

Case Report

The first reported Sri Lankan case of Sirenomelia, the Mermaid Syndrome

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Introduction

Sirenomelia is a fatal congenital defect characterized by varying degrees of lower limb fusion, spinal anomalies, sacral agenesis, genitourinary and anorectal atresia¹. It was first described by Rocheus in 1542¹ and has an incidence of 0.8-1/100,000 births² where only around 300 cases have been reported from all around the world³. The etiology of this syndrome is not well-known; however, maternal diabetes, maternal drug abuse, genetic predisposition, and vascular hypoperfusion have been suggested as possible causative factors³. There is a strong association with maternal diabetes where the relative risk is 1: (200-250)^{3,4}. We report a case of sirenomelia in Sri Lanka, resulting in fetal demise at 34 weeks of gestation.

Case report

A 30-year-old primiparous mother delivered a macerated fetus with multiple congenital abnormalities at 34 weeks of gestation. The parents were not consanguineous; their family history was unremarkable, and the mother had no comorbidities during

pregnancy. Ultrasound scan at 22 weeks revealed severe growth restriction and oligohydramnios with multiple anomalies, including lemon-shaped skull, small cerebellum, partial sacral agenesis, pelvic kidney with moderate hydronephrosis, and single umbilical artery. Parents were counselled regarding the poor prognosis.

The mother got admitted with reduced fetal movements at 34 weeks of gestation, and the ultrasound scan revealed an intrauterine death. She delivered a macerated fetus with a birth weight of 900 g. The most characteristic finding was the fused lower limbs and feet, with ten toes. The baby also lacked external genitalia, and there was an imperforate anus, narrowed thorax, and a single umbilical artery (Figure 01). Furthermore, the baby was found to have typical Potter's facies, inclusive of an elongated head, low-set ears, prominent bilateral epicanthic folds, and a flattened nose (Figure 02). The X-ray revealed two femurs, two tibias, two fibulas, fused feet with ten phalanges, partial sacral agenesis, and lung hypoplasia (Figure 03). The parents declined an autopsy.



Figure 01 - posterior view of the fetus showing fused lower limbs and imperforated anus



Figure 02 - anterior view of the fetus showing absent external genitalia, fused lower limbs, potter's facies, and a narrowing chest



Figure 03 – X-ray of the fetus

Discussion

Early medical literature categorizes sirenómelia as the most severe form of caudal regression syndrome⁵. However, recent literature specifies it as a similar but distinct disorder⁵. The vascular steal hypotheses and the defective blastogenesis hypotheses are the closest to describing the pathogenesis of sirenómelia as the precise pathogenesis is still unknown⁶.

Scarce blood flow to the caudal mesoderm results in agenesis of lower abdominal structures such as the lumbosacral spine, pelvis, genitourinary and anorectal structures and subsequently leading to approximation and fusion of lower limbs according to the vascular steal hypotheses whereas, according to the blastogenesis hypothesis, the principal defect in the development of caudal mesoderm is attributed to a teratogenic factor during early embryonic development causing the defective formation of caudal structures⁶.

Principal features of sirenómelia are fusion of lower extremities, single umbilical artery, and persistent vitelline artery⁵. Our patient displayed all these findings. Sirenómelia is also associated with Potter's facies which include large low-set ears, flat nose, hypertelorism, prominent epicanthic folds, oligohydroamniosis, and lung hypoplasia⁷. All these features were present in our patients as well. *Stocker et al* classified Sirenómeliac infants depending on the presence or absence of bones within the lower limb into seven categories (Type I to Type VII)⁸. Our patient could be classified as Type I as all thigh bones and leg bones are present.

Sirenómelia is usually incompatible with life where most babies die within hours to days of birth due to associated visceral abnormalities such as renal agenesis and severe lung hypoplasia⁷, which is also the case in our patient who presented as an IUD at 34 weeks of gestation. The few cases that

survived had less visceral involvement where a series of surgical procedures were essential to separate the lower limbs and correct the other defects⁸. In conclusion, Sirenómelia is a rare congenital anomaly with a poor prognosis. Antenatal diagnosis is possible with an accurately detailed ultrasound scan. Even though the exact etiology has not been identified, good blood sugar control and avoidance of exposure to teratogens during pregnancy might help prevent this lethal anomaly.

References

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