

## Sirenomelia- A Case Report

<sup>1\*</sup>Dr. Priyanka Dhavjekar, <sup>2</sup>Dr Pia Muriel Cardoso

<sup>1</sup>Junior Resident, Dept Of Obstetrics And Gynaecology, Goa Medical College.

<sup>2</sup>Associate Professor, Dept Of Obstetrics And Gynaecology, Goa Medical College.

### I. Introduction

Sirenomelia, more commonly known as ‘mermaid syndrome’ is an extreme example of caudal regression syndrome. The characteristic feature of this rare congenital fetal anomaly is the complete or partial fusion of the lower limbs.<sup>1</sup>



The incidence of sirenomelia is 0.8-1case/100,000 births with a male to female ratio of 3:1<sup>2</sup>. There are approximately 300 cases reported in literature, 15% of which are associated with twinning, most frequently monozygotic. There is a strong association with maternal diabetes and approximately 22% of the foetuses born with this anomaly were associated with diabetes in the mother<sup>3</sup>.

### II. Case Study

The patient was a 24years old Primigravida who was referred to our institute from a Karwar health centre at 35wks gestation as a case of severe oligohydramnios for further management. The patient was otherwise healthy, with no history of drug abuse or genetic illness in the family. She had documented reduced liquor in a prior scan at 20-22wks gestation. Her OGCT 75g glucose (DIPSI guidelines) was increased 140mg%, which was done at 23 weeks gestation. Fasting and post prandial values were 150 and 203mg%. On clinical examination liquor was grossly reduced, with no demonstrable liquor pocket on ultrasound. Fetal MRI was performed in the evaluation of this severe oligohydramnios, in which both fetal kidneys and bladder were not visualised, suggestive of bilateral fetal renal agenesis. The patient and relatives were prognosticated regarding the poor fetal outcome. Subsequently the patient wished to follow up in her home town and was referred to us again when she came in labor at 38weeks. She was already in the second stage of labor with cephalic presentation and a fetal heart rate of 120 beats per minute. She delivered an infant weighing 1.8kg, with an APGAR score of 3 and 1 at 0 and 5 minutes. Subsequently the baby expired within 30minutes of life. On physical examination the baby had a narrow chest, bilateral fused index and middle fingers and the lower limbs were fused together having two feet. The feet had 10 toes and were rotated externally. Clinical examination revealed that both femoral bones were present, with two tibia and one fused fibula, thus classifying our patient as type II according to the Stocker and Heifetz classification. The infant also had features of Potter’s facies including prominent infra-orbital folds, small slit like mouth, receding chin and low set ears. The infant had imperforate anus, absent urinary orifice and undetermined sex (no external genital organs). Autopsy was declined by the parents. The intrapartum and postnatal course of the mother was uneventful.



### III. Discussion

Sirenomelia is one of the most severe forms of caudal regression syndrome. Affected infants are born with partial or complete fusion of the legs. Additional malformations may also occur including genitourinary abnormalities, gastrointestinal abnormalities, anomalies of the lumbarsacral spine and pelvis and absence or underdevelopment (agenesis) of one or both kidneys. Affected infants may have one foot, no feet or both feet, which may be rotated externally. The tailbone is usually absent and the sacrum is partially or completely absent as well. Additional conditions may occur with sirenomelia including imperforate anus, spina bifida, and heart (cardiac) malformations. Sirenomelia is often fatal during the newborn period.

The primary molecular defect resulting in sirenomelia is yet to be unshrouded. There are two main hypothesis of its pathogenesis and they are the vascular steal hypothesis and the defective blastogenesis hypothesis. According to the vascular steal hypothesis<sup>4</sup>, there is deficient blood supply to the caudal mesoderm resulting in agenesis of midline structures and thus resulting in abnormal approximation of the lower limb structures and the fusion of the two lower limbs. However in defective blastogenesis hypothesis<sup>5</sup> the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage. Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures.

Some of the environmental risk factors implicated in sirenomelia are maternal diabetes, smoking, heavy metal exposure and retinoic acid.<sup>6</sup> Sirenomelia is associated with severe life-threatening complications and is often fatal in the first years of life. However, survival beyond infancy into later childhood or young adulthood has been reported in a handful of cases. The characteristic finding of sirenomelia is partial or complete fusion of the lower legs. The degree of severity is highly variable. Affected infants may have only one femur (the long bone of the thigh) or may have two femurs within one shaft of the skin. Affected infants may have one foot, no feet or both feet, which may be rotated so that the back of the foot is facing forward.

Affected infants may also have a variety of urogenital abnormalities<sup>8</sup> including absence of one or both kidneys (renal agenesis), cystic malformation of the kidneys, an absent bladder, narrowing of the urethra (urethral atresia). In addition, they may have an imperforate anus, a condition in which a thin covering blocking the anal opening or the passage that normally connects the anus and lowest part of the large intestine (rectum) fails to develop.

Infants with sirenomelia may also have abnormalities affecting the sacral and lumbar spine<sup>9</sup>. In some patients, abnormal front-to-back curvature of the spine (lordosis) may occur. Affected individuals may also lack external genitalia. Absence of the spleen and/or the gallbladder has also been reported. Defects affecting the abdominal wall may also occur such as protrusion of a portion of the intestines through a hole near the bellybutton (omphalocele). Some individuals with sirenomelia may have a meningomyelocele, a condition in which the membranes that cover the spine and, in some cases, the spinal cord itself protrude through a defect in the spinal column. Congenital heart defects and respiratory complications such as severe underdevelopment of the lungs (pulmonary hypoplasia) can also be associated with sirenomelia.

The facial abnormality usually found in sirenomeliac infants known as Potter's facies<sup>7</sup>, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. In both of our cases, features of Potter's facies were present. When features of Potter's facies are combined with oligamnios and pulmonary hypoplasia it is known as Potter's syndrome, which was present in our second case. In our first case, the right thumb was hypoplastic, which was also previously reported. Stocker and Heifetz<sup>10</sup> classified Sirenomeliac infants from Type I to Type VII according to the presence or absence of bones within the lower limb.

Type	Femur	Tibia	Fibula	Description
I	2	2	2	All bones present
II	2	2	1	Single fused fibula
III	2	2	0	Fibula absent
IV	Partially fused	NA	Fused	Femur partially fused; fibula fused
V	Partially fused	NA	Not fused	Partially fused femur
VI	1	1	NA	Single femur; single tibia
VII	1	0	NA	Single femur; tibia absent

#### IV. Conclusion

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in preconceptional period and in first trimester should be maintained to prevent this anomaly.

#### References

1. Valenzano M, Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. Sirenomelia. Pathological features, antenatal ultrasonographic clues, and a review of current embryogenic theories. *Hum Reprod Update*. 1999;5:82–6.
2. Reddy KR, Srinivas S, Kumar S, Reddy S, Hariprasad Irfan GM. Sirenomelia a rare presentation. *J Neonatal Surg*. 2012;1:7.
3. Aslan H, Yanik H, Celiklaslan N, Yildirim G, Ceylan Y. Prenatal diagnosis of Caudal regression syndrome: A case report. *BMC Pregnancy Childbirth*. 2001;1:8.
4. Sadler TW, Rasmussen SA. Examining the evidence for vascular pathogenesis of selected birth defects. *Am J Med Genet A*. 2010;152A:2426–36.
5. Duesterhoeft SM, Ernst LM, Siebert JR, Kapur RP. 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:3175–84.
6. Naveena S, Mrudula C. Sirenomelia — The mermaid syndrome: A case report. *IOSR J Dent Med Sci*. 2013;7:01–4.
7. Dharmraj M, Gaur S. Sirenomelia: A rare case of foetal congenital anomaly. *J Clin Neonatol*. 2012;1:221–3.
8. Baxi L, Warren W, Collins MH, Timor-Tritsch IE. Early detection of caudal regression syndrome with transvaginal scanning. *Obstet Gynecol*. 1990;75:486–489.
9. Mariani AJ, Stern J, Khan AU, et al. Sacral agenesis: An analysis of 11 cases and review of the literature. *J Urol*. 1979;122:684.
10. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. *Perspect Pediatr Pathol*. 1987;10:7–50.

\*Dr. Priyanka Dhavjekar. "Sirenomelia- A Case Report." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)* 16.11 (2017): 01-03