www.jmscr.igmpublication.org
Impact Factor (SJIF): 6.379
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v6i3.71

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

A Case Report: Sirenomelia

Authors
Jaswir Singh, Anuj Bansal, Ankur Jain
Corresponding Author
Ankur Jain
Address: S-4, Majithia Apartments, Majithia Enclave, Patiala
Email: *dr_ankur.jain@hotmail.com*

Abstract
Sirenomelia is a rare and lethal congenital anomaly with an incidence of one in 60,000 to 100,000 pregnancies .The characteristic features of sirenomelia is complete fusion of lower limbs and commonly associated with renal agenesis, absent external genitalia, gastrointestinal defects (blind ending colon and imperforate anus); small, absent, fused or poorly formed pelvic bones. The characteristic and distinguished anatomic findings in Caudal Regression Syndrome are presence of single umbilical, persistent vitelline artery. Birth defects in upper body sometimes occur and include abnormality in Ears, Lungs, Anus, Spine and Brain. Death of the child occurs most commonly due to underdeveloped and immature lungs caused due to oligohydroamnios.

Introduction

Sirenomelia Sequence or Mermaid Syndrome was originally described by Rocheus and Palfyn in 16th century and named after the mythical Greek sirens.[1,2] It is a congenital abnormality characterized by fused lower extremities or a single lower limb.[3,4] It is also very commonly associated with features like genitourinary and anorectal defects.[2,4] The prevalence of this syndrome is 0.1-0.25:10,000 in normal pregnancies .This condition is strongly associated with maternal diabetes with a relative risk of 1:250. Other factors associated are tobacco, alcohol consumption, genetic factors, heavy metal exposure to mother, more common in male child, being one twin of identical twin during gestation.

Case Report

A new born infant, born of a non-consanguineous marriage, to a known primigravida, non-complaint gestational diabetic mother, 24 years of age, presented to the department of Pediatrics at Government Medical College, Patiala, India, in November 2016 with Potter's face, complete fusion of the lower limbs from the perineum to ankle, imperforate anus, sacral agenesis, supernumerary toes, deformed left ear, with single umbilical artery and vein and rudimentary phallus with no urethral opening. Antenatal ultrasonography revealed renal agenesis and oligohydramnios.

Till the date of delivery, the pregnancy was uneventful except for the mother's poorly controlled diabetes mellitus. There was no history of any exposure of tobacco smoking, alcohol

consumption or any exposure to other drugs or toxic substances. She underwent normal vaginal delivery of a live, malformed male fetus of 1700gm. There was history of delayed cry at birth. Child developed severe respiratory distress in the form of increased respiratory rate, retractions. Resuscitation was given in the form of Bag and Mask ventilation and due to non-improving status, endotracheal intubation was done. However, the child deteriorated and expired two hours after birth.

Figure 1: Above pictures shows Potter's face, complete fusion of the lower limbs from the perineum to ankle, rudimentary phallus, supernumerary toes, deformed left ear in sirenomelic child.

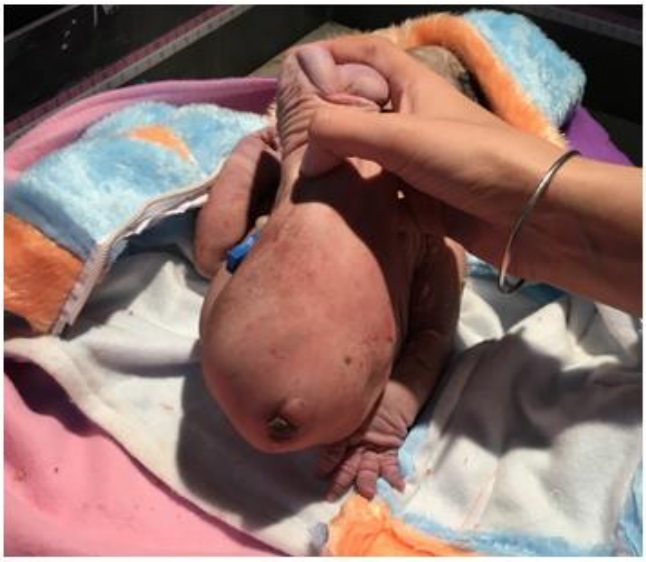

Figure 2: Above pictures shows imperforate anus in sirenomelic child.

Discussion

Anomalies observed in sirenomelia are described as the most severe form of Caudal Regression Syndrome.[5] Fusion of the lower extremities, presence of single umbilical and persistent vitelline artery are the major features of sirenomelia.[6] Sirenomelia in humans is thought to be associated with deficient Bmp signaling and excessive RA signaling in caudal region of body. According to clinical studies, there are two pathogenic hypothesis for sirenomelia: the vascular steal hypothesis and defective blastogenesis hypothesis. Vascular hypothesis[7]: deficient blood flow and nutrient supply to caudal mesoderm results in agenesis of midline structures, abnormal approximation and fusion of both lower limb fields. Defective blastogenesis hypothesis[8] states that teratogenic (retinoic acid, cadmium and cyclophosphamide) events during gastrulation stage results in such type of deformity. Maternal diabetes, alcohol consumption, tobacco use, heavy metal exposure, retinoic acid, other environmental and genetic factors are possible causative factors.

There are approximately 300 cases[5] reported in literature and about 13-15 cases are reported in India .It is a fatal disorder and the child dies within a day or two after the birth due to various abnormalities associated with Kidneys, Bladder, Lungs and Gastrointestinal system. Most of the cases are diagnosed after birth, but it can be diagnosed as early as 13 weeks antenatally by using colour doppler sonography[9,10]. There is a case of Tiffany Yorks, who in 1958 underwent surgery to separate her legs but she continues to suffer from issues of her immobility. Till date she is the longest surviving Sirenomelia patient.

As in our case, the facial abnormality, usually found in such cases is Potter facies which include large low set ears with deformed ear lobe (typically seen in our case), prominent epicanthal fold, flat nose, receding chin, hypertelorism. According to Stocker and Heifetz Sirenomelic infants are classified from Type I to TypeVII[11], according to presence or absence of bones in the lower limbs. Our case is probably of type I, based on external

examination.

Summary

Sirenomelia being lethal congenital anomaly should be terminated when diagnosed antenatally by USG. Maternal blood glucose monitoring, avoiding tobacco, alcohol, exposure to heavy metals, antenatal genetic examination should be done. Due to increase in trends of alcohol consumption, cigarette smoking, increased heavy metal exposure due to an increase in pollution among young woman can lead to increase in the incidence of Sirenomelia in future.

References

1. Van Keirsblick J, Cannie M, Robrechts C, de Ravel T, Dymarkowski S, Va den Bosch T, et al. First trimester diagnosis of Sirenomelia. Prenat Diagn 2006; 26: 684-8.
2. Schiesser M, Holzgreve W, Lapaire O, Willi N, Luthi H, et al. Sirenomelia, the mermaid syndrome-detection in the first trimester. Prenat Diagn 2003; 23: 493-5.
3. Ladure H, D"herve D, Loget P, Poulain P. Prenatal diagnosis of Sirenomelia. J Gynecol Obstet Biol Reprod (Paris) 2006; 35: 181-5.
4. Akbayir O, Gundorduk K, Sudolmus S, Gulkilik A, Ark C. First trimester diagnosis of Sirenomelia: a case report and review of the literature. Arch Gynecol Obstet 2008; 278: 589-92.
5. Duhamel B. From the Mermaid to Anal Imperforation: The syndrome of Caudal regression. Arch Dis Child. 1961;36:152–5.
6. Twickler D, Budorick N, Pretorius D, Grafe M, Currarino G. Caudal regression versus sirenomelia: Sonographic clues. J Ultrasound Med. 1993;12:323–30
7. Sadler TW, Rasmussen SA. Examining the evidence for vascular pathogenesis of selected birth defects. Am J Med Genet A. 2010;152A:2426–36
8. Duesterhoeft SM, Ernst LM, Siebert JR, Kapur RP. Five cases of caudal regression with an aberrant abdominal umbilical artery: Further support for a caudal regression-sirenomelia spectrum. Am J Med Genet A. 2007;143A:3175–84.
9. Vijayaraghavan SB, Amudha AP. High-resolution sonographic diagnosis of sirenomelia. J Ultrasound Med. 2006; 25:555–7.
10. Sahu L, Singh S, Gandhi G, Agarwal K. Sirenomelia: A case report with literature review. Int J Reprod Contracept Obstet Gynecol. 2013;2:430–2.
11. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol. 1987;10:7–50.