Sirenomelia- The Mermaid Syndrome: A Case Report

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Abstract: Sirenomelia, Mermaid syndrome, is a very rare and fatal congenital anomaly. In Sirenomelia lower limbs are fused and atrophied. The lower body looks like tail of a fish and the upper body is like human . Hence they are known as mermaid syndrome.
Musculoskeletal anomalies, renal agenesis , absent external genitalia and gastrointestinal defect or cardiovascular anomalies are usually associated with this syndrome.
The Incidence of sirenomelia is one in 60,000 – 70,000 pregnancies. Our aim to report these cases is to add information to the existing knowledge to emphasize on early screening and termination of pregnancy.

Keywords: Sirenomelia, gestational diabestes, congenital anomaly, malformation

INTRODUCTION :

Sirenomelia, Mermaid syndrome, is a very rare fatal congenital anomaly. It is characterized by complete or partial fusion of lower limbs giving an appearance of human head and upper body with tail of a fish. It is commonly associated with musculoskeletal anomalies, renal agenesis, absent external genitalia and other cardiovascular and gastrointestinal defects. 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.

It has a reported incidence of one in 60,000 – 70,000 pregnancies. It has strong association with maternal diabetes. It has been seen that relative risk is 1:200-250 and upto 22% of fetuses with this anomaly will have diabetic mothers. Maternal drug use and rheumatoid factor are also identified as the causative factors. Antenatal diagnosis of sirenomelia is difficult. It is usually suspected during second trimester with severe oligohydramnios associated with renal dysgenesis or agenesis. The first trimester ultrasound diagnosis of Sirenomelia can also be made and the earliest reported case is in the 9th week of gestation .

Sirenomelia carries with it a very poor prognosis. Survival is largely dependent on the extent of visceral anomalies. So a voluntary termination of pregnancy is advisable in order to avoid the physical and psychological stress to the parents and family.

More than half the cases of sirenomelia result in stillbirth[1]. Only four cases of a surviving infant with sirenomelia have been reported . Here we report a case of termination of pregnancy done on the basis of ultrasound diagnosis of bilateral renal agenesis with no liquor volume.

Case report :

Case 1:

A 24 years old G3A2 at 27 weeks of gestation was admitted for termination of pregnancy because of severe oligohydraminos that was incompatible with life. She had 2 spontaneous abortions at 2 and 3 months of amenorrhoea which were followed by suction and evacuation. In this pregnancy she had an ultrasound examination at 7 weeks gestation which showed no apparent abnormality in the fetus. There were no medical or obstetric risk factors. There was no personal history for diabetes mellitus or congenital anomaly in relatives; except that her father was diabetic and was on oral hypoglycemic. Routine ultrasound examination revealed a live single fetus corresponding to 25 weeks gestation, weighing 852 grams and with absent movements. There were no single measurable pocket of amniotic fluid (severe oligohydramnios). Fetal kidneys, urinary bladder and stomach were not visualized. A large intra abdominal cyst of 5 x 5 cm was noted.Repeat ultrasound was done which showed severe oligohydramnios due to which both lower limbs and lumbar spine could not be visualized properly. Termination was done after proper consent. The procedure was carried out by vaginal misoprostol (E1) 200mg 4 hrly. Patient went into labour after 3 doses and delivered a fetus having fused lower limbs including both feet and compressed fetal head with low set ears. The anal opening was absent

and foetal sex could not be identified. The foetus weighed 800 grams and placental weight was 85 grams. The examination of umbilical cord revealed a single umbilical artery and an umbilical vein. The above findings were consistent with the diagnosis of sirenomelia. The couple declined for autopsy.

Case 2:

A 31 year old G2P1L1 at 24 weeks period of gestation was counseled for termination of pregnancy due to severe oligohydramnios associated with renal agenesis and sacral agenesis. She has a 4 year male child delivered by emergency caesarean section done for non progress of labour. The child is healthy with normal developmental milestones. Her medical history, family history and personal history are not significant.

In this pregnancy patient got her first ultrasound done at 24 weeks of pregnancy which revealed a single live intrauterine pregnancy of 24 weeks gestation (weighing 950 grams) with severe oligohydramnios. Fetal kidneys and urinary bladder was absent. Sacral agenesis was noted.Fetus was crumpled up so the lower limbs could not be visualized properly. A repeat ultrasound revealed similar findings. After proper consent termination was done by mifepristone 600 mg oral followed by misoprostol after 48 hours. Patient went into labour after 2 doses of misoprostol(200 mg 4 hourly) and delivered a male fetus weighing 1 kg with fused lower limb and feet. The anal opening was absent.The examination of umbilical cord revealed a single umbilical artery and an umbilical vein. Radiograph of the fetus showed two femur, two tibia but fibula of each side was fused. The coupled declined for autopsy.

DISCUSSION

Sirenomelia is a rare and lethal congenital anomaly with unknown etiology showing abnormal development of caudal region of the body.This malformation is mostly lethal, survival is very rare in literature. Death usually results from obstructive renal failure due to renal agenesis or dysgenesis, with survival depending on adequate kidney functioning and renal outflow. The etiology of this multisystemic human malformation is unknown and no teratogens have been found in humans. Retinoic acid and heavy metals have been described as important environmental risk factors for caudal malformations. There is a strong association of Sirenomelia with maternal diabetes . Upto 22% of fetuses with this anomaly will have mothers with diabetes[2].

The etiopathogenesis of this syndrome has been subject to a lot of debate over time. Stevenson et al proposed the vascular steal theory. This theory suggests that there is shunting of blood via abnormal abdominal artery arising high up in the aorta. This leaves the caudal part of the embryo poorly perfused. The defective blastogenesis theory regards sirenomelia as part of the caudal regression syndrome (CRS). CRS is hypothesized to arise from primary defect of caudal mesoderm. A teratogenic event during the gastrulation stage i.e. 3rd gestational week, may interfere with the formation of notochord, resulting in abnormal development of caudal structures .

Stocker and Heifetz classified the Sirenomelia sequence into 7 types .

1. All thigh and leg bones are present

2. Single fibula

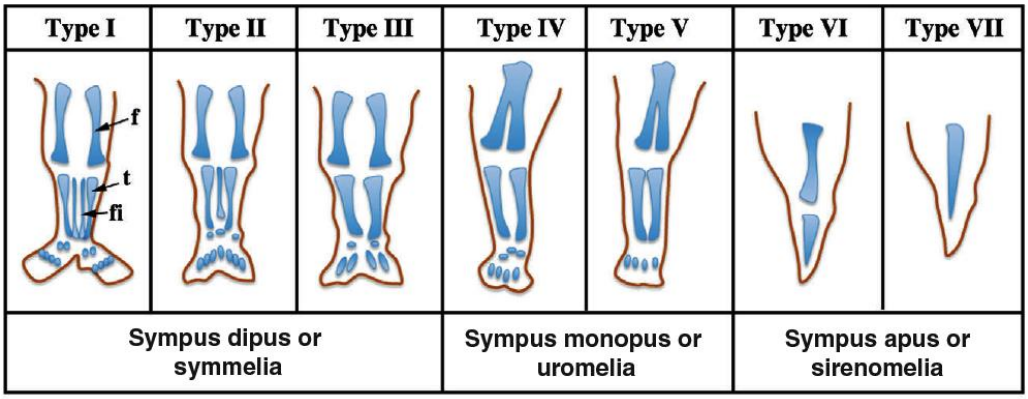
3. Absent fibula

4. Partially fused femurs and fused fibulae

5. Partially fused femurs

6. Single femur and single tibia

7. Single femur, absent tibia



Symelia apus – most common type: feet are absent

Symelia unipus/monopus – presentation of one foot

Symelia dipus – two distinct feet but are malrotated

In case 1 we did not have any radiographs and could therefore not classify our patient into any of these categories. Even though external palpation was in favour of a Type III. In case 2, the radiograph was suggestive of Type III sirenomilia.

Conclusion:

Sirenomelia is a rare and fatal congenital anomaly and has strong association with maternal diabetes. Early prenatal diagnosis by first trimester scan should be the aim to minimise trauma related to termination of pregnancy at advanced gestation. In addition, where possible, a second ultrasound scan should be performed 4-6 weeks after the initial 8-9 weeks scan so that gross structural anamolies are detected and termination of pregnancy be considered earlier.

References:

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Case 1

Case 2