Sirenomelia: A rare anomaly and challenges of ultrasound diagnosis

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Abstract

Background: Sirenomelia also known as mermaid syndrome is a rare congenital anomaly, characterised by disorder of the lower spine and the lower limbs.
Methods: A review of medical records.
Results: A neonate was delivered with fusion of the lower limbs, absence of external genitalia, imperforate anus and a single umbilical artery. The upper half was grossly normal.
Conclusion: This case demonstrates the rarity of sirenomelia and the difficulty of ultrasound diagnosis after first trimester of pregnancy.

Keywords: Sirenomelia, Mermaid syndrome, Oligohydraminous, Ultrasound diagnosis

Highland Med Res J 2016;16(2):99-101

Introduction

The term Sirenomelia (also known as mermaid deformity, symmelia, symposia, sympus, uromelia and monopodia) is derived from the Greek word siren and melia or limb. Public interest in Sirenomelia was aroused by the successful operation of the limbs of a neonate with Sirenomelia in Lima Peru. This is in addition to about a few known survivors of this condition beyond the early neonatal life. Caudal regression is a rare syndrome which represent a spectrum of congenital malformations ranging from lumbosacral spine agenesis to the most severe cases of sirenomelia with characteristic lower limb abnormalities, and its associated gastrointestinal and urogenital malformation.[1] Sirenomelia is rarely compatible with life, most are stillbirth and a few survive beyond the neonatal life. The incidence of this condition ranges from 1:60,000 – 100,000 live births with a male to female ration of about 3:1, with the occurrence of Sirenomelia in a singleton fetus (as in this case), is about 150 times rare compared to a monozygotic twins[2]

In Sirenomelia the lower limbs are fused together, sometimes with a single femur. Associated anomalies include absent external genitalia, imperforate anus, lumbosacral vertebrae, pelvic anomalies, and renal agenesis. There are various classification of this condition, however the division into three is perhaps most practical. (I) Apus; (No feet, only one femur and one tibia, as seen in this case. (II) Unipus; One foot, two femur, two tibia, two fibulae and (III) Dipus – two feet usually fused at the heel, two fused legs giving the appearance of a flipper. The later is commonly referred to as Mermaid syndrome.[3]

Case Report

A booked 26 year old Po^{+1} lady presented to the hospital with complain of lower abdominal pain and slight bleeding per vagina. Her gestational age (GA) was estimated to be about 34weeks by L.M.P (Last Menstrual Period). An ultrasonographic evaluation done at 26weeks of pregnancy had shown severe oligohydramnios and a low lyng placeta. There was no demonstrable skeletal or other fatal organ anomaly. Past medical history was unremarkable. She was not a known diabetic and there was no history of drug intake or malformation in the family. She was admitted to the hospital for bed rest on account of placenta previa. Another sonographic evaluation done while on admission at 35weeks showed a Type II placenta previa with severe oligohydramnios and no demonstrable skeletal anomaly. She had significant vaginal bleeding at 36 weeks gestation necessitating an emergency caesarian section. She was delivered of a 1.7kg neonate with severe birth asphyxia (Apgar score of 3 at one and five minutes respectively). The neonate had no facial dimorphism but, there was a gross anomaly (Figure 1) identified as Sirenomalia, characterized by fusion of the lower limbs, absence of external genitalia, imperforate anus and a single umbilical artery. The upper half was grossly normal. It lived for about 5 hours. Radiograph of the neonate (Figure 2) allowed categorization on the basis of skeletal deformity. The x-ray showed a subtotal sacrococygeal agenesis and a single dysmorphic lower limb, thus a type 1 Apus form of sirenomelia was made.

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Figure 1. Sirenomelia Baby

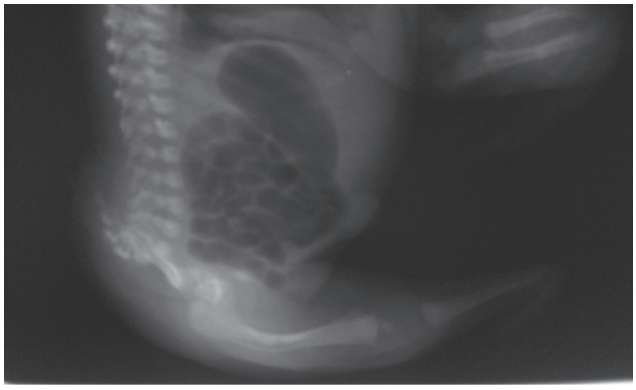

Figure 2. X-ray demonstrating single femur and tibia

Discussion

The specific anomaly of sirenomelia is based on the presence of lower limb fusion, associated with other skeletal and limb deformities. There is usually varying degree of associated anomalies of gastrointestinal, genitor-urinary, cardiovascular and central nervous systems. There is some overlap with the VACTERL constellation of anomalies with respect to these other association. The case presented has associated gastrointestinal(micro colon) and genitourinary (bilateral renal agenesis) systems abnormalities.

The pathogenesis of this condition is not well understood however, various mechanism have been postulate. The vascular steal theory of Saddler et al is widely accepted but not unanimously[4]. The vascular steal phenomenon suggest a single aberrant umbilical artery (vitelline) stealing blood supply from the lower torso and limbs to the placenta, leading to hypoplasia of the lower aorta and mal-development of lower limbs. A different mechanism is however suggested in diabetic. The pattern of anomalies in diabetics is probably due to a general alteration of mesodermal cell migration in primitive streak period, with resultant altered oxidative metabolism resulting in increased production of free oxygen radicals in the developing embryo, which may be teratogenic.[5]

Sirenomelia is a lethal condition because of its bilateral renal agenesis/dysgenesis which causes oligohydramios and lung hypoplasia. Since diminished or lack of amniotic fluid.(oligohydramnios) decreased the sonographic image resolution and diagnosis with certainty, as a result the anomaly was not diagnosed in the two sonographic evaluation done. The oligohydramnios in Sirenomelia is due to poor fetal urine production which contributes significantly to the production of amniotic fluid and for normal pulmonary development. The lack of urine, secondary to renal agenesis or digenesis is a major factor in the development of the oligohydramnios and fetal deformation. Essentially there is a compression of the developing fetus due to lack of space in utero and this compression may cause lung hypoplasia, abnormal facies (potters) or abnormal positioning of the hand and feet. In more than 50% of cases, Sirenomelia is difficult to diagnose and may be missed in late 2nd and 3rd trimester of pregnancy.[6] In late second and third trimester, there is difficulty in making diagnosis of sirenomelia on ultrasound. This was the major sonograhic challenge in this case as two separate ultrasound down did not diagnose sirenomelia.

This limitation of ultrasound in the second and third trimester of pregnancy for the diagnosis of Sirenomelia can be overcome by ultrasonographic evaluation in the first and early second trimester of pregnancy. At this stage the fetus is surrounded by amniotic fluid, as its production then is primarily by the amniotic membrane covering the placenta and cord, therefore detailed scanning is possible. A transvaginal ultrasonography diagnosis of sirenomelia can be made as early as 9th postmenstrual week. There may be size for date discrepancy with a shorter crown – rump length, short femur and humerus, shorter and thinner radius, ulna, tibia, and fibula can also be visualized at about 10th week gestation.[7] In early second trimester the inability to show separate lower limbs or identification of a single femur, or using an high frequency probe of about 5-12mHz to demonstrate continuous skin line over on the thigh, over both femur in both coronal and transverse scan is indicative of a fused thigh.[8] If diagnosed is made, the termination of pregnancy can be offered to the mother. This would be less emotionally traumatic to the parents compared to detection at delivery as it turned out in this case made at delivery.

Colour and Power Doppler ultrasound can be diagnostic in some cases of Sirenomelia with the demonstration of a large single aberrant intra-abdominal arterial blood vessel which continue into the two vessel umbilical cord.[9]

Other non-routine antenatal imaging modality used in the diagnosis of this condition includes, Magnetic resonance imaging (MRI), a non-ionizing, non-invasive imaging modality, can be used to further confirm suspected case of sirenomelia, from ultrasound at any gestational age. MRI is not limited by oligohydramnios which is the major challenge present in this congenital anomaly[10]. However MRI is not readily available and relatively expensive in our environment.

Conclusion

This case report demonstrates that, sirenomelia is a very rare congenital anomaly with challenges of ultrasound diagnosis in the late second and third trimesters of pregnancy, due to olygohydramnios from renal agenesis. Antenatal sonographic evaluation done in the first or early second trimester of pregnancy and MRI performed any time in the course of pregnancy can be used to overcome these Challenges.

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