Neonatal mermaid syndrome-Sirenomelia

Author(s): Sriram P, Venkatesh C, Srijit R, Sreenivasa Rhagavan, Vishnu Bhat

Vol. 14, No. 1 (2010-01 - 2010-06)

Sriram P, Venkatesh C, Srijit R, Sreenivasa Rhagavan, Vishnu Bhat

Department of Pediatrics, J.I.P.M.E.R., Puducherry, India

Abstract

Sirenomelia is an uncommon congenital malformation. We describe a newborn with clinical features of sirenomelia with fused lower limbs, anal atresia, bilateral renal agenesis and a single umbilical artery.

Keywords: Sirenomelia, Mermaid syndrome, Renal agenesis
Accepted July 25 2009

Introduction

Sirenomelia, alternatively named mermaid syndrome is an exceptionally rare congenital deformity in which the legs are fused together, giving the appearance of mermaid's tail. This deformity is variably called as symmelia, sympodia monopodia and to its resemblance to the mermaid of Greek and Roman mythology called as mermaid baby. Sirenomelia has prevalence of 1.5-4.0 cases per 100,000 live births [1]. About 300 cases of this lethal anomaly have been reported in the world literature [2]. It is a lethal condition characterized by fusion of the lower limbs, single umbilical artery, and severe malformations of the uro-genital and lower gastrointestinal tract. It can occur as a separate entity or it is commonly associated with caudal regression syndrome. It is usually fatal within a day or two of life because of the complications associated with abnormal kidney and bladder development and function.

Case report

A 21-year old primigravida had a normal antenatal course until 26 weeks when a sonogram revealed oligohydramnios and a single fetus with bilateral renal agenesis and a single fused lower limb. A detailed evaluation of the fetal spine was not possible due to the presence of severe oligohydramnios. The remainder of the antenatal screening studies, including the maternal serum alfa-fetoprotein level, was within normal limits. Her past medical history and family history was unremarkable. In particular there was no history of maternal diabetes. A 1670-g, full term infant, small for gestational age and of undetermined sex was born by spontaneous vaginal delivery and the Apgar score was one at 1 and 5 minutes and 7 at 10 minutes. The infant had flattened facies and a single fused lower limb, with absence of toes with a deformed structure almost like a tail in appearance. There was also single umbilical artery, anal atresia, and a skin tag in the anterior perineal area representing the external genitalia (Fig 1). Skeletal survey showed soft tissue fusion of the lower limb, rudimentary sacrum with single hip bone and acetabular space. There was a single femur with broad distal metaphysis, single tibia with absence of fibula. There were two ossification centres at the lower end of femur. There was a single tubular bone in the rudimentary foot. There were no associated vertebral anomalies (Fig. 2). Since the par-ents were not willing to continue any treatment in the hospital further workup and investigations were not possible.

Fig. 1: Single limb with mermaid appearance

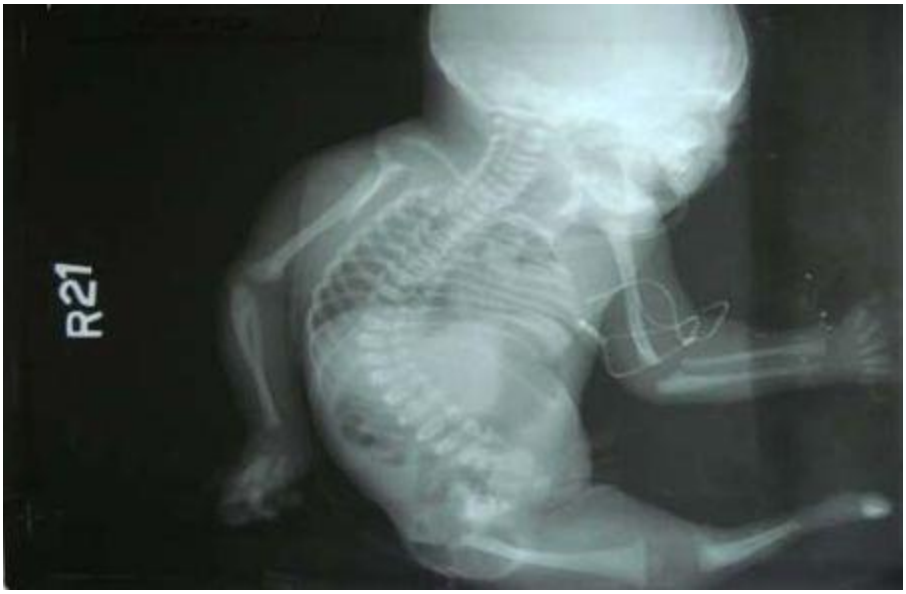

Fig 2: Ssingle femur and Tibia with 2 ossification centres

Discussion

The infant described in our case has all typical features of sirenomelia.The term come from “siren” or “mermaid” resulting from the characteristic fusion of the lower ex-tremities. With the fusion of legs there may be no feet (sirenomelia apus), one foot (sirenomelia monopus) and both feet (sirenomelia dipus). It is an extremely fatal condition because of the presence of bilateral renal agenesis, severe pulmonary hypoplasia and these are associated with potter’s facies [3].

Sirenomelia may be caused by the abnormalities in blastogenesis that affect the distribution of blood to the caudal region of the fetus. The embryological injury is at the caudal mesoderm between the 28-32 days of life. It is presumably the consequence of a wedge shaped early deficit of the posterior axis mesoderm. The fused lower limb is due to failure of lateralization of the lower limbs which usually occurs at four weeks of intrauterine life. There is increased risk of this condition in diabetic pregnancies. A single umbilical artery is observed in these patients which is in direct continuity with abdominal aorta.

Stevenson, Jones, Phelan [3] proposed in their study the “Vascular steal theory”. The common feature in their study was the presence of a single large artery assuming the function of umbilical arteries. Arteries below this steal vessel were hypoplastic. The underdevelopment of these vessels is responsible for the deformities of the caudal structures leading to sirenomelia. This explains the associated renal, genital and gastrointestinal anomalies [4,5]. There are other theories like scanty liquor amnii and its pressure effect, failure of the development of caudal somites of embryo and it has also been described as a variant of caudal regression syndrome [6]. Fusion of the lower limbs, rudimentary feet, bilateral renal agenesis, severe deformity of the bony pelvis, single femur and tibia, single umbilical artery, absence of external genitalia and anal atresia were present in our case and fit the criteria proposed by Duhamel for the mermaid syndrome [7]. The skeletal survey showed an increased width of the metaphysis with two centres of ossification at the lower end of femur. The other associated anomalies like Neural tube defects, vertebral anomalies, exomphalos major [6], sacral agenesis and pulmonary hypoplasia were absent.

There is currently no known serum marker that may be used for antenatal diagnosis of sirenomelia. Prenatal ultrasonography is helpful to diagnose sirenomelia as early as twenty weeks of pregnancy [9]. In the setting of intrauterine growth retardation, oligohydramnios and bilateral renal agenesis, the observation of lower fetal extremity fusion is the key to the antenatal diagnosis of sirenomelia. Sirenomelia is a lethal condition and an early antenatal diagnosis is important in order to allow prenatal counseling for possible pregnancy termination.

References

1. Buyse ML. Birth defects encyclopedia, 1st edn. Massachusetts 02142, USA. Blackwell scientific publication 1990, pp 1541-1542.
2. Gupta R, Gupta M, Gupta S. Sirenomelia bipus (mermaid) with meningocele. Indian J Pediatr 1992, 59: 387-388.
3. Stevenson RE, Jones KL, Phelan MC. Vascular steal, the pathogenetic mechanism producing sirenomelia and the associated defects of the viscera and soft tissues. Pediatrics 1986; 78:451-7. MEDLINE.
4. Stocker JT, Heifetz SA. Sirenomelia: a morphological study of 33 cases and review of the literature, perspect pediatr pathol 1986; 10:7.MEDLINE.
5. Sirtori M, Ghidini A, Romero R, Obbins JC. Prenatal diagnosis of sirenomelia. J Ultrasound Med 1989; 8: 83-8. MEDLINE.
6. Bhatt VM, Venkataramani TK. Sirenomelia (mermaid): Congenital deformity, a case report. J Obstet Gynecol India 1969; 19: 250-254.
7. Duhamel B. From the mermaid to anal imperforation: The syndrome of Caudal regression. Arch Dis Child 1961; 36: 152-154.
8. Bhandari B, Bhandari PC, Mathur A. Sirenomelia with exomphalos associated with absent hindgut. Indian Pe-diatr 1979, 16: 1045-1046.
9. Kampmeier OF. On sireniform monsters, with a con-sideration of the causation and the predominance of the male sex among them, Anact Rec 1927; 34: 365-89.

Correspondence:
Vishnu Bhat
Neonatal unit, Department of Pediatrics
J.I.P.M.E.R, Puducherry 605006, India
E-mail: drvishnubhat(at)yahoo.com

Curr Pediatr Res 2010; 14 (1): 67-68