Case Report

Conjoined legs: Sirenomelia or caudal regression syndrome?

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Abstract

Presence of single umbilical persistent vitelline artery distinguishes sirenomelia from caudal regression syndrome. We report a case of a12-year-old boy who had bilateral umbilical arteries presented with fusion of both legs in the lower one third of leg. Both feet were rudimentary. The right foot had a valgus rocker-bottom deformity. All toes were present but rudimentary. The left foot showed absence of all toes. Physical examination showed left tibia vara. The chest evaluation in sitting revealed pigeon chest and elevated right shoulder. Posterior examination of the trunk showed thoracic scoliosis with convexity to right. The patient was operated and at 1 year followup the boy had two separate legs with a good aesthetic and functional results.

Key words: Caudal regression syndrome, ectromelia, sirenomelia, conjoined legs

Introduction

Sirenomelia was originally described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens.[1,2] It is a rare, lethal, congenital anomaly characterized by fusion of the lower extremities or a single lower limb[3,4] with single umbilical artery along with severe malformations of the gastrointestinal, genitourinary, cardiovascular, and Musculo skeletal systems.

Caudal regression syndrome (CRS) is a pathology caused by anomaly of spinal trunk "ending," and encompasses a wide range of anomalies of the hind end of the trunk, including partial agenesis of the thoracolumbosacral spine, associated pelvic deformities, imperforate anus, malformed genitalia, bilateral renal dysplasia or aplasia, pulmonary hypoplasia, and in the most severe deformities, extreme external rotation and fusion of the lower extremities (sirenomelia). It is also associated with femoral hypoplasia, clubbed feet, and flexion contractures of the lower extremities.[5] Kahilogullari *et al.*[6] has reported a case of CRS who was diagnosed at the age of 16.

Controversy exists in the literature regarding whether sirenomelia occurs as a separate entity or is an extreme form of CRS. The purpose of this report is to describe a patient with clinical symptoms that mimic and contrast CRS and sirenomelia.

Case Report

A 12-year-old boy presented with fusion of lower legs above the ankle joint. Bilateral femur and upper two-third of tibia and fibula were separated. There was fusion of both legs in the lower one third of leg [Figure 1]. The boy weighed 23 kgs at the time of admission. The parents were nonconsanguineous. There was no history of maternal diabetes and patient had a breach presentation with immediate birth cry. Both knees had flexion deformities (right: 10° and left: 25° as measured by 180° goniometer). Both legs were short (left shorter than right). Both feet were rudimentary. The right foot had a valgus rocker-bottom deformity. All toes were present but rudimentary. The left foot showed absence of all toes. Physical examination showed left tibia vara. The chest evaluation in sitting revealed pigeon chest and elevated right shoulder. There was expansion of 2, 3, and 3 cm in axilla, nipple, and xiphisternum levels, respectively. Posterior examination of the trunk showed thoracic scoliosis with convexity to right. He was having no other physical anomalies in the skull, face, and lumbar region.

Magnetic resonance imaging of spine showed no abnormality in soft tissue planes that is expected from CRS

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or sirenomelia-like blunted sharp ending of spinal cord or hypoplastic sacrum. Ultrasound of abdomen and pelvis ruled out any visceral anomalies. Color Doppler imaging for both lower limbs showed that anterior tibial, posterior tibial, and peroneal arteries were small in caliber and nontraceable throughout their extent but showed normal triphasic flow pattern in visualized segments. Bilateral external iliac, common femoral, superficial femoral, profunda femoris, and popliteal arteries were normal in caliber and blood flow.

The superficial and deep veins were competent in both sides and showed spontaneous phasic flow with adequate response to valsalva and augmentation. There was no evidence of deep venous thrombosis (DVT), arterial venous malformation (AVM) and aneurysm.

Operative procedure

A multidisciplinary surgical team, composed of a plastic surgeon, a vascular surgeon, and an orthopedic surgeon, achieved complete separation of the lower limbs. The typical zigzag incisions were marked between the limbs, Z-plasty of skin was performed [Figure 2] with independent vascular and nerve supplies. The vascular surgeon identified the anteriortibial, posterior tibial, and peroneal arteries on both the limbs. Intraoperatively, it was found that femurs, tibias, and fibulas were normally represented. Muscles seemed to be present and tonic. After separation, the left side tibia was found to be posterolaterally angulated for which a corrective osteotomy was done and fixed with K-wire to make the feet plantigrade [Figure 2]. Pulse oximeters were helpful during the surgical procedure to assess the viability of the separated limb. Wound closure was performed with split thickness skin grafting from anterolateral thigh. The skin-graft donor site was closed with absorbable sutures. A generous amount of antibiotic ointment was directly applied to the skin graft, and the incisions were followed by a nonadherent dressing material. Dressing was changed after 72 h. The boy had two separate legs with a good aesthetic and functional result [Figure 3].

Postoperative physiotherapy consisted of active movements after the skin grafts had settled in by 6 days. The child was instructed to massage the scars to soften them. Once the

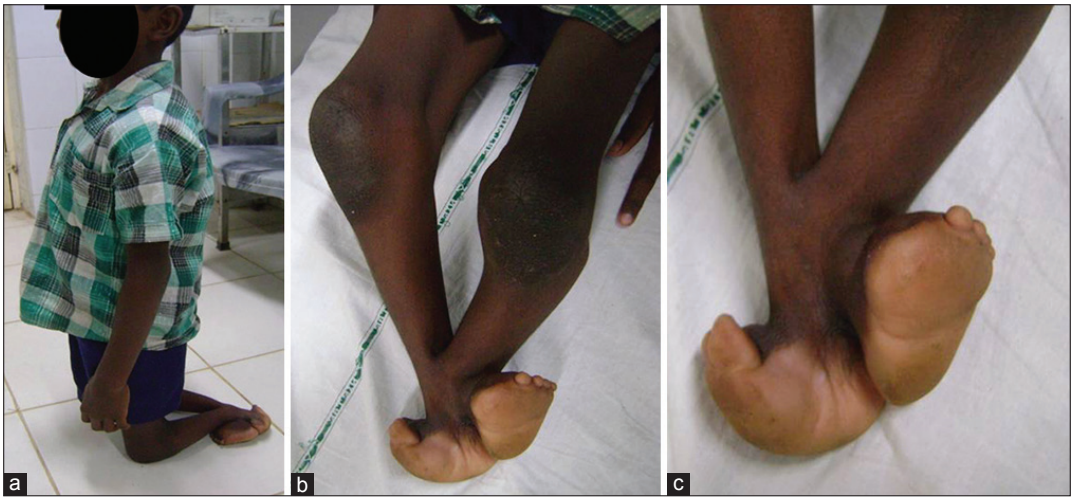

Figure 1: Clinical photograph of a 12-year old boy showing (a) fusion of lower legs above ankle joint and the way child use to stand and walk (b) fusion of lower legs with separate femurs and divided upper tibia and fibula (c) closer view

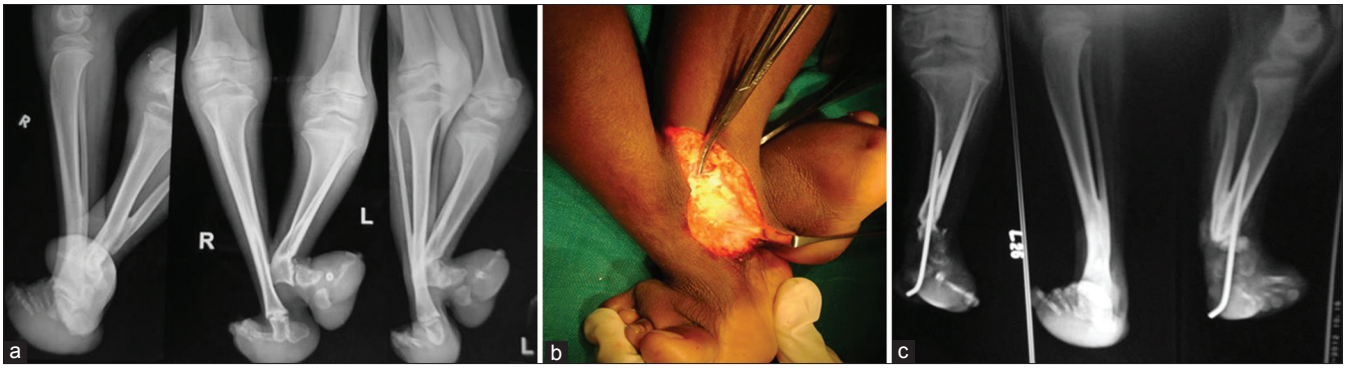

Figure 2: X-ray of both lower limbs showing (a) fusion at distal part of legs (b) peroperative photographs showing incision for Z-plasty of skin (c) X-ray of leg bones showing osteotomy and K-wire fixation of tibia

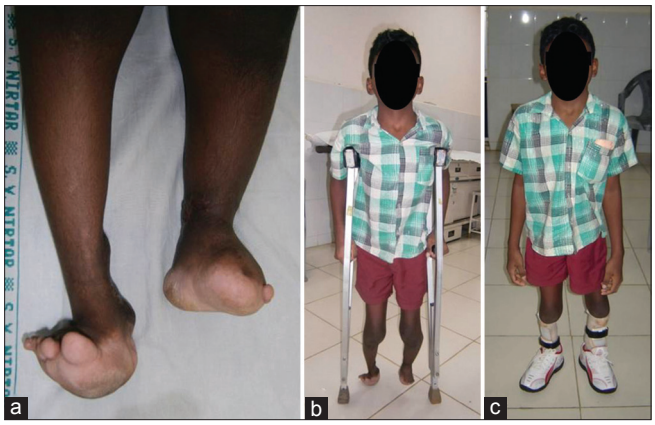

Figure 3: Clinical photographs showing (a) separation of both lower limbs (b) patient standing with B/L axillary crutches, 5 months after the surgery (c) patient standing independently with prosthesis, 6 months after surgery

scar was well established, a cast was applied to stretch the knee flexion contracture which was mostly of soft tissue origin. Strengthening exercises, balance retraining, and gait reeducation were also introduced as part of therapy.

After the surgery, the child demonstrated grade 3 muscle strength in bilateral quadriceps. The arm span was 141 cm but the child measured only 114 cm in total vertical height[7] The hip range of motion was within normal limits and the child was able to stand on both feet and was able to walk with prosthesis [Figure 3]. The prosthesis prescribed was partial type prosthesis for right side and Syme's type of prosthesis for left side along with distal weight bearing socket having soft lining and ethaflex extension for foot and a pair of normal shoes. Anterior strap with pad was given above the bulbous end for suspension.

At 6 months' followup after the operation, the child presented with excellent general condition and good state of healing of the skin scars at the lower extremities, without any evidence of skin retraction. At the time of this writing, the boy was 13-year-old and growing normally. The parents were instructed to go for a detailed assessment to rule out the possibility of any urologic, congenital heart defects and any neuroblastoma that might have coexisted.

Discussion

The prevalence of this syndrome is 0.1-0.25:10,000 in normal pregnancies and male to female ratio is 3:1.[1,8] More than half the cases of sirenomelia result in still birth.[9,10] This condition is 100 times more likely to occur in identical twins and is hypothesized to result from a failure of normal vascular supply from the lower aorta in utero. 22% of fetuses with this anomaly will have diabetic mothers.[11,12] Death of these patients is due to visceral anomalies and maximum duration of survival has been reported as 27 months.[13] About 300 cases have been reported globally with nine from India.

Taor *et al*.[14] has classified sirenomelia into three categories depending upon the time of presentation: simpusapus: no feet, one tibia, one femur, simpusunipus: one foot, two femur, two tibia, two fibula, simpusdipus: two feet and two fused legs (flipper like) – this is called a mermaid. Mirza[15] had reported a case with a single lower limb and two feet which she mentioned as a case of simpus dipus variant.

Diversion of blood flow away from the caudal portion of the embryo through the abdominal umbilical artery/"vascular steal" has been proposed as the primary mechanism leading to Sirenomelia.[1,2,10] In contrast, CRS is hypothesized to arise from a primary defect of the caudal mesoderm.[10] A teratogenic event during the gastrulation stage, i.e., the 3rd gestational week, may interfere with the formation of notochord, resulting in abnormal development of the caudal structures.[8] The presence of two umbilical arteries, renal anomalies compatible with life, divided lower limbs, abdominal wall defects, anomalies of tracheoesophageal tree, neural tube, and heart, allows differentiating the CRS from sirenomelia.[16-20] The presence of bilateral umbilical arteries in the described patient directs the patient to a diagnosis of CRS while fusion at the lower leg points toward sirenomelia. These types of clinical features have not been reported before and do not fit into any of the classified variety.

The reason for sirenomelia is an abnormal blood supply. There are lots of associated anomalies, making two identical cases look different. Because of the vascular insufficiency and reduced blood supply to the caudal part of the fetus, anomalies of spinal cord, gastrointestinal, and urogenital tracts usually take place. The cases of fixed lower extremities (with fusion of soft tissues of lower extremities), single lower extremity (bones fusion), and abnormal number of lower limbs are described.[21,22] A single thick hip is described in cases of bone fusion.[23] Feet can be absent, entirely or partially fused into one foot of the unusual appearance.

Anomalies of kidneys, significant oligohydramnios, and concomitant hypoplasia of the lungs make a very unfavorable prognosis for sirenomelia. Some milder forms of sirenomelia with survival have been described in the literature. Usually those had only soft tissue fusion of the lower legs.[24] Some authors consider sirenomelia to be a clinical manifestation of CRS because of abnormal development of fetal caudal mesodermal structures before the 4th week of gestation, that later extends to different craniocaudal levels.[25] It also leads to the absence of genitalia and renal agenesis if paramesonephric and mesonephric

ducts are involved. If the mesonephric ducts had developed enough, and joined metanephric blastema, the kidneys may develop as well.[26] Survival depends upon visceral anomalies instead of sirenomelia. The rudimentary foot may be hypothesized to the smaller and not fully traceable anterior tibial, posterior tibial, and peroneal arteries noted in the case report presented. Being the derivative of the axial artery, the peroneal artery is never fully absent but the Doppler reveals not fully traceable peroneal artery in this case.

To conclude, we feel that our case can be labelled as distal sirenomelia with no visceral anomalies. Till 2006, six cases of surviving infant with Mermaid syndrome were reported. This case is 12-year-old and after thorough literature search, it was found that the child was the longest surviving child till today. It needs a multidisciplinary approach for management.

References

1. Van Keirsbilck J, Cannie M, Robrechts C, de Ravel T, Dymarkowski S, Van 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, Lüthi H, Lopez R, *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, Gungorduk 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. Stroustrup Smith A, Grable I, Levine D. Case 66: Caudal regression syndrome in the fetus of a diabetic mother. Radiology 2004;230:229-33.
6. Kahilogullari G, Tuna H, Aydin Z, Vural A, Attar A, Deda H. Caudal regression syndrome diagnosed after the childhood period: A case report. Neuroanat 2005;4:16-7.
7. Brown JK, Whittemore KT, Knapp TR. Is arm span an accurate measure of height in young and middle-age adults? Clin Nurs Res 2000;9:84-94.
8. Valenzano M, Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. Sirenomelia. Pathological features, antenatal ultrasonographic clues, and a review of current embryogenic theories. Hum Reprod Update 1999;5:82-6.
9. Chandran S, Krishnan L, Christo GG. Sirenomelia sequence. Indian Pediatr 1991;28:1069-71.
10. Reddy KR, Srinivas S, Kumar S, Redy H, Prasad G, Irfan MM, *et al*. Sirenomelia a rare presentation. J Neonat Surg 2012;1:7.
11. Aslan H, Yanik H, Celikaslan N, Yildirim G, Ceylan Y. Prenatal diagnosis of Caudal regression syndrome: A case report. BMC Pregnancy Childbirth 2001;1:8.
12. González-Quintero VH, Tolaymat L, Martin D, Romaguera RL, Rodríguez MM, Izquierdo LA. Sonographic diagnosis of caudal regression in the first trimester of pregnancy. J Ultrasound Med 2002;21:1175-8.
13. Guidera KJ, Raney E, Ogden JA, Highhouse M, Habal M. Caudal regression: A review of seven cases, including the mermaid syndrome. J PediatrOrthop 1991;11:743-7.
14. Taori KB, Mitra K, Ghonga NP, Gandhi RO, Mammen T, Sahu J. Sirenomelia sequence (mermaid): Report of three cases. Indian J Radiol Imaging 2002;12:399-401.
15. Mirza B. Sirenomelia: Adding something new. J Neonatal Surg 2012;1:9.
16. Gürakan B, Karaaslan E, Balci S. Sirenomelia in an infant of a diabetic mother. A case report. Turk J Pediatr 1996;38:393-7.
17. Houfflin V, Subtil D, Cosson M, Valtille E, Carpentier F, Bourgeot P, *et al*. Prenatal diagnosis of three caudal regression syndromes associated with maternal diabetes. J Gynecol Obstet Biol Reprod (Paris) 1996;25:389-95.
18. Romero R, Pilu G, Jeanty P, Ghidini A, Hobbins JC. Bilateral Renal Agenesis. Prenatal Diagnosis of Congenital Anomalies. Norwalk, Connecticut/San Mateo, CA: Appleton and Lange; 1988. p. 259-60.
19. Harris RD, Alexander RD. Ultrasound of the Placenta and Umbilical Cord. Ultrasonography in Obstetrics and Gynecology. In: Callen PW, editor. 4thed. Philadelphia, PA: W.B. Saunders Company; 2000. p. 597-625.
20. Zaw W, Stone DG. Caudal Regression Syndrome in twin pregnancy with type II diabetes. J Perinatol 2002;22:171-4.
21. Honda N, Shimokawa H, Yamaguchi Y, Satoh S, Nakano H. Antenatal diagnosis of sirenomelia (sympusapus). J Clin Ultrasound 1988;16:675-7.
22. Sirtori M, Ghidini A, Romero R, Hobbins JC. Prenatal diagnosis of sirenomelia. J Ultrasound Med 1989;8:83-8.
23. Chenoweth CK, Kellogg SJ, Abu-Yousef MM. Antenatal sonographic diagnosis of sirenomelia. J Clin Ultrasound 1991;19:167-71.
24. Solovyov O, Goncharova YO, Masliy YV, Sudoma IO. Sirenomelia, first trimester. © Solovyov2009-12-08-14. Available from: http://www.sonoworld.com/TheFetus/page.aspx?id=2739 [Last accessed on October 2012].
25. Källén B, Winberg J. Caudal mesoderm pattern of anomalies: Transrenal agenesis to sirenomelia. Teratology 1973;9:99.
26. Barr M. Comments on "Origin of abnormality in a human simelian foetus as elucidated by our knowledge of vertebrate development". Teratol (Lett) 1988;38:487-8.

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