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Case Report

Sirenomelia (mermaid baby)

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ABSTRACT

Sirenomelia is a rare congenital malformation wherein the two lower limbs are fused to give an appearance of a mermaid. It is almost always lethal and is associated with several other defects like anorectal malformations, renal agenesis, umbilical cord vessel abnormalities and gastrointestinal defects. Reporting this case of sirenomelia from our institution in a preterm baby born alive to a primigravida at 36 weeks of gestation, who presented in labour.

Keywords: Sirenomelia, Congenital malformations, Mermaid baby

INTRODUCTION

Sirenomelia was originally described in the 15th century with its name derived from the Greek mythological creatures, "sirens" which had a single lower limb resembling a fish tail. This lethal malformation is reported to have an incidence of about 0.8 to 1 in 1,00,000 live births.[1] It is more common among male fetuses with a sex ratio of 3:1.[2] It is found to be 100 times more commoner in twins than singleton pregnancies. The subsequent pregnancies have no increased risk as the disorder is sporadic. Maternal diabetes has a strong association with sirenomelia. It has various anatomical variants as described by Stocker and Heifetz (7 subtypes).

CASE REPORT

A 23-year-old primigravida belonging to upper middle class was first admitted at our institution at 36 weeks in view of preterm labour. She had a non-consanguineous marriage. Her first trimester was uneventful with no known teratogenic exposure or febrile illness. The mother had no co morbidities. An antenatal ultrasound done at 26 weeks of gestation revealed anhydramnios and fetal parts were not clearly visualized. Examination of abdomen revealed less liquor and a growth restricted fetus with a symphysiofundal height of 32 cm and a normal fetal heart rate. An ultrasound was repeated to confirm the findings and labour was augmented with oxytocin. She delivered a late preterm baby vaginally. Baby did not cry at birth. There were minimal respiratory efforts after positive pressure ventilation. Examination of the baby revealed a single fused lower limb with a single bone and no feet. There was no genitalia or anal opening (Figure 1). No facial abnormalities were noted. No associated umbilical cord abnormalities were detected. The baby was shifted to the nursery and was supportive care was given. The baby developed bradycardia and hypoxia and had a cardiac arrest after six hours of birth. CPR was initiated but the baby could not be revived. Autopsy could not be done as parents were not consenting.

DISCUSSION

Sirenomelia is the most severe form of congenital limb defects. Various etiological mechanisms have been proposed for this malformation. Carlos et al. in their study mention that it may be idiopathic or there may be an underlying genetic basis. It has been demonstrated in mice that deficiency of an enzyme (cyp26a1) that degrades retinoic acid or defective bone metalloproteinase is responsible for this disorder. The

other mechanisms suggested are primary mesodermal defect (defective blastogenesis) and abnormal vascularity of the caudal region (vascular steal hypothesis).[3]

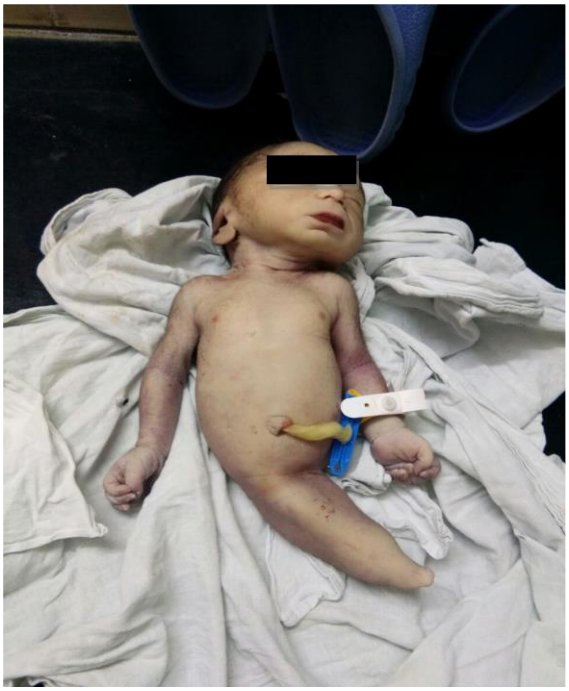

Figure 1: Neonate showing features of sirenomelia.

The incidence is as high as 1 in 350 among babies born to mothers with diabetes. The risk is 200 times more when compared to general population.[4] Most of the babies with sirenomelia were stillborn or end up in early neonatal demise. An important factor that decides survival is the functioning of kidney in such neonates.[5-8] Diagnosis can be established easily in first trimester and early second trimester by ultrasound showing a rudimentary single lower limb as the amniotic fluid volume may be adequate. Colour doppler can be used to demonstrate single large umbilical artery and absence of renal arteries. It is almost invariably associated with abnormalities in the kidneys like bilateral renal agenesis or multicystic dysplastic kidneys. This makes the diagnosis difficult in late second trimester due to associated oligohydramnios. Rarely infants with minor renal abnormalities or normal kidneys have been reported.[5] The baby born with sirenomelia usually has potters facies characterised by hypertelorism, low set ears, flat nose and a receding chin. The case described here did not have these characteristic facies. The mother had not been screened for diabetes as she presented to us in labour. Other anomalies commonly associated could be lumbosacral malformations and anorectal malformations.[6] There have been reported cases of sirenomelia associated with complete anterior abdominal wall agenesis.[7]

CONCLUSION

Here we have brought out the etiopathogenesis, clinical features, and ultrasonographic features of sirenomelia from the relevant literature. Thus, the aim is to make an early diagnosis of these lethal malformations and offer legal termination to avoid psychological trauma.

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