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# SIRENOMELIA: A RARE CASE OF FETAL CONGENITAL ANOMALY

Gynaecology

**Dr Kanika Kinra*** MBBS, Resident Internal Medicine, SGL Charitable Superspeciality Hospital, Garha Road Jalandhar, Punjab *Corresponding Author

**Dr Neelu Khanna Suri** MBBS, MS Obstetrics & Gynaecology , SGL Charitable Superspeciality Hospital, Garha Road Jalandhar, Punjab

**Dr Pawan Kumar Suri** MBBS, MD, DM Cardiology , SGL Charitable Superspeciality Hospital, Garha Road Jalandhar, Punjab

**Dr Anuj Sangwan** MBBS, Medical Officer, Sonipat HCMS-1, Haryana Government

## ABSTRACT

Sirenomelia was first described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek God sirens1. This rare syndrome has the incidence of 0.8-1 case/100,000 births with male to female ratio being 3:12. We here report a rare case of primigravida with 28weeks of pregnancy who delivered a live sirenomeliac baby with identifiable risk factors being Cytomegalovirus (CMV) Infection (raised titres), Toxoplasma IgG avidity positive, Hypothyroidism, Vitamin D deficiency and herbal infertility treatment taken by male partner. The baby had typical potter facies, fused legs typically Sirenodipus and no external genitalia.

## KEYWORDS

Sirenomelia, Cytomegalovirus, Hypothyroidism, Toxoplasma, Vitamin D

## INTRODUCTION:

Sirenomelia was first described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek God sirens[1]. This rare syndrome has an incidence of 0.8-1 case/100,000 births with male to female ratio being 3:1[2] and is characterized by fusion of lower limbs, single umbilical artery, severe malformation of urogenital and lower gastrointestinal tract. Resultant oligohydraminos, leads to these infants having Potter's facies and pulmonary hypoplasia. Another important feature is the presence of a single large artery, arising high in the abdominal cavity that assumes the function of the umbilical arteries and diverts nutrients from the caudal end of the embryo distal to the level of its origin. By 4 th week of gestation formation of the sacrum/lower back and corresponding nervous system is usually nearing completion. According to some studies perturbations of mesoderm specification, epithelial-mesenchyme transition, and mesodermal cell migration can lead to such structural birth defects. [3]

The malformation sequence consists of varying degrees of lower limb fusion bearing a resemblance to the mermaid of ancient Greek mythology. Oligohydramnios secondary to severe renal dysplasia is universal [4-6].

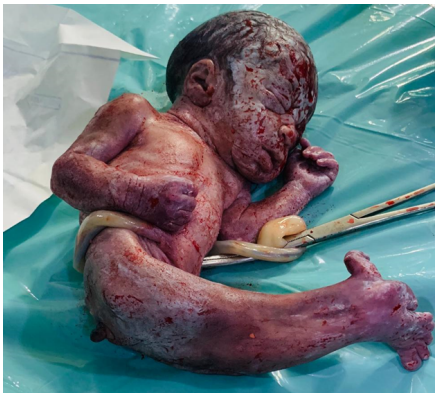
## CASE REPORT:

Mrs X 28 Y Primigravida ,married for 2 and half years, unbooked patient presented to us at 28 weeks of gestation with preterm labour pains with USG documented showing SLIUF of average gestational age 28weeks+5days with anhydramnios, single umbilical artery, absent fetal bladder, non-visualization of kidneys in renal fossa & large stomach bubble, her old records showed that she was tested positive for cytomegalovirus in her 5th month of pregnancy but she didn't took any treatment for that , also she had Toxoplasma IgG avidity positive ,also she was diagnosed with hypothyroidism during 4th month of her gestation , was started on thyroxin supplementation , she had low Vitamin D levels during her antenatal period, patient delivered live sirenomeliac baby with fused legs , no genitalia was formed. Baby had an APGAR score of 5/10 and baby survived for one hour. Past history that Her husband had oligospermia for which he was taking some herbal / ayurvedic treatment .They told that after starting treatment of infertility by husband she conceived within 15 days of his treatment initiation.

The identifiable risk factors in our patient were that she developed hypothyroidism during her gestation, also she had CMV infection which might have caused the congenital malformation, avidity test suggesting past toxoplasma infection & may be those non documented herbal medicines that her husband took for oligospermia would have led to the above mentioned causation. Also there were low Vitamin D levels whose consequences cannot be ignored and is rather an upcoming area of research.

There was no history of exposure to any known teratogenic agent during the antenatal period. Sirenomeliac babies have an association with maternal diabetes and monozygotic twins,however our patient did not have any such history. On examination, the neonate had a flat facial profile (Potter's facies). The lower limbs were fused from the hip downwards and two feet were seen, (sirenomelia dipus); external genitalia were absent (Fig. 1)

A small phallus like structure or rudimentary tail was seen in the back , the anal orifice was absent(fig 2)



## DISCUSSION:

Sirenomelia is characterized by fused lower limbs, severe renal anomalies with oligohydramnios and is now considered to be a separate entity. It is frequently associated with VACTERL syndrome. [7]

Swader reported the first surviving infant in 1989. Till 2006, 6 cases of surviving infants with mermaid syndrome were reported. The etiology

and pathogenesis of this malformation is unknown. Duhamel in 1961 stated that Sirenomelia and anorectal malformations represent the two extremes of a single comprehensive syndrome arising from an embryonal defect in the formation of the caudal region. He called it the Syndrome of Caudal Regression [8-12]

Sirenomelia has been classified into three types according to the number of lower limb bones present:

1. Sirenomelia apus: No feet only one tibia and one femur.
2. Sirenomelia unipus : One foot, two femurs two tibia, and two fibula.
3. Sirenomelia dipus: Two feet and two fused legs giving the appearance of a flipper.

Sirenomelia dipus, also called as Mermaid syndrome, has the most favorable outcome.[9-13]

Our above reported case was terminated on the basis of last ultrasound diagnosis of bilateral renal agenesis with no liquor volume. The foetus was identified to have characteristic features of Sirenomelia at the time of termination.

## CONCLUSION:

We report this case due to its rarity and term live birth with associated maternal CMV infection, past toxoplasmosis , Vitamin D deficiency & hypothyroidism, signifying early detection and treatment of such dreaded infections can lead to early diagnosis and appropriate management of such babies, either in form of early termination or infection treatment, either ways decreasing morbidity and DALYs. It is our hope that this report will add various associations to the existing knowledge and data about the condition.

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**Conflicts of interest**
There are no conflicts of interest.

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