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Case Report

Mermaid baby: Fairy tale in practical

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Abstract:

Sirenomelia, also called Mermaid syndrome is a rare and fatal anomaly which is characterized by a single lower extremity featuring a Mermaid like appearance where head and trunk is like humans and lower extremity is like tail of a fish. It is associated with other gastro-intestinal, genito-urinary and other organ abnormalities. Most patients die shortly after birth. The exact cause of Sirenomelia is unknown. Here we present a newborn baby with Sirenomelia. Mother had bad obstetric history, antenatal ultrasonography revealed severe oligohydramnios and renal agenesis. On examination there was dysmorphic facies, fusion of lower limbs, single leg with two foot and ten toes. There was no identical external genitalia and anus.

Key words:

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Introduction:

A single lower extremity is a characteristic of the deadly defect known as Sirenomelia, commonly called Mermaid syndrome featuring a Mermaid like appearance where head and trunk is like humans and lower extremity is like tail of a fish.[1] This disease is fatal because it is associated with number of the congenital defects.[2]Mermaids are often referred to as Sirens according to the Greek Mythology. Siren is a creature with the body of a bird and the head of a human.[3] Over time the Siren was described as Mermaid in folklore who is an aquatic creature with the head and upper body of a female human and tail of a fish.[1] It is a fatal congenital defect that is extremely rare and characterized by the apparent merging both lower limbs into a single limb. It is connected to other birth defects like thoraco-lumber spinal anomalies, sacrococcygeal agenesis, genito-urinary and anorectal atresia.

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The earliest evidence for the Mermaid syndrome was traced back to the 16th century.[1,2] It was first described by Rochesus in 1542 and Palfyn in 1953.[2] Sirenomelia affects 0.8-1 case /100,000 live births, with a male to female ratio 3:1.[3] This syndrome has a substantial correlation to maternal Diabetes mellitus with a relative risk 1:200-500 and 22% fetuses having mother with the disease. [4,5] Intake of haloperidol, cadmium, lead, vitamin-A and vascular hypoperfusion may be causative factors.[6]

Case report:

A newborn baby, weighing 1.8 kg, 2nd issue of non-consanguineous parents, got admitted into the NICU of a tertiary medical college hospital with the complaints of not cried after birth, severe respiratory distress and multiple congenital anomalies. The baby was delivered by LUCS at 35 weeks of gestation due to less fetal movement.

Mother, 28 years old lady was on regular antenatal checkup. There was apparently no history of fever, rash, hypertension, diabetes mellitus or any other chronic ailment and her health condition was apparently good. She did not take any offending drugs during pregnancy. Mother had bad obstetric history (first issue-IUFD). In this pregnancy, ultrasonography of pregnancy profile with anomaly scan showed severe oligohydramnios and kidneys, renal arteries, stomach and urinary bladder were not visualized.

On examination baby was lethargic, dyspneic, had a dysmorphic face with hypertelorism, flat nose, low set ear, lower limbs were fused which is flattened antero-posteriorly with dorsum of fused feet facing posteriorly and ten toes. There was a skin tag in the lower abdomen, external genitalia, urethral and anal opening were absent. A small sacral pit was present in the back. The baby died three hours after admission. We failed to do any investigations because family did not give permission. So further evaluation was not possible. The parents were counselled to follow-up for genetic counseling during next pregnancy.

Figure-1

Figure-2,

Figure-3

Discussion:

With normal fetal karyotyping, Sirenomelia is an unusual disorder.[7] Sirenomelia anomalies are considered the most severe manifestations of Caudal regression syndrome.[2] The main characteristics of Sirenomelia include fusion of the lower extremities, a single umbilical artery and a persisting vitelline artery.[8] The pathophysiology of Sirenomelia includes approximation and merging of the hind limbs beginning from the posterior side. This occurs if the components in between them, such as midline structures like the urogenital sinus and cloaca on ventral side, and somites and neural tube on the dorsal side do not develop properly and resulting in dysmorpology.[9]

The vascular malformations seen in Sirenomelia are caused by a disruption in the development of the vitelline and umbilical arteries. The most frequent defect is single umbilical artery with an aberrant origin that branches off high after the celiac branch from abdominal aorta, narrows abnormally and lacks circulation to the lower limbs, kidneys, urogenital tract and large intestine.[7,9,10]

The vascular steal phenomenon and the blastogenic hypothesis are the two primary pathogenic hypotheses based on several clinical research describing the potential mechanism of Sirenomelia. The two mechanical ideas include medial compression by overextension of the neural tube and lateral compression of the caudal body by membrane folds.[9]

Sirenomelia is characterized by a number of visceral abnormalities, including entire renal agenesis, ectopic renal tissues, nonexistent external genitalia or genital tag of tissue and undamaged gonads. Among gastrointestinal anomalies are a blind terminating colon and imperforate anus. The other related anomalies include malformed vertebrae, hemivertebrae and sacral agenesis.[11]

Antenatal high-resolution ultrasonography can diagnose Sirenomelia by detecting severe oligohydramnios, renal agenesis and other defects.[1] Distinctive features of Sirenomelia can be easily seen on prenatal MRI, even when in oligohydramnios is present.[11]

Sirenomelia in babies exceedingly difficult to treat and outcome is poor. Only a very small percentage of patients survive past the neonatal period. For anorectal and genitourinary abnormalities as well as limb reconstruction in the babies who survive, extensive procedures are required.

Genetic counseling of the parents is also very important because recurrence risk is 3% -5%.[12]

Conclusion:

Due to pulmonary hypoplasia, renal agenesis and other complicated birth anomalies, Sirenomelia is fatal within a few days to hours. Different studies have demonstrated a significant correlation between Sirenomelia and maternal diabetes, while exact cause is still unknown. To

identify the molecular level of defects of this fatal disease, more and more researches are required. Early diagnosis is crucial so that the decision of termination of pregnancy can be made.

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