Case Report: Sirenomelia (Mermaid Syndrome)

Kaushik Mahajan[1], RK Praneshwari Devi[2], Leimapokpam Roshan Singh,[3] Kabita Athokpam[4]

ABSTRACT

Introduction: Sirenomelia (Mermaid syndrome) is a rare congenital malformation consists of varying degrees of lower limb fusion, appears like a mermaid's tail. Sirenomelia has an incidence of 0.8 to 1 case per 1,00,000 births.This is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function.

Case report: A 19 years old mother, unbooked, G_2P_{0010} presented to RIMS Emergency labour room at 34 days 4 days gestations with history of discharge per vagina and pain abdomen. There was only one antenatal checkup at primary health center and no ultrasound done during her antenatal period. There was no significant medical history, no history of tobacco intake before and during pregnancy and there was no history of diabetes, hypertension, thyroid disorder or fever in first trimester. Husband was 29 years old. There was no history of genetic or congenital anomaly in the family. Fundal height corresponds to 26-28 week size and liquor appeared clinically reduced. She went into labour spontaneously and delivered a 1.5 kg a single live baby with normal upper part of the body, deformed pelvic bones with fused lower limbs, absent external genitalia and imperforate anus. Baby was resuscitated by paediatrician, but baby could not be revived and was declared dead two hours later. Patient party was advised karyotyping of neonate but they denied. Mother's postpartum period was uneventful and was discharged after 48 hours with advice of birth spacing of minimum 1 year, mandatory antenatal visits, follow-up and peri-conceptional folic acid intake.

Conclusion: Sirenomelia is a rare congenital anomaly.High risk pregnencies should be identified. A targeted level scan should be done at 18-20 weeks and to be repeated at 24 weeks to exclude anomalies.More emphasis should be given on proper prenatal diagnosis and care with a possible termination of pregnancy proposed as an option if detected early.

Keywords: Sirenomelia, Mermaid Syndrome, Preterm Labour, Neonatal Death

INTRODUCTION

Sirenomelia (Mermaid Syndrome) is a rare and lethal multisystemic human congenital malformation. It is characterised by malformation of lower limbs as complete or partial fusion of lower limbs into a single lower limb which gives an appearance of a mermaid's tail.[1] Sirenomelia has an incidence of 0.8 to 1 case per 1,00,000 births.[2] Sirenomeliac baby can not survive more than a day or two of birth as it is associated with abnormal kidney and urinary bladder development and function.[3] This syndrome has strong association with maternal Diabetes mellitus with relative risk 1: 200-250 and 22% of foetuses will have diabetic mother.[4] It invariably presents with lower limb fusion, sacral and pelvic bony anomalies, absent external genitalia, and renal agenesis or dysgenesis.[5] Most of the cases of sirenomelia leads to stillbirth and this condition found 100 times more commonly found in identical twins than in single births or fraternal twins.[6]

CASE REPORT

A 19 years old mother, unbooked, G_2P_{0010} presented to RIMS Emergency labour room at 34 days 4 days gestations with history of discharge per vagina and pain abdomen. There was only one antenatal checkup at primary health center and no ultrasound done during her antenatal period. There was no significant medical history, no history of tobacco intake before and during pregnancy and there was no history of diabetes, hypertension, thyroid disorder or fever in first trimester. Husband was 29 years old. No previous history of genetic or congenital anomaly were found in the family. Fundal height corresponds to 26-28 week size and liquor

Figure-1: Showing the complete picture of the Sirenomeliac baby (Mermaid baby).

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Figure-2: Picture showing fused limbs and absent genetalia of the Mermaid baby.

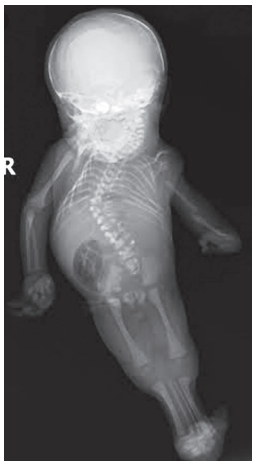

Figure-3: Showing Xray of the delivered baby.

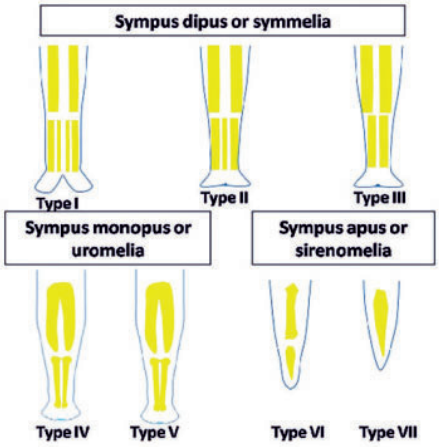

Figure-4: Showing Stocker and Heifetz classification of sirenomeliac baby.[12]

appeared clinically reduced. Labour was spontaneously progressed and she delivered a 1.5 kg a single live baby with normal upper part of the body, deformed pelvic bones with fused lower limbs, absent external genitalia and imperforate anus. Baby was resuscitated by paediatrician, but baby could not be revived and was declared dead two hours later. Patient party was advised karyotyping of neonate but they denied. Mother's postpartum period was uneventful and was discharged after 48 hours with advice of birth spacing of minimum 1 year, mandatory antenatal visits, follow-up and peri-conceptional folic acid intake.

DISCUSSION

In the way back in the sixteenth century Rocheus and Polfyr first medically described Sirenomelia. In 1961 Duhamel described Sirenomelia is the most severe form of caudal regression syndrome and is incompatible with life. Around 300 cases reported in the world literature, of which 13 have been from India.[7] Most of them were diagnosed after birth and no definite antenatal diagnosis was possible in all of these cases.[6] In antenatal period, Sirenomelia can be diagnosed as early as by using high resolution or colour Doppler sonography.[8] Oligohydromnios 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 lumber spine deformities. In addition bilateral renal agenesis, heart and abdominal wall defects are seen on scan.[4] Sirenomelia does not recur in families and no chromosomal abnormalities are observed.[7]

Primary molecular defect causing sirenomelia not clear but two main pathogenic hypothesis i.e the vascular steal hypothesis and the blastogenesis hypothesis has been explained. Vascular steal hypothesis explains that due to deficient blood flow and nutrient supply to the caudal mesoderm, limbs get fused and also results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields.[9] Whereas according to defective blastogenesis hypothesis, during the gastrulation stage the primary defect in development of caudal mesoderm resulting in a teratogenic event. This type of defect impede with the formation of notochord and causes abnormal development of caudal structures.[10] Maternal diabetes,tobacco use,retinoic acid and heavy metal exposure are possible environmental factors.[11] But in our case there was no such history of Maternal diabetes,tobacco use,retinoic acid and heavy metal exposure before and during pregnancy.

According to the presence or absence of bones in lower limb Stocker and Heifetz classified Sirenomeliac infants from Type I to Type VII [Figure 4].[12] Our case belonged to Type 4 of this classification.

CONCLUSION

High risk pregnencies should be identified. A targeted level scan should be done at 18-20 weeks and to be repeated at 24 weeks to exclude anomalies. Early diagnosis and termination should be done. Patients present with oligohydramnios along with fetal growth retardation either with or without history of leaking, we have to look for fetal kidneys and fetal bladder along with fetal vasculature. Sirenomelia associated with severe visceral anomalies has poor survival rate. Sirenomelia only associated with the fusion of lower limbs is less fatal. A very small portion of surviving sirenomeliac baby require a multidisciplinary approach of treatment. More emphasis should be given on proper prenatal diagnosis and care with

a possible termination of pregnancy proposed as an option if detected early.

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REFERENCES

1. Garrido-Allepuz C, Haro E, Gonzalez-Lamuno D, Martinez –Frias ML, Bertocchini RMA. A Clinical and experimental overview of Sirenomelia: insight into the mechanisms of congenital limb malformations. Dis Models Mech.2011;4:289-99
2. Kallen B, Castilla EE, Lancaster PA, Mutchinick O, Knudsen LB, Martinez-Frias ML, Mastroiacovo P, Robert E: The cyclops and the mermaid: an epidemiological study of two types of rare malformation. J Med Genet 1992;29:30-35.
3. Taori KB, Mitra K, Ghonga NP, Gandhi RO, Mammen T, Sahu J. Sirenomelia sequence (mermaid): report of three cases. Indian J Radiol Imaging 2002;12:399- 401.
4. Aslan H,Yanik H, Celikaslan N, Yildirim G, Ceylan Y. Prenatal diagnosis Caudal Regression Syndrome: a case report. BMC Pregnancy and Childbirth 2001;1:8.
5. Assimakopoulos E, Athanasiadis A, Zafrakas M, Dragoumis K, Bontis J. Caudal regression syndrome and sirenomelia in only one twin in two diabetic pregnancies. Clin Exp Obstet Gynecol 2004;31:151-3.
6. Sammons, Mary Beth (November 26, 2009).10 year old girl born with legs fused together AOL. Archived from the original on November 26, 2009.
7. Duhamel B. From the Mermaid to Anal Imperforation: The Syndrome of Caudal Regression. Arch Dis Child 1961;36:152-5.
8. Vijayaraghavan SB, Amudha AP. High-resolution sonographic diagnosis of sirenomelia. J Ultrasound Med.2006;25:555-7.
9. Sadler TW, Rasmussen SA. Examining the evidence for vascular pathogenesis of selected birth defects. Am J Med Genet A. 2010;152A:2426-36.
10. 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 Genet A. 2007;143A:3175–84.
11. Naveena S, Mrudula C. Sirenomelia – The mermaid syndrome: A rare case report. IOSR J Dent Med Sci. 2013;7:01-4.
12. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol. 1987;10:7–50.

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