Rare Case Report of Sirenomelia: The Mermaid Baby

Sonal Prasad[1], J Anupama[2]

ABSTRACT

Introduction: Sirenomelia or MERMAID BABY is rare syndrome characterized by fusion of lower limbs, urogenital and gastrointestinal tract malformations, Potter's facies and pulmonary hypoplasia. The incidence is 0.8-1 case/100,000 births with male to female ratio being 3:1.

Case report: A 30 year old G2P1L1 with 34 weeks of gestational age with previous one vaginal delivery was admitted in labor room in second stage of labor. She was from poor socioeconomic background with no antenatal care or investigations except ultrasound indicating intrauterine fetus of 25 weeks with severe oligohydraminos (AFI 3). There was history of tobacco intake. Her pregnancy was uneventful with no history of medical disorder in mother or congenital anomaly in family. Husband was 42 years old. A 1.5 kg baby with single fused lower limbs was delivered. The infant had Potter's facies, narrow chest, fused lower limbs with a single femur and an incomplete tibia with no foot. Tibia flexed anteriorly. External genitalia and anal opening were absent, and umbilical cord had single umbilical artery. The baby died within 45 min postbirth. X-rays revealed lower limbs were fused completely into single limb and one femur and one tibia were seen. Ultrasound abdomen showed absence of both kidneys, urinary bladder and stomach. Normal four chambered heart was seen with patent ductus arteiosus. Autopsy was declined by the parents. Intrapartum and the postpartum period of mother was uneventful.

Conclusion: Sirenomelia is a rare and lethal congenital anomaly. Our case belonged to Type VI of Stocker and Heifetz Sirenomeliac infants classification.

Keywords: Sirenomelia, Mermaid Baby, Potter's Facies

INTRODUCTION

Sirenomelia was first described by Rocheus in 1542 and Palfyn in 1553 and called after the mythical Greek sirens.[1] This rare syndrome has the incidence of 0.8-1 case/100,000 births with male to female ratio being 3:1[2] and is characterized by fusion of lower limbs, single umbilical artery, severe malformation of urogenital and lower gastrointestinal tract. Resultant oligohydroamnios, leads to these infants having Potter's facies and pulmonary hypoplasia. 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.

This syndrome has strong association with maternal Diabetes mellitus with relative risk 1:200-250 and 22% of foetuses will have diabetic mother.[3] Intake of haloperidol and tobacco antenatally is considered a cause for Sirenomelia. Other proposed teratogens are cadmium, lead, vitamin A. Nutritional deficit, vascular hypoperfusion has also been proposed as a possible etiological factors.

CASE REPORT

A 30 year old G2P1L1 with 34 weeks 5 days of gestational age with previous one vaginal delivery was admitted in labor room in second stage of labor. She was from poor socioeconomic background with no antenatal care or investigations except ultrasound done at 25 weeks of gestation indicating single live intrauterine fetus of 25 weeks with severe oligohydraminos (AFI 3).No comment was made on the presence/absence of malformations. There was history of tobacco intake before and during pregnancy. Her pregnancy was otherwise uneventful and there was no history of diabetes, hypertension, thyroid disorder or fever in first trimester. Husband was 42 years old. There was no history of genetic or congenital anomaly in the family.

She delivered a 1.5 kg baby with single fused lower limbs and multiple congenital anomalies as shown in figure 1. The Apgar score was 3 at 1' and 0 at 5 min. On physical examination, the infant showed narrow chest indicating lung hypoplasia, fused lower limbs with a single femur and an incomplete tibia with no foot. Tibia was flexing anteriorly. External genitalia were absent, there was no anal opening and umbilical cord had single umbilical artery [Figure 1]. There were also prominent epicanthal folds, hypertelorism, downward curved nose, receding chin, low-set soft dysplastic ears and small slit-like mouth suggestive of Potter's facies [Figure 2]. The baby died within 45 min post birth. X-rays revealed lower limbs were fused completely into single limb and one femur and one tibia were seen [Figure 3]. Ultrasound abdomen showed bilateral agenesis of kidney and urinary bladder along with absence of stomach. Four chambered view of heart was seen with patent ductus arteiosus. Autopsy was declined by the parents. Intrapartum and the postpartum period of mother was uneventful.

DISCUSSION

In literature approximately 300 cases of sirenomielia are reported worldwide of which 14 are from India.[4] Though described as a rare lethal anomaly, 9 mermaid syndrome

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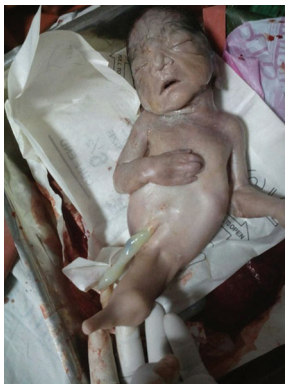

Figure-1: Showing the complete picture of the mermaid baby with fused legs and absence of genitalia

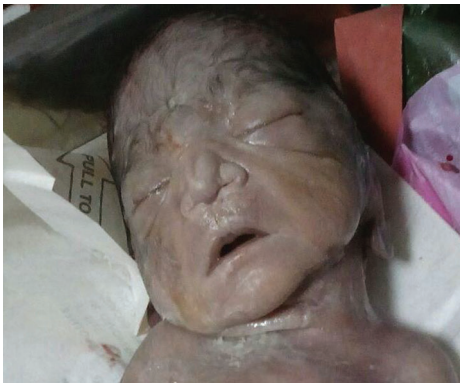

Figure-2: Showing Potter Facies

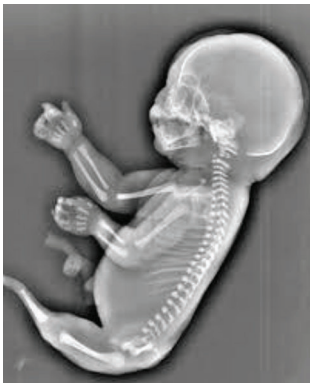

Figure-3: Showing X ray of the delivered baby

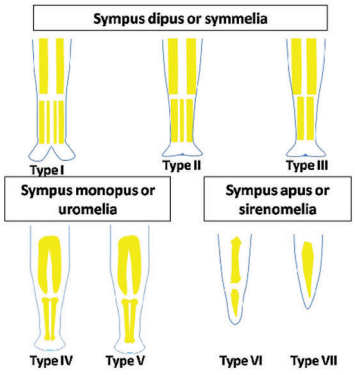

Figure-4: Stocker and Heifetz classification of Sirenomeliac infants[10]

cases have been reported surviving after reconstructive surgery.[5] The most important characteristics of survival of the affected newborn is the presence of functional kidney. In most of the cases the diagnosis was performed after birth. In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or color Doppler sonography.[6] Oligohydraminos acts as an alerting sign and diagnosis is done by the presence of fused femur and decreased distance between two femurs and decreased or absent mobility of two lower limbs along with associated with skeletal and lumbar spine deformities. In addition bilateral renal agenesis, heart and abdominal wall defects are seen on scan.[7]

The primary molecular defect resulting in sirenomelia remains unclear. However, two main pathogenic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. Vascular steal hypothesis states that fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields.[8] According to defective blastogenesis hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage. Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures.[9] Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors. In our case, there was history of tobacco use before and during pregnancy.

Stocker and Heifetz classified Sirenomeliac infants from Type I to Type VII [Figure 4] according to the presence or absence of bones within the lower limb.[10] Our case belonged to Type VI of this classification.

CONCLUSION

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Early diagnosis and termination are targeted. Optimum maternal blood glucose level in preconceptional period and in first trimester along with prevention from exposure to teratogenic drugs should be maintained to prevent this anomaly. In addition a second ultrasound should be performed after 4-6 weeks after the initial first trimester (8-9 weeks) scan. Early onset oligohydramnios is an alerting sign and detailed anolmaly scan can pick the condition up so that termination of pregnancy can be planned.

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