Original Article

INTERNATIONAL JOURNAL OF CLINICAL PHARMACOKINETICS AND MEDICAL SCIENCES

Published by Pharma Springs Publication Journal Home Page: https://pharmasprings.com/ijcpms

Sirenomelia-mermaid syndrome, its types and causes

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Article History:

Received on: 03 Nov 2024
Revised on: 03 Dec 2024
Accepted on: 10 Dec 2024

Keywords:

Sirenomelia,
Mermaid Syndrome,
Types,
Causes,

Abstract

Sirenomelia, also known as mermaid syndrome, is a rare congenital abnormality characterized by the fusion of the lower limbs, giving the appearance of a mermaid's tail. This condition is extremely uncommon, with only a few hundred cases reported worldwide.Historically, sirenomelia was first described in the 16th century. It is classified into different types based on the degree of limb fusion and associated abnormalities. These include complete and partial sirenomelia, with the latter involving some level of limb separation.Signs and symptoms of sirenomelia include fused legs, malformed internal organs, and, in many cases, renal and gastrointestinal defects. The exact cause remains unclear, though it is believed to result from abnormal development during early embryonic stages.Risk factors include maternal diabetes, advanced maternal age, and certain genetic conditions. Diagnosis is typically made through prenatal imaging such as ultrasound.The outlook for individuals with sirenomelia is generally poor, as many affected fetuses do not survive to term. For those who do, the prognosis depends on the severity of associated organ malformations.

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eISSN: 2583-0953
DOI: https://doi.org/10.26452/ijcpms.v4i4.690

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INTRODUCTION

A medical disorder called "mermaid syndrome" causes a baby's legs to fuse. Mermaid syndrome is also known as:

Sequence of Sirenomelia

Syndrome of Sirenomelia

A relatively uncommon condition known as sirenomelous mermaid syndrome affects 0.8 to 1 kid out of every 100,000 births.

This disease is three times more common in male babies than in female babies [1].

HISTORY:

In 1542, sirenomelia was first documented. Otto Kampmeier identified the link between sirenomelia and a single umbilical artery in 1927. Prominent individuals. Few people with renal tissues that were still functional made it through the newborn stage [2].

Figure 1 Sirenomelia Syndrome

Classification:

The presence or lack of bones in the lower limb is used to categorize sirenomelia.
I) All thigh and lower leg bones are present;

II) The fibula is united

III) Absence of fibula

IV) Fused fibula and partially fused femur

V) Femur that is partially fused

VI) Joined tibia and femur

VII) Absent tibia, fused femur

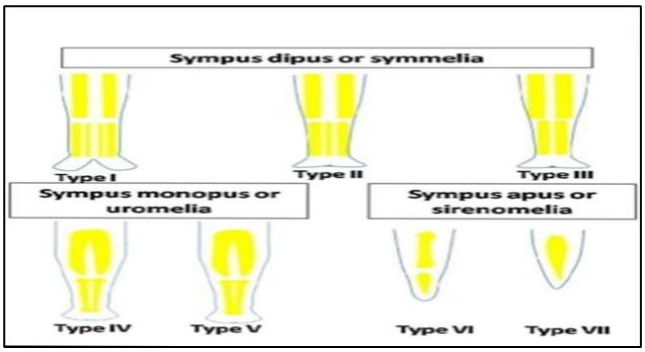

Figure 2 Sympus Dipus or Symmelia

The skeletal structure of the lower leg is used to classify sirenomelia; class I includes all bones and only the soft tissues are fused, whereas class VII includes only a fused femur. It has also been categorized as a type of caudal regression syndrome and an extended component of the VACTERL relationship [3].

SIGNS AND SYMPTOMS :

A variety of deformities, some potentially fatal and others less severe, can be present in babies with mermaid syndrome. A partial or total fusion of the lower limbs is the main feature, which could result in a baby having just one femur, the long bone at the front of the thigh. Additionally, babies may have two rotated feet or neither at all[4].

Other features of mermaid syndrome include:

Abnormalities that are absent or improperly developed in the bladder, kidneys, urethra, or anus genitals, both internal and external issues with the skeletal system and spine's development concerns with the abdominal wall, such as protruding intestines, heart troubles, and lung problems [5].

CAUSES :

The specific cause of mermaid syndrome is unknown to researchers and medical professionals. Nonetheless, it appears likely that several elements are involved. Given that most cases arise randomly and lack a clear etiology, novel mutations are expected to play a role in the illness. These can range from person to person. Perhaps the disease is triggered by a particular environmental factor, and some people are genetically predisposed or vulnerable to it. It seems that the early development of the blood circulatory system has gone incorrect in certain infants with mermaid syndrome. Why this occurs is still a mystery to scientists[6].

OUTLOOK FOR PEOPLE WITH SIRENOMELIA:

Outlook

Despite being extremely rare, mermaid syndrome frequently results in death. Despite treatment, the majority of babies with this illness are stillborn or pass away within a few days of delivery. Few newborns have made it past the newborn stage anywhere in the world.
Mermaid syndrome can occasionally be identified as early as 13 weeks of pregnancy (Trusted Source), and in these cases, some women may decide to terminate their pregnancy [7].
Milagros Cerron, a 2-year-old girl with mermaid syndrome, made her first steps following surgery, according to a 2006 BBC story. According to the same account, Tiffany Yorks, who was 17 at the time, was the only other person to survive years

following surgery. However, mermaid syndrome complications have since claimed the lives of both of these survivors.

Figure 3 Mermaid Syndrome

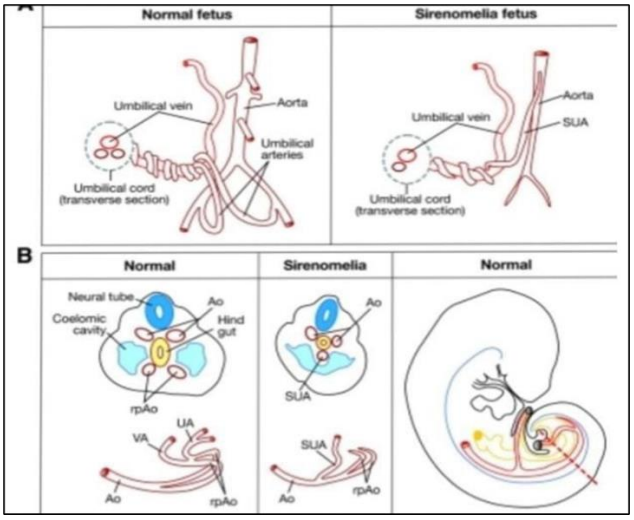

Figure 4 Difference between Normal and Sirenomelia fetus

RISK FACTORS:

The presence of diabetes in the biological mother, which affects 22% of fetuses with this disorder, is one potential risk factor for mermaid syndrome; it's possible that the mother's proper blood glucose management can lower the risk. Reliable Source exposure to chemicals known as teratogens, which raise the risk of congenital disabilities. Being an identical twin—of the 300 cases of mermaid syndrome that medical journals have documented, 15% are twins, most of whom are similar—the mother being under 20 years old; genetic variables being male since the condition is 2.7 times more likely to affect males than girls [8].

PATHOGENESIS:

It has been suggested that the pathophysiology of sickle cell disease is a vascular steal phenomenon in which a single, aberrant umbilical artery steals the lower limbs' blood supply. The diagnosis of sirenomelia is more likely than caudal regression if there are oligohydramnios.In this case, AMNIOINFUSION aids in defining the anatomy [9].

DIAGNOSIS:

It is usually made antenatal during the second semester by fetal ultrasonography.

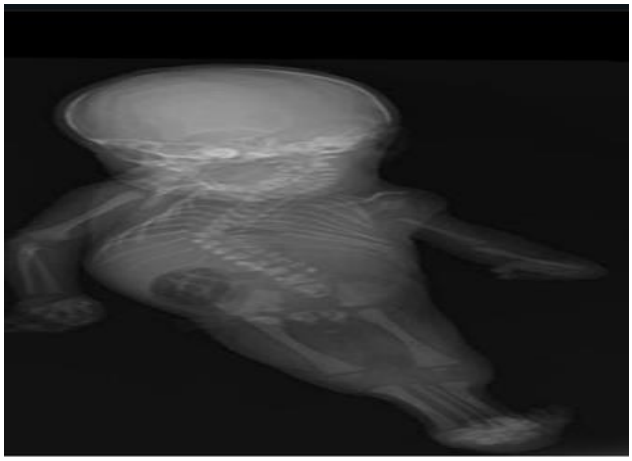

Figure 5 Ultrasonography of Sirenomelia Syndrome

TREATMENT:

A baby with mermaid syndrome typically requires treatment from several different medical specialties. The consequences of the syndrome are what the professionals rely on. Orthopedic surgeons have separated joined limbs. However, mermaid syndrome is usually fatal after the newborn period, even with treatment. After this age, children with mermaid syndrome frequently have brittle leg bones prone to breaking. Multiple procedures can be necessary for their lower body and other hereditary problems, like renal issues [10].

PREVENTION:

Prevention can be achieved and should be the aim. To prevent congenital anomalies, it is essential to maintain optimal maternal blood glucose levels during the preconception period and the first trimester through routine prenatal checkups.

CASE REPORT:

At 39 weeks and 5 days of gestation, a 27-year-old unbooked G3P1L1A1 who had previously experienced one live vaginal birth and one spontaneous abortion in the first trimester was

brought to the labor department due to abdominal pain. She came from a lower socioeconomic tribal group and had no history of previous prenatal treatment. Both prenatal and postpartum tobacco usage were documented. There was no known family history of genetic or congenital abnormalities, and the child was otherwise healthy [11].

Upon examination, her cephalic presentation and standard fetal heart rate indicated that she was in the second stage of labor. She gave birth to a 2.5 kg term baby who had several congenital disabilities. At 1', the Apgar score was 3; at 5 minutes, it was 0. Despite the neonatologist's attempts at resuscitation, the infant passed away within 30 minutes of birth. The newborn had a small chest, bilateral hypoplastic thumb, fused lower limbs (one foot and five toes), no external genitalia, an imperforate anus, and an umbilical cord with a single umbilical artery, according to the medical examination. Potter's face was also characterized by pronounced epicanthic folds, hypertelorism, a downward-curved nose, a receding chin, low-set soft dysplastic ears, and narrow, slit-like lips. The parents refused to have an autopsy. The mother's postpartum and intrapartum periods went smoothly [12].

Figure 6 Child Birth with Sirenomelia Syndrome

CONCLUSION:

For the detection of sirenomelia as well as other congenital abnormalities, intentional ultrasonography is crucial in high-risk situations such as maternal diabetes, oligohydramnios, and heavy metal exposure. The incidence of sickle cell disease has been linked to maternal diabetes. The sporadic aspect of this illness may be due to a mix of environmental and hereditary factors. Generally speaking, this illness is lethal during the prenatal stage, and the few survivors require interdisciplinary care.

ACKNOWLEDGEMENT

I want to thank the management from Saastra College of Pharmaceutical Education & Research, Near Varigonda Jwalamukhi Temple, Muthukur Road, Kakupalli, Nellore-524311, Andhra Pradesh, India, for their encouragement and support in completing this review work.

AUTHOR CONTRIBUTION

All authors made substantial contributions to the conception, design, acquisition, analysis, or interpretation of data for the work. They were involved in drafting the manuscript or revising it critically for important intellectual content. All authors gave final approval of the version to be published and agreed to be accountable for all aspects of the work, ensuring its accuracy and integrity.

Conflict of Interest

The authors declare no conflict of interest, financial or otherwise.

Funding Support

The authors declare that they have no funding for this study.

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