SIRENOMELIA

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ABSTRACT

A case of sirenomelia was presented in a new born with perinatal death. The fetus had abnormal appearance of the head and face, single lower extremity, enlarged ears, small eyes and flattened nose. The external genitalia and anus was absent. The lungs were hypoplastic; the kidneys were agenetic. There was a single umbilical artery. A blind pouch was noted at the sigmoid colon and imperforate anus was present. The genitalia was absent; the tubes and fimbria was noted and the uterus could not be well identified. Babygram revealed malformed ischium and pubic bones. Asymmetric femurs, single tibia, and single fibula was noted.

INTRODUCTION

Sirenomelia or the mermaid syndrome of lower extremity fusion or sympus apus, is a severe manifestation of the caudal regression syndrome and has a reported incidence of about 1 in 60,000.[1,2] Sirenomelia is uniformly fatal due to its association with other major anomalies including (a) rotation and fusion of the lower limb; medial position, fusion, or absence of the fibulas; (b) pelvic bone anomalies; (c) renal agenesis; (d) absence of an anus and external genitalia, blind - ending colon; (e) oligohydramnios; (f) single umbilical artery; (g) vascular steal has been proposed as the pathogenic mechanism producing sirenomelia and associate visceral and soft tissue defects.[3]

CASE REPORT

The patient was a product of term pregnancy with unremarkable antenatal period. It was delivered by Caesarian section due to breech presentation and fetal distress. Perinatal death was encountered. At physical examination, the fetus had abnormal appearance of the head and Potter's facies, central cyanosis and a single lower extremity. The ears were enlarged, the eyes were smaller and the nose was flattened. The chest and abdomen appeared normal. There was no external genitalia, nor anus. The hip joints were constantly flexed and knees were extended. Varus deformity of both feet was observed. At autopsy, the lungs were hypoplastic; both kidneys were agenetic. There was a single umbilical artery. A blind pouch was noted at the sigmoid colon and imperforate anus was present. The genitalia was absent; the tubes and fimbria were noted and the uterus was not well identified. The appearance of the fetus was shown on figure 1. Roentgenography was performed shortly after death (Fig. 2).

DISCUSSION

Caudal regression syndrome (CRS) was termed by Duhamel[4] as a spectrum of congenital

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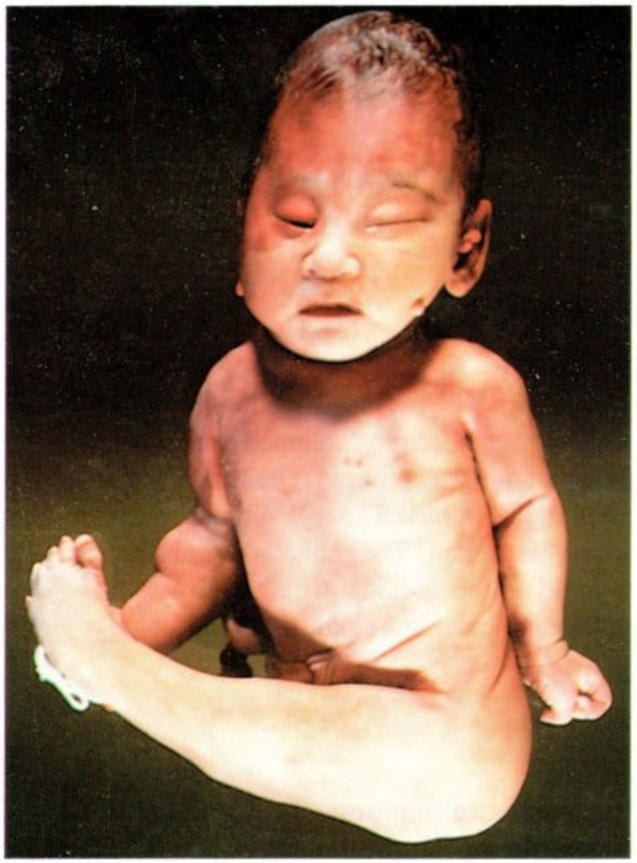
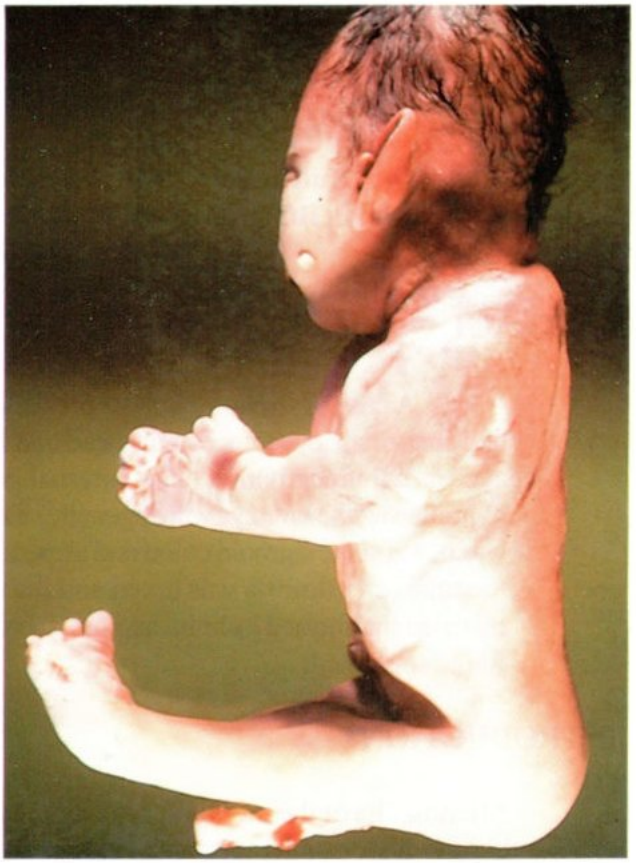
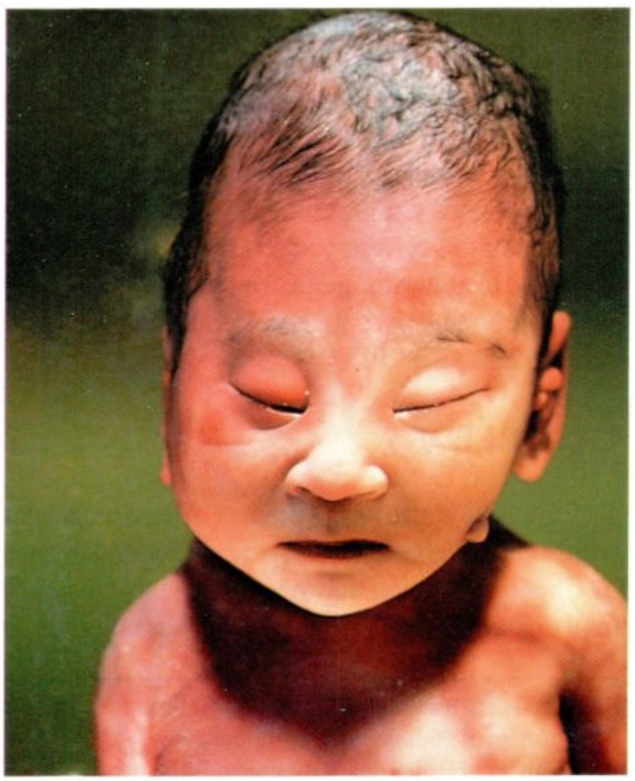


1 (a)



1 (a)

Fig. 1 (a). general appearance of the whole fetus
(b). facial and cranial part



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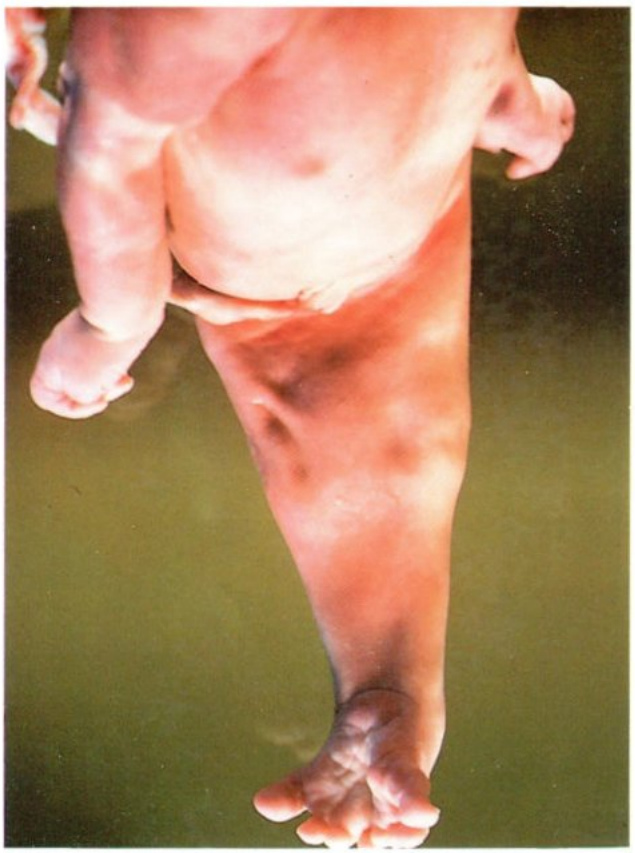

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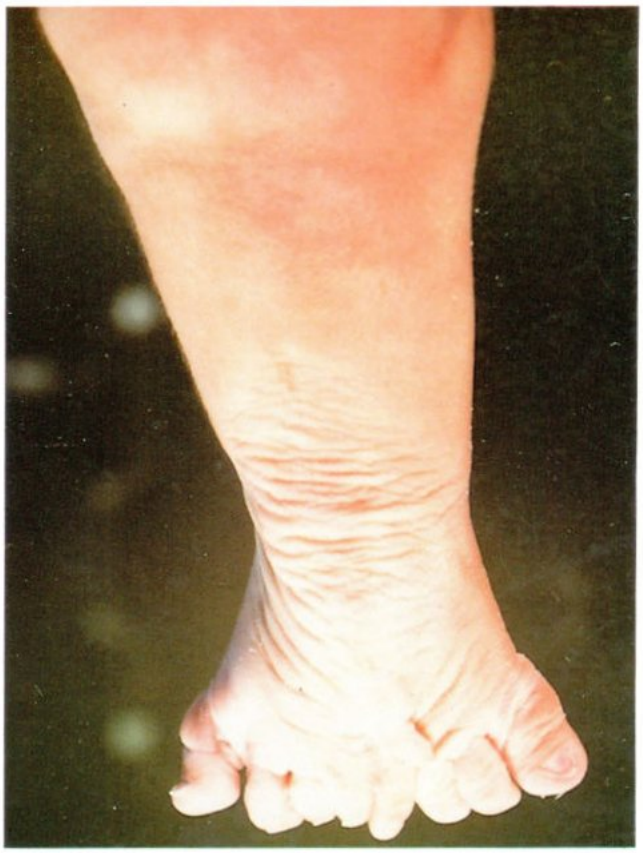
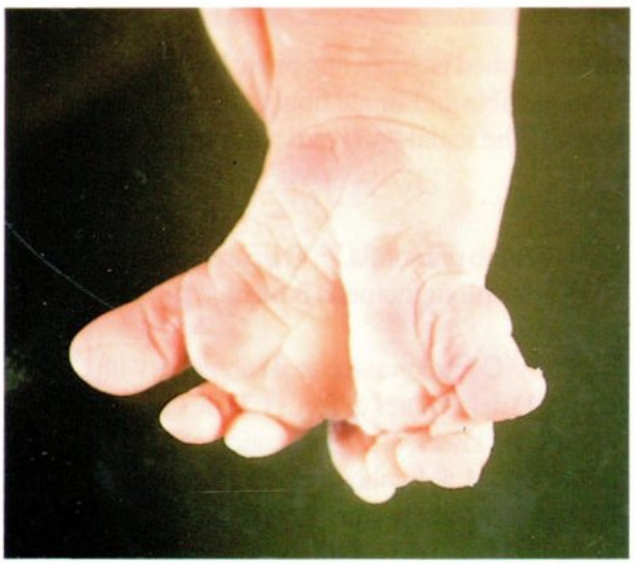


1 (d)



1 (e)

Fig. 1 (c). trunk and lower extremity
(d). lower limb
(e). feet

malformations ranging from simple anal atresia, to absence of the sacrum, lumbar and lower thoracic vertebrae (caudal aplasia/dysgenesia), to the most severe form with fusion of the lower extremities and major visceral malformations known as sirenomelia.[5]

The embryologic insult occurs at the mid-posterior axis mesoderm and/or caudal blastema,[5] but the etiology remains unclear. Factors influencing caudal vertebral development operate before 4 weeks of gestation.[7] Association between maternal diabetes mellitus and CRS was described.[8-11] Although a wide spectrum of vertebral and limb malformations similar to caudal regression were produced in animal models by different teratogens, the only agents associated with this syndrome in humans included organic fat solvents and the appetite suppressant diethylpropion.[12-16] CRS may result from the combination of maternal diabetes and a genetic predisposition.[17]

Vascular hypoperfusion is favored in the pathogenesis of the extreme form of CRS,

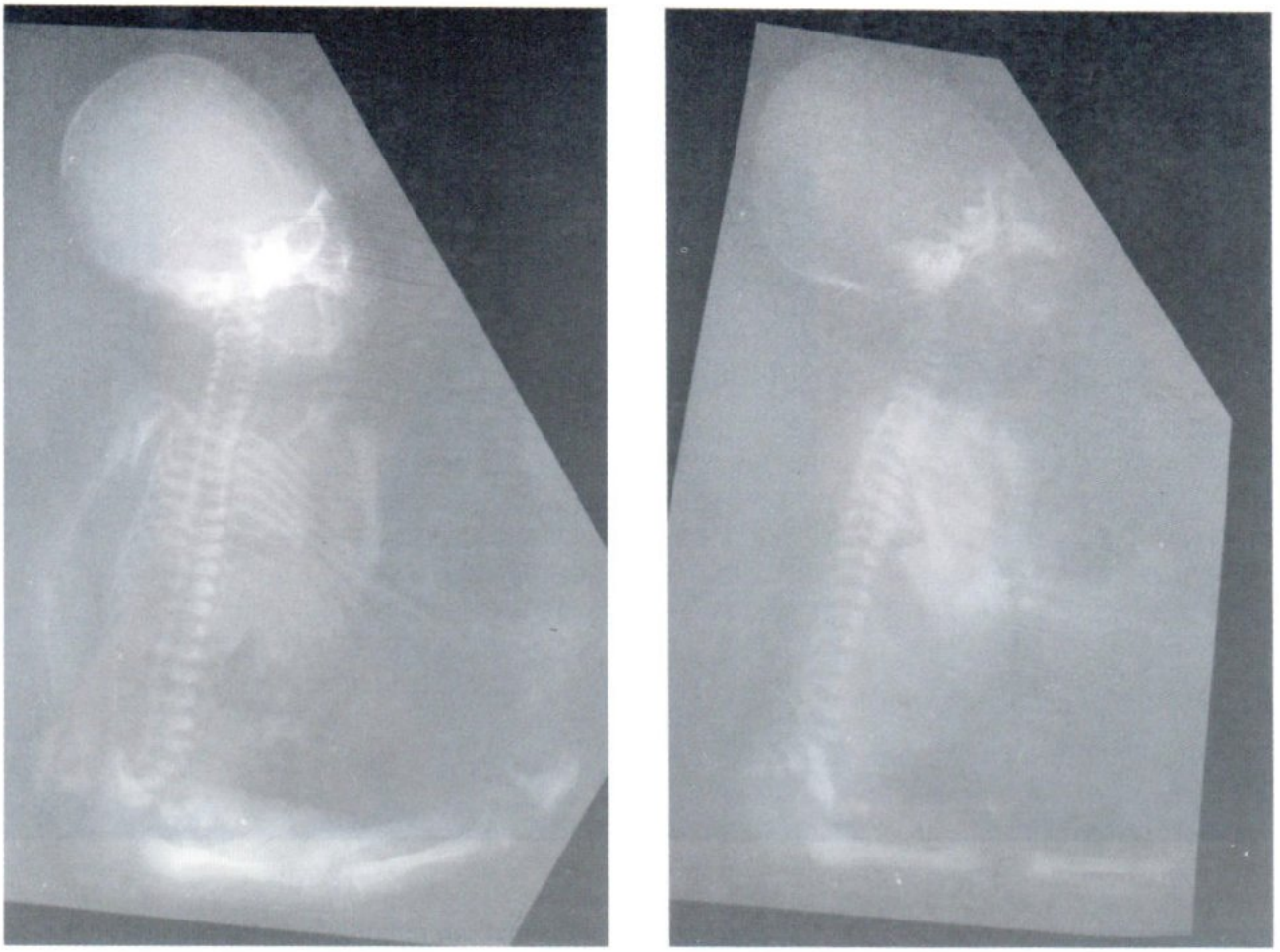


Fig. 2 Babygram immediate post perinatal death showed malformed ischium and pubic bones. Two femurs were noted and with asymmetric size. There were single tibia and single fibula. The bony parts of the feet was difficult to evaluate. The head was enlarged. The facial bony parts, the spinal column, the ribs and the upper extremities appeared normal.

sirenomelia.[18] The nutritional deficiency theory was proposed and supported based on the findings at dissection of the abdominal vasculature in cases of sirenomelia.[19,20] The vascular steal theory indicates that a single large artery assumes the function of the umbilical arteries and diverts blood flow from the caudal portion of the embryo to the placenta, producing nutritional deprivation and maldevelopment of the caudal structures. Examination of umbilical cords in 95 infants with sirenomelia revealed a single umbilical artery in all cases.[21]

The single lower extremity seen in sirenomelia results either from failure of cleavage of the lower limb bud [20] or from the posterior fusion of the hindlimb primordia. [22] The association of maternal diabetes with sirenomelia is much less pronounced .[21]

Common associated anomalies with CRS [23-26] are: (1.1) deformities of feet, (1.2) flexion contractures of hips and knees, (1.3) dislocation of hip, (1.4) pelvic deformity, (1.5) kyphoscoliosis, (1.6) absence of ribs; (2.1) anorectal atresia, (2.2) inguinal hernia, (2.3) abdominal wall defect, (2.4) malrotation of gut, (2.5) tracheoesophageal fistula, rectovaginal fistula, rectourethral fistula; (3.1) vesicoureteral reflux, (3.2) hydronephrosis, (3.3) fused kidneys, (3.4) renal agenesis, (3.5) ectopic ureters, (3.6) transposition of external genitalia, (3.7) mullerian duct agenesis; (4.1) neural tube defects, (4.2) congenital heart disease,

(4.3) midline facial cleft, (4.4) strabismus.

To correlate sonographic with pathologic findings, two distinct groups exist within the continum of CRS [27] Caudal regression would be diagnosed if there is lumbosacral agenesis of the spine, associating with normal amniotic fluid volume and a three-vessel umbilical cord. The diagnosis of sirenomelia was confirmed in the presence of oligohydramnios, bilateral renal agenesis, and lower extremities fusion and a two-vessel cord is usually seen. Amniofusion enables a more accurate sonographic evaluation of fetal anatomy in severe oligohydramnios.[28] MRI was said to be helpful in detecting anomaly with cases of oligohydramnios.[29] Lower extremity abnormalities in caudal aplasia range from hypoplasia to the complete fusion seen in sirenomelia. The persistent side by side relationship of fetal femurs, that does not change over time, during sonographic examination is suggestive of this abnormality.[5]

Obstetric management depends on gestational age at diagnosis, severity of the lesion, associated malformations, and parental wishes. The chromosomes in CRS are usually normal;[30] however, there are isolated reports of pericentric inversion.[31] Recurrence risk with a positive family history of the anomaly are about 3 to 5 per cent.

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