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Sirenomelia in twin pregnancy: A case report

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Abstract
Sirenomelia also known as mermaid syndrome since Greco-Roman period, is a lethal disorder characterized by a single lower extremity, with concomitant presence of several anomalies of urogenital and gastrointestinal system. Sirenomelia is a syndrome of merging, malrotation and dysgenesis of lower extremity. Merging is more correct term to use because fusion refers to joining of two processes after breakdown of intervening epithelia. Here, we present a case of dichorionic diamniotic twin pregnancy, where one foetus was diagnosed with sirenomelia associated with hemivertebra and absent right radius and ulna and the co-twin was apparently healthy.

Keywords: Sirenomelia, mermaid syndrome, twin gestation

Introduction
Sirenomelia is an uncommon and fatal congenital defect characterized by varying degrees of lower limb fusion, agenesis or dysgenesis of kidneys, absent external genital, imperforate anus, lumbosacral and pelvic bone anomalies and single umbilical artery. Sirenomelia was considered as a severe form of caudal regression but Twickler *et al.* has provided evidence that caudal regression and sirenomelia are two different entities [1]. The risk of sirenomelia is nearly 100-150 times more in monozygotic twins as compared to dizygotic twins or singleton pregnancy [2]. Index case was an extremely rare case of sirenomelia in dichorionic diamniotic twins (DCDA) that was diagnosed after birth. First twin diagnosed with sirenomelia associated with hemivertebra and absent right radius and ulna and the co-twin was syndromic for downs syndrome.

Case report
A 25 years primigravida at period of gestation 36 weeks with diamniotic dichorionic twins, (DCDA) in labour was referred to our hospital. She was a booked case at primary health centre. Her antenatal period was uneventful and routine investigations were normal. She was normotensive and non- diabetic. There was no history of intake of alcohol, tobacco or any other teratogenic medicine. Two antenatal ultrasounds at 19 weeks and 30 weeks of gestation done in a peripheral institution, revealed DCDA twin gestations, with normal amniotic fluid pocket, but were not able to detect congenital malformations. She had normal vaginal delivery within 6hours of hospitalization. First twin, a still-born fetus, weighing 1100 gram was delivered. On physical examination, wide philtrum, low-set ears, limb abnormalities, complete fusion of lower extremities with four toes in single foot (Figures 1a) were detected. Single umbilical artery, imperforate anus, no external genital organs along with spina bifida occulta was identified (Figure 1b). The infant was clinically diagnosed as sirenomelia and classified as sympusunipus (one-foot present), as per classification by Foster *et al*. Radiological examination revealed partial fusion of both femurs, fusion of fibulae, tibia seen, supernumerary ribs (B/L 13), sacral agenesis, hemivertebrae, right radius and ulna were absent. Right hand had 3 metacarpals and left 4 metacarpals (Figure 2). Stocker and Heifetz classified sirenomelia into seven sub-types, findings in our case were consistent with type IV sirenomelia.
Second twin was female with a birth weight of 1700 gram, Apgar was 8 at 1 min and 9 at 5 minutes. She had mongoloid slant of eyes, depressed nasal bridge, generalized hypotonia, simian crease and sandal gap. Placenta was normal with 3 vessels in umbilical cord. The findings were suggestive of Downs's syndrome, evaluation of the baby planned but had early neonatal death. Parents denied any further investigation including the autopsy of the twins.

Discussion

Pathogenesis of sirenomelia have been explained by two main hypothesis vascular steal hypothesis and the defective blastogenetic hypothesis. According to vascular steal hypothesis [3], an alteration in early vascular development leading to, "vitelline arterial steal", in which blood flow is diverted from caudal region of the embryo to placenta leading to hypoperfusion. In our case, single umbilical artery could have created a state of hypoperfusion and resulted in multiple anomalies. Caudal eminence gives rise to lower limb buds as well as perineum somite and vertebrae, during blastogenesis hence, any teratogenic insult to caudal neuropore could give rise to spectrum of anomalies seen in sirenomelia [4]. This case was DCDA twin pregnancy with sirenomelia of which limited studies have been reported.

Sirenomelia can be diagnosed in first trimester, between 9 to 10 weeks of gestation using 2D, 3D and color Doppler ultrasound [5-8]. It is easier to diagnose at this time, as amniotic fluid volume is normal. As amniotic fluid is secreted by amniotic membrane in first trimester. At this time findings include thickened nuchal translucency, fused extremities and single umbilical artery. With the help of trans-vaginal ultrasonography, all the long bones of the fetus can be visualized at 10 weeks of gestation [9-10]. In second and third trimester if severe oligohydramnios, ultrasonographic diagnosis may be difficult and MRI may be needed to confirm the diagnosis of sirenomelia.

(a) Ventral View (b) Dorsal View

Fig 1: Sirenomelic baby with fused lower extremities, one foot and no genitalia.

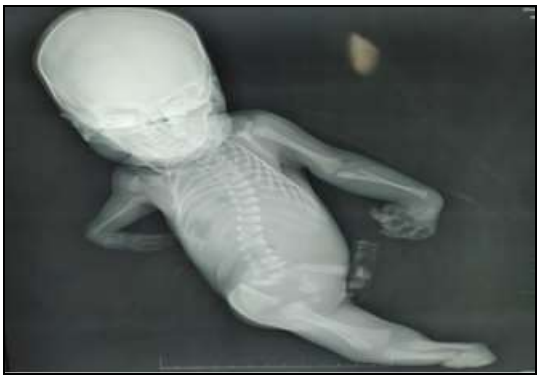

Fig 2: Lower limb X-ray showing incomplete fusion of both femurs.

Conclusion

The incidence of congenital malformation is appreciably higher in multifetal gestation compared with that in singleton pregnancy, hence anomalies scan in such high-risk cases should be done by experienced radiologist. Most preferable and accurate timing of diagnosis of sirenomelia is first trimester. As transvaginal and three-dimensional sonography and magnetic resonance imaging (MRI), play a key role in early diagnosis of sirenomelia they should be used judiciously, so that option of first trimester pregnancy termination or voluntary fetal reduction in case of twin gestation is available to the parents.

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