



Mermaid syndrome, Sirenomelia: A case report and review of literature

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ABSTRACT

Sirenomelia is a rare anomaly of caudal region of the body presented with fusion of lower limbs in which Genito-urinary, Gastrointestinal, Cardiovascular and Neural tube anomalies are found in most cases. We introduce a case of severe oligohydramnios, which had characteristic aspects of Sirenomelia at the time of delivery and review the pertinent articles. All in all it is our aim to present another case of sirenomelia to add to existing knowledge and data about the condition in order to make an earlier diagnosis and put an end to pregnancy safely in advance.

Introduction

Sirenomelia or Mermaid syndrome was originally stated by Rocheas in 1542 and Palfya in 1553 and named after the mythical Greek Siren.^{1,2,3} For the first time, Homer explained in the tempting singing creatures that seduced sailors to their deaths.⁴ Historical texts of around 2500 BC found in several European countries included references to female hybrids.⁴ It occurs about 1 in 100,000 live births.⁵ The most striking yet inconstant view of this malformative disorder is the complete or partial fusion of the lower limbs. It is a congenital anomaly including renal agenesis, ambiguous external genitalia, imperforate anus and blind intestinal loop.^{6,7} Occasionally

double inferior Vena cava and angiomatous lumbosacral myelocystocele are reported as well.^{8,9} Most of the Sirenomelia come to an end as stillbirth.⁶ Only a few numbers of whom have cited to be alive after delivery.^{10,11} Interestingly it has been said that there is an association with maternal diabetes.^{1,12} Hence we aim to present a fetus with features of mermaid syndrome in our department for first time and review proper articles.

Case presentation

A 34 years old primigravid pregnant woman at 25 weeks of gestation is counseled to terminate her pregnancy because of marked

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oligohydramnios that was incompatible with life. She had undergone an Ultrasound examination at 10 weeks gestation without apparently any reported abnormality in fetus. There were no medical or obstetric risk factors. There was no personal or family history for Diabetes Mellitus or congenital anomalies in relatives.

Routine Ultrasound examination, at 24 weeks of gestation revealed a live single fetus corresponding to 21 weeks gestation, weight: 270 gram. There were no single measurable pocket of amniotic fluid (severe Oligohydramnios); moreover, fetal kidneys, urinary bladder and stomach were not visualized.

A repeat Ultrasound was done in another institute and it turned out to be quite dextrocardia and left diaphragmatic hernia. Since there was severe oligohydramnios, the sonographer was not able to precisely assess both lower limbs and lumbar spines. Eventually after couple's consent termination is done at 25 weeks.

During autopsy procedure, fusion of the whole lower limbs from the hips to the ankles was founded out (Figure-1). In addition, it was realized syndactyly of the left foot with soft tissue union of the first and second toes.

The head was compressed and showed low set ears.

The face displayed bilateral complete lip and palate clefts. The lumbar spine showed a defect at the level of $L_3 - L_4$. There was no anal opening and no discernable genitalia.

The umbilical stump revealed just one artery and one vein. Complete absence of genitourinary system was evident. Kidneys, ureters and bladder were absent too. There was no stomach, and the midgut loop was blindly ended. The heart was located in the right side of the chest cavity (dextrocardia) (Figure-2) The lungs were non-flatted. The mentioned findings

were consistent with the diagnosis of sirenomelia.



Figure 1. Mermaid fetus, showing fused lower limbs and lip and palate clefts

Microscopic examination revealed just partially autolysed organs and generally not special pathologic changes.



Figure 2. Mermaid fetus, Dextrocardia

Discussion

Sirenomelia is a rare and lethal congenital anomaly with unknown etiology showing abnormal development of the caudal region of the body involving varying degrees of fusion of the lower limbs with or without bony defects (Table-1).

Other visceral defects such as hypoplastic lungs, cardiac agenesis, absent genitalia, digestive defects, absent kidney and bladder,

Table1. Antenatal cases of mermaid syndrome diagnosed in the first trimester

| Author | Year of publication | Number of references |
|--|---------------------|----------------------|
| Duhamel B | 1961 | 26 |
| Von Lennep E | 1985 | 19 |
| Steven RE, Hibelink DR | 1986 | 17,18 |
| Stocker JT | 1987 | 27 |
| Sirtori M | 1989 | 21 |
| Tang TT, Rodriguez JI | 1991 | 13,14 |
| Sarpong S, Kallen B | 1992 | 20,5 |
| Lynch SA | 1995 | 24 |
| Jaiyesimi F | 1998 | 23 |
| Aslan H | 2001 | 12 |
| Taori KB , Stanton MP | 2002 | 7,11 |
| Browne M | 2004 | 9 |
| Pinette MG | 2005 | 15 |
| Bakhtar O, Romano S, Vankeirsblick j, Ladure H | 2006 | 22,4,2,3 |
| Akbayir O | 2008 | 29 |
| Sikandar R | 2009 | 1 |
| Fadhlaoui A | 2010 | 25 |
| Ugwa RO | 2011 | 6 |
| Our current case | 2012 | - |

vertebral and central nervous system defect are also reported.^{13, 14} The poor prognosis is usually forecasted depending on some kind of the visceral anomalies. Death usually stems from renal agenesis or dysgenesis¹⁵, which there likely seems to be happened to our case.

Risk factors

Many different kinds of theories have been suggested such as maternal diabetes^{6, 7, 8, 16}; however, our mother was not known to be diabetic. Genetic and environmental factors are also noted.^{7, 17} Hibelink et al pointed out that teratogenic agents like cadmium element and lead may cause sirenomelia in the golden hamster.¹⁸ The likely potential teratogenic effect of vitamin A is mentioned by Vonlennep et al¹⁹ and cocaine²⁰ or irradiation exposure²¹ as well. Association of the sirenomelia with new reproductive technologies, namely with ICSI (Intra Cytoplasmic Sperm Injection) was also described.²²

Besides, it has been reported in twins. Reports indicate a 100–150 times higher incidence in monozygotic twins relative to dizygotic twins or singletons.²³ In our patient, no twin gestation or family history of twinning was found out.

Another huge concern is genetic transmission. Lynch *et al*²⁴ realized an autosomal type of caudal dysgenesis, but no chromosomal defects are discovered in mermaid syndrome. This was an encouraging view for our mother to have sort of genetic consultation. Genetic counseling should even be suggested because the mentioned risk of recurrence is fairly 3–5%.²⁵ All abnormalities in our case like imperforate anus, a neural tube defect, limbs abnormality, cardiac dextrocardia and renal agenesis are frankly compatible with concept of sirenomelia in other similar related articles as well.²⁵

Pathogenesis

The pathogenesis of this syndrome has been discussed over time. Numerous theories have

been proposed to explain its origin. Five pathogenetic theories of sirenomelia are described:

1. An embryological insult¹⁵
2. Vascular steal theory¹⁷
3. As part of the caudal regression syndrome (CRS)²⁶
4. As part of the VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities)²³
5. External forces acting on the caudal extremity.²⁵

From an embryological point of view, the sirenomelia sequence comes from an 'caudal mesoderm damage taking place between days 28–32 of fetal life (first theory).¹⁵ Therefore there are renal agenesis, lack of genital organs, imperforated anus, vertebral dysgenesis, and lower limb atrophy.²⁵

Second theory, Sirenomelia is probably a consequence of an abnormal blood supply of caudal parts of the embryo. A diverged abnormal vessel carries blood through the umbilical cord into the placenta. The vessel "steals" blood from the structures located below its origin from the tissues of fetal caudal part.¹⁶

A third theory regards sirenomelia as part of the caudal regression syndrome (CRS), a rare congenital anomaly described by Duhamel²⁶; including sacrum dysgenesis, spinal cord defect, urinary incontinence and misplaced lower limbs. Some inconstant features are renal dysgenesis and imperforate anus.

According to the fourth theory syndrome in the literature, sirenomelia could be a member of the VACTERL syndrome. VACTERL syndrome consists of vertebral, anal, cardiovascular, trachea-esophageal fistula, renal and limb dysgenesis. A major overlap has been realized in the phenotypic features of sirenomelia and VACTERL.²³

A fifth theory is the pressure theory stating hypoplasia of caudal extremity is owing to

external forces.²⁵ However not accepted on the whole.²⁵ Other less appropriate theories have also been proposed. Yet the overlap in these syndromes/theories is a matter of debate in the scientific world as to the uniqueness or varieties of these syndromes.

Classification

Stocker and Heifetz²⁷ classified the sirenomelia sequence into 7 types as below:

1. All thigh and leg bones are present
2. Single fibula
3. Absent fibula
4. partially fused femurs fused fibulae
5. partially fused femurs
6. single femur single tibia
7. single femur absent tibia

We did not have any radiographs of the lower limbs, but our case turned out to be pretty type six.

Diagnosis

It has been reported the possible first trimester diagnosis of mermaid fetus.³ Although the pregnant mother was supervised, it didn't take place in our case. The use of color Doppler imaging and MRI is claimed to have beneficial influence on in advance assessment.^{1, 2, 28}

Prognosis

Sirenomelia is fatal in most cases because of the characteristic pulmonary hypoplasia and renal agenesis. In children borned alive death will occur rather within a couple of days following birth²⁷. Presence of kidney is mandatory to survive. Murphy et al²⁴ reported a survived sirenomelia neonate. Laparotomy and colostomy were done, and eventually lower extremities were separated. It stands to reason that its managing is tough and rather costly with unforeseeable results in the case described by Stanton.¹¹ The patient had five interventions

between the age of 15 days and 4 years (she has been bedridden and dependent).

Conclusion

Sirenomelia is a kind of scarce and deadly congenital malformation. As far as we are concerned it is pretty much uncommon condition, so giving elaborated information may provide clinicians with a great chance to terminate the pregnancy in advance much more safely.

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Conflict of Interest

None declared.

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