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Mermaid Syndrome- A Systematic Review

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ABSTRACT

Mermaid Syndrome, or Sirenomelia, is an extremely rare congenital disorder characterized by the fusion of the lower limbs, often accompanied by severe urogenital and gastrointestinal malformations. This systematic review aims to provide a comprehensive overview of the etiology, clinical presentation, diagnostic challenges, and management strategies for Mermaid Syndrome. A detailed analysis of case reports and research studies published in medical and scientific journals was conducted, focusing on cases reported over the past four decades. The review highlights that the etiology of Sirenomelia remains unclear, with theories suggesting multifactorial causes, including genetic, environmental, and vascular abnormalities. Prenatal diagnosis using ultrasonography and advanced imaging techniques is critical for early detection. Management is complex, requiring a multidisciplinary approach encompassing surgical intervention, supportive care, and rehabilitation. While survival rates remain low due to associated complications, advancements in neonatal care and reconstructive surgeries have shown promise in improving outcomes for select cases. The findings emphasize the need for further research to better understand this enigmatic condition and enhance treatment modalities.

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Introduction

Mermaid Syndrome was initially described by Rocheus et al in 1542, and then by Palfyn et al in 1543. Mermaid syndrome, also known as sirenomelia, is an extremely uncommon congenital condition in which the lower limb is completely absent and resembles the perianal due to an evolutionary error in the caudal area with variable degrees of leg adhesion [1,2]. One newborn out of every 100,000 has this abnormality. There have been 300 cases of this uncommon aberration reported worldwide thus far. The ratio of males to females is 3:1. [3]. The incidence was 150–200 times higher in monozygote twins, 200 times higher in a newborn whose mother had diabetes, and 15% of pregnant women had gestational diabetes mellitus [4,5]. Exact cause of mermaid syndrome is unknown. The only condition in mothers that is known to be linked to mermaid syndrome is gestational diabetes mellitus. Mothers under the age of 20 and those over 40 are particularly at risk. The fundamental reasons why mermaid syndrome occurs are amniotic band disruption and hyperthermia. In addition, mother-to-drug contact with cocaine, tobacco, and alcohol cigarettes, as well as exposure to teratogenic variables including air pollution, are contributing factors. The mermaid syndrome is also brought on by fetal exposure to cadmium, lithium, phenytoin, sodium valproate, carbamazepine, warfarin, methylergonovine, diethylpropion, trimethoprim, and ochratoxin (a form of fungus).(6). Sirenomelia is classified into following types :Type 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: single femur, single tibia; VII: single femur, absent tibiae. Ultrasonography at the end of the first trimester can be used to diagnose Mermaid Syndrome .If the fetus has a serious abnormality that is inconsistent with survival, the pregnancy may be terminated.

Pathophysiology

The Vitelline Artery Steal Hypothesis

The vitelline artery steal hypothesis suggests that an abnormal single umbilical artery (SUA) redirects blood flow from the caudal fetal limb buds to the placenta, compromising the regional blood supply during critical stages of development. In the early stages of embryogenesis, the vitelline artery complex serves as a primary circulatory network, supplying the yolk sac. However, abnormal development of umbilical vasculature in Sirenomelia (SML) leads to the formation of an enlarged, aberrant aorta-like artery that originates from the vitelline artery and is positioned high in the developing abdominal cavity.

This abnormal vascular structure disrupts normal blood flow, leaving critical caudal regions inadequately perfused. As a result, normal branches of the aorta, including the celiac artery, may be hypoplastic, absent, or disconnected from this abnormal arterial structure. The diversion of blood away from the lower fetus to the placenta by this aberrant channel results in restricted blood flow to the caudal mesoderm, leading to developmental defects in tissues relying on these arterial branches. Autopsy findings often reveal varying degrees of underdevelopment, malformation, or regression in caudal fetal tissues dependent on these arteries.

Defective Blastogenesis Hypothesis

The defective blastogenesis hypothesis posits that SML arises from disruptions during blastogenesis, specifically in the later stages of gastrulation. During the third week of pregnancy, disturbances in embryonic development result in defective angiogenesis and insufficient blood flow to the caudal region during the tailbud stage. This inadequate vascularization disrupts normal growth and differentiation in the caudal mesoderm and associated structures.

In experimental models, mutations in genes such as Cyp26a1 and Bmp7 have been associated with the development of

mermaid-like phenotypes in mice. In these cases, disruptions in bone morphogenetic protein (BMP) signaling, coupled with excessive secretion of retinoic acid (RA), lead to caudal malformations and severe cardiovascular anomalies. Loss of BMP signaling in the ventral caudal embryonic mesoderm impairs the development of major arteries and the caudal body. Furthermore, RA and BMP are critical regulators of each other's pathways, and their imbalance can result in profound developmental abnormalities.

In humans, while specific genetic mutations directly causing SML remain unclear, chromosomal abnormalities such as triploid mosaicism and balanced translocations, including those involving chromosome 16, have been implicated. These findings suggest that a complex interplay of genetic, molecular, and environmental factors underlies the pathogenesis of SML, requiring further investigation to fully elucidate its mechanisms.

Diagnosis

Early and accurate diagnosis of sirenomelia, commonly known as mermaid syndrome, requires regular prenatal ultrasound examinations. These ultrasounds can reveal abnormalities in the lower limbs, reduced craniocaudal length, vertebral defects (such as gaps in the dorsal, lumbar, or sacral regions), and absence of fetal movements. However, synchronized oligohydramnios and intrauterine growth restriction may pose challenges in imaging. Transvaginal ultrasonography can enhance anatomical visualization, offering better clarity. Ultrasound, especially in the first or early second trimester, is a crucial tool for detecting sirenomelia. Additionally, three-dimensional sonography and magnetic resonance imaging (MRI) are instrumental in achieving a comprehensive assessment of the fetus. Postnatal imaging, including X-rays, along with autopsy, is recommended for confirmation.

Differential diagnoses include conditions such as a single umbilical artery, bilateral renal agenesis, megacystis, VACTERL/VATER association (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities), and caudal regression syndrome (CRS).

Clinical Presentation

Sirenomelia is marked by significant vascular anomalies, typically involving a single umbilical artery and vein rather than the normal two arteries and one vein. Other associated abnormalities include sacral agenesis, absence of the gallbladder and spleen, omphalocele, lordosis, malformed vertebrae (hemivertebrae), and central nervous system anomalies.

Potter's facies, a characteristic facial anomaly, is frequently observed in newborns with sirenomelia and is defined by a broad epicanthic fold, low-set ears, hypertelorism, a flat nasal bridge, and a receding chin. Additional abnormalities may include cleft palate, cervical and upper thoracic vertebral anomalies, pulmonary hypoplasia, and cardiac malformations.

Renal and urethral abnormalities are common, ranging from total renal agenesis to ectopic or cystic kidneys. Urethral atresia and bladder absence are also prevalent. Genital anomalies primarily affect the external structures, with gonads typically unaffected. Intestinal abnormalities such as rectal atresia, imperforate anus, and a blind-ended colon are frequently observed.

Treatment

The treatment of sirenomelia is challenging, expensive, and often yields limited success. Management primarily involves surgical and medical interventions to address systemic and structural anomalies. In cases where infants with sirenomelia survive, surgical separation of the fused lower limbs may be considered. Subcutaneous tissue expanders, such as saltwater-filled balloons, are often used to prepare and stretch the skin before surgery. The expanded skin facilitates the separation procedure.

Conclusion

Mermaid syndrome, or sirenomelia, is an exceedingly rare congenital anomaly characterized by partial or complete fusion of the lower limbs. It is often accompanied by severe visceral anomalies that are incompatible with life outside the uterus. Early detection through prenatal ultrasonography is critical for diagnosis, enabling timely decision-making. If confirmed, pregnancy termination is usually recommended due to the poor prognosis and high complexity of treatment.

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Availability of Data and Materials

The data and materials used in this study are available from the corresponding author upon reasonable request.

Ethics Approval and Consent to Participate

This study was conducted in accordance with the Declaration of AUC, Hyderabad, and received approval from the Department of Pharmacy Practice, Anwarul Uloom College of Pharmacy, New Mallepally, Hyderabad, Telangana, India (500001). The patient was fully informed about the study, and informed consent was obtained in their native language before publication decisions were made. Documentation of consent has been provided at the time of submission.

Consent for Publication

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images. A copy of the signed consent is available for review with Journal.

Competing Interests

The authors declare that they have no inancial or non-inancial competing interests.

References

1. Duesterhoeft S M Et Al Five cases of caudal regression with an aberrant abdominal umbilical artery: further support for a caudal regression-sirenomelia spectrum. Am J Med Genet A. 2007;143A(24):3175–3184. doi: 10.1002/ajmg.a.32028. [DOI] [PubMed] [Google Scholar].

2. Garrido-Allepuz Et al A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations. Dis Model Mech. 2011;4(03):289–299. doi: 10.1242/dmm.007732. [DOI] [PMC free article] [PubMed] [Google Scholar].

3. Reddy K R Et Al Sirenomelia: a rare presentation. J Neonatal Surg. 2012;1(01):7. [PMC free article] [PubMed] [Google Scholar].

4. Taori K B Et Al Sirenomelia sequence (mermaid): report of three cases. Indian J Radiol Imaging. 2002;12(03):399. [Google Scholar]

5. Sikandar R Et Al Sirenomelia, the mermaid syndrome: case report and a brief review of literature. J Pak Med Assoc. 2009;59(10):721–723. [PubMed] [Google Scholar].

6. Nadereh taee Et Al Mermaid Syndrome: A Case Report of a Rare Congenital Anomaly in Full-Term Neonate with Thumb Deformity doi: 10.1055/s-0038-1669943.

7. Rita Prasad Verma Et Al Genetics of Sirenomelia, the Mermaid Syndrome DOI: 10.5772/intechopen.97555.

8. Mehmet Zeki Yaşar Et Al Mermaid syndrome: A case report in Somalia https://doi.org/10.1016/j.amsu.2022.103533

9. Annie F. Siddiqui Sirenomelia or "Mermaid Syndrome" in a Twin Pregnancy: A Case Report .

10. Morales Rosello Jose Et Al Sirenomelia, case report and review of the literature. Mar;2022 J Matern Fetal Neonatal Med. 35:1203–1206. doi: 10.1080/14767058.2020.1742693. [DOI] [PubMed] [Google Scholar]

11. Şirin Küçük Et Al Sirenomelia (Mermaid Syndrome): A Case Report doi: 10.5146/tjpath.2020.01491.

12. Qodri Santosa Et Al Sirenomelia or “mermaid syndrome”: an extremely rare case in Indonesia Case Report DOI: 10.14238/pi63.1.2023.45-50.