

Sirenomelia (Mermaid Syndrome) In an Infant of a Diabetic Mother

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Abstract: *Abnormalities of carbohydrate metabolism occur frequently during pregnancy. Unfortunately, carbohydrate intolerance during pregnancy causes significant increase in fetal and maternal morbidity. The consequences to the fetus are more serious than those to the mother. Caudal regression syndrome is rare, but most specific congenital anomaly associate with diabetic pregnancy. We present such a case of neglected uncontrolled diabetic mother with a malformed baby having sirenomelia.*

I. Introduction

Caudal regression is a rare syndrome which represents a spectrum of congenital malformations ranging from lumbosacral spine agenesis to the most severe cases of sirenomelia with lower extremities fusion characteristics.¹ A case of this defect has been encountered from an uncontrolled diabetic mother. In Sirenomelia, 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. The condition has been thought to be part of the caudal regression spectrum.²

II. Case Report

A 33-years-old pregnant woman (G5 P1 L1 A3) presented to our hospital with the chief complaint of labor pain. Her gestational age (GA) was estimated 26 weeks by LMP (Last menstrual period) and sonography. She had a sonographic report at late second-trimester of pregnancy which had shown severe oligohydramnios, with no evidence of skeletal or other organ anomaly. Since diminished or lack of amniotic fluid volume (oligohydramnios) disturbs sonographic image resolution, anomalies had not been diagnosed by the radiologist. Several hours after her admission in labor room, she delivered an infant with cephalic presentation and Apgar Score of 3/0 at 1 and 5 minutes. The newborn baby had gross anomaly known as sirenomelia (Fig. 1), characterized as fusion of lower limbs. Absence of external genitalia and imperforate anus were also apparent.



Figure 1

III. Discussion

The sacrum is a bone at the base of the spinal column that is formed following the fusion of 5 vertebrae. Sacral agenesis is a condition that exists when either part or all of the sacrum is absent.³ Total agenesis of the sacrum is manifested by a narrowed pelvis with the medial portion of the iliac ala almost in contact and is included in the group of malformations known as caudal regression syndrome.⁴

The etiology of Sirenomelia remains unclear. Diversion of blood flow away from the caudal portion of the embryo through the abdominal umbilical artery/ "vascular steal" has been proposed as the primary mechanism leading to Sirenomelia. In contrast, CRS is hypothesized to arise from primary defect of caudal mesoderm. A teratogenic event during the gastrulation stage i.e. 3rd gestational week, may interfere with the formation of notochord, resulting in abnormal development of caudal structures. Altered oxidative metabolism from maternal diabetes may cause increased production of free oxygen radicals in the developing embryo, which may be teratogenic. There is a strong association of Sirenomelia and Caudal Regression Syndrome with maternal Diabetes. The disorder occurs in about one in 350 infants of diabetic mothers, representing an increase of about 200-fold over the rate seen in the general population.⁵

IV. Conclusions

Sirenomelia is a rare and fatal congenital anomaly and has strong association with maternal Diabetes. Early prenatal diagnosis by first trimester scan should be the aim to minimize the trauma related to the termination of pregnancy at advanced gestation. In addition, where possible, a second US scan should be performed 4-6 weeks after the initial 8-9 weeks scan so that gross structural anomalies are detected and termination of pregnancy be considered earlier.

References

- [1]. Dordoni D, Freeman PC. Sirenomelia sequence. *The Fetus* 1991; 1: 7553. 1-3.
- [2]. Tanha FD, Googol N, Kaveh M. Sirenomelia in an infant of diabetic mother. *Acta Medica Iranica* 2003; 41(1): 69-72.
- [3]. Schiesser M, Holzgreve W, Lapaire O, Willi N, Luthi H, et al. Sirenomelia, the mermaid syndrome-detection in the first trimester. *Prenat Diagn* 2003; 23: 493-5.
- [4]. Ladure H, D'herve D, Loget P, Poulain P. Prenatal diagnosis of Sirenomelia. *J Gynecol Obstet Biol Reprod (Paris)* 2006; 35: 181-5.
- [5]. 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; 143A: 3175-84.