Case Report

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Sirenomelia- A rare congenital anomaly: Case report

Dipanjali Thombare, Prachi Dixit, Ankit Chavan[1], Ashwini Najan

Abstract:

Sirenomelia is an extremely uncommon congenital deformity in which neonate has fused lower limbs associated with various visceral malformations including urogenital and gastrointestinal tract, pulmonary hypoplasia, and potter's facies. The prevalence is 0.8 to 1 per 100,000 newborns. About 300 cases were recorded till date. We describe a case of 31-year G4P1L1A2 with previous lower segment caesarean section with gestational age of 22 weeks 5 days with anomaly scan suggestive of single live intrauterine pregnancy of 21 weeks 5 days with bilateral renal agenesis with placenta previa and no demonstrable amniotic fluid pocket seen. Her pregnancy was uneventful without any medical disorder, drug history, and no congenital anomaly in the family. Termination of pregnancy done by emergency hysterotomy and delivered anomalous foetus with fused lower limb with absent toes, low set ears, absent left ear ostium, imperforate anus, and absent external genitalia. Sirenomelia is fatal congenital anomaly with unclear etiology. Early antenatal diagnosis and termination of pregnancy is the treatment.

Keywords:

Mermaid syndrome, oligohydramnios, renal agenesis, single umbilical artery, sirenomelia

Introduction

Sirenomelia alternatively known as mermaid syndrome which is an exceptionally uncommon and lethal congenital deformity results in varying degrees of lower limb fusion associated with lethal urogenital and gastrointestinal malformations.[1,2] The prevalence of this condition is 0.8-1 in 100,000 neonates. About 300 cases of this lethal congenital malformation have been reported till date. Males are affected more as compared to female in the ratio of 2.7:1.[3] The affected infant has features like mermaid: human-like head and trunk with fish like tail.[4] These are associated with single umbilical artery, severe deformities of the urogenital and lower gastrointestinal system, renal agenesis, and caudal regression syndrome.[5] Most cases die intrauterine or soon after delivery from complications brought on by many visceral abnormalities. Diabetes mellitus have a strong association with this syndrome with relative risk of 1:200-250.

Case Presentation

A 31-year-old G4P1L1A2 with previous lower segment caesarean section had normal antenatal course till gestational age of 22 weeks 5 days when an anomaly scan revealed single intrauterine pregnancy of 21 weeks 5 days with bilateral renal agenesis with placenta previa and no demonstrable amniotic fluid pocket seen. Tiny echogenic focus was noted in left ventricle and a single lower limb of left side was seen. Due to significant oligohydramnios, it was not possible to perform a thorough sonographic evaluation of the right limb and spine. In her past history, she had two first trimester spontaneous abortions in which dilatation and evacuation was done. In her third pregnancy, she delivered full-term healthy female baby of 2.8 kg by lower segment caesarean section for protracted first stage. In present pregnancy, first trimester

Department of Obstetrics and Gynecology, NKPSIMS and RC and LMH, Nagpur, Maharashtra, India, [1]Department of Orthopedics, NKPSIMS and RC and LMH, Nagpur, Maharashtra, India

Address for correspondence:
Dr. Prachi Dixit, Department of Obstetrics and Gynecology, NKPSIMS and RC and LMH, Nagpur, Maharashtra - 440 019, India.
E-mail: drprachidivish@gmail.com

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ultrasound was normal, antenatal screening blood investigations were normal, and no medical illness and drug history. There was no history of consanguinity in her marriage. No family history of congenital anomaly. The condition of current pregnancy was explained to the patient and relatives and patient was admitted for termination of pregnancy. Unfortunately, patient went in spontaneous labour and started fresh bleeding per vaginum. As she was case of previous lower segment caesarean section and low-lying placenta in present pregnancy, emergency hysterotomy was done. A live anomalous foetus was delivered which had early neonatal death within 10 minutes of delivery. Postoperative period was uneventful. The detailed foetal examination revealed anomalies like single fused lower limb with absent toes and looks like a tail, low set ears, absent left ear ostium, imperforate anus, and absent external genitalia [Image 1]. Plan infantogram shown in Image 2 which shows single femur and tibia, missing lower vertebrae and underdeveloped pelvis. Karyotyping and autopsy revealed normal male karyotype with bilateral renal agenesis, malrotation of intestine, and absent urinary bladder.

Discussion

Sirenomelia baby has fused legs giving the appearance of Mermaid's tail. Its likeness to the mermaid of Greek and Roman mythology has given it the name "mermaid baby".[6] Different names for this condition include sympus dipus or symmelia, sympus monopus or uromelia, and sympus apus or sirenomelia. It is a fatal anomaly characterized by fusion of the lower limbs, severe urogenital and lower gastrointestinal system abnormalities, and single umbilical artery. This condition can occur alone or in conjunction with caudal regression syndrome.[7] In our situation, the fetus had all the symptoms of sirenomelia. Males are more often affected than females in the ratio of 2.7:1. This condition can be detected prenatally by ultrasound at 18-20 weeks. In our case, proper prenatal skeletal evaluation was not possible due to severe oligohydramnios.

Etiology is controversial; genetic and environmental factor may play the role. Mostly it occurs sporadically. The karyotype of newborns with mermaid syndrome is frequently normal. Diabetes mellitus shows association with this syndrome with relative risk of 2%.[7] Kallen reported that diabetes could be a factor for this condition which modifies action of one or more teratogens.[8] One of the hypotheses regarding pathogenesis is "vascular steal theory", proposed by Stevenson, Jones, Phelan, in which blood is diverted into the single umbilical vitelline artery from malformed or hypoplastic pelvic arteries. This decreased blood supply distal to the aberrant artery results in abnormal development of caudal part of the embryo; this explains association of renal, genital, and gastrointestinal anomaly.[9] Another hypothesis in caudal body malformation is primary defect in the formation of mesoderm.[7] Sirenomelia is also sometimes associated with potters facies[10] (flat nose, chin recession, low set ears, with absent pinna, the eyes showed hypertelorism, a wide interocular distance, and prominent epicanthal folds). Stoker and Heifetz given classification of sirenomelia which divide it into seven types from type I to type VII.[11] This classification based on the existence of skeletal components in the leg and thigh. In our case, type VI was present [Table 1].

Table 1: Stoker and Heifetz classification of sirenomelia

	Classification
Type I	All thigh and leg bones are present
Type II	Single fibula
Type III	Absent fibula
Type IV	Partially fused femurs, fused fibulae
Type V	Partially fused femurs
Type VI	Single femur, single tibia
Type VII	Single femur, absent tibia

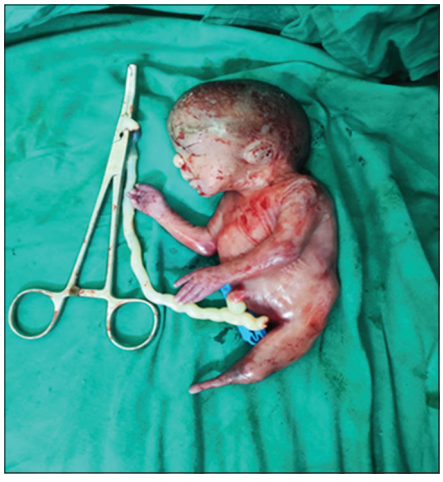

Image 1: Fish tail mermaid appearance

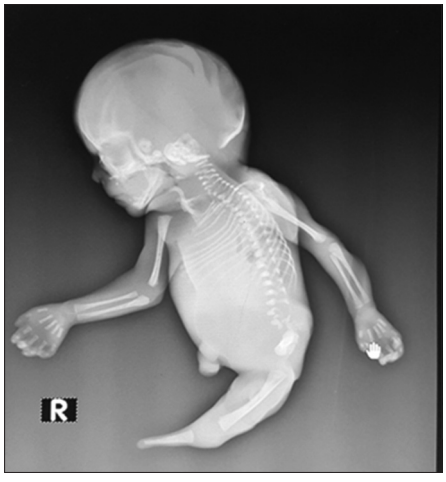

Image 2: Infantogram - single femur and tibia

Conclusion

Serinomelia is rare and fatal congenital anomaly. As the etiology is unclear, early diagnosis and termination, as indicated in the MTP ACT, is targeted. To plan for the termination of the pregnancy, the diagnosis must be made before 20 weeks of gestation. Maintenance of optimal blood glucose level and exposure to teratogen should be prevented to avoid this anomaly. Oligohydramnios with early onset is a warning sign and a thorough early anomaly scan can detect the condition.

Ethical approval

Ethical approval was taken from ethics committee of the institute prior to the study. Ethical code number- ECR/88/Inst/MH/2013/RR/19.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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