the AFP and the chromosomes are usually normal. The diagnosis of sacral agenesis should be made with knowledge of the normal rate of ossification of the sacral spine not with standing this, the diagnosis has been made correctly suspected at 9 weeks using transvaginal ultrasound by finding a shortened crown rump length in a diabetic mother. Even so the diagnosis was not confirmed until 17th week of gestation. In sagital section the normal curve of the sacrum is lost and the spine appears shortened with an abrupt termination (12,13). If there is caudal regression, the lower limbs may be hypoplastic or fused (as in sirenomelia) and the bladder may be large. In more severe cases the bones of the pelvis may be absent. Also there may be anorectal atresia, tracheo-oesophageal fistula and genitourinary anomalies (14-16). The earlier the caudal regression is detected the more likely it is that there will be major associated defects. The prognosis is ultimately governed by these associated defects.

Sacral agenesis has fewer associated anomalies than caudal regression and carries a greater chance of survival. On the other hand sirenomelia is uniformly fatal. In sirenomelia third trimester ultrasonography for diagnosis is usually impaired by severe oligohydramnios related to bilateral renal agenesis, whereas during the early second trimester the amount of amniotic fluid may be sufficient to allow diagnosis. Early antenatal sonographic diagnosis is important in view of the dismal prognosis and allows for earlier, less traumatic termination of pregnancy (l7,18).

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