Sirenomelia (mermaid syndrome): A rare anomaly

Sir,

Sirenomelia (mermaid syndrome) is a rare congenital anomaly in which the lower limbs are fused together, sometimes with a single femur. Associated malformations include absent external genitalia, imperforate anus, lumbosacral vertebral and pelvic abnormalities and renal agenesis.[1] This condition has been thought to be part of caudal regression syndrome.[2] The prevalence of this syndrome is 0.1-0.25:10,000 in normal pregnancies. There is strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of foetuses with this anomaly will have diabetic mothers.[2] We have encountered a case with this rare anomaly born to an uncontrolled diabetic mother unaware of her condition.

A full term baby was delivered by normal vaginal delivery with cephalic presentation and Apgar score was 3/0 at 1 and 5 minutes respectively. The newborn baby had gross anomaly known as sirenomelia, characterised by fusion of lower limbs and absence of external genitalia, with imperforate anus also apparent on examination [Figures 1 and 2]. X-rays of the neonate revealed narrow chest (lung hypoplasia), deformed sacrum, fused lower limbs and absence of both fibular bones [Figure 3]. Ultrasonography of abdomen revealed bilateral renal agenesis. The baby died after 30 minutes of delivery. The mother of the neonate was 26 years of age, a primigravida and not a known diabetic. On investigation in postpartum period her serum glucose levels were 260 mg% and 350 mg/dl at two consecutive times suggestive of diabetes mellitus.

Caudal regression syndrome is a rare congenital defect in general population with a prevalence of 0.1-0.2:10,000 in normal pregnancies but a relative risk of 200-250 in diabetes.[2,3] Sirenomelia is the most severe condition of this syndrome. The specific anomaly of sirenomelia is based on the presence of lower limb fusion, associated with other skeletal and lumbar



Figure 2: Clinical photograph of dorsal aspect of baby showing absence of anal opening



Figure 1: Clinical photograph of ventral aspect of baby showing absence of external genitalia and fusion of lower limbs



Figure 3: Infantogram showing narrow chest, narrow pelvis and fused lower limbs

deformities. The aetiology of sirenomelia remains unclear. This pattern of anomalies is probably due to a generalised alteration of mesodermal cell migration in primitive streak period.[2] Altered oxidative metabolism from maternal diabetes may cause increased production of free oxygen radicals in the developing embryo, which may be teratogenic.[2] Another hypothesis of pathogenesis of sirenomelia in nondiabetic cases has been proposed to be vascular steal phenomenon with single, aberrant, umbilical artery stealing blood supply from the lower torso and limbs.[4]

Diagnosis of this condition can be made in the first trimester by noting the short crown - rump length on ultrasonography.[5] If diagnosed in first trimester termination of pregnancy can be offered relatively safely to the mother. This would also be less emotionally traumatic to the parents than if detected later in pregnancy as delivery of a baby with sirenomelia at term, as in the present case, can scar their psyche.

It is a lethal anomaly because of bilateral renal agenesis which leads to severe oligohydraminos and lung hypoplasia. There is no treatment available for sirenomelia. However prevention is possible and should be the goal. Because of strong association of sirenomelia and maternal diabetes mellitus there should be optimum maternal blood glucose level in preconceptual period and in first trimester to prevent this anomaly.

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