

Laporan Kasus

Sirenomelia (Mermaid Syndrome)

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Abstract

Objective: To report a rare fetal abnormalities case

Method: Case report

Case: Reported case of a 37-year-old patient came to the emergency room at RSIA Siti Hawa Padang with complaints of inpartu signs in the last 8 hours before entering the hospital. After the examination, the diagnosis was G3P2A0L3 35-36 weeks of preterm parturient active phase of first stage + twice previous CS + breech presentation. Then, the patient was planned for an emergency CS at 07.50 am. Born babies with genitalia form was not identified, BW: 2030 grams, BH: 30 cm, Scoring Apgar: 3/5, there are congenital abnormalities in the lower extremity of the baby like mermaid form. After observing the baby's room, the baby was declared dead at 09.50 am.

Conclusion: Sirenomelia is a rare congenital defect that has a fatal impact, characterized by varying degrees of fusion in the lower limbs, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary and anorectal atresia. Because of the poor prognosis, management of sirenomelia will be very difficult with unexpected results.

Key words : Fetal abnormalities, Sirenomelia, Mermaid syndrome.

Sirenomelia (Sindroma Mermaid)

Abstrak

Tujuan: Melaporkan sebuah kasus jarang tentang kelainan janin

Metode: Laporan kasus

Kasus: Pasien 37 tahun datang sendiri ke IGD RSIA Siti Hawa Padang dengan keluhan nyeri pinggang menjalar ke ari-ari semakin lama semakin kuat dan sering sejak 8 jam sebelum masuk rumah sakit. Setelah dilakukan pemeriksaan ditegakkan diagnosis G3P2A0H3 parturien preterm 35-36 minggu kala I fase aktif + bekas SC 2x + letak sungsang. Kemudian, pasien direncanakan untuk dilakukan SC emergensi pukul 07.50 WIB. Dilahirkan bayi dengan JK : tidak teridentifikasi, BB : 2030 gram, PB : 30 cm, A/S : 3/5, terdapat kelainan kongenital pada ekstermitas bawah bayi. Setelah dilakukan observasi di ruang bayi, kemudian bayi dinyatakan meninggal pukul 09.50 WIB.

Kesimpulan: Sirenomelia adalah cacat bawaan yang jarang dan menimbulkan dampak yang fatal, ditandai dengan berbagai tingkat fusi pada tungkai bawah, anomali tulang belakang thoracolumbar, agenesis sacrococcygeal, genitourinari dan atresia anorektal. Karena memiliki prognosis yang buruk, pengelolaan sirenomelia akan menjadi sangat sulit dengan hasil yang tidak terduga.

Kata Kunci : Kelainan janin, Sirenomelia, Mermaid syndrome.

Introduction

Recent literature shows the improvement achieved in the detection of fetal abnormalities as early as possible by using ultrasound to assess fetal brain malformations generally performed at 19-21 weeks' gestation.¹ For assessment of the fetal brain, assessment of second trimester anomalies includes the axial plane, such as; transthalamic, transventricular and transcerebellar have become standard approaches.^{2,3} However, several major developments occur in the second half of the pregnancy period, including neuronal proliferation, migration and organization, and the same applies to acquired lesions, such as bleeding and tumors that usually occur in advanced gestational age.^{3,4} Therefore, although it is possible to detect most fetal brain anomalies in the second trimester, impaired migration, proliferation and organization, and acquired lesions become clear only in the third trimester.⁵

In Greek mythology, Sirens are three creatures with the head of a woman and the body of a bird from a wing down. Over time, the female bird was described as a more aquatic creature and finally with a mermaid-like appearance.⁶ Sirenomelia is a rare and fatal congenital disorder characterized by varying degrees of fusion of the lower limbs, thoracolumbar spinal anomalies, sacrococcygeal agents, genitourinary and anorectal atresia which is characterized by varying degrees of fusion of the lower limbs, thoracolumbar spinal anomalies, sacrococcygeal agents, genitourinary and anorectal atresia, as well as Woffian or Mullerian duct agenesis, abnormalities in the gonads are usually not found. As a result of the oligohydramnios produced, babies with this disorder are most often found to have Potter facies and pulmonary hypoplasia. The rarity of this case is clear from the fact that many obstetricians may not find a case of sirenomelia.⁷ Sirenomelia "Mermaid

Syndrome" was found to be around 1: 1.00000 live births and was fatal on the first day of life, due to abnormal kidney complications, urinary system and anorectal development.⁸

First described by Rocheus in 1542 and Palfyn in 1553. This syndrome has a strong relationship (22%) with diabetes mellitus in the mother and a relative risk of 1: 200-250. Intake of haloperidol, cadmium, vitamin A, vascular perfusion can be a number of causative factors.⁹ The facial deformity commonly found in sirenomelia babies is known as Potter's facies, which includes large and low ears, protruding epicanthic folds, hypertension, a flat nose and a shrunken chin. The Potter facies feature is present. When the features of Potter's facies were combined with oligohydramnios and hypoplasia of the lungs known as Potter's syndrome was present. The right thumb was hypoplastic, which was also reported previously. Stocker and Heifetz classified sirenomelia babies from Type I to Type VII based on the presence or absence of bone in the lower extremities.¹⁰

Sirenomelia is fatal in most cases due to characteristic pulmonary hypoplasia and renal agenesis. About 50% of children are born alive after eight or nine months of pregnancy. Death occurs within five days of birth. Postnatal management requires the presence of kidneys, even if dysgenesis. Murphy et al reported one case in which a child born with sirenomelia survived; the infant was neurologically normal and had lower limb fusion, imperforate anus, colonic atresia, bilateral pelvic kidneys with renal dysplasia and sacral dysplasia, and genital abnormalities. Laparotomy and colostomy were performed, and finally a plan for separation of the lower extremities. Clarke et al. Reported on infants aged three months whose sirenomelia was diagnosed prenatally. The baby is neurologically normal and has lower limb fusion with associated renal dysplasia, imperforate anus, pelvic and sacral dysplasia, and genital abnormalities.

Anomalies of the infant compatible with life and surgical separation of the lower limbs were planned. Managing sirenomelia is difficult and quite expensive, requiring multiple interventions, the results of which are unpredictable (in the case described by Stanton¹, the patient had five interventions between the ages of 15 days and 4 years; he was bedridden and dependent).¹¹

Case Report

A 37-year-old patient came to the emergency room at RSIA Siti Hawa Padang with complaints of inpartu signs in the last 8 hours before entering the hospital. After the examination, the diagnosis was G3P2A0L3 35-36 weeks of preterm parturient active phase of first stage + twice previous CS + breech presentation. Previously, the patient had 2x pregnancy controls to an obstetrician at 2 months and 3 months of gestation due to complaints of vaginal bleeding. An ultrasound examination was performed at 2 months of gestation, and no abnormalities were found. The next control was carried out at the primary health care until 6 months of gestation. However, in the last 7 months of gestation, the patient has never had pregnancy control due to the COVID-19 pandemic.

There is no history of heart disease, lung, liver, kidney, diabetes, hypertension and allergies. The patient has never used any other medicines besides pregnancy vitamins. Then, the patient was planned for an emergency CS at 07.50 am. Born babies with genitalia form was not identified, BW: 2030 grams, BH: 30 cm, Scoring Apgar: 3/5, there are congenital abnormalities in the lower extremity of the baby like mermaid form. After observing the baby's room, the baby was declared dead at 09.50 am.

Discussion

Sirenomelia is reported to be more common

in one of 2 monozygotic twins. Most of these cases were stillborn or soon after birth. Sirenomelia is a rare inherited disorder, so far around 300 cases have been reported in the literature. Death is usually due to renal agenesis, Swader reported the first survivor in 1989.^{12,13}

The exact cause of sirenomelia is unknown. Researchers believe that both environmental and genetic factors can play a role in the development of disorders that occur. Most of the cases seem to occur randomly for no apparent reason (sporadically) indicating environmental factors or new mutations.^{13,14} Most likely sirenomelia is multifactorial which means that several different factors can play a causative role. In addition, different genetic factors can contribute to disorders in different people (genetic heterogeneity).¹⁵

Duhamel in 1961 stated that sirenomelia and anorectal malformation represented two extremes of one comprehensive syndrome arising from embryonal defects in the formation of the caudal region called the Caudal Regression Syndrome.¹⁶ Stevenson et al, dissected abdominal arteries from 11 cases of sirenomelia and showed a pattern of vascular abnormalities that explains the defects that are usually found in this condition. They show a single large artery (steal vessel) arising from a high intraabdominal cavity and diverting nutrients from the tail end of the embryo.¹⁷ There is a strong relationship between sirenomelia and diabetes in the mother, where up to 22% of fetuses with this anomaly are known to have diabetic mothers. There is a strong relationship with maternal diabetes where the relative risk is 1:200-250. Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors.^{17,18}

Stocker and Heifetz classify sirenomelia in seven types, with the presence or absence of bone in the lower limbs.¹⁸

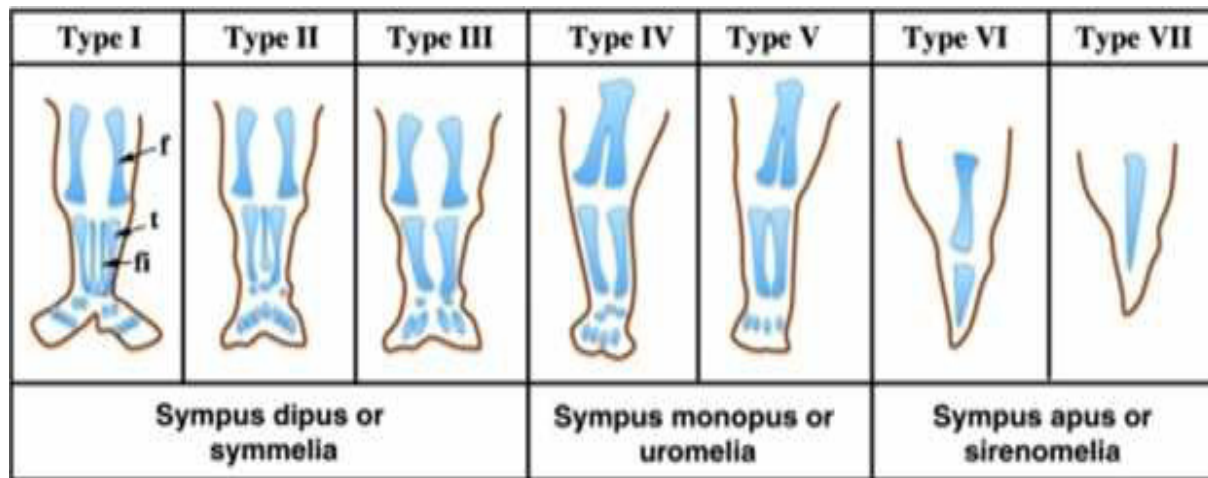


Figure 1 Classification of Sirenomelia.¹⁸

- I) All the femur and lower legs formed
- II) Fibula fused
- III) There is no fibula
- IV) Femur partially fused, fibula fused
- V) A part of the femur is fused
- VI) Femur and tibia fused
- VII) Femur fused, tibia is not formed

In this case, the diagnosis is made after birth. In the antenatal period, sirenomelia can be diagnosed most quickly at 13 weeks of gestation using color Doppler sonography. Facial abnormalities are usually found in sirenomelia infants known as Potter's facies which include large and low ears, prominent epicanthic folds, hypertelorism, flat nose and shrinking chin. When Potter's facies are combined with oligohydramnios and pulmonary hypoplasia, it is known as Potter syndrome.^{19,20,21}

In the literature, diagnosis is generally suspected to be slower. The main warning sign is usually oligohydramnios, related to dysgenesis or urinary tract agents that appear from the end of the first trimester, when fetal urine excretion usually becomes significant in forming amniotic fluid. However, after the appearance of oligohydramnios, assessment of fetal morphology will become more

difficult. Van Zalen Sprock et al, reported 2 cases of diagnosis that could be diagnosed at the age of the first trimester of pregnancy.



Figure 2 Photos of Sirenomelia Baby in this Case.

One embryo is obtained with hygroma as a primary warning sign at 14 weeks of gestation, and the diagnosis of sirenomelia can be established accurately using transvaginal ultrasonography. In other cases, the diagnosis is suspected at 16 weeks of gestational age.²²

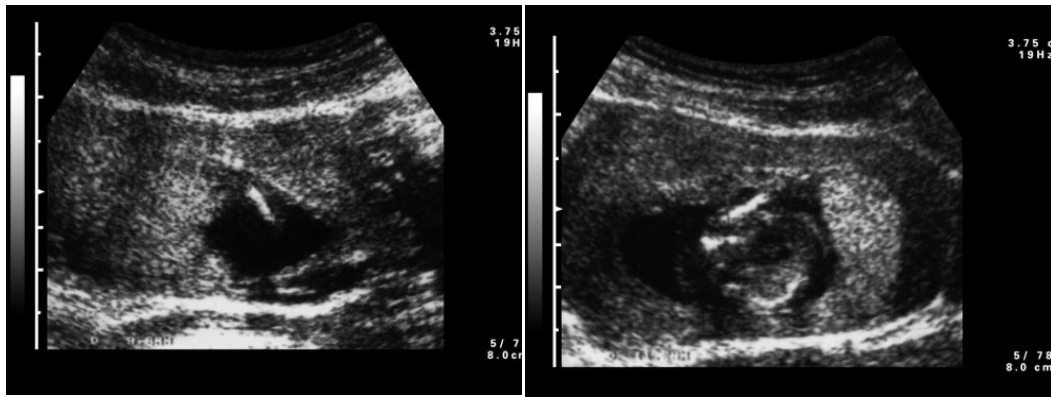


Figure 3 Views of the Lower Extremity of the Fetus and Aspect of the Pelvic Bone²³

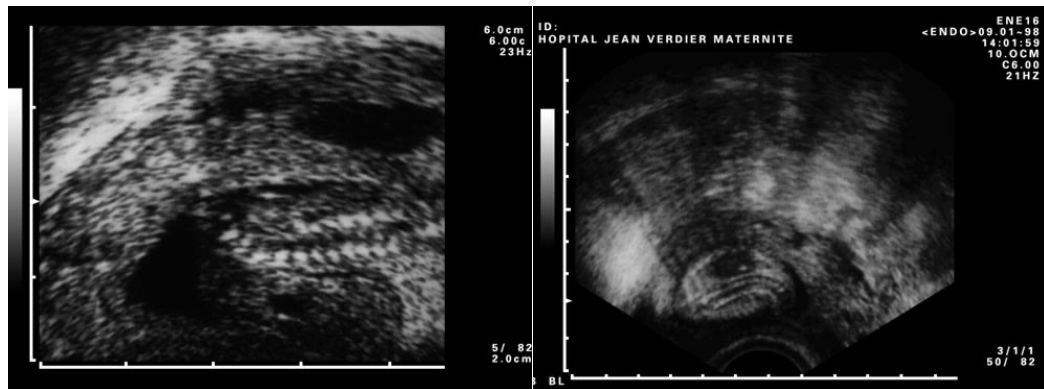


Figure 4 Views Through the Abdomen and the Pelvis Showing the Ultra-Sound Picture of the Lower Part of the Spine (Sacral Agenesis) And Hyperechoic BoSwel.²³

Sirenomelia may be suspected in a routine ultrasound scan at 12-13 weeks of gestation if all four extremities are assessed. In suspected cases, the examination should be completed by transvaginal ultrasound, which allows early diagnosis of sirenomelia before the appearance of oligohydramnios.²³

Renal anomalies, significant oligohydramnios and pulmonary hypoplasia occur simultaneously. Concurrent anomalies, such as anorectal defects, urinary tract defects, pelvic anomalies and external genitalia have a very poor prognosis and almost always cause very significant death or disability. The therapeutic decision depends on the gestational age at which the diagnosis of sirenomelia is performed, as well as the severity of the malformative syndrome and

the desires of the parents. Genetic counseling must be done in the next pregnancy, because the risk of recurrence from sirenomelia is estimated at 3-5%.²⁴ In this case, we know that the baby just died after several hours.

Conclusion

Sirenomelia is a rare congenital defect that has a fatal impact, characterized by varying degrees of fusion in the lower limbs, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary and anorectal atresia. Sirenomelia has long been considered the main form of caudal dysplasia sequences or caudal regression syndromes. The exact cause of sirenomelia is unknown. Researchers believe that environmental and

genetic factors can play a role. Most cases occur randomly for no apparent reason. Because it has a poor prognosis, management of sirenomelia will be very difficult with unexpected results.

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