



Case Report

Sirenomelia Sequence (Mermaid Syndrome) - A Rare Case

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ABSTRACT

Sirenomelia is a rare, congenital sporadic defect which occurs in newborn infants, more commonly seen in males. It is an extreme form of caudal regression, with extensive damage in the caudal portion of body resulting from an insult occurring before the twenty first day of gestation. Its most remarkable feature is external rotation with fusion of lower extremities. The other defects of caudal axis include imperforated anus, lower vertebral defects and genitourinary anomalies associated with cardiovascular, respiratory and upper gastrointestinal tract malformations in 20-30% of cases. Renal agenesis or cystic renal dysplasia occurs in virtually all cases leading to Potter sequence & pulmonary hypoplasia. We present a rare case of sirenomelia in a 20weeks abortus received for autopsy where almost all classical features of this defect were seen like fusion of the lower limbs, bilateral renal agenesis with Potter's facies, absent uterus, imperforated anus, malrotation and atresia of gut, narrow thorax with pulmonary hypoplasia.

Key words: Sirenomelia, Caudal regression, Potter sequence

INTRODUCTION

Sirenomelia also known as the “mermaid syndrome” as it resembles the mystical mermaid. The term “sirenomelia” was derived from the sirens of Greek and Roman mythology. [1] It is a rare and fatal congenital anomaly with an incidence of 0.8 to 1 case per 1,00,000 births. Male to Female ratio is 3:1. [2] The time of insult resulting in the malformations is known to occur between 28 to 32 days of gestation but the exact pathogenesis remains elusive, with some studies implicating caudal regression or vascular steal phenomenon. [1, 3] The malformation sequence is composed of varying degrees of lower limb fusion along with multiple malformations of the

gastrointestinal, genitourinary, cardiovascular and musculoskeletal systems. Oligohydramnios secondary to severe renal dysplasia is universal. [3, 4]

CASE REPORT

A 20 year old primigravida with 5 months amenorrhea was registered for her antenatal check up in our institute. Her routine investigations were within normal limits. On further investigations her pre-natal ultrasound (USG) showed congenital anomalies of foetus which were not compatible with life. There was no evidence of diabetes mellitus, drug abuse or consanguineous marriage. The patient underwent therapeutic abortion and fetus

was sent for autopsy. On autopsy examination external examination of the foetus showed fusion of lower limbs resulting in single fused lower limb with a short thigh & a short leg having a rudimentary foot. There was kyphoscoliosis (Figure 1), absent genitalia (Figure 2), flat facial profile i.e Potter's facies (Figure 3) with high arched palate, imperforate anus (Figure 4). On internal examination there was bilateral renal agenesis, absent urinary bladder, absent urethra & ureter, hypoplastic

lungs (figure 5) and blind loop intestine (figure 6). Postmortem total body anteroposterior & lateral radiographs demonstrated hemivertebrae with kyphoscoliosis, absent sacrum, single rudimentary femur with absent tibia & fibula & rudimentary tarsal bones (Figure 7).

Considering these malformations of lower limb & visceral abnormalities diagnosis of sirenomelia (STOCKER TYPE VII LEG DEFORMITY) was made.



Fig 1 : Kyphoscoliosis



Fig 2 : Absent Genitalia



Fig 3 : Potter's Facies



Fig 4 : Imperforated Anus



Fig 5 : Hypoplastic Lung



Fig 6 : Blind Loop Intestine



Fig 7: Postmortem Radiogram: Hemivertebrae, Kyphoscoliosis, Single Femur, Absent Tibia, Fibula & Sacrum

DISCUSSION

Rocheus and Polfy in the sixteenth century gave the first description of sirenomelia but it was Duhamel who in 1961 defined the various anomalies associated with mermaid syndrome and labelled it as the most severe form of caudal regression syndrome. The etiology of this syndrome is still unclear and a strong association with maternal diabetes and drug abuse has been observed. [2, 5] No chromosomal anomalies have been detected. [3, 6] It is three times more common in males with a higher incidence in identical twins. The risk of its occurrence depends on the maternal age of

less than 20 years and older than 40 years. [1] Sirenomelia shows a great variability in the spectrum of the malformations. It was Stocker and Heifetz (1987) who classified the leg abnormalities into Types I to VII depending on the presence or absence of bones Type I being the least severe with presence of all bones as compared to the most severe Type VII which consists of a lower extremity with a fused femur and absence of both tibia and fibula. [3, 7] Another way of classifying sirenomelia is according to the number of lower bones present into three different types i.e A) Sirenomelia apus: No feet only one tibia and one femur. B) Sirenomelia unipus : One foot, two femurs two tibia, and two fibula. C) Sirenomelia dipus: Two feet and two fused legs giving the appearance of a flipper. [2] There are various theories about the etiology of the mermaid syndrome. Two major theories are currently speculated. [1] Duhamel explained the caudal regression syndrome to explain various congenital anomalies with sirenomelia being the most severe form. He explained the malformations as a result of defect in blastogenesis and disturbances in the caudal mesodermal axis of the embryo prior to 4th week of gestation. [5] Stevenson 1986 proposed an alternate theory of vascular

steal which is currently the most acceptable theory for the mermaid syndrome. He postulated that blood is diverted from the caudal region of the embryo to the placenta due to shunting via an abnormal abdominal artery, which gives rise to a nutritional deprivation and abnormal development of the caudal structures. The site at which the purposed steal occurs determines the severity of the anomalies. The mechanism of this variation is not fully understood.^[3] Both of these theories appear to be oversimplifications of the true etiology of sirenomelia as neither of them explains the non-caudal anomalies, which have been observed now and then.

CONCLUSION

Sirenomelia is a severe form of caudal regression, the etiology of which is still unclear. As this malformation is uniformly fatal, early antenatal diagnosis with ultrasonography justifies therapeutic termination of pregnancy. Severe oligohydramnios may limit the evaluation resulting in fetal miscarriage. In such circumstances autopsy study helps to diagnose this syndrome.

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