SIRENOMELIA: A RARE CONGENITAL ANAMOLY

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INTRODUCTION: Sirenomelia or "mermaid syndrome" represents a severe form of caudal regression. It is a rare congenital malformation that is incompatible with life[1].

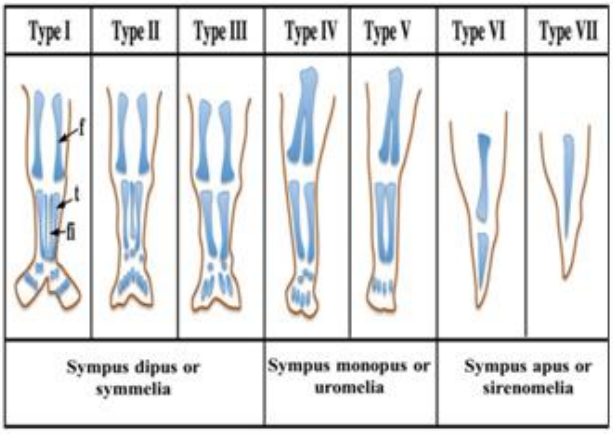
In Greek mythology, the Sirens were three creatures with the head of a woman and the body of a bird from the wings down. They were dangerous to sailors, whom they narcotized with their enchanting music and voices to later kill them.

Over time, these bird-women were portrayed as more aquatic creatures, and eventually with a full mermaid-like appearance[2, 9]. It is likely that creatures of classical and medieval mythology were inspired by the observation of real cases of human malformations and it is likely that Sirens were similarly inspired.

However, despite the present perception of Sirens as romantic and cute creatures, the sirenomelia human malformation is a severe condition[6].

Ultrasound may be useful in the early antenatal detection of this anomaly. This is the first case of sirenomelia reported in the black race.

Sirenomelia has been classified by Stoker and Heifetz into 3 types according to the number of lower limb bones present.



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

CASE STUDY: A 27 years old pregnant female, $G_2P_1L_1$ came for routine antenatal scan at 21 weeks gestation.

Ultrasound examination at 14 weeks showed a single viable foetus with gestational age 2 weeks less than that with LMP.

There was no history of diabetes, hypertension, and consumption of teratogenic drugs, tobacco or alcohol.

Amniocentesis for chromosomes was scheduled at 21 weeks but was not performed due to severe oligohydramnios.USG of the abortus revealed bilateral renal agenesis.

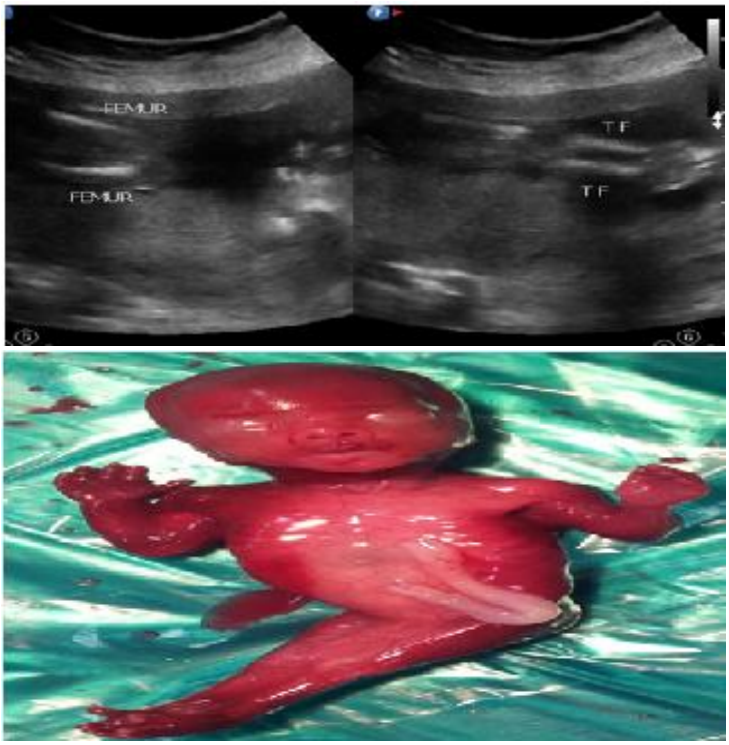
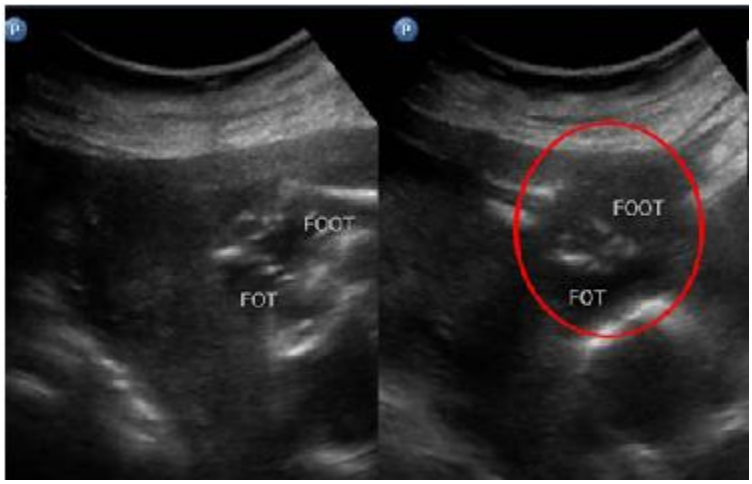


Figure1 - USG and Gross images shows fused lower limbs with Talipes Equino Varus deformity in both feet with both heels in close proximity to each other and absent external genitalia.

TALIPES EQUINOVARUS (CLUB FOOT)



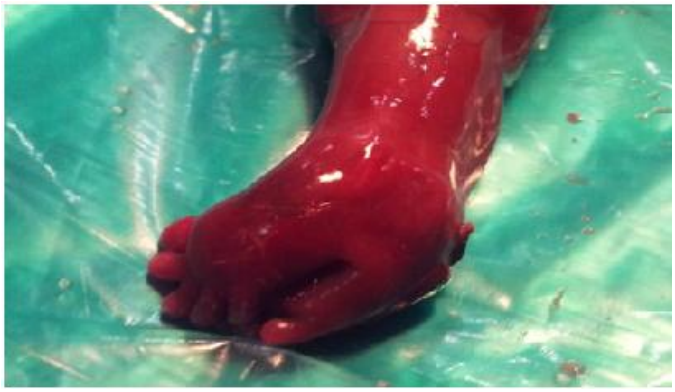


Figure 2 - Talipes Equinovarus deformity in both feet with both heels in close proximity to each other.

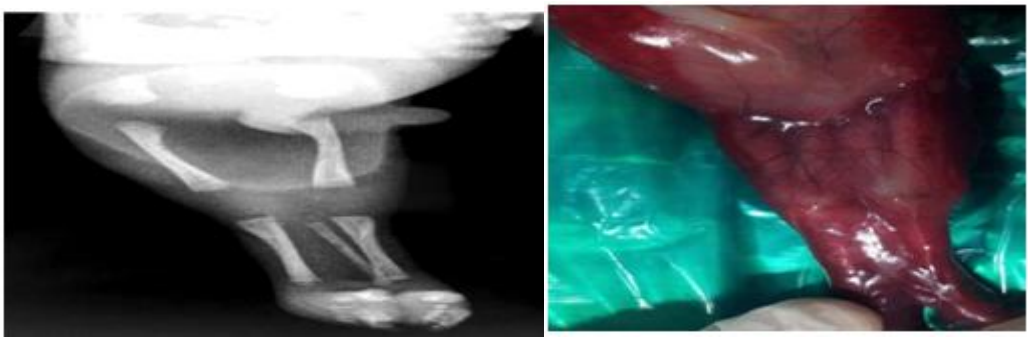


Figure 3 - X ray and Photograph of the specimen showing two femurs, two tibiae and fibulae and two feet- Sirenomelia dipus

SPINA BIFIDA OCCULTA

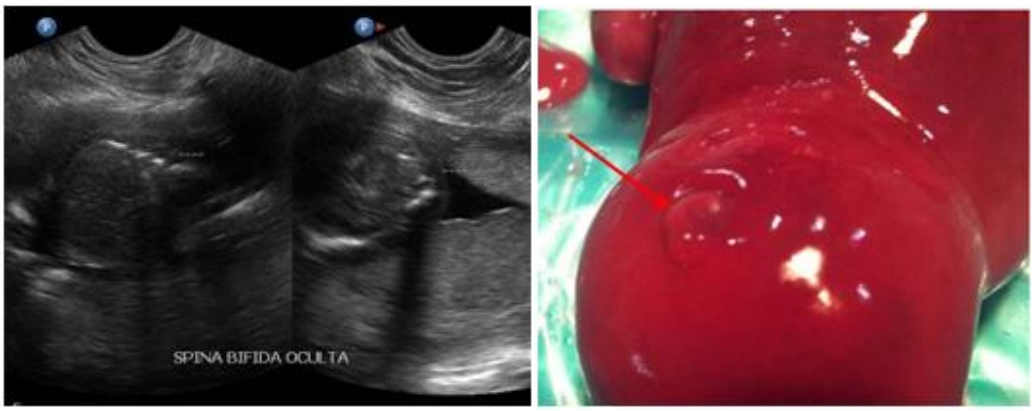


Figure 4-(a) USG showing absence of posterior vertebral elements with intact overlying skin suggestive of spina bifida occulta (b) Photograph of the foetus showing a dimple over the sacral region.

VENTRICULAR SEPTAL DEFECT

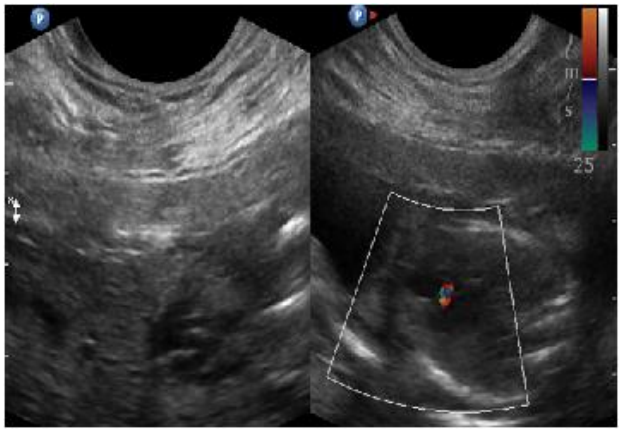


Figure 5- B-Mode and Color Doppler Antenatal fetal echocardiography showing a small fetal VSD.

BILATERAL RENAL AGENESIS

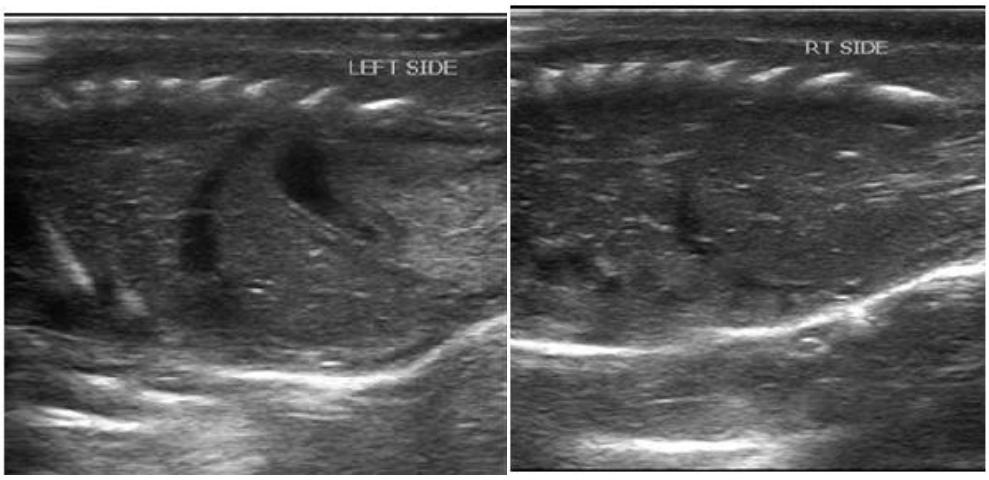


Figure 6- Antenatal sonography showing absence of renal tissue on both sides suggestive of Bilateral renal agenesis.

DISCUSSION: Sirenomelia is a rare and fatal congenital anomaly with an incidence of 0.8 to 1 case per 1,00,000 births with male to female ratio of3:1.This malformation sequence consists of varying degrees of lower limb fusion bearing a resemblance to the mermaid of ancient Greek mythology[3].It is a lethal condition and death is usually due to renal agenesis which is incompatible with life[4]. Oligohydramnios secondary to severe renal dysplasia is universal.

The first medical description of Sirenomelia was by Rocheus and Polfyr way back in the sixteenth century. Duhamel in 1961 defined all the anomalies of mermaid syndrome and described it as the most severe form of caudal regression syndrome (CRS).CRS is thought to be the result of injury to the caudal mesoderm early in gestation.

The etiology of a Sirenomelia is unknown, but it is not believed to be hereditary[5].It presents with lower limb fusion (of different degrees), Sacral and pelvic bony anomalies, absent external genitalia, imperforate anus, renal agenesis or dysgenesis[8] (which leads to severe oligohydramnios

and pulmonary hypoplasia), malformed vertebrae and hemivertebrae, CNS anomalies and cardiac defects[4, 7.Fetuses] with sirenomelia almost invariably exhibit single umbilical artery.

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