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Case Report

Sirenomelia - Mermaid syndrome with oesophageal atresia: A rare case report

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ABSTRACT

Sirenomelia or Mermaid syndrome is a very rare congenital deformity in which legs are fused together and commonly associated with abnormal kidney development, genital, and rectal abnormalities. In this present case, sirenomelia was associated with oesophageal atresia, which is a rare association and occurs in about 20-35% of cases.

KEY WORDS: Potter's syndrome, oesophageal atresia, sirenomelia

INTRODUCTION

Sirenomelia is a very rare congenital deformity found in approximately 1 out of 1,00,000 live births. More than half of cases result in stillbirth [1]. It presents with lower limb fusion, sacral, and pelvic bone anomalies. Prognosis is very poor because of the variable major anomalies, including renal agenesis, sacral agenesis, and imperforate anus [2]. Another rare association is oesophageal atresia [3]. There is strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of fetuses with this anomaly will have diabetic mothers [4,5]. Sirenomelia predominantly affects males and is 100 times more likely to occur in identical twins [6-8]. More than half of the cases result in stillbirth and those born alive usually die within a day or two of birth. Only a few cases with sirenomelia have been reported to have survived beyond the neonatal period [9-11]. About 300 cases have been reported in the world literature, so far of which eight have been reported in India [12].

CASE REPORT

A primigravida delivered a stillborn term baby, who had not undergone any antenatal checkup. We received an infant weighing 1900 g with crown-heel length 45 cm, head circumference of 32 cm, chest circumference 31 cm, abdominal circumference 27 cm, upper segment measuring 28 cm, and lower segment 17 cm. Examination of the infant revealed; the face showed all the features described by Potter - flattening of nose, recession of chin, soft flat ears with apparent absence of the cartilage and low position of the ears, the eyes showed hypertelorism (a wide inter-ocular distance), and prominent epicanthal folds [Figure 1]. Spine was normal. The right upper limb was normal and there was the absence of metacarpal bone in the left thumb. Lower limb was fused from pubis to foot and feet showed nine toes. External genitalia could not be made out [Figure 2]. There was no anal opening. The umbilical cord measuring 7 cm × 1 cm, with presence of only 2 vessels, 1 artery, and 1 vein.

An X-ray of the infant revealed hemivertebra, sacral agenesis, the absence of the metacarpal bone on left thumb, absence of fibula on left side [Figure 3].

Internal examination revealed presence of prominent blind loop of the large intestine [Figure 4], oesophageal atresia [Figure 5], the absence of kidney and urinary bladder. Other organs were within normal limits. Small gonads were found.

Microscopic examination confirmed the presence of one umbilical artery and one umbilical vein. Section from gonads

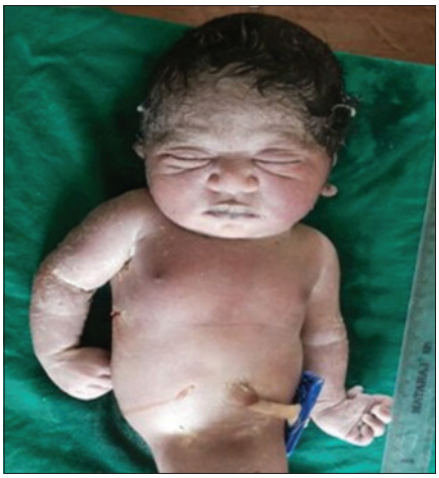

Figure 1: Potter's facies - flat nose, recession of chin, soft flat ears with apparent absence of the cartilage and low position of the ears, hypertelorism (a wide inter-ocular distance), and prominent epicanthal folds.

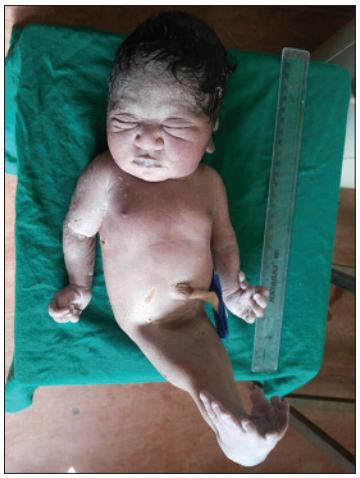

Figure 2: Mermaid syndrome with fused lower limb, absence of external genitalia.

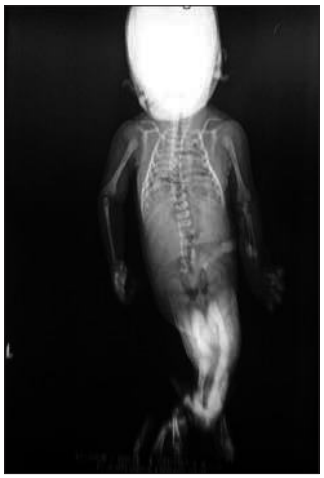

Figure 3: X-ray showing hemi vertebra, sacral agenesis, absence of metacarpal bone on left thumb, absence of fi bula on left side.

showed structure of ovary and fallopian tube. Sections from other organs were within normal histological limits.

Figure 4: Prominent blind loop of the large intestine with absence of rectum (arrow), normal liver, normal lung, and normal heart.

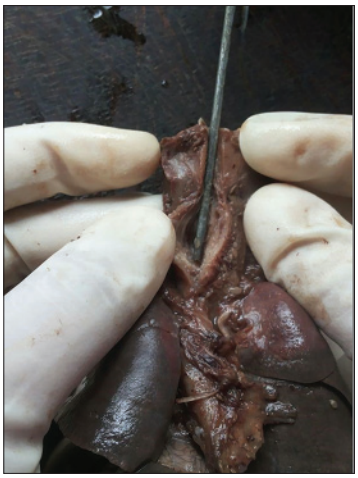

Figure 5: Oesophageal atresia.

DISCUSSION

Sirenomelia also known as mermaid syndrome is a rare and lethal congenital anomaly. The cause of sirenomelia remains unclear, however, maternal diabetes mellitus, genetic predisposition, environmental factors, and vascular steal phenomenon with the single vitteline umbilical artery diverting blood supply and nutrients from the lower body and limbs have been proposed as possible causative factors [9,10]. A normal fetus has two umbilical arteries, which pump blood from the fetus to the placenta, and one umbilical vein, which returns blood from the placenta to the fetus. In the pelvis, the umbilical arteries branch off the iliac arteries, which supply the legs and pelvic organs, such as genitalia. The "vascular steal theory" indicates that a single large artery assumes the function of the umbilical arteries, thus diverting blood from the caudal portion of the embryo to the placenta [10,11].

Sirenomelia was formerly thought to be an extreme form of the caudal regression syndrome, however, it is reclassified to be considered a separate condition. The spectrum of disease ranges from fusion of lower extremities, bilateral renal agenesis,

absence of sacrum, hemivertebrae, absence of rectum, and bladder [12,13]. The presence of two umbilical arteries, non-lethal renal anomalies, non-fused lower limbs, abdominal wall defects, and abnormalities of tracheo-oesophageal tree, neural tube, and heart differentiates caudal regression syndrome from sirenomelia [3]. Confusion still exists on whether sirenomelia is a severe form of vertebral defects, anorectal atresia, cardiac abnormalities, tracheo-oesophageal fistula, renal, and limb abnormalities (VACTERL) anomalies [12,13].

There is strong association of sirenomelia and caudal regression syndrome with maternal diabetes. The disorder occurs in about one in 350 infants of diabetic mothers, representing an increase of about 200-fold over the rate seen in the general population [Table 1] [2-4].

Sirenomelia sequence is classified into three groups according to the number of feet present. Sympus apus, in which both legs are merged completely into a single lower extremity. In this condition both feet is absent or rudimentary. Symelia unipus shows a presentation of one foot, two femur, tibia, and fibula. Symelia dipus shows two distinct feet are present but are malrotated and resemble fins [2].

Sirenomelia predominantly affects males (sex ratio 2.7:1) and is frequent among one of the two monozygotic twins [1,5,6,8,11]. In this study infant sex was confirmed microscopically as female, as the normal structures of the ovary and fallopian tube were identified from the sections of gonads. Oesophageal atresia is a rare association seen with sirenomelia, which is seen in this case. It occurs in about 25-30% of the cases [3]. Sirenomelia is fatal because of renal agenesis and associated severe pulmonary hypoplasia [1,2,5,10].

Potter's syndrome, which consists of (1) Potter's facies (large, low-set ears, prominent epicanthal folds, hypertelorism, flat nose, and receding chin), (2) oligohydramnios, and (3) pulmonary hypoplasia [2,9,13]. This case demonstrated the characteristic Potter's facies.

Skeletal abnormalities which are seen in this case are hemivertebra, sacral agenesis, absence of metacarpal bone on left thumb, absence of fibula on left side

Our case was compatible with symelia apus associated with Potter's facies and Type II according to the classification of sirenomelia by Stocker and Heifetz.

Table 1: Classification of sirenomelia by Stocker and Heifetz [14]

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 diagnosis is obvious at birth on examination of a baby, but prenatal diagnosis can also be made as early as the first trimester by an ultrasound [1,4,9,10,13]. Color Doppler imaging can be helpful in identifying the single large vitteline artery and the absence of renal arteries and 3D-sonography and magnetic resonance imaging may complement the 2D-sonographic findings [4]. Diagnosis of this fatal anomaly prior to 24 weeks gestation is useful so that the option of pregnancy termination may be given to the parents [9,10,13]. This was not possible in our case as the mother's pregnancy was unsupervised. The therapeutic decision depends on the gestational term at the time of diagnosis, and on the severity of the malformative syndrome and the parents wish. Genetic counseling should be proposed because of the re-occurance risks [13].

Few cases with sirenomelia have been reported to have survived beyond the neonatal period. The spectrum of disease ranges from fused lower abnormalities, pelvic dysplasia, an osseous fusion of the calcanei, imperforate anus, and genitourinary anomalies [6-8].

CONCLUSION

Sirenomelia is a rare and fatal congenital anomaly associated with a various spectrum of disease. The present case highlights about the rare associations with sirenomelia like oesophageal atresia, absence of the metacarpal bone, absence of the fibula, VACTERL anomaly. Early prenatal diagnosis by first trimester scan should be the aim to minimize the trauma related to the termination of pregnancy at advanced gestation.

REFERENCES

1. Ugwu RO, Eneh AU, Wonodi W. Sirenomelia in a Nigerian triplet: A case report. J Med Case Rep 2011;5:426.
2. Al-Haggar M, Yahia S, Abdel-Hadi D, Grill F, Al Kaissi A. Sirenomelia (symelia apus) with Potter's syndrome in connection with gestational diabetes mellitus: A case report and literature review. Afr Health Sci 2010;10:395-9.
3. Sathe PA, Ghodke RK, Kandalkar BM. Sirenomelia with oesophageal atresia: A rare association. J Clin Diagn Res 2014;8:163-4.
4. Sikandar R, Munim S. Sirenomelia, the Mermaid syndrome: Case report and a brief review of literature. J Pak Med Assoc 2009;59:721 3.
5. Morfaw FL, Nana PN. Sirenomelia in a Cameroonian woman: A case report and review of the literature [v2; ref status: Indexed, http://f1000r.es/QcTFxB]. F1000Res 2012;1:6.
6. Sammons MB, Shiloh Pepin: The mermaid girl [Last accessed on 2009]. Available from: http://www.aolhealth.com/health/mermaid-syndrome-sirenomelia.
7. Stanton MP, Penington EC, Hutson JM. A surviving infant with sirenomelia (Mermaid syndrome) associated with absent bladder. J Pediatr Surg 2003;38:1266-8.
8. Clarke LA, Stringer DA, Fraser GC, Yong SL. Long term survival of an infant with sirenomelia. Am J Med Genet 1993;45:292-6.
9. Goodlow OG, Sibley RI, Allen BG, Kamanda WS, Gullattee AC, Rayfield WC. Sirenomelia: Mermaid syndrome. J Natl Med Assoc 1988;80:343-6.
10. Das BB, Rajegowda BK, Bainbridge R, Giampietro PF. Perinatal/neonatal case presentation. J Perinatol 2002;22:168-70.
11. Garrido-Allepuz C, Haro E, González-Lamuño D, Martínez-Frías ML, Bertocchini F, Ros MA. A clinical and experimental overview of sirenomelia: Insight into the mechanisms of congenital limb malformations. Dis Model Mech 2011;4:289-99.

12. Taori KB, Mitra K, Ghonga NP, Gandhi RO, Memmen T, Sahu J. Sirenomelia sequence (mermaid): Report of three cases. Indian J Radiol Imaging 2002;12:399-401.
13. Fadhlaoui A, Khrouf M, Gaigi S, Zhioua F, Chaker A. The sirenomelia sequence: A case history. Clin Med Insights Case Rep 2010;3:41-9.
14. 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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