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Sirenomelia: Mermaid Syndrome - Fantasy or Fact

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Abstract: ***The mermaid syndrome (Sirenomelia) is an extremely rare anomaly, with an incidence of 0.8 - 1in 100, 000 live births, in which a newborn is born with legs fused together featuring a mermaid like appearance, an evolutionary defect in caudal region with degrees of leg adhesion, causing complete absence of lower limb. About 300 cases have been reported in the world literature so far, with 14 reported from India.***

Keywords: sirenomelia, mermaid syndrome, congenital anomaly, antenatal care, fused legs, potter's facies

1. Introduction

Sirenomelia is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia. The rarity of the case is obvious from the fact that many gynecologists might not have come across a case of sirenomelia in their whole professional career. There is a strong association with maternal diabetes where relative risk is 1: 200 - 250 and up to 22% of fetuses with this anomaly will have mothers with diabetes

2. Case Report

A 25-year-old multigravida reported to labour room with complaints of pain abdomen. She was 34 weeks gestation with previous LSCS, Rh positive, euglycemic and normotensive. Hers was consanguineous marriage with first cousin, with no personal or family history of diabetes/ substance abuse.

Patient had erratic antenatal checkups with mid trimester anomaly scan suggestive of Dysplastic kidneys, severe oligohydraminos with non visualisation of urinary bladder. Patient was advised termination yet she continued her pregnancy. Growth scan in 3rd trimester showed features of sacral agenesis, dysplastic kidneys, Fetal Growth Restriction with absent liquor

Figure 1

Figure 2

On examination fundal height was less than the gestational age, breech presentation with clinically decreased liquor. She delivered vaginally a fresh stillborn, which showed fused lower limbs with a single foot and 5 toes, absent external genitalia (Fig - 1), imperforate anus (Fig - 2) with single umbilical artery (Fig - 3), with features S/O Potter's facies.

Unfortunately, autopsy was declined by the parents.

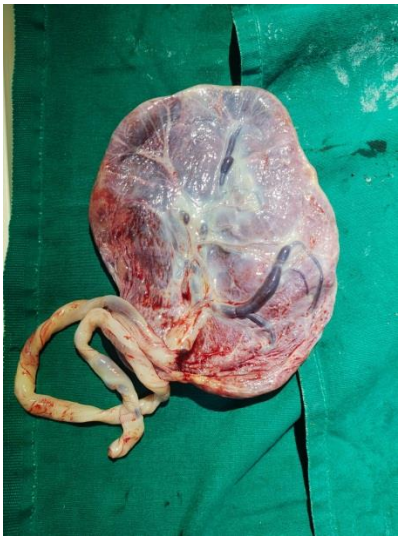

Figure 3

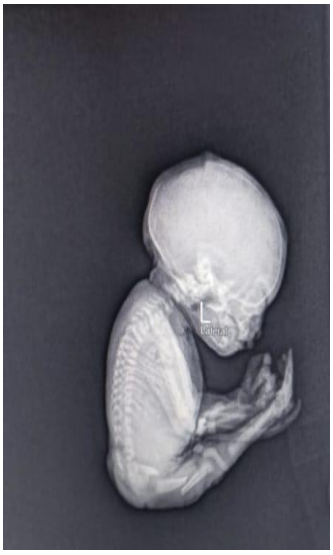

Figure 4

3. Discussion

In 1961, Duhamel classified the mermaid syndrome as type 5 caudal regression syndrome (CRS) for the similarity with CRS anomalies.7 But nowadays, mermaid is a separate syndrome, and the diagnostic key is the presence of single umbilical artery and renal agenesis, which is a stable clinical syndrome in mermaid, while in the CRS, there is a dysfunction and no deadly kidney anomalies.

In 1987, Stocker and Heifetz introduced the theory of vitelline artery steal and reported that all patients with sirenomelia had a large umbilical artery, separated from the upper abdominal artery aorta slightly below the celiac artery, and the branches of other aorta had not evolved. This suggested that due to the lack of blood supply and inadequate nutrition, the growth of the lower part of the body was stopped and led to sacral agenesis, lower limb fusion, imperforated anus, rectal agenesis, internal and external absence of genitalia, and renal agenesis.

Stocker and Heifetz classified sirenomelia in seven types

1) All thigh and leg bones present.
2) Single fibula.
3) Absent fibulae.
4) Partially fused femurs, fused fibulae.
5) Partially fused femurs, absent fibulae.
6) Single femur, single tibia.
7) Single femur absent tibiae.

Although the main factor for mermaid syndrome is unknown, Several hypothesis for pathogenesis include - maternal diabetes, Vitelline artery steal theory, defective blastogenesis hypothesis, exposure to teratogens /unknown genetic defects in Cy26a1 and BMP7 genes.

The Cyp26a1 gene is responsible for coding the enzyme that breaks down retinoic acid (the metabolite of vitamin A). Retinoic acid temporarily increases the vasculature in the caudal region of the embryo. Disruption of the Cyp26a1 gene and incomplete development of the caudal region of the embryo result in a mermaid syndromeinmice. Bone morphogenic protein7) is an important protein that plays an

important role in angiogenesis in vitro. By stimulating endothelial cells of the caudal region, vascular and tissue production leads to normal growth of the lower limbs in the fetus.

The exposure to teratogenic factors, such as air pollution, and mother - to - drug contact with cocaine and tobacco and alcohol cigarettes, and radionuclide are also among the causes. Fetal exposure to cadmium, lithium, phenytoin, sodium valproate, carbamazepine, warfarin, methylergonovine, diethylpropion, trimethoprim, and ochratoxin (a type of fungus) are also the causes of the birth of the mermaid syndrome in other articles.

4. Conclusion

The precise etiology of sirenomelia is unknown. Sirenomelia is a rare and lethal congenital anomaly. Termination should be off ered when diagnosed antenatal, however, prevention is possible and should be the goal. Regular antenatal check - up with optimum maternal blood glucose level in pre - conception period and in first trimester should be maintained to prevent this anomaly.

Conflict of Interest: None

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