ISSN: 2755-9890

Journal of Spine Research & Reports

SCIENTIFIC Research and Community

Case Report

Open Access

Clinical Imaging, Anatomical and Embryological Classification of Sirenomelia, A Case Report

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Abstract

Sirenomelia or mermaid syndrome is a Very Rare congenital Defect (VRD) with a reported prevalence of 0.98 per 100,000 births, where the normally paired lower limbs are replaced by a single midline limb. With few exceptions, sirenomelia is a lethal condition in the perinatal period associated with genital, large bowel, urinary, cardiac or central nervous system defects along with other VRDs such as bladder exstrophy, cyclopia/holoprosencephaly and acardia-acephalus. Antenatal diagnosis of sirenomelia by ultrasound is often impaired by oligohydramnios resulting from the frequently associated bilateral renal a/dysgenesis. A case report describes a live birth by normal vaginal delivery presenting with a single midline limb, two feet and absence of external genitalia/anal orifice. Antenatal ultrasound scans revealed moderate to severe oligohydramnios, and poorly visualised foetal kidneys. This study describes the clinical presentation, associated malformations, and classification of sirenomelia.

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Received: June 06, 2024; **Accepted:** June 13, 2024; **Published:** June 20, 2024

Keywords: Sirenomelia, Mermaid Syndrome, Anatomical Classification

Introduction

Sirenomelia, also known as mermaid syndrome, is an exceptionally rare congenital defect (VRD) with a reported prevalence of 0.98 per 100,000 births [1]. The condition is characterised by the existence of paired lower limbs, which are replaced by a single midline limb. With few exceptions, Sirenomelia tends to be a life-threatening condition in the perinatal period associated with genital, large bowel, urinary, cardiac or central nervous system defects. Additionally, it may coincide with other VRDs such as bladder exstrophy, cyclopia/holoprosencephaly and acardia-acephalus. Detecting sirenomelia through prenatal ultrasound diagnosis can be particularly challenging due to the presence of oligohydramnios, which frequently results from concurrent bilateral renal abnormalities. This review was previously presented as a meeting abstract at the 2018 BACA Winter Scientific Meeting on December 13, 2018.

We report a referred case of a new-born diagnosed with Sirenomelia presented to the Obstetrics and Gynaecology department at Arif Memorial Hospital, Lahore, Pakistan on day one of life. In our discussion, we will delve into the physical characteristics, classification, and the underlying causes (aetiology) of this syndrome. This case report was reported in accordance with ethical guidelines and obtained approval from Rashid Latif Medical University (IRB: IRB/2018/003). Patient provided informed consent by signing a waiver, agreeing to the collection of history and images for the case report.

Case Presentation

A 29-year-old woman G3P2, 32 weeks of gestation. No documented history of exposure to any known teratogenic agent during her pregnancy, and no history of maternal diabetes or hypertension. No other relevant past medical history. An antenatal scan revealed moderate to severe oligohydromnios and the fetal kidneys were poorly visualized. The baby was born by normal vaginal delivery. Apgar score was 1 at 1, 5, and 10 minutes respectively. In spite of the medical intervention, the baby died a day later.

The physical examination of the new born revealed an obvious lower limb fusion into a single limb terminating with 10 toes; absence of anal orifice and genitalia. On further examination it was noted that the baby had a flaccid posture and there was an opening on the back from which urine was dribbling.

Figure 1: Fusion of lower limbs into a single limb. Distal portion of the single limb appears to be partially separated and ends in 2 feet with 5 toes each. There is also absence of genitalia.

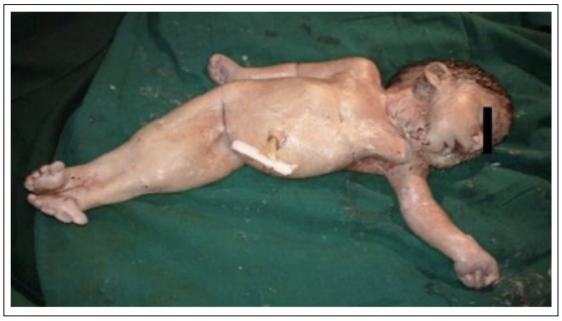

Figure 2: Note the presence of a single femur attached to an underdeveloped pelvis. Distal to the Femur are 2 separate long bones, likely to be tibia bones with no associated fibular. There is obvious bowel loop dilation in the abdomen and presence of Dextrocardia in the thorax

Discussion

Sirenomelia was initially categorized into three types based on the number of lower limbs, which were given the names sympusapus (none), monopus (one), and dipus (two) (2,7). However, a more comprehensive classification was proposed by Stocker and Heifetz in 1987, which delineated seven distinct types (Figure 3): I:all thigh and leg bones 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.

This revised classification provides a more nuanced understanding of the variations seen in sirenomelia cases, considering the presence or absence of specific leg bones and their degree of fusion or separation.

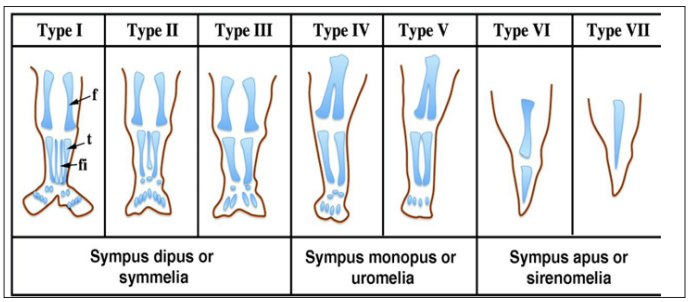

Figure 3: Classification of Sirenomelia Stocker et al,1987

There is limited knowledge of the embryological development of Serinomelia in humans, however extensive studies have been conducted in animal models to gain insight into the mechanisms underlying the development of this disorder Morphologically in a mouse, each lower limb derives from a different paramedial developmental field determined at gastrulation. This differentiation is primarily driven by the expression of Pitx1-dependent Tbx4. This developmental process occurs over 25 days, commencing at around the fourth week following fertilization, and by the end of the eighth week, the limb is perfectly formed. Lower limbs are delayed 2 days in comparison to forelimbs. Fibroblast growth factors 8 and 10 are theorised to play a role, however the precise trigger for limb bud initiation is still unknown.

Abnormal development in sirenomelia is characterised by two main non-mutually exclusive hypotheses: vascular and blastogenetic. The blastogenetic hypothesis postulates a primary anomaly in the development of the caudal axial mesoderm, while the vascular hypothesis is centered around an abnormal development of the umbilical vessels resulting in a deficient blood supply of the caudal part of the embryo [2-7]. This latter hypothesis is supported by observations of caudal "vascular steal" through a persistent single umbilical artery (SUA) which redirects blood flow from the lower limb to the placenta (Figure 4).

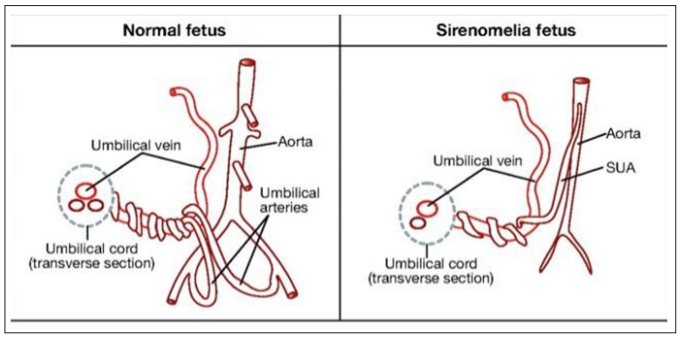

Figure 2: Persitent single umbilical artery found in most cases of sirenomelia. As described by Carlos Garrido-Allepuz et al

Conclusion

This case report highlights a rare condition, 'Sirenomelia' to document its incidence and further understand embryological processes resulting in its occurrence. It serves as a valuable contribution in shedding light, on this rare condition . It becomes increasingly evident that an improved and more comprehensive classification system for sirenomelia is imperative. The existing classification systems, though informative, lack an extensive distortion focused rationale, offering little more than discrete categorizations within a broader spectrum of anomalies. Consequently, there could be a diverse types of Sirenomelia that remain unaccounted for in the current classifcations. In our case, the presence of a single femur and 2 separate tibias, as seen on X-ray, does not fall within the seven- type classification system described by Stocker et al (Figure 3). This underscores the complexity and variability of sirenomelia presentations, necessitating a more comprehensive and adaptable classification approach.

Several environmental factors have been linked to the development of sirenomelia including maternal diabetes , drug abuse and heavy metal poisoning . However, it is noteworthy that in our particular case, none of these risk factors were identified during the evaluation. It is essential to continue investigation into environmental factors associated with the condition. This is especially crucial in regions of Asia where the prevalence of sirenomelia is notably elevated. A comprehensive understanding of the environmental factors contributing to sirenomelia is essential

for early detection, prevention, and management of this rare congenital anomaly in high-risk areas.

References

1. Iêda M Orioli (2011) Sirenomelia: An Epidemiologic Study in a Large Dataset from the International Clearinghouse of Birth Defects Surveillance and Research, and Literature Review. Am J Med Genet C Semin Med Genet 157C: 358-373.
2. Foerster A (1861) Die Missbildungen des Menschen. nebst einem Atlas (German) Manke Jena Friedrich https://archive.org/details/bub_gb_FHczQwMIscgC/mode/2up.
3. Stocker JT, Heifetz SA (1987) A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol 10: 7-50.
4. Kampmeier O F (1927) On sireniform monsters, with a consideration of the causation and predominance of the male sex among them. Anat Rec 34: 365.
5. Kallen B, Castilla EE, Lancaster PA, Mutchinick O, Knudsen L B, et al. (1992) The cyclops and the mermaid: an epidemiological study of two types of rare malformation. J Med Genet 29: 30-35.
6. Stocker JT, Heifetz SA (1987) Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pathol 10: 7-50.
7. Carlos Garrido Allepuz, Endika Haro, Domingo González Lamuño, María Luisa Martínez Frías, Federica Bertocchini, et al. (2011) A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations Disease Models & Mechanisms 4: 289-299.