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Sirenomelia

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Key words : - Mermaid syndrome, Vitelline artery steal.

Introduction

Sirenomelia is a rare congenital anomaly characterized by abnormal development of the caudal region of the body with variable degree of fusion of lower extremities. It derives its name by its resemblance to the Mermaid (Siren) of Greek mythology.

About 300 cases have been reported in the world so far out of which 9 have been reported in India. Sirenomelia is also known as Sirenomelia sequence, Symmelia, Symposia, Uromelia and Monopodia.

Case report:

A twenty two year old primigravida came at twenty eight weeks of unsupervised pregnancy. There was no history of consanguinous marriage and any illness or medication during pregnancy.

Antenatal Ultrasound showed breech presentation with severe oligohydramnios. Lower fetal spine could not be traced and fetal kidneys were not visualised. The fetal brain appeared normal with normal fetal cardiac activity.

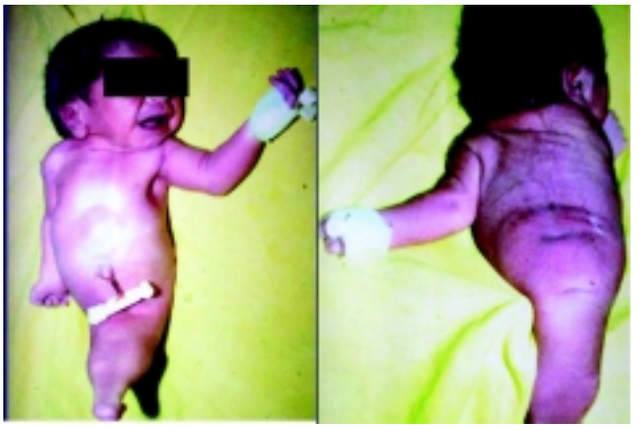

Fig. 1: Photographs of the baby showing fusion of both lower limbs with single rudimentary flap like foot.

Patient gave birth to a preterm baby of 26-28 weeks of gestational maturity weighing 1.4 kg. The baby cried immediately after birth and survived for 49 hours.

Physical examination [Fig.1] of the baby showed fusion of both lower limbs with single rudimentary flap like foot. There was no anal or urethral openings with no external genitalia. The upper segment of the body was normal. There was persistent frothing from the mouth. The umbilical cord showed single umbilical artery.

Whole body radiographs [Fig.2&3] of the baby showed fused soft tissue of the lower limbs with fused femorae in upper two-third. There were separate normal tibia with absent fibula on both sides. There was a single common foot like structure having two metatarsals and one phalynx. Spine showed multiple hemivertebrae in cervicodorsal and lumbar region with open spinal canal. Only part of sacrum was present, lower sacrum and coccyx was absent. The right lung was hypoplastic with right sided pneumothorax. The inserted nasogastric tube was seen coiled in upper blind pouch of oesophagus with gas shadow present in stomach suggestive of 'C' type of Tracheo-oesophageal fistula.(Acc.to GROSS' classification).

Ultrasound of the baby showed both kidneys, urinary bladder and internal genitalia were absent.

Due to cultural constraints autopsy of the baby was not done.

Discussion

The first case of sirenomelia was reported in 1542. Duhamel [1] coined the term Caudal regression syndrome in 1961 to describe the association of sirenomelia with anorectal, genitourinary and vertebral anomalies. Later a distinction was made between Siernomelia sequence and Caudal regression syndrome in the belief that the former

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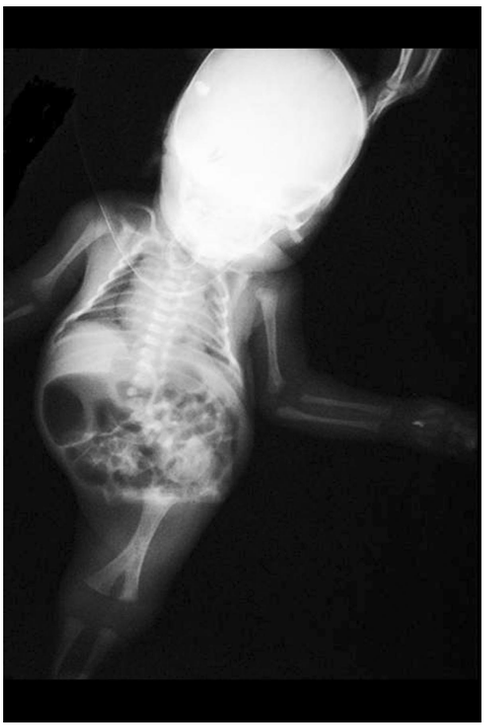

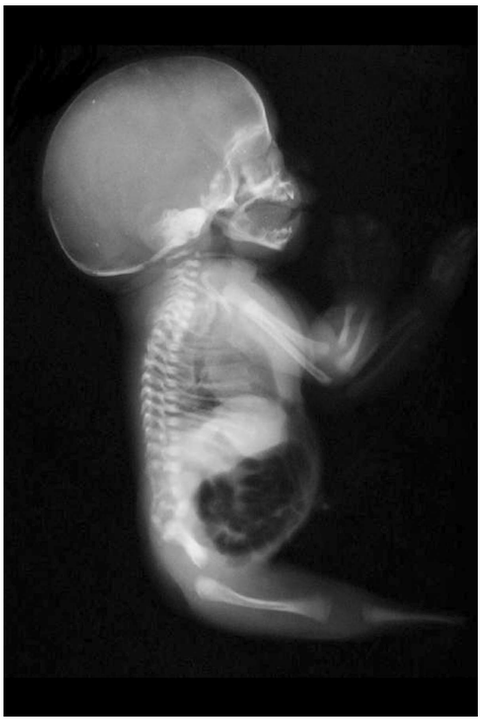

Fig. 2&3: Whole body radiographs of the baby showing fused femorae in upper two-third with separate normal tibia & absent fibula on both sides.

had a specific pathogenetic factor namely arterial steal phenomenon, whereas the later was probably a heterogenous group with Diabetic embryopathy as a single most frequently suspected etiological factor. In 1973 Quan and Smith [2] proposed the acronym VATER to denote the non-random association of Vertebral defects, Tracheo-oesophageal fistula with esophageal atresia, Renal and Radial limb defects. It was suggested that categerisation as VATER required presence of atleast 3 of this defects.The acronym was later expanded to VACTERL in order to emphasize the importance of associated Cardiac and non-radial limb defects. Two major publications done on these malformation are :(1) Duncan et al [3] : a series of 50 pts out of which 48 showed at least 3 of the 6 VACTERL anomalies. (2) Stocker and Heifetz [4] : All 80 Sirenomelics had at least 3 of the 6 VACTERL anomalies.

The exact etiopathogenesis is uncertain. The proposed theories are Vascular steal or Vitelline artery steal theory by Stevenson [5] which is the most accepted. Others include teratogenic effects of lead, cadmium and vit.A, primitive streak defect, axial mesodermal dysplasia sequence.Maternal diabetes mellitus and genetic predisposition is also implicated. Vitelline artery steal theory [5] states that an aberrant vessel(derived from the vitelline artery) arises from high abdominal aorta in the affected fetus and functions as the vessel returning blood through the umbilical cord to placenta. This vessels steals blood from structures distal to its origin.

Hence this is a lethal condition due to associated renal agenesis and its complication leading to prolonged severe oligohydramnios and hence pulmonary hypoplasia.

Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks, a 13 year old girl who was born with fused legs.Over the years she has undergone numeruos operations to separate her lower extremities.

Prenatal diagnosis by demonstrating the fused femorae or decreased distance between the two femurs is desired in early pregnancy so that termination of pregnancy can be offered at the earliest after proper parental counselling.

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