ISSN 2598-3180 (Online)
JOURNAL OF MIDWIFERY
Research and Practice
Accredited by SINTA 4th

Article

Sirenomelia: A Case Report

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SUBMISSION TRACK

Recieved: May 12 2022
Final Revision: June 09 2022
Available Online: June 29 2022

KEYWORDS

Sirenomelia. Single lower limb, single umbilical artery, thumb deformity

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A B S T R A C T

Sirenomelia is an extremely rare anomaly, an incidence of 1 to 4.2 in 100,000 births, in which a newborn born with legs joined together featuring a mermaid-like appearance (head and trunk like humans and tail like fish), and in most cases die shortly after birth. Gastrointestinal and urogenital anomalies and single umbilical artery are clinical outcome of this syndrome. There are two important hypotheses for pathogenesis of mermaid syndrome: vitelline artery steal hypothesis and defective blastogenesis hypothesis

The cause of the sirenomelia is unknown, but there are some possible factors such as age younger than 20 years and older than 40 years in mother and exposure of fetus to teratogenics. Here, we introduced 39 year old mother's first neonate with Sirenomelia syndrome. The mother had gestational diabetes mellitus and neonate was born with single lower limb, ambiguous genitalia, and thumb anomalies, and 4 days after birth, the neonatedied due to multiple anomalies and imperforated anus.

I. INTRODUCTION

Sirenomelia (mermaid syndrome) is a rare congenital fetal anomaly (Samal, et al., 2015; Taee, et al., 2018), in which a newborn born with legs joined together featuring a mermaid-like appearance (head and trunk like humans and tail like fish), and in most cases die shortly after birth. Gastrointestinal and urogenital anomalies and single umbilical artery are clinical outcome of this syndrome. There are two important hypotheses for pathogenesis of mermaid syndrome: vitelline artery steal hypothesis and defective blastogenesis hypothesis. The cause of the mermaid syndrome is unknown, but there are some possible factors such as age younger than 20 years and older than 40 years in mother and exposure of fetus to teratogenics (Taee, et al., 2018). Although, this syndrome is incompatible with life due to the association of several congenital visceral abnormalities; however, there are few reports of surviving infants (Samal, et al., 2015)

The prevalence of this anomaly is 1 in 100,000 births (Taee, et al., 2018). Verma noted that the prevalence of SML is to be between 1.1 and 4.2 per 100,000 births (Verma, 2021). Sofar, 300 patients with this rare anomaly have been reported in the world. The incidence of male to female is 3 to 1 (Reddy et al., 2012). The incidence in monozygote twins were reported 150 to 200 times; 200 times more likely to be seen in a newborn with diabetes mothers; 15% of mothers had gestational diabetes mellitus during pregnancy (Taori et al., 2002; Sikandar et al., 2009).

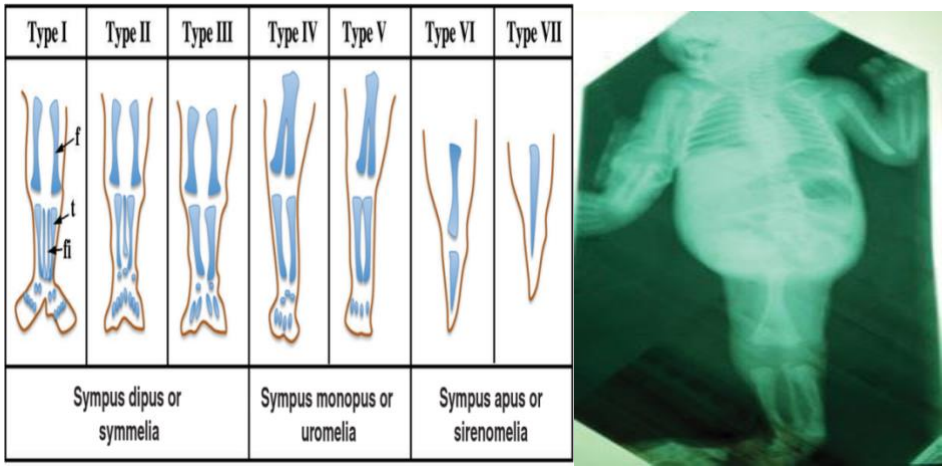

Fig 1: (Fused thighs and legs with two femoral and two tibial bones within single soft tissue and muscular compartment (Type V) and Radiography of a neonate with sirenomelia (Das et al., 2002).

I.1 Case Report

A term neonate with ambiguous genitalia and single lower limb was born to a 39 year old mother with a history of gestational diabetes mellitus. The neonate was the first child of the family and parents did not have a familial relationship. The mother did not have any health care and medical treatment at the time of pregnancy. The neonate's birth weight was 3,500 g. In the first examinations, the head, face, and neck were normal. In the upper limb, the thumb was connected with a small connector to left hand. The auscultation of heart and lung were normal. There was no problem with the chest wall. The abdomen was clearly turgid, but the liver and the lungs were not big at the touch **(Fig. 2a).** The umbilical cord had an artery and a vein **(Fig.2b).** At the end of the abdomen,

there was a deviant limb in front facing at the rear, with two heels, followed by two thumbs and six fingers, and with a relatively tall and nail like arrangement **(Fig. 2a).**

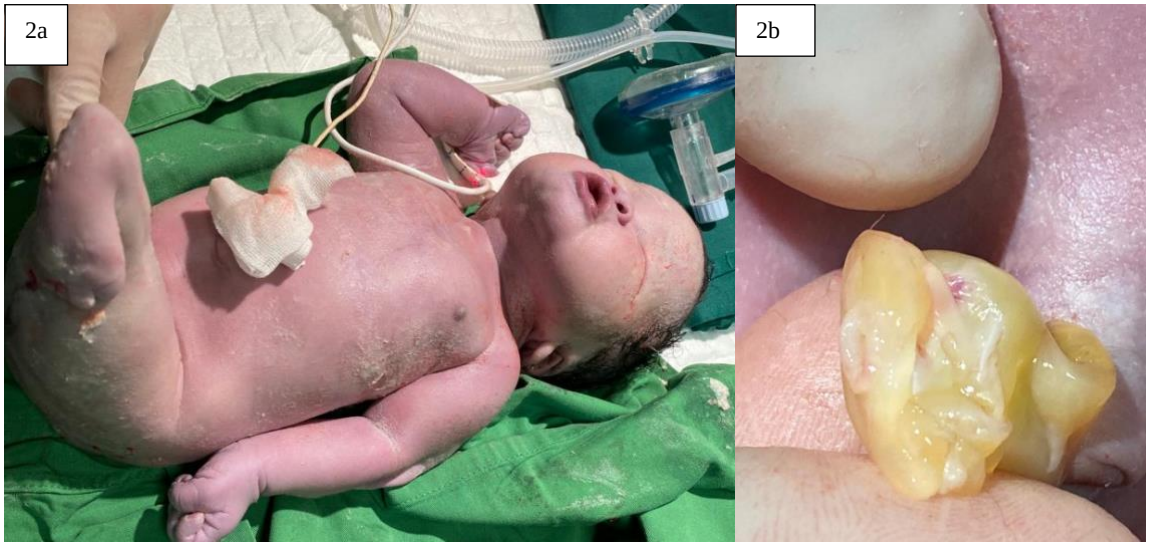

Fig 2 : (Anterior view of a neonate with sirenomelia (a) and The umbilical cord had an artery and avein (b))

There was a dentin the back of the neonate at the end of the spine of the lumbar region. Sacral bone was not touchable, and there was no pelvic bone on the left. At the junction of the spine, there was an isolated single limb of the hole in the closed face and 3 cm lower than the anterior cavity of the ellipsoidal shape that was seen locally. The bottom of the cavity was duct. Urinary tract was not seen. However, urinary excretion dropped out of the occipital cavity **(Fig. 3).** In examining the auscultation, the heart and heart vessels were normal. In sonography, the liver, the bile ducts were reported to be normal. The left kidney and the right kidney was larger than normal and the bladder was small and abnormal. Internal genital organs including testicular gonads, ovaries, and veins were not observed. The umbilical artery with a large diameter of the abdominal aortic artery originated, and the rest of the abdominal aortic pathway after the umbilical artery had irregular branches and hypoplastic.

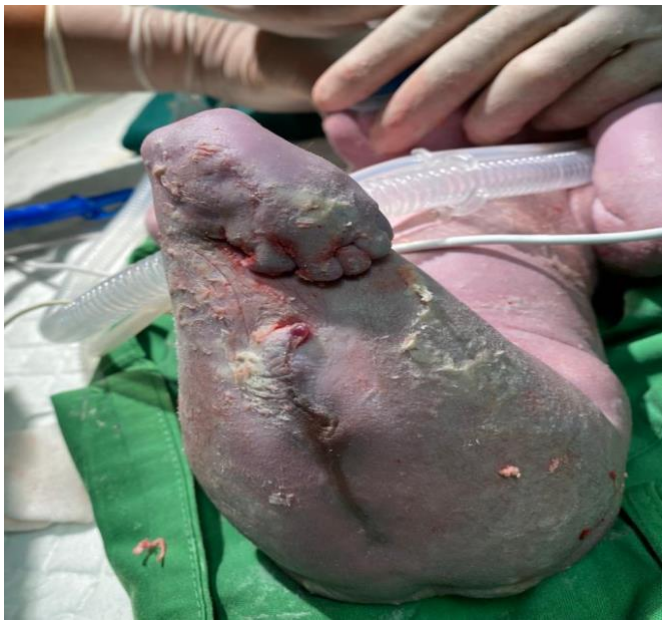

Fig 3: (Posterior view of a neonate with sirenomelia)

During patient hospitalization, the neonate had urinary excretion as a droplet, but no excretion of stool was observed. The neonate developed shortly after birth, abdominal distension, bile vomiting, and oral intolerance. The neonate was fed intravenously during life. Neonate bile vomiting continued with fecal material removal, and after that, the neonate died 4 days after birth with a block of bowel obstruction and generalized peritonitis .

II. DISCUSSION

Sirinomelia syndrome is an extremely rare anomaly that was first reported in 1,542 by Rocheus et al and later by Palfyn et al in 1543. Mermaid means trunk looks like human and rear looks like a fish (Sikandar et al., 2009; Taee, et al., 2018). Duhamel classified the mermaid syndrome as type 5 caudal regression syndrome (CRS) for the similarity with CRS anomalies (Fig.1) (Duhamel, 1961). Stocker and Heifetz report a more accurate classification of sirenomelia: I, all thigh and leg bones are present; II, single fibula; III, absent fibulae; IV, partially fused femurs, fused fibulae; V, partially fused femurs, absent fibulae; VI, partially fused femurs, fused fibulae; VII, partially fused femurs, absent fibulae; VIII, partially fused femurs, absent fibulae fibulae; VI, single femur, single tibia; VII, single femur, absent tibiae (Yaşar, et al., 2022). By the way, The classic Sirenomelia sequence includes a uniform spectrum of caudal malformations, spinal defects, and a single umbilical artery. SML is postulated to be due to a genetic predisposition, unmasked by biochemical or environmental triggers. Primary developmental defects in the formation of caudal mesoderm or embryonic caudal vessels with resultant local tissue hypoperfusion are proposed hypotheses for its pathogenesis (Verma, 2021).

SML occurs sporadically in humans, presumably due to a spontaneous mutation, and is speculated to have an autosomal dominant inheritance pattern. In mutant mice, specific defects in Cyp26a1 and Bmp 7 genes are demonstrated to produce offsprings with SML. Bmp 7 is a signaling protein, which belongs to the transforming growth factor-β (TGF β) superfamily. Tsg 1, a Bmp and chordin-binding protein, functions as an activator-inhibitor of Bmp signaling in the

embryonic caudal region (ECR). Loss of Bmp7 genes combined with a complete loss or half-dose of Tsg 1 is demonstrated to produce an invariable SML phenotype. SML is also demonstrated to occur with increased Retinoic acid (RA) signaling in the ECR. The Cyp26a1 gene is involved in coding for an enzyme, which expresses in ECR and degrades RA. A specific defect in this gene leads to excess local RA concentration and SML generation with a reported 20% penetrance in mutant mice. However, the mutational screening of Cyp26a1 and Bmp 7genes has failed to confirm their involvement in mankind and the molecular defect and genetic inheritability of SML in humans remain undefined (Verma, 2021).

Environmental and teratogenic factors, such as cocaine, retinoic acid, heavy metals, cyclophosphamide, and certain antibiotics, have been linked to SML in humans and animal models. In addition, nicotine, alcohol, radionuclides, diethylpropion- an appetite suppressor, organic solvents of fats, and even air pollution have been associated with SML and caudal regression syndrome, which is controversially considered as its minor form. Other authors have reported fetal exposure to cadmium, lithium, phenytoin, sodium valproate, carbamazepine, warfarin, methylergonovine, diethylpropion, trimethoprim, and ochratoxin-a type of fungus as possible triggers for the anomaly. Some of the maternal complications, such as diabetes mellitus, hyperthermia during the 1st trimester of pregnancy, amniotic bands, and age below 20 years or over 40 years at conception, have also been implicated in the pathogenesis of SML (Verma, 2021)

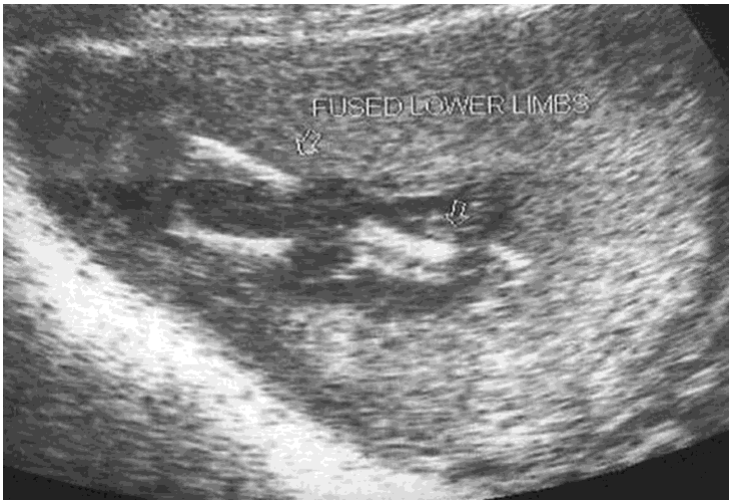

Fig 4: (Ultrasound Features)

On ultrasound examination, we found fused thighs and legs with two femoral and two tibial bones within a single soft tissue and muscular compartment . There was an absence of fibular bones in the legs; both feet were fused posteriorly and externally diverging anteriorly, and they looked like the fins of a fish. The fetal urinary bladder was minimally filled, and located slightly to the left. Fetal left renal pelvicalyceal system was mildly dilated , and fetal right kidney was not visualized. Only a single umbilical artery was present, and severe oligohydramnios was seen .

Anomalies that are commonly seen with the mermaid syndrome include: cleft palate pulmonary hypoplasia, cardiac defects, omphalocele, pentalogy of Cantrell, and meningomyelocele (Garrido-Allepuz et al., 2011; Moosa et al., 2012).Mermaid syndrome occurs sporadically. Neonates born with mermaid syndrome often have normal karyotype (Orioli et al., 2011).

III. CONCLUSION

According to the findings of the Sirinomelia syndrome, caudal area hemorrhagia has been identified as a major cause and other factors include gestational diabetes mellitus has been reported. Blood glucose control in pregnant women and prevention of contact of pregnant mother with teratogenic substances is recommended.

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