CASE REPORT

Sirenomelia: A Rare Entity

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Abstract

Sirenomelia also called as Mermaid Syndrome, is a rare congenital malformation of uncertain aetiology. It is characterised by fusion of the lower limbs and commonly associated with severe urogenital and gastrointestinal malformation. It should be suspected in antenatal period in cases presenting with severe oligohydramnios and intra-uterine growth retardation. Here, we report a case of sirenomelia in a 26th weeks foetus.

Key Words

Sirenomelia, Caudal Regression Syndrome

Introduction

Sirenomelia or the mermaid syndrome is an extreme example of the caudal regression syndrome (1). It is a rare congenital anomaly in which the lower limbs are fused together, sometimes with a single femur (2). Associated malformations include absent external genitalia, imperforated anus, lumbosacral , vertebral and pelvic abnormalities and renal agenesis (2,3). Because of the resultant oligohydramnios, these infants most often have potter facies and pulmonary hypoplasia (4). The prevalence of this syndrome is 0.1 - 0.25:10,000 in normal pregnancies and it has very strong association with maternal diabetes where the relative risk is 1:200 - 250 and 22% of foetuses with this anomaly will have diabetic mothers (5). Here we present a case of sirenomelia in a 26 weeks foetus.

Case report

A 25 years old primigravida married since 4 years came to the hospital with history of labour pains since 4 hours. Her general physical examination revealed her to be B +ve with heamoglobin 9.0 gm% with blood sugar levels 86 mg/dl. On abdominal examination, she was 26 weeks pregnant with uterine contractions. Vaginal examination showed the patient in active labour with cervix 6 cms dilated and fully effaced. She had no significant past medical history. During the prenatal period at 19 weeks, ultrasonography of the patient revealed severe oligohydramnios with intrauterine growth retardation and absence of foetal kidneys.

She remained in labour for 4 hours and then she delivered a 500 gms foetus. Gross examination of the foetus displayed multiple external deformities. There was fusion of both lower limbs, absence of external genitalia with imperforate anus (*Fig 1*) & (*Fig 2*). The foetus was subjected to radiological examination which showed deformed sacrum, absence of pelvic bones, fused lower limbs showing single femur and tibia and absence of both fibular bones. Ultrasonography of the foetus revealed bilateral renal agenesis.

Discussion

Sirenomelia is a rare congenital malformation with an incidence of 1.5 - 4.2 per 100,000 births.[3] It is three times more common in males having a higher incidence

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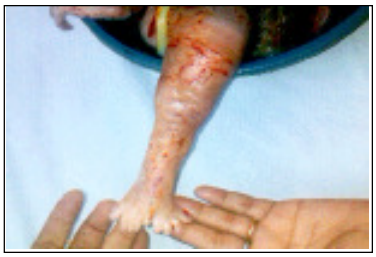

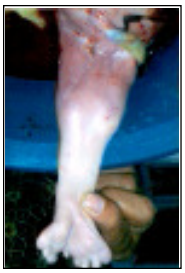

Fig 1 &2 Showing Fusion of Both Lower Limbs with Absence of External Genetalia & Imperforate Anus in Sirenomelic Baby

in identical twins, increase in risk with maternal diabetes and maternal age of less than 20 years and older than 40 years (3,4). Initially thought to be fatal , however there are reports of four children surviving with sirenomelia (4).

The aetiology of sirenomelia remains unclear.Various theories have been postulated to explain the aetiology of sirenomelia. Altered oxidative metabolism from maternal diabetes may cause production of free oxygen radicals in the developing embryo, which may be terratogenic (5) Another hypothesis given by Stevenson et al. explains that in mermaid syndrome, blood is diverted from the caudal region of the embryo to the placenta producing a nutritional deprivation and abnormal development of caudal structures. The site at which the steal occurs determines the severity of the anomalies (6) Absence of chromosomal anomalies and familial inheritance has been noted in almost all cases.

Sirenomelia is invariably fatal because of bilateral renal agenesis which leads to severe oligohydramnios and lung hypoplasia. There is no treatment available for sirenomelia and prevention should be the goal. So prenatal diagnosis is very important. It can be diagnosed as early as 13 weeks of pregnancy (7,8). Third trimester ultrasonographic diagnosis is impaired by severe oligohydramnios whereas during the second trimester the amount of amniotic fluid is sufficient to allow diagnosis. If diagnosed early, termination of pregnancy can be offered relatively safely to the mother. This would be less traumatic both physcially and mentaly to the parent than if detected later in pregnancy and moreover because of its association with maternal diabetes mellitus, the maternal blood glucose levels should be optimal in preconception period and in first trimester to prevent this anomaly.

References

1. Tanha FD, Googol N, Kaveh M. Sirenomelia (mermaid syndrome) in an infant of a diabetic mother. *Acta Medica Iranica* 2003; 41:69-72
2. Mysorekar VV, Rao SG, Sundari N. Sirenomelia:a case report. *Ind J Pathol Microbiol* 2007; 50 (2): 359-61
3. Gouri MSR, Kumari KMK, Rao SG, Devi U. Sirenomelia. *Ind J Pathol Microbiol* 2009; 52:579-80
4. Browne M, Fitchev P, Adley B, *et al*. Sirenomelia with an angiomatous lumbosacral myelocystocele in a full term infant. *J Perinatol* 2004; 24:329-31
5. Kedian YS, Duhan N, Rattan KN, *et al.* Sirenomelia (mermaid syndrome): a rare anomaly. *African J Pediatr Surg* 2008; 5:105-06
6. Stevenson RE, Jones KL, Phelan MC, *et al*. Vascular Steal: The Pathogenetic mechanism producing Sirenomelia. *Pediatr* 1986;78:451-57
7. Van KJ, Cannie M, Robrechts C, *et al.* First trimester diagnosis of Sirenomelia. *Prenat Diagn* 2006; 26(8):684-88
8. Contu R, Zoppi MA, Axiana C, *et al*. First trimester diagnosis of Sirenomelia by 2D and 3D ultrasound. *Fetal Diagn Ther* 2009; 26(1):41-4.