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Research Article

SIRENOMELIA: A RARE CONGENITAL ANOMALY

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ABSTRACT
Sirenomelia is a rare congenital anomaly characterized by variable degree of fusion of lower limbs resembling a mermaid of Greek mythology. Literature is scarce and is available only as few case reports. A 24 year old was referred at 33 weeks of pregnancy with severe oligohydramnios. She delivered a stillborn baby with multiple congenital anomalies and joined lower limbs. Oligohydramnios obscured the antenatal diagnosis by USG. High suspicion of the condition is the key to diagnosis in a fetus with multiple congenital abnormalities.

INTRODUCTION
It is a rare congenital abnormality associated with abnormal development of caudal region of body with variable degree of fusion of lower limb. It may be associated with sacrococcygeal agenesis, genitor-urinary and gastro-intestinal malformations. Diagnosis by antenatal USG in presence of oligohydramnios is difficult. MRI can help in these cases. It is almost fatal. Very few surviving cases of sirenomelia have been reported.

CASE
A 24 year old antenatal woman, multipara was referred at 33 weeks gestation on 31.10.2017 from a primary health center to S.D.N. hospital, a multispeciality hospital, for further management of severe oligohydramnios. Her present, past and family history was unremarkable. Her routine antenatal tests including GDM screening were normal.

Detailed ultrasonogram revealed a single live fetus of 30 weeks with severe oligohydramnios. Fetus had fused lower limbs, multiple congenital anomalies, asymmetric IUGR and curvature of spine was not maintained. Details of congenital anamolies could not be elicited due to severe oligohydramnios. She delivered a macerated stillborn baby weighing 1710 gm on 5.1.18. The baby had joined lower limbs, imperforate anus, ambigious genitalia with pelvic and sacral dysplasia, skull malformation with cleft lip and cleft palate. Right ear lobe of baby was absent (Fig.1).

Figure 1: Macerated Baby with Sirenomelia

DISCUSSION

Sirenomelia is a limb anomaly in which the normally paired lower limbs are replaced by a single midline limb. The most impressive phenotypic appearance of sirenomelia is the presence of a 180°-rotated, axially positioned, single lower limb (Boer *et al.,* 2017). Associated thoracolumbar spinal anomalies, gastrointestinal and genitourinary anomalies are almost always present. Anomalies reported are imperforate anus, spina bifida, renal agenesis, cystic malformation of kidneys,absent bladder, urethral agenesis, pulmonary hypoplasia and cardiac abnormalities. Renal agenesis or renal dysgenesis lead to oligohydramnios.

Incidence is approximately 0.99 to 1 in 100,000 births (Kallen, 1992). Prevalence is more in monozygotic twins (Kallen,1992). Due to the absence of external genitalia and infrequent information on gonadal or chromosomal sex, data on sex distribution are very scarce. Nonetheless, the limited information available indicates male sex preference (Duesterhoeft *et al.*, 2007)

Exact etiology of sirenomelia is not known. A multifactorial etiology has been proposed. A manifestation of the caudal regression syndrome as a consequence of abnormal development of structures derived from the caudal mesoderm of the embryo before 4th week of gestation leading to this rare congenital abnormality has been proposed (Duesterhoeft *et al.,* 2007). A strong association with maternal diabetes mellitus has been found (Kallen, 1992). Genetic predisposition has been proposed as a possible causative factor (Sahu, 2013). The term VACTERL denotes association of vertebral defects, anorectal anomalies, tracheo-esophageal fistula, renal and radial limb defect which can be associated with sirenomelia. (Kallen *et al.*, 1992).

Vascular steal theory proposed also explains its etiology, that a single artery leaving the aorta high up in the abdomen beyond which aorta and its branches are hypoplastic, assumes the function of umbilical artery diverting blood supply and nutrients in the caudal portion of the embryo to give rise to malformed leg (Stevenson,1986).

Depending upon the number of feet present, sirenomalia has been classified into 3 types *i.e.,* simpus apus with no feet, simpus unipus with one feet and simpus dipus with two feet. Our case was of simpus dipus type (Sharma, 2017).

Antenatal USG can be diagnostic. Fused femur, decreased distance between two femurs and decreased or absent mobility of the two lower limbs with respect to each other suggests sirenomelia (Taori, 2002). Usually associated oligohydramnios obscures the diagnosis as in our case. However, there is a narrow window, between weeks 8 and 16 of gestation, that is, when the limb structures are visible to ultrasound, and the amniotic fluid still depends mainly from maternal production, when visualization of sirenomelia is possible. Fetal MRI can be helpful where evaluation is difficult due to oligohydramnios (Sawhney, 2006).

In majority of cases sirenomelia is fatal. 50% are stillbirth babies (Stevenson, 1986) and the rest 50% do not live more than 2 to 24 hours after birth. Survival depends on the visceral anomalies associated and not only on sirenomelia. If diagnosed earlier pregnancy can be terminated. If the newborn survives for 24 hours and has good functioning kidney, then surgical intervention is required to separate the joined legs and rebuild the defects present. Even after several successful surgeries the prognosis is not favorable.

Very few survivors with sirenomelia have been reported in literature.

Murphy *et al*., (1992) reported one case where a child born with Sirenomelia survived; the infant was neurologically normal and had fused lower extremities, an imperforate anus, colon atresia, bilateral fused pelvic kidneys with renal dysplasia and sacral dysplasia, and genital abnormalities. Laparotomy and colostomy were performed, and an eventual separation of the lower extremities was done (Murphy *et al.*, 1992).

Clarke *et al.,* (1993) report on a three-month-old infant whose sirenomelia was diagnosed prenatally. The infant was neurologically normal and has fusion of the lower limbs with associated renal dysplasia, an

imperforate anus, pelvic and sacral dysplasia, and genital abnormalities. The infant's anomalies were compatible with life and surgical separation of the lower limbs was done (Clarke, 1993).

Pinette has reported a case of surviving sirenomelia, born 1999 from Maine, She was on dialysis from the age of 3-4 months and had undergone renal transplant at the age of 2 years. Attempt to separate fused legs was not done because of concerns of potential disruption of blood supply to internal organs. She was the only survivor who did not have surgery for separation of legs (Pinette, 2005).

Little mermaid, (born 2004) of Peru has been reported as a surviving sirenomelia. She was operated for separation of legs in 2005. A kidney transplant was done in 2012. She will require many more operations to reconstruct her digestive, urinary and sexual organs (Dailymail, 2014).

Another survivor at 123 days has been reported from Kuwait. She had an assisted breech delivery and had multiple congenital anomalies. She was managed medically and surgically and was discharged after 123 days of hospitalization (Al Hadhoud *et al.*, 2017).

Conflict of interest – None

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