Sirenomelia in an Iraqi twin: a case report

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Abstract:

Background: Sirenomelia is a rare congenital malformation characterized by fusion of the lower limbs giving a characteristic mermaid-like appearance to the affected fetus. It is commonly associated with gastrointestinal, genitourinary, cardiovascular and musculoskeletal system defects.

Objective : To report the clinical manifestations of an extremely rare and complex malformation along with the associated anomalies.

Case report: A case of sirenomelia was reported in a one of a set of twin delivered at term by caesarean section to a 39 year old Iraqi mother. According to the search that has been done, it seems that this is the first reported case in this country. The following associated anomalies (imperforate anus, absence of external genitourinary orifices, esophageal atresia with tracheo-esophageal fistula, intestinal atresia and bilateral renal agenesis) were observed.

Conclusion: Sirenomelia is a rare and fatal congenital malformation. Still there is some controversy regarding its etiology, however there is an increasing belief that this complex malformation is distinct from the caudal regression syndrome. Survival depends on the presence of normal renal function

Keywords: Sirenomelia, mermaid, associated anomalies.

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Introduction:

Sirenomelia, also known as Mermaid syndrome is a very rare congenital anomaly in which the legs are fused together giving them the appearance of a mermaid's tail. The condition is found in approximately 1 in 100.000 live births (1) with a male/female ratio of 3:1 (2), and is more common among one of two monozygotic twins (3).

Several malformations of the gastrointestinal, genitourinary, cardiovascular and musculoskeletal systems are usually associated. Oligohydramnious secondary to severe renal dysplasia is universal (2).

The first medical description of Sirenomelia was by Rocheus and Polfyr in the sixteenth century. Duhamel in 1961 defined all the anomalies of mermaid syndrome and described it as the most severe form of caudal regression syndrome (4).

More than half the cases of Sirenomelia result in stillbirth and those born alive usually die within a day or two of birth. Death is usually a result of urinary malformations incompatible with life, such as renal agenesis (1).

Case report

A two hour old term newborn, of undetermined sex and with multiple congenital anomalies, was referred to the Pediatric Surgical Department at Basrah Children's Specialty Hospital. The baby was one of a set of twin who was delivered at term by cesarean section. The other twin was alive and well.

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The mother was 39 year old and the father 42 year old and they were not relatives. There was no previous history of congenital anomalies in the family.

The mother had history of hypertension during pregnancy. No history of diabetes mellitus, drug ingestion or exposure to radiation, with irregular antenatal follows up.

The baby had a birth weight of 2 kg and according to the referral documents the Apgar score was 1 at 1 minute and 5 at 5 minutes.

On examination, the following congenital malformations were documented: Fusion of the lower extremities, imperforate anus, absence of the external genitalia and absence of urinary orifice. Figure(1,2 &3).

The baby was admitted to the neonatal intensive care unit. Investigations were done and revealed blood urea of 3.5 mmol/L and blood sugar of 2.5 mmol/L at time of admission. Abdominal ultrasound revealed non-visualization of both kidneys and urinary bladder and echocardiography was normal. Erect abdominal X-Ray revealed picture suggestive of intestinal atresia. During resuscitation, the baby was noted to have excessive frothy secretions. Nasogatric tube was inserted and coiled and chest x-ray confirmed the presence of esophageal atresia with tracheo-esophageal fistula.

At 2 day of age both blood urea and serum creatinine have elevated (11.6 mmol/L and 170µmol/L) respectiyely. The family was informed that their baby had severe multiple congenital anomalies and that corrective surgery will be of no benefit due to the presence of bilateral renal agenesis. The baby died at 55 hours of age.

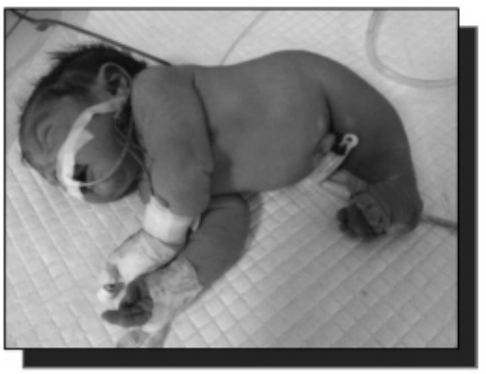

Fig.1 Sirenomelia dipus

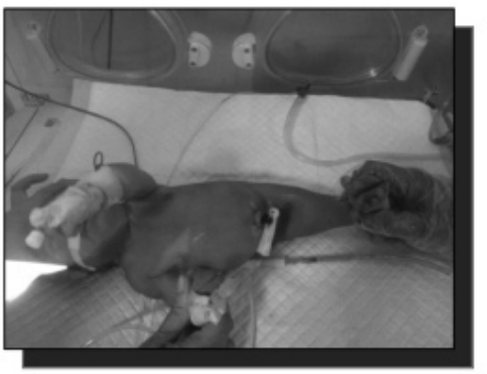

Fig.2 Absence of external genitourinary orifices

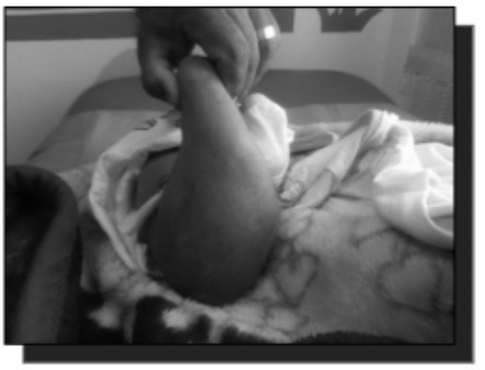

Fig.3 Imperforate anus

Discussion:

Sirenomelia or mermaid syndrome is an abnormal development of the caudal region of the body involving varying degrees of fusion of the lower limbs with or without bony defects. It is usually associated with other visceral defects such as hypoplastic lungs, cardiac agenesis, absent genitalia, digestive defects, absent kidney and bladder, vertebral and central nervous system defects (3,5,6). Usually the prognosis is poor. Survival depends on the nature of the visceral anomalies and the presence of adequate kidney functioning and renal outflow(7). Death usually results from obstructive renal failure due to renal agenesis or dysgenesis.

The cause of sirenomelia remains unclear, however, maternal diabetes mellitus (8), genetic predisposition, environmental factors and vascular steal phenomenon with the single vitelline umbilical artery diverting blood supply and nutrients from the lower body and limbs (9) have been proposed as possible causative factors. In this case the mother was not diabetic. The syndrome is also reported to be associated with twins.

Reports indicate a 100–150 times higher incidence in monozygotic twins relative to dizygotic twins or singletons (10). Moreover, about 20% of cases are derived from products of twin pregnancies. In our case the baby was one of a set of twin.

Sirenomelia was formerly thought to be an extreme form of caudal regression syndrome (CRS), however it is reclassified to be considered a separate condition (10). Specific anomalies are common to both conditions, but aside from fusion of the lower extremities, an aberrant abdominal umbilical artery/"persistent vitelline artery" has been invoked as the chief anatomic finding that distinguishes Sirenomelia from caudal regression syndrome (11).

Sirenomelia has been classified into 3 types according to the number of lower-limb bones present: apus (no feet, 1 tibia, and 1 femur), unipus (1 foot, 2 femurs, 2 tibias, and 2 fibulas), and dipus (2 feet, 2 tibias, 2 fibulas,2 femurs)(12). This case belongs to the dipus variety. Figure(1).

Conclusion:

Sirenomelia is a rare and fatal congenital malformation. Controversies on its etiopathogenesis persist even though it is increasingly believed to be distinct from the caudal regression syndrome. The associated visceral anomalies are usually incompatible with life. However surviving sirenomelic cases have been described. To our best knowledge, this is the first reported case in our locality. We hope it will bring the attention of gynaecologists, radiologists, pediatricians and pediatric surgeons to enhance high index of suspicion specially among gestations at high risk.

References:

1- *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.*

2- *Reddy KR, Srinivas S, Kumar S, Reddy S, Hariprasad, Irfan GM. Sirenomelia a rare presentation. J Neonat Surg 2012;1:7*

3- *MorfawLI,NanaPN(2012)SirenomeliainaCameroonian woman: a case report and review of the literature [v1; ref status: Indexed, http://f1000r.es/OlDGLR].*

4- *Duhamel B. From the Mermaid to Anal Imperforation: The syndrome of caudal regression. Arch Dis Child 1961; 36: 152-155.*

5- *Tang TT, Oechler HW, Hinke DH, Segura AD, Franciosi RA. Limb body-wall complex in association with sirenomelia sequence. Am J Med Genet 1991; 41(1): 21–25.*

6- *Rodriguez JI, Palacios J, Razquin S. Sirenomelia and anencephaly. Am J Med Genet 1991; 39(1): 25–27.*

7- *Pinette MG, Hand M, Hunt RC, Blackstone J, Wax JR, Cartin A. Surviving sirenomelia. J Ultrasound Med 2005; 24(11): 1555–1559.*

8- *Rosemary O Ugwu*, Augusta U Eneh , Woroma Wonodi. Sirenomelia in a Nigerian triplet: a case report. Journal of Medical Case Reports 2011, 5:426*

9- *Stevenson RE, Jones KL, Phelan MC, Jones MC, Barr M Jr, Clericuzio C. Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics 1986, 78:451-457.*

10- *Rozina Sikandar, Shama Munim. Sirenomelia, the Mermaid Syndrome: case report and a brief review of literature. J Pak Med Assoc. Vol. 59, No. 10, October 2009*

11- *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; 143: 3175-3184.*

12- *Strocker JT, Heifetz SA. Sirenomelia, a morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol.1987;10:7–50.*