Sirenomelia: A Rare Entity

Sapna Gupta, Anil Mehta, Belu Sharma, Nirupam Gupta*, Jyotsana Lamba

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
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
ultrasoundographic 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 physically and mentally 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

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