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Systematic Review | Obstetrics & Gynaecology

SIRENOMELIA (MERMAID SYNDROME): UNEXPECTED INTRAPARTUM DIAGNOSIS IN AN UNBOOKED PREGNANCY WITH A PRISMA-BASED SYSTEMATIC REVIEW OF PATHOGENESIS, ANTENATAL DIAGNOSIS AND OUTCOMES.

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ABSTRACT Sirenomelia, commonly referred to as mermaid syndrome, is a rare and typically lethal congenital malformation characterized by fusion of the lower limbs and severe anomalies of the urogenital, gastrointestinal, and vascular systems. Despite advances in prenatal imaging, cases continue to present unexpectedly in labour wards, particularly in resource-limited settings. We describe the clinical journey of a preterm stillbirth with sirenomelia diagnosed intrapartum in an unbooked pregnancy and integrate this experience into a systematic review of contemporary and historical literature. This review synthesizes current evidence on embryological mechanisms, maternal risk factors, antenatal diagnosis, phenotypic classification, associated anomalies, prognosis, and preventive strategies. By bridging bedside observation with evidence-based literature, this manuscript highlights persistent gaps in antenatal care and underscores the importance of early screening, multidisciplinary counseling, and public health interventions.

KEYWORDS : Sirenomelia, Mermaid syndrome, Caudal regression spectrum, Prenatal diagnosis, Lower limb fusion, Stillbirth Systematic review.

INTRODUCTION

Rare congenital anomalies presenting unexpectedly during labour represent critical failures of antenatal surveillance systems, particularly in resource-limited healthcare settings.

The delivery of a fetus with a lethal congenital anomaly represents one of the most challenging intersections of clinical medicine, ethics, and emotion. Sirenomelia occupies a unique place among congenital malformations due to its striking phenotype and uniformly poor prognosis. The condition derives its name from Greek mythology, where sirens were depicted as beings possessing fused lower limbs resembling a mermaid's tail. Sirenomelia represents a complex spectrum of caudal developmental failure rather than an isolated limb anomaly[1]. The reported incidence ranges from approximately 0.8–1 per 100,000 births, with a male predominance and a strong association with stillbirth and early neonatal mortality[2,3].

Although advances in fetal ultrasonography permit diagnosis as early as the first trimester, unexpected labour ward presentations continue to occur, particularly among women lacking antenatal care access[4,5].

This manuscript follows a clinical journey beginning with an unbooked preterm labour admission culminating in post-delivery diagnosis of sirenomelia. Using this experience as a narrative anchor, a systematic review of literature was undertaken to explore evolving concepts of pathogenesis, diagnosis, and outcomes.

Case Presentation: Labour Ward Encounter

A 35-year-old Muslim female, gravida 3 para 1 live 1 abortion 1 (G3P1L1A1), presented to the labour ward of Kasturba Hospital, Delhi, at 20:40 hours on 21 January 2020 with severe labour pains. The pregnancy remained completely unbooked with no antenatal investigations or ultrasonography performed. There was no documented history of teratogenic drug exposure or consanguinity. Maternal glycemic status could not be assessed due to absence of antenatal investigations. Gestational age was estimated at 31 weeks and 3 days based on last menstrual period.

Abdominal examination revealed a uterus corresponding to preterm gestation with breech presentation. Fetal heart sounds were not localized despite repeated attempts.

Rapid labour progression resulted in vaginal delivery at 21:37 hours of a stillborn fetus weighing 1.3 kg.

External examination revealed:

Complete fusion of lower limbs forming a single limb mass.
Absence of anal opening.

Ambiguous external genitalia.
Gross urogenital malformation.

Upper limbs and craniofacial structures appeared grossly normal.

Based on classical phenotypic features, a diagnosis of sirenomelia was established.

Absence of fetal autopsy represents a limitation due to inability to confirm internal vascular anomalies.

The unexpected diagnosis prompted reflection on missed antenatal opportunities for early anomaly detection and counseling.

Figure 1: Showing complete fusion of lower limbs forming a single limb mass while upper limbs and craniofacial structures appeared grossly normal in a case of Sirenomelia.

Figure 2: Showing Ambiguous external genitalia in Sirenomelia

Materials and Methods

Study Design

This manuscript combines a single case report with a

systematic literature review conducted according to Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) recommendations.

Ethical principles for retrospective anonymized case reporting were followed. Patient identifiers were excluded.

Selection bias was minimized through predefined eligibility criteria and duplicate screening.

Literature Search Strategy

Electronic databases searched included: PubMed, Scopus, Google Scholar

Search period: January 2010 — March 2025.
Search terms included combinations of: "Sirenomelia", "Mermaid syndrome", "Lower limb fusion", "Caudal regression", "Prenatal diagnosis", "Vascular steal"
Boolean operators "AND" / "OR" were applied.
Manual reference cross-checking was also performed.

The search was designed and executed in accordance with PRISMA 2020 guidelines.

Figure 3. PRISMA flow diagram showing literature selection process .

Eligibility Criteria

Inclusion Criteria: Peer reviewed articles, English language publications, Case reports, Case series, Reviews and Observational studies.

Exclusion Criteria: Animal studies, Conference abstracts without full text, Duplicate publications.

Data Extraction

Extracted variables included:
Embryological theories
Maternal risk factors
Antenatal diagnosis
Associated anomalies
Survival outcomes

RESULTS :

Systematic Review

Embryological Basis

Two dominant theories exist.
The vascular steal hypothesis proposes persistence of an abnormal vitelline artery diverting blood flow from the caudal embryo leading to hypoperfusion[6].

Alternatively, defective blastogenesis suggests a primary gastrulation defect affecting caudal mesoderm formation[7].
Recent molecular evidence implicates retinoic acid dysregulation and altered BMP and WNT signalling pathways[8–10].

Phenotypic Classification

Stocker and Heifetz described seven skeletal types ranging from partial fusion to complete absence of lower limb bones[11].
The present fetus represents a severe phenotype.

Associated Anomalies

Renal agenesis occurs in over 90% of cases resulting in oligohydramnios and pulmonary hypoplasia[12].

Imperforate anus, genital anomalies, and vertebral defects are frequently described[13,14].

Maternal Risk Factors

Maternal diabetes mellitus remains the strongest association[15].
Additional factors include:
Extremes of maternal age.
Monozygotic twinning.
Teratogen exposure.
Poor antenatal access[16,17].

Antenatal Diagnosis

First trimester ultrasound can identify sirenomelia between 9–12 weeks gestation[4,18].
Markers include:
Absent kidneys.
Severe oligohydramnios.
Single umbilical artery[19].
Color Doppler supports vascular steal diagnosis[20].
Late gestation diagnosis becomes difficult due to oligohydramnios[21].

Prognosis

Most fetuses are stillborn or die shortly after birth due to renal failure and pulmonary hypoplasia[12].
Rare survivors require extensive reconstructive procedures[23].

Table 1. Key Findings From Included Studies on Sirenomelia

Sl No.	Author	Year	Study Type	Key Findings	Antenatal Diagnosis	Outcome
1.	Stevenson RE et al.	1986	Observational study	Proposed vascular steal hypothesis explaining caudal hypoperfusion	Not applicable	Established major pathogenetic theory
2.	Stocker JT & Heifetz SA	1987	Morphologic classification	Seven-type skeletal classification of sirenomelia spectrum	Not applicable	Standard classification still used
3.	Martínez-Frías ML	2012	Epidemiological study	Identified association with caudal regression spectrum and developmental field defect	Limited prenatal detection in older era	High stillbirth rate reported
4.	Garrido-Allepuz C et al.	2011	Experimental developmental biology review	Demonstrated role of retinoic acid imbalance and molecular signaling defects	Experimental models	Supported blastogenesis theory
5.	Tang TT et al.	2014	Case series	First trimester ultrasound may detect fused lower limbs and renal agenesis	Yes — early ultrasound	Early counseling possible

6.	Twickler DM et al.	2013	Imaging review	Doppler and targeted sonography improve vascular anomaly detection	Yes — anomaly scan	Prenatal diagnosis improved management
7.	Bulbul A et al.	2021	Case report	Severe oligohydramnios secondary to renal agenesis limits late imaging	Partial	Neonatal mortality within hours
8.	Ramasamy S et al.	2023	Case report with review	Maternal diabetes identified as major risk factor	Yes — late diagnosis	Poor neonatal survival
9.	Khatami F et al.	2024	Systematic review	Early anomaly scan remains most reliable diagnostic window	Yes — first trimester	Majority stillborn or early neonatal death
10.	Orioli IM & Castilla EE	2010	Epidemiologic registry analysis	Increased incidence in monozygotic twins and diabetic pregnancies	Variable	Uniformly poor prognosis

DISCUSSION

This case illustrates sirenomelia as both a biological anomaly and a systemic healthcare gap.

Despite technological advances, lack of antenatal registration continues to delay diagnosis. Early anomaly scanning allows counseling, psychological preparation, and legal termination options where applicable[4,5].

Despite advances in fetal imaging, disparities in antenatal care access remain a major contributor to delayed diagnosis in developing healthcare systems. Similar challenges have been reported across South Asia and Sub-Saharan regions, emphasizing the need for community-based early pregnancy registration[5,24].

From a public health perspective, strengthening antenatal outreach programs remains critical[24].

Clinical Learning Points

1. Sirenomelia remains strongly associated with severe oligohydramnios and renal agenesis.
2. First trimester ultrasound remains the most reliable diagnostic window.
3. Unbooked pregnancies continue to account for delayed diagnosis.
4. Multidisciplinary counseling is essential following antenatal detection.

CONCLUSION

Sirenomelia remains a rare but devastating congenital anomaly with profound clinical, emotional, and ethical implications. This journey-style systematic review demonstrates how a single labour ward encounter reflects broader challenges in antenatal care delivery. Strengthening early pregnancy surveillance, improving access to ultrasonography, and fostering multidisciplinary counseling remain essential to mitigating the impact of this lethal condition.

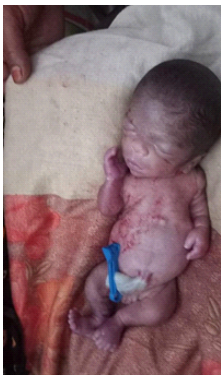

Figure 1: Showing complete fusion of lower limbs forming a single limb mass while upper limbs and craniofacial structures appeared grossly normal in a case of Sirenomelia.

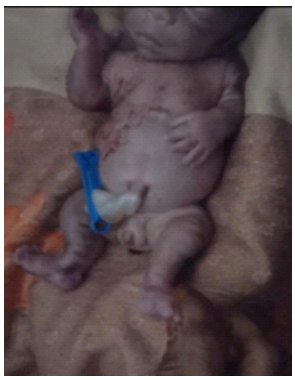

Figure 2: Showing Ambiguous external genitalia in Sirenomelia.

Figure 3 : PRISMA flow diagram showing literature selection process .

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