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SIRENOMELIA: A RARE CASE REPORT

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ABSTRACT Sirenomelia is a rare congenital fetal anomaly , also known as mermaid syndrome with characteristic feature of complete or partial fusion of lower limbs (1). In this syndrome, fetus dies in utero or survive for few hours after birth as this anomaly is incompatible with life. In this case report ,we have 19 yr old primigravida with h\o chewing ghutka (tobacco) since her teenage which she continued to have till her first trimester of pregnancy and resulted in delievering baby with mermaid syndrome .

KEYWORDS : mermaid Syndrome, Potterfacies ,caudal Regression Syndrome ,sirenomelia

INTRODUCTION

Sirenomelia is a rare congenital anomaly with incidence of 0.8 -1 case per 1 lakh birth . the incidence of this syndrome is more in male as compared to female 3:1 (2). Around 300 cases has been noted till now.It has been found that it is associated with maternal diabetes seen in 22% of babies,in monozygotic twins (3) It is also found in mothers exposed to cocaine , tobacco and drugs like sodium valproate ,phenytoin and lithium. (4)

CASE REPORT

A 19 yrs old female came in casualty as referred case of primigravida with 36 weeks of gestation with severe oligohydraminos. On retrospective history ,pt was married since 10 months and conceived spontaneously . Pt was a booked case at subcentre with 5 antenatal visits with history of receiving 5 doses of iron sucrose .pt had history of chewing ghutka (tobacco) since her teenage ,there is no history of intake of any drugs ,no history of radiation exposure during pregnancy .no previous history of any disease or similar family history . On reviewing her usg scan , her first scan done at 19 weeks of gestation was normal, no anomaly has been noted in 28 weeks and 36 weeks scan. Pt was taken for emergency cesarean section i\v\o severe oligohydraminos with breech with fetal distress in labor . A preterm anomalous baby delievered out with birth weight 2.6 kg having anomalous skull with increased A-P diameter , prominent occiput , low set ears , flattened nasal bridge (potter facies),umblical cord consisting of single umblical artery (2 vessel cord), sex could not be determined, compleyte lower limb was fused and anal opening was not present . Baby had weak cry after birth and was intubated but could not be revived back and died after half n hour.

Figure 1 Showing fusion of both lower limbs

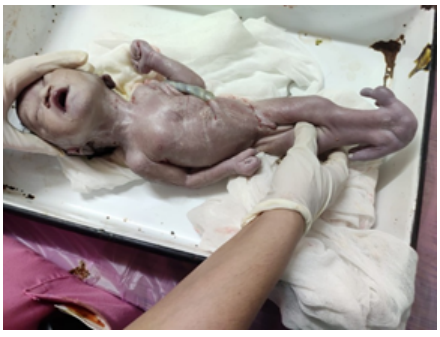

Figure 2 Sirenomelia baby with Potter's facies and ambiguous genitalia with fused lower limbs

Figure3 Showing Mermaid Baby

DISCUSSION

Sirenomelia is a lethal congenital anamoly and is the most severe form of cordal regression syndrome (5). It has the features of fusion of lower limb, single umbiical artery, and any visceral anomalies. There are two hypothesis for explaining the cause of sirenomelia, first is vascular steal hypothesis which states that fusion of the lower limb occured due to deficient blood flow to the caudal mesoderm resulting in agenesis of midline structures and fusion of the lower limbs (6). Second is defective blastogenesis hypothesis in which there is defect in development of caudal mesoderm due to teratogenic event during gastrulation stage (7). The risk factor associated with sirenomelia is maternal dibetes, tobacco use, heavy metal exposure, intake of drugs like phenytoin and lithium. This anomaly can be diagnosed as early as thirteen weeks by high resolution USG and during subsequent anamoly scan (8) (9). The facial abnormality in sirenomelia is

known as potter's facies having features of large, low set ears, prominent epicanthic folds, hypertelorism, flat nose and receding chin. When potters facies are present along oligohydramnios and pulmonary hypoplasia, it is known as Potter's syndrome (10).

However, genetic defects in humans are still unknown in thesirenomelia, two defects in the Cyp26a1 and BMP7 genes in mice result in the birth of a mermaid neonate. The Cyp26a1 gene is responsible for coding the enzyme that breaks down retinoic acid (the metabolite of vitamin A). Retinoic acid temporarily increases the vasculature in the caudal region of the embryo. Disruption of the Cyp26a1 gene and incomplete development of the caudal region of the embryo result in a mermaid syndrome in mice. Bone morphogenic protein7) is an important protein that plays an important role in angiogenesis in vitro. By stimulating endothelial cells of the caudal region, vascular and tissue production leads to normal growth of the lower limbs in the fetus (11) (12) (13). Stocker and Heifetz classified Sirenomeliac infants from Type I to Type VII according to the presence or absence of bones within the lower limb (14).

CONCLUSIONS

Sirenomelia is a lethal disorder. If we can diagnose this syndrome during antenatal period, patient should be adviced termination of the pregnancy. This can be prevented by regular anenatal checkups and by taking proper history of patient regarding association with risk factors. Blood sugar levels in the antenatal visits should be done and maintained in optimum level. Anomaly scan should be done. As it is said " Prevention is better than cure".

REFERENCES:

1. Sirenomelia. Pathological features, antenatal ultrasonographic clues, and a review of current embryogenic theories. Valenzano M, Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. s.l. : Hum Reprod Update, Vols. 1999;5:82–6.
2. Reddy KR, Srinivas S, Kumar S, Reddy S, Hariprasad Irfan GM. Sirenomelia a rare presentation. J Neonatal Surg. 2012;1:7. [PMC free article] [PubMed] [Google Scholar].
3. Aslan H, Yanik H, Celikaslan N, Yildirim G, Ceylan Y. Prenatal diagnosis of Caudal regression syndrome: A case report. BMC Pregnancy Childbirth. 2001 and Scholar], 1:8. [PMC free article] [PubMed] [Google.
4. Naveena S, Mrudula C. Sirenomelia — The mermaid syndrome: A case report. IOSR J Dent Med Sci. 2013 and Scholar], 7:01–4. [Google.
5. Duhamel B. From the mermaid to anal imperforation: the syndrome of caudal regression. Arch Dis Child. 1961;36(186):152-5
6. Sadler TW, Rasmussen SA. Examining the evidence for vascular pathogenesis of selected birth defects. Am J Med Genet A. 2010 and 152A:2426-36.
7. Duesterhoeft SM, Ernst LM, Siebert JR, Kapur RP. Five cases of caudal regression with an aberrant abdominal umbilical artery: Further support for a caudal regression sirenomelia spectrum. Am J Med. 2007.
8. Vijayaraghavan SB, Amudha AP. High-resolution sonographic diagnosis of sirenomelia. J Ultrasound Med. 2006 and Scholar], 25:555–7. [PubMed] [Google.
9. Sahu L, Singh S, Gandhi G, Agarwal K. Sirenomelia: A case report with literature review. Int J Reprod Contracept Obstet Gynecol. 2013 and Scholar], 2:430–2. [Google.
10. Dharmraj M, Gaur S. Sirenomelia: a rare case of foetal congenital anomaly. J Clin Neonatol. 2012.
11. Zakin L, Reversade B, Kuroda H, Lyons K M, De Robertis E M. Sirenomelia in Bmp7 and Tsg compound mutant mice: requirement for Bmp signaling in the development of ventral posterior mesoderm. Development. 2005 and Scholar], 132(10):2489–2499. [PubMed] [Google.
12. Ribes V, Fraulob V, Petkovich M, Dollé P. The oxidizing enzyme CYP26a1 tightly regulates the availability of retinoic acid in the gastrulating mouse embryo to ensure proper head development and vasculogenesis. Dev Dyn. 2007 and [Goog, 236(03):644–653. [PubMed].
13. Sheng N, Xie Z, Wang C et al.Retinoic acid regulates bone morphogenic protein signal duration by promoting the degradation of phosphorylated Smad1. Proc Natl Acad Sci U S A. 2010 and Scholar], 107(44):18886–18891. [PMC free article] [PubMed] [Google.
14. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol. 1987 and Scholar], 10:7–50. [PubMed] [Google].