

SIRENOMELIA – A CASE REPORT

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ABSTRACT

Mermaid baby also known as sirenomelia is a rare and usually lethal congenital abnormality with multisystem involvement. Characteristic feature consists of varying degrees of lower limb fusion which in severe form results in single lower limb with non-discernible limb bones. We herein report a case of sirenomelia with variable malformation.

Key-Words: Sirenomelia, Mermaid Syndrome

Introduction

Sirenomelia is a rare and usually lethal congenital abnormality with multisystem involvement. The reported incidence is 1 in 100000 pregnancies.^[1] Cases have been reported from all ethnic groups worldwide^[2], and the Male: Female ratio is 2.7:1.^[3] The most prominent feature is partial or complete fusion of lower limbs. This external appearance of the infant resembles the mermaids or sirens of Greek mythology.^[4] We herein report a case of sirenomelia which had various morphological and visceral abnormalities.

Case Reports

Mrs X, 23 years, Primigravida at 31 weeks of gestation according to her last menstrual period presented with complain of pain abdomen since last four hours. There was no personal or family history of diabetes, any congenital malformation or teratogenic drug exposure; there was no history of any chronic medical disorder. Her present pregnancy was poorly followed up in local health centre with only one visit at around 30 weeks of gestation. Ultrasonography done then showed single live fetus at 30 weeks 2 days gestation with severe oligohydramnios (amniotic fluid index= 2 cm), three chambered fetal heart, bilateral hydronephrotic kidneys and dilated ureters. Musculoskeletal system showed normal long bones but digits could not be visualised due to overcrowding of fetal parts. On admission patient was hemodynamically stable. On per abdominal examination, fundal height corresponded to thirty weeks of gestation with cephalic presentation with normal fetal heart rate pattern with mild uterine contractions. On per vaginal examination cervix was 3 centimetres dilated with bulging fetal membranes. She was put on conservative management and was given

betamethasone cover. She gradually went into active labour and delivered a 1250 grams baby with following morphological abnormalities: (i) Swollen face with depressed nasal bridge; (ii) Absent external genitalia with an indistinct tag of tissue; (iii) Absent anal opening; (iv) Absent urethral meatus; (v) Fused lower segment of the body below the pelvis resulting into a single lower limb; (vi) Umbilical cord with single umbilical artery. Baby needed resuscitation after birth with positive pressure ventilation and expired within 5 minutes in spite of resuscitative efforts. Infantogram was done which showed sacral agenesis, hypoplastic pelvis, fused lower extremities with single femur, tibia and fibula. Parents refused autopsy of the baby and post-mortem ultrasonography. Mother got discharged on the second post natal day with an advice to follow up.



Figure-1: Anterior view of baby showing single lower limb and absent external genitalia



Figure-2: Infantogram showing hypoplastic pelvis, single femur, tibia and fibula

Discussion

Sirenomelia is a rare congenital malformative disorder with uncertain aetiology. The clinching diagnostic point at birth is the phenotypic appearance with varying degrees of fused lower extremities. It is more commonly seen in monozygotic twins as compared to dizygotic twins and singletons, the reported incidence being 100-150 times higher.^[5] It is usually associated with other visceral defects such as hypoplastic lungs, cardiac agenesis, absent genitalia, digestive defects, absent kidney and bladder, vertebral and central nervous system defects.^[6,7] Most babies with sirenomelia have single umbilical artery as seen in our case.^[8] Stocker and Heifetz classified the sirenomelia sequence into 7 types^[9]:

Table-1: Stocker and Heifetz classification of sirenomelia

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

The exact cause is still unknown. Stevenson et al.^[8] proposed a vascular steal hypothesis to explain the defects associated with sirenomelic fetus. It proposed that the single umbilical artery of vitelline origin, which originates high in the abdomen, diverts blood flow towards placenta. This leaves the lower body part poorly perfused with inadequate nutrient supply, which in turn results in agenesis of midline structures and subsequent abnormal approximation of lower limb fields.

Another well-known theory proposes that sirenomelia is a primary defect of blastogenesis that occurs during the final stages of gastrulation at the tail bud stage, corresponding to the third gestational week in human.^[10] Experimental data suggests that sirenomelia has a genetic basis resulting from a defect in retinoic acid or bone morphogenetic protein signalling in the caudal embryonic region.^[11,12] Maternal diabetes mellitus^[13] and heavy metal exposure^[14] have also been linked to it.

Survival basically depends on the degree and type of visceral abnormalities specially the presence of a functioning kidney. The diagnosis, quite obvious at birth can also be established by antenatal ultrasonography. Sonography clues include oligohydramnios, renal agenesis and lower limb abnormalities.

In our case, the mother had no associated risk factor but

due to the poor antenatal follow up, antenatal diagnosis could not be made on time but based on ultrasonography, the parents were counselled regarding the congenital anomaly in child.

Conclusion

Sirenomelia is usually incompatible with life depending on the underlying visceral abnormalities while the phenotype does not affect survival. With advances in reconstructive surgery, minor phenotypic defects can be corrected. Early antenatal diagnosis by anomaly scan can prevent the trauma of advanced termination of pregnancy or stillbirth.

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