A Rare Case Report of Sirenomelia

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Sirenomelia, which is also known as mermaid syndrome, is an extremely rare congenital developmental disorder characterized by anomalies of the lower spine and the lower limbs. Affected infants are born with partial or complete fusion of the legs. Additional malformations may also occur including genitourinary abnormalities, gastrointestinal abnormalities, anomalies of the lumbar-sacral spine and pelvis and absence or underdevelopment (agenesis) of one or both kidneys. Affected infants may have one foot, no feet or both feet, which may be rotated externally. The tailbone is usually absent, and the sacrum is partially or completely absent as well. Additional conditions may occur with sirenomelia including imperforate anus, spina bifida, and heart (cardiac) malformations. The incidence of sirenomelia is 8-1 case/100000¹ births with male to female ratio being 3:1.²

Sirenomelia has no definite cause, almost all cases occur without any reason.

Previously, some literatures mentioned that sirenomelia is the severe form of caudal regression syndrome and has many developmental disorders. Recently much literature mentioned sirenomelia is similar but a different disorder. NORD has a different report on caudal regression syndrome.
PRESENTATION OF CASE

A 31 yr old G4P3L3 at 38 wks, 04 days of GA with previous three live vaginal births was admitted in the labour room at 2.26 AM with pain abdomen. She had a history of prior antenatal care at the sub-centre level irregularly and without any gynaecologist consultation. She belongs to a tribal community with lower socioeconomic status. She had no history of any addiction before and during pregnancy. She was otherwise healthy with no known history of genetic or congenital malformation in her family or her previous pregnancy.

On antenatal investigation - blood group- O+, Hb% 10 gm %, HVL, HbsAg- negative, Urine (routine, microscopic)- WNL, RBS- 92 mg %, VDRL- non reactive. She had not done any NT scan or TIFFA scan. No OGTT was done between 24-28 wks GA. Severe oligohydramnios (AFI-03CM) was seen on ultrasonography which was done 2 days before delivery.

On examination, she was in the 2nd stage of labour with a cephalic presentation with a fully effaced and fully dilated cervix, head at station 0. She delivered a 2.256 kg term stillbirth baby at 3.05 AM with a fused upper part of both legs. On physical examination, the infant showed fused lower limbs upper part, two feet, 5 toes in each foot, imperforate anus, absent external genitalia, beaked nose, prominent epicanthic fold, low set ears, potters facies, loose skin in hands, single umbilical artery, right foot everted and left foot rotated. An autopsy was declined by the parents. The intrapartum and postpartum period of the mother was uneventful.

CLINICAL DIAGNOSIS

According to the clinical and physical presentation, this is a case of sirenomelia or mermaid syndrome.

DIFFERENTIAL DIAGNOSIS

Physical signs of the following disorders can be a differential diagnosis.

1. Caudal regression syndrome- Characterized by the absence of sacrum, defects of the variable portion of lumbar spines associated with anomalies from different organs.
2. VACTREL- Consists of (V) = Non random association of malformation including vertebral abnormalities; (A) = anal atresia; (C) = cardiac (heart) defects; (T) = tracheo esophageal fistula; (R) = renal (kidney) abnormalities; and (L) = (other) limb abnormalities.

Signs and Symptoms

Characterised by partial or complete fusion of lower legs. Severity is highly variable. Some infants have only one femur or have two femurs within one shaft of the skin. Some have one foot/no foot or both feet which may be rotated so that back of the foot is facing forward. In my case, two femurs with one shaft of skin and two rotated feet were present.

There are some urogenital abnormalities like the absence of one/both kidneys, polycystic kidney, absent bladder, imperforate anus, and lower part of the large intestine (rectum) not developed. Severe oligohydramnios is also present.

PATHOLOGICAL DISCUSSION

The aetiology of sirenomelia is unknown. It believes that both environmental and genetic factors may play a role. Sirenomelia disorder gene carries a person who may not have developed sirenomelia unless otherwise this particular gene is triggered or activated under some circumstances such as a particular environmental factor. A teratogen is any substance that can disrupt the development of an embryo. Although molecular defects resulting in sirenomelia remain unclear, two main pathogenic hypotheses are probably responsible for the disease.

In some individuals, sirenomelia develops as the result of irregularities in the early development of the blood circulating system (Defective vascular system development) within the embryo. Some affected individuals have found a single large artery arising from the higher part of the abdominal aorta in the abdominal cavity without the usual two arteries that normally branch at the lower part of the aorta which carries blood to the tail (caudal) end of the embryo. A single artery is present (called a “steal” vessel) since it essentially steals blood from the lower portion of the embryo. As a result of this abnormal routed blood flow, the steal vessels also divert nutrients away from the blood-deprived portion of the embryo. Thus the ‘steal’ vessel redirects the blood flow to the placenta without ever reaching the tail (caudal) end of the embryo. As consequence, this caudal area is underdeveloped so that tissues and organs dependent upon them for nutrient supply fail to develop, and are malformed in some incomplete stage. It varies from case to case depending upon the arterial supply. This outcome results in the lower limb bud of the embryo failing to divide into two legs. The underlying reason for these irregularities is unknown.

Sirenomelia is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function. In literature, approximately 300 cases [3] are reported worldwide. In most cases, the diagnosis was done after birth. In the antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high-resolution or colour Doppler sonography.[4,5] The condition is usually incompatible with life due to visceral abnormalities, especially that of the renal system.

Diagnosis

Diagnosis of sirenomelia can be made prenatally, mostly during the second trimester, by foetal ultrasound. A foetal ultrasound can detect some of the defects associated with sirenomelia.

Management

Sirenomelia is a rare and lethal congenital anomaly. If antenatally diagnosed, termination should be done. However,
prevention is possible and it should be the goal. A regular antenatal check-up is mandatory. Evaluating the blood glucose level in the preconception period and the first trimester with optimum maternal blood glucose level in the preconception period and the first trimester should be maintained to prevent this anomaly.

After delivery, if the baby is alive then treatment may require a team of specialists like paediatricians, surgeons, cardiologists, orthopedicians, nephrologists and other health care personnel to systematically and comprehensively plan for the affected child.

Sirenomelia is usually fatal in the newborn period despite treatment.

**DISCUSSION**

In sirenomelia, there are a wide range of physical malformations and the specific findings can vary greatly from one individual to another.

In a comparison study, an increase in sirenomelia prevalence with maternal age less than 20 years and more than 40 yrs was statistically significant. (Kallen et al, 1992). The proportion of twinning was 9 %, higher than the 1 % expected.

The proportion of cases born alive, premature, and weighing less than 2,500 g were 47 %, 71.2 %, and 88.2 %, respectively. In my case, the patient’s age was 31 yrs, single pregnancy with premature delivery and 2085 gm. weight.

A more adequate classification of sirenomelia is that of Stocker and Heifetz [1987] in which seven types are defined: I- all thigh and leg bones present; II - single fibula; III- absent fibulae; IV- partially fused femurs, fused fibulae; V- partially fused femurs, absent fibulae; VI- single femur, single tibia; VII- single femur, absent tibiae. In my case, it was type I/II. Because the upper part (Thigh) was fused, the lower part was free, with a single fibula.

Zakin et al. (2005) have shown that the lack of bone morphogenetic protein 7 (Bmp 7) in combination with half a dose or complete loss of twisted gastrulation (Tsg) protein causes sirenomelia in mice. A genetic study could not be done in my case due to the refusal of its parents.

One-fifth of published sirenomelia cases were delivered to diabetic mothers, the offspring of whom were reported to have a prevalence of one in 200 births for a sirenomelia/caudal regression (CRS) infant (Gurukan et al., 1996; Martínez-Frias et al., 1998a; Al-Haggag et al., 2010)]. In my case, mother’s blood sugar level was within the normal limit. However, in a 15-year pathology series, Bruce et al. (2009) found a history of maternal diabetes in three out of nine CRS cases, but in none of six cases of sirenomelia.

Association of sirenomelia/CRS with component anomalies of the Potter sequence is expected due to the an/oligohydramnios produced by the usually associated severe and lethal renal /dysgenesis (Savader et al., 1989; Al-Haggag et al., 2010). A typical Potter facie was present in my case.

Another potential similarity of sirenomelia/CRS with VATER association is the imperforate anus, the A in the VATER acronym, which Duhamel (1961) proposed as the mildest end of the CRS which was also present in my case.
CONCLUSIONS

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed termination should be done. However, prevention is possible and should be the goal. Regular antenatal checkups always maintain the optimum maternal blood glucose level in the preconception period and in the first trimester to prevent the sirenomelia.

REFERENCES


