



Mermaid Syndrome : Sirenomelia – A case report.

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Introduction

- Sirenomelia or mermaid syndrome is a rare congenital anomaly showing partial or complete fusion of lower limb bones. It is also associated with renal agenesis and incomplete development of the genitourinary system with oligohydramnios.
- Sirenomelia is at times also found to be associated with thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia. Mermaid syndrome , being a rare anomaly , is found in about 0.8-1 case / 100,000 births with a 3:1 male is to female ratio , with around 300 cases studied in literature.

Case Report

- A 30 years Primigravida , registered pregnancy , presented with her anomaly scan of 19 weeks 5 days with bilateral renal agenesis, absent bladder , single lower limb with a probability of fused femur , two leg bones seen and anhydramnios.
- She had no complains and had no significant medical or surgical history.
- This was a spontaneous conception with no history of diabetes mellitus, limb or renal defects in her or her family members.
- There was no history of drug intake or radiation exposure. Her blood sugars in this pregnancy were normal.
- On explaining the poor prognosis of the baby the couple opted for medical termination of pregnancy with T Mifepristone 200mg and T. Misoprostol 200mcg.
- The foetus delivered had a single lower limb , with one foot and fused toes.
- There was a skelektal deformity at level of sacrum , no external genital organs were seen , thus sex could not be determined.
- Features of potter facies with low set ears , prominent epicanthal folds , flattened nose and recessed chin were also seen.
- A baby gram was done which showed a single femur and two lower limb bones which were unfused. The sacral bone was not completely formed and the remaining pelvic bone was absent.



Figure 1 Single lower limb with fusion of last two toes.



Figure 2 showing low set ears



Figure 3 features of potter facies

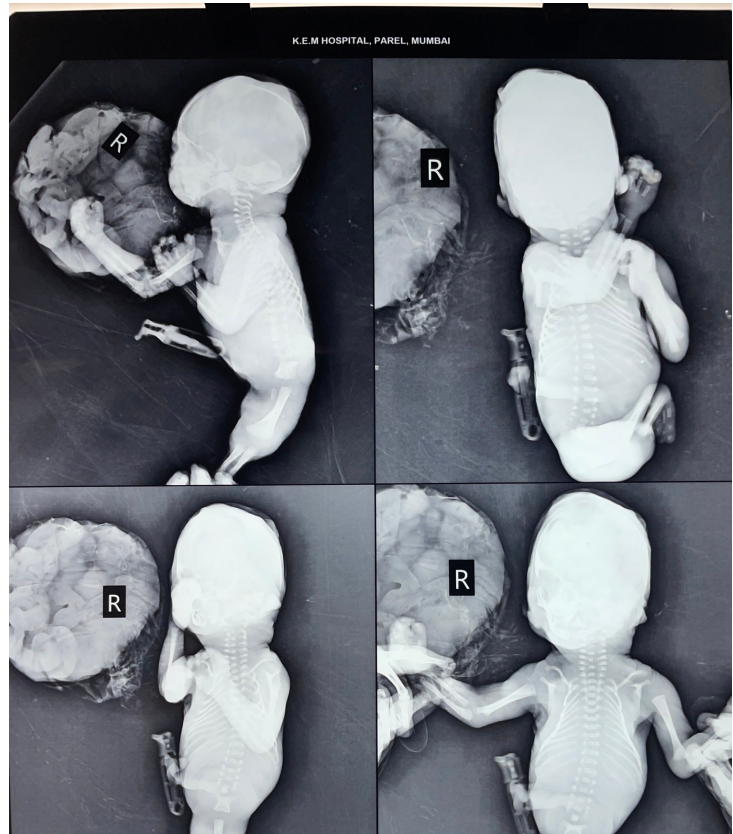


Figure 4 shows a baby gram

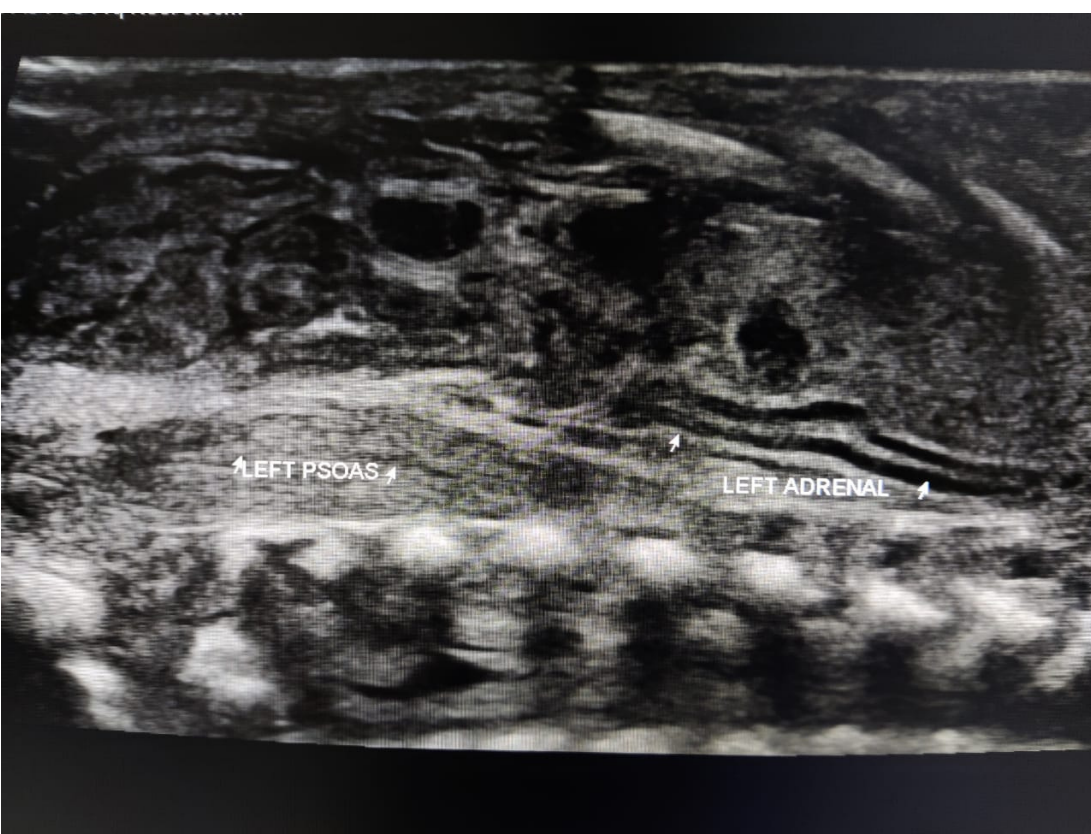
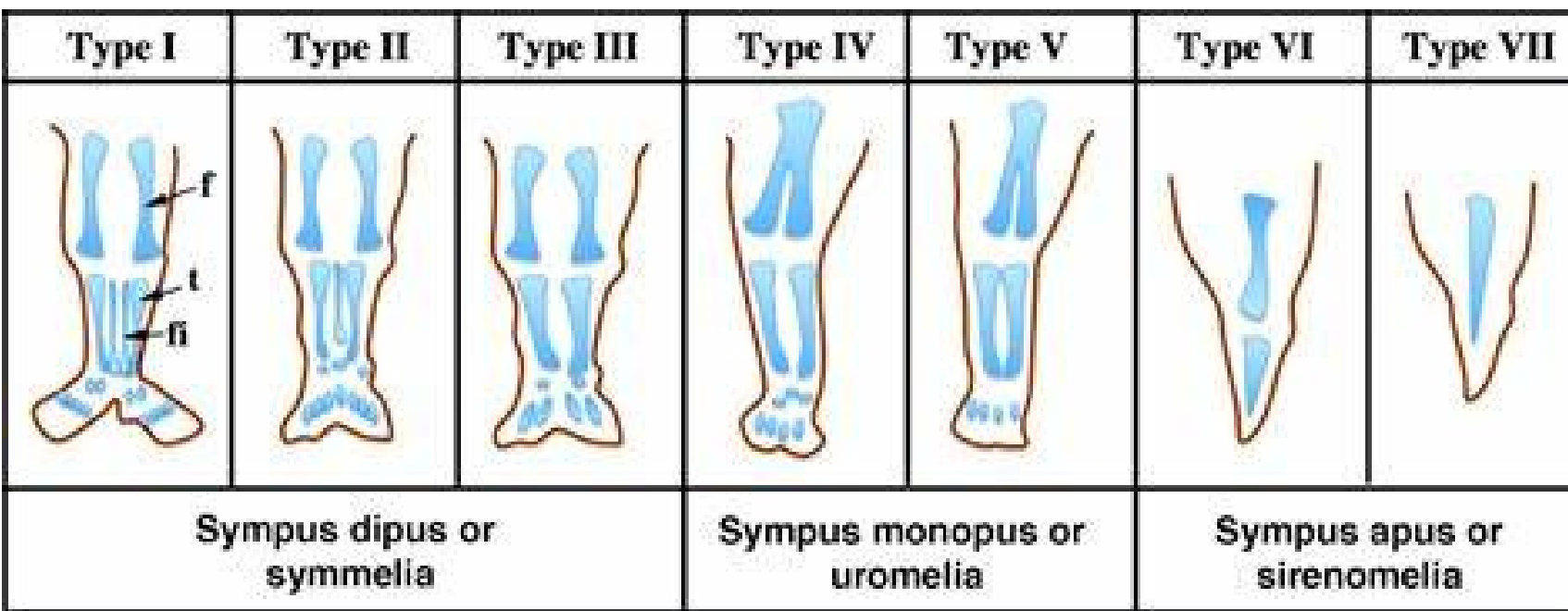


Figure 5 is a postnatal ultrasound

- Post natal ultrasound examination of the fetus showing bilateral lying down adrenals with bilateral renal agenesis. Autopsy was declined by the patient and her relatives.

Discussion and Conclusion

- Sirenomelia is also shown to portray strong association with maternal diabetes mellitus .It is theorized ,the oxygen free radicals due to maternal deranged sugars may be teratogenic.
- This foetus showed features of potter’s syndrome.
- Stocker and Heifetz classified sirenomelia into 7 types by the degree of fusion of lower extremities’ bones
- Vascular steal hypothesis implies that due to the limbs being fused there is deficiency of blood supply and nutrient deficiency leads to agenesis of midline structures.



- Defective blastogenesis hypothesis implies that a teratogenic event in gastrulation stage causes abnormal notochord and caudal medoderm formation thus causing defect in caudal structures formation.
- 14 cases have been documented in India so far. Sirenomelia should be detected on ANC scan. Screening for Diabetes Mellitus is a must. It is not a preventable disorder but early diagnosis through ultrasonography is a key to reduce both maternal and foetal morbidity.

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