



Sirenomelia: A Rare Malformation

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Abstract

Sirenomelia, also known as 'mermaid syndrome', is a very rare congenital syndrome with an overall incidence of 1 in 1 lakh live births. It is a polymalformation disorder in which both the lower extremities of the baby are fused. There is no urinary bladder, no rectum, no vagina, no uterus; a colon of few inches is present. It is fatal by second or third day of life in most cases.

Keywords: Mermaid baby; Sirenomelia; Caudal regression syndrome

Case Presentation

A twenty two year old primigravida, at thirty four weeks gestation delivered a 1.7 kg baby by emergency lower segment cesarean section. Cesarean section was done in view of severe intrauterine growth retardation and severe oligohydramnios. There was no history of diabetes mellitus or gestational diabetes in the mother, no history of any chronic medical illness, any drug intake or consanguinity in mother and no history of any genetic disorder in family. Only one prenatal ultrasound was done in first trimester which was reported to be normal. Baby did not cry at birth and required resuscitation. APGAR score was 5, 7 and 7 at 1, 5 and 10 minutes respectively. Baby had two umbilical arteries.

Baby was born with both lower extremities fused and two feet present distally (Figure 1). There were no genitalia or urethral opening. Only a tuft of hair was present at the genital area. An anal opening was present in which the nasogastric tube could be passed to a length of 3 cm, with no meconium staining. A sacral dimple was present 1.5cm above the anal opening. Dysmorphic facies were present in the form of dysplasia of the left mandible and dysplastic ears. There was no deformity in bilateral upper limbs. Abdomen and cardiovascular examination was normal. Within few minutes of birth the baby was put on mechanical ventilator due to respiratory failure. Baby succumbed at 12 hours of life and parents did not give consent for postmortem dissection (Figure 1).

Discussion

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Sirenomelia, is a malformation disorder involving the caudal half of the body [1]. Severe oligohydramnios leads to potter's facies and pulmonary hypoplasia in majority of the babies making it a fatal condition with most of them are stillborn or die within first few hours of birth. Only four surviving cases are reported till date [2]. It results from defective vascular supply to the lower extremities and genitals by descending aorta. The defect occurs in the gastrulation phase of the embryonic development which occurs around 3rd week of intrauterine life.

Etiology of sirenomelia is uncertain, although maternal diabetes mellitus, genetic predisposition, environmental factors and single vitelline umbilical artery causing vascular steal phenomena are proposed to be some of the causative factors [3]. It is considered as one of the most severe forms of the caudal regression syndromes specific to the babies born to mothers with diabetic mellitus. However in our case etiology is unclear as there is no associated maternal diabetes, neither there was single umbilical artery making vascular steal phenomena unlikely.

Managoli S et al. [4] reported an association of amniotic band disruption sequence and sirenomelia. Adhesions of amniotic bands had disrupted the fetal parts especially anteriorly in the midline, causing multiple anomalies (midline disruption and adhesion of the lips by amniotic strings, a disrupted mandible, a bifid tongue, amputated fingers and toes, hands attached to each other and to the chest wall in the midline at the sternum, feet that were attached to the symphysis is in the midline by bands, body wall defects [4]. However no such features of amniotic band disruption were present in our case. Sirenomelia sequence is classified into three groups depending on the number of feet present. The most common of them is symelia apus, in which both legs are completely fused into a single lower extremity. In this condition both feet are either absent or rudimentary. Symelia unipus



Figure 1: Sirenomelia: A Rare Malformation.

presents with one foot, two femora, tibiae and fibulae. Whereas in symelia dipus, two distinct feet are present but are malrotated and resemble fins, like in our case [5].

Lynch and Wright reported a case in which the mother was diabetic and the baby was born with sirenomelia with renal agenesis and an absent right radius. There was also a hypertrophic cardiomyopathy and a bicuspid pulmonary valve [6]. Perez-Aytes, et al. [7] reported an unusual case with partial fusion of lower limbs with absent feet with other features of caudal regression and a single aberrant umbilical artery [7].

Progressive oligohydramnios due to renal agenesis is the earliest sign of diagnosing this condition in the first trimester. In late second

trimester and third trimester however it is easily diagnosable in ultrasound. It can be confirmed by helical three dimensional CT scan [8].

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