Sirenomelia: A Rare Case of Foetal Congenital Anomaly

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ABSTRACT

Sirenomelia, alternatively known as ‘mermaid syndrome’ is a very rare congenital deformity in which the legs are fused together, giving them the appearance of the tail of a mermaid. Other birth defects are always associated with sirenomelia, most commonly abnormalities of the kidneys, large intestines, and genitalia. The present case is a one of sirenomelia associated with an absent right kidney, mild left hydronephrosis, single umbilical artery, and severe oligohydramnios. We discuss the findings, relative to the present literature and related etiopathogenesis.

Key words: Mermaid syndrome, oligohydramnios, sirenomelia

INTRODUCTION

This condition is found in approximately one out of every 100,000 live births[1] and is usually fatal within a day or two of birth because of complications associated with abnormal development and function of the kidneys and urinary bladder. More than half the cases of sirenomelia result in stillbirth, and this condition is 100 times more likely to occur in identical twins than in single births or fraternal twins.[2] It results from a failure of normal vascular supply from the lower aorta in utero. Maternal diabetes has been associated with caudal regression syndrome and sirenomelia.[3]

CASE REPORT

A 22 year’s old primipara underwent an ultrasound examination for routine fetal wellbeing. On ultrasound examination, we found fused thighs and legs with two femoral and two tibial bones within a single soft tissue and muscular compartment [Figure 1]. There was an absence of fibular bones in the legs; both feet were fused posteriorly and externally diverging anteriorly, and they looked like the fins of a fish. The fetal urinary bladder was minimally filled, and located slightly to the left. Fetal left renal pelvicalyceal system was mildly dilated [Figure 2], and fetal right kidney was not visualized. Only a single umbilical artery was present, and severe oligohydramnios was seen [Figure 3]. Fetal age was 26 weeks by ultrasound and the last menstrual period. Five days later, spontaneous delivery occurred at a private nursing home. Due to cultural constraints, a photograph of the delivered baby could not be taken. We saw completely fused lower limbs, rudimentary external male genital organs, and absent anus.

DISCUSSION

The sirenomelic resembles the mermaid of Greek and Roman mythology, which was depicted as having the head and upper body of a human and the tail of a fish. It is a severe form of caudal regression syndrome that results in a fusion of the lower extremities, which is not compatible with life. A spectrum of anomalies affects primarily the musculoskeletal, genitourinary, and gastrointestinal systems. This is a rare malformation with a range of 0.1 to 1% of all malformed infants. The etiology is unknown, but maternal drug abuse may have been an etiologic factor in the present case. Caudal regression syndrome consists of a spectrum of anomalies that range from an ectopic anus to sirenomelia. An embryologic insult to the caudal mesoderm that occurs between 28 to 32 days gestation is felt to be responsible for these malformations.[4]

According to a study by Garrido-Alllepuz et al.,[5] the causes of this malformation remain unknown, although the discovery that it can have a genetic basis in mice represents an important step toward the understanding of its pathogenesis. Sirenomelia occurs in mice lacking Cyp26a1, an enzyme that degrades retinoic acid (RA), and in mice that develop with reduced bone morphogenetic protein (Bmp) signaling in the caudal embryonic region.

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Access this article online

Quick Response Code: www.jcnonweb.com

DOI: 10.4103/2249-4847.106006
Clinical studies of sirenomelia have given rise to two main pathogenic hypotheses. The first hypothesis, based on the aberrant abdominal and umbilical vascular pattern of affected individuals, postulates a primary vascular defect that leaves the caudal part of the embryo hypoperfused. The second hypothesis, based on the overall malformation of the caudal body, postulates a primary defect in the generation of the mesoderm. This review has combined experimental and clinical information on sirenomelia together with the necessary background to understand how deviations from normal development of the caudal part of the embryo might lead to this multisystemic malformation.

Kallen suggested that diabetes may be a cofactor that modifies the action of one or more unknown teratogens. Raabe et al. reported the sonographic antenatal diagnosis of sirenomelia at 18 weeks’ gestation as evidenced by (i) oligohydramnios, (ii) bilateral renal agenesis (nonvisualization of the kidneys and bladder on serial prenatal examinations), and (iii) fusion of the lower extremities. Therapeutic abortion was implemented. Caution should be exercised to avoid inaccurate diagnosis, especially when attempting to document renal agenesis because enlarged or abnormally shaped adrenals may be mistaken for kidneys. Diagnosis of this fatal anomaly prior to 20 weeks’ of gestation is useful so that the option of termination of pregnancy may be given to the parents.

Fusion of the lower limbs, right renal agenesis, deformity of the bony pelvis, single umbilical artery, absent external genitalia, and imperforate anus were present in this case and fit the criteria proposed by Duhamel for ‘mermaid syndrome’. Instead of bilateral renal agenesis, in the present case, right renal agenesis with mild left hydronephrosis was noted. This anomaly is a new observation in association with sirenomelia.

Horikoshi et al. reported two cases of sirenomelia sequence which showed oligohydramnios and a single umbilical artery. The first case was of a single fetus with symelia apus and only one leg. In our case, the fetus had symelia dipus with two fused lower extremities. The second case was of a dichorionic-diamniotic twin pregnancy in which one fetus had symelia dipus with two fused lower extremities. Prenatal diagnosis of the condition was not made. The case presented here is a single pregnancy and diagnosed prenatally. In Horikoshi et al. reported both the cases absence of a urinary tract, imperforate anus, and spine deformity were confirmed.

Potter’s syndrome, consists of (i) Potter’s facies (large, low-set ears, prominent epicanthal folds, hypertelorism, flat nose, and receding chin), (ii) oligohydramnios, and (iii) pulmonary hypoplasia. This syndrome is almost invariably present with bilateral renal agenesis. The case presented here
does not demonstrate the characteristic Potter’s facies. Although a strong association between caudal regression syndrome and maternal diabetes has been described, no such relationship exists between Potter’s syndrome and diabetes. However, Al-Haggar et al.[12] reported one case of a fetus of sirenomelia sequence with Potter’s syndrome which showed oligohydramnios and symelia apus. The infant showed absent urinary tract and external genitalia; the legs were fused by skin, and had separate bones associated with Potter’s syndrome. The mother had a history of gestational diabetes mellitus. The present case was neither associated with a diabetic mother nor with Potter’s syndrome.

The study of Orioli et al.[13] describes the prevalence, associated malformations, and maternal characteristics among cases with sirenomelia. Data was obtained from 19 members of the International Clearinghouse for Birth Defect Surveillance and Research, and were reported according to a single pre-established protocol. The cases were clinically evaluated locally and reviewed centrally. A total of 249 cases with sirenomelia were identified among 25,290,172 births, for a prevalence of 0.98 per 100,000, with higher prevalence in the Mexican registry. An increase of the prevalence of sirenomelia with maternal age less than 20 years was statistically significant. The proportion of twinning was 9%, higher than the 1% expected. Sex was ambiguous in 47% of the cases, and no different from the expectation. The proportion of cases born alive, premature, and weighing less than 2,500 g were 47%, 71.2%, and 88.2, respectively. Half of the cases with sirenomelia also presented with genital, large bowel, and urinary defects. About 10-15% of the cases had lower spinal column defects, single or anomalous umbilical artery, upper limb, cardiac, and central nervous system defects. There was a greater than expected association of sirenomelia with other very rare defects such as extrophy of bladder, cyclopia/ holoprosencephaly, and acephalus acardia. The application of the new biological network analysis approach, including molecular results, to these associated very rare diseases is suggested for future studies.

Morfaw and Nana[14] reported a case of sirenomelia occurring in a 19 year old Cameroonian woman following premature rupture of membranes and associated cord prolapsed, the first documented case of sirenomelia in Cameroon. They highlighted some of the cultural myths associated with this disorder and discussed their findings relative to the present literature and the related controversies on its etiopathogenesis.

CONCLUSION

Although sirenomelia is usually associated with bilateral renal agenesis and absent urinary bladder, the present case has a mild hydronephrotic single kidney and partially filled urinary bladder. Sirenomelia is strongly associated with Potter’s syndrome and a diabetic mother, but this case was neither associated with a diabetic mother nor with Potter’s syndrome. Here we highlighted some of the findings from associated literature.

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