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Surgical Management of Sirenomelia: A Case Study

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Background: Sirenomelia is a rare congenital condition characterized by fusion of the lower limbs. Patients with sirenomelia generally do not survive long after birth because the condition is associated with multisystem organ dysfunction due to developmental anomalies. Considering the low incidence and few cases surviving the neonatal period, there is minimal understanding regarding the surgical management of sirenomelia. We present a unique case of an infant born with type 1 sirenomelia, absence of external genitalia, presence of a cloaca, absence of the bladder, and presence of an imperforate and vestigial anus, who not only survived the birth process, but, at the age of 11 months, was determined to be a candidate for surgical separation of the lower extremities.

Methods: This case was approached much like a dorsal rectangular flap syndactyly release. Large Z-plasty flaps were designed and raised, and the soft tissue within the skin bridge was meticulously dissected to preserve anatomy and to provide adequate skin flaps without perineal skin grafting. A quadrangular flap was designed to reconstruct the perineum and produce a neo-vulva using de-epithelialization.

Results: Successful lower extremity separation was achieved. There were no major postoperative complications. The patient progressed with lower extremity function, and eventually achieved independent ambulation.

Conclusions: Management of sirenomelia is incredibly challenging, and data to guide surgical management are limited. This report details our approach to a successful lower extremity separation, repair, and neo-vulvar reconstruction in a case of type I sirenomelia. *(Plast Reconstr Surg Glob Open 2023; 11:e5275; doi: 10.1097/GOX.0000000000005275; Published online 15 September 2023.)*

INTRODUCTION

Sirenomelia is incredibly rare and almost universally lethal. Colloquially referred to as "mermaid syndrome," sirenomelia is characterized by the manifestation of a single caudal appendage resembling a mermaid's tail.[1] The estimated incidence is 1:60,000-100,000, with increased frequency in monozygotic twin pregnancies.[2,3] Given the rarity and lethality of cases, there is a limited understanding surrounding the etiology and management of sirenomelia.

In addition to the signature presentation of fused lower limb, aberrances in virtually all systems have been associated with cases of sirenomelia, including urogenital, gastrointestinal, neural tube, cardiac, and vascular malformations. Common abnormalities include absent or atypical genitalia, renal agenesis, absent bladder, imperforate anus, and a single umbilical artery.[4,5] Furthermore, resultant oligohydramnios from renal dysplasia can result in potter-like facies, lung hypoplasia, and upper limb deformities.[4] A large epidemiological study by Orioli et al estimated that only half of sirenomelia cases survive a live birth, and only 1% survive the first week of life.[2] Typically, prognosis is determined by renal function and extent of lower limb skeletal fusion.[6,7] Treatment involves a multidisciplinary approach to correcting the variety of congenital malformations that may be present. Ultimately, the levels of organ dysfunction and skeletal fusion determine how to proceed with therapy.

Here, we present a case of an infant born with sirenomelia and associated genitourinary, renal, and gastrointestinal abnormalities, who not only survived the neonatal period, but continued to progress with medical

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management at 15 months of age. At the age of 11 months, she underwent a novel procedure in which the fused lower extremities were separated and repaired.

CASE REPORT

The patient is an 11-month-old female child who was born with fusion of the lower limbs. **[See figure, Supplemental Digital Content 1**, which displays the preoperative photographs (A, B), CTA (C), and 3D CT (D) of an 11-month-old female patient with sirenomelia. **http://links.lww.com/PRSGO/C767**.] Lower extremity fusion was initially identified prenatally via ultrasound. Additional congenital malformations at the time of birth included an imperforate anus and a single perineal channel draining urine, with complete absence of external genitalia. Ultrasound demonstrated a right solitary kidney and no definitive bladder. Considering the patient had significant multisystem comorbidities, surgical management was highly collaborative. A multidisciplinary team consisting of general surgery, plastic surgery, orthopedic surgery, and urology met regularly to discuss timing and priority of interventions.

Initial patient management revolved around investigating and intervening upon visceral and gastrointestinal issues, given their severity and potential lethality. Within 2 days of birth, exploratory endoscopy and laparotomy were performed to investigate and address gastrointestinal abnormalities, and further investigate abnormal pelvic anatomy. The distal colon was found to terminate in an atretic end. This portion was removed, and a diverting colostomy was placed. The ostomy demonstrated successful expression of stool, and oral feeding was resumed. The patient experienced feeding difficulties, and a diagnostic esophagram revealed esophageal stricture. The patient underwent esophagoscopy with dilation to address the stricture, but ultimately required percutaneous endoscopic gastrostomy placement at age of 4 months due to continued difficulty with oral feeding. This procedure was complicated by esophageal perforation, pneumothorax, and pneumomediastinum, requiring readmission for nearly 4 weeks, and multiple tube thoracotomies, for resolution.

After stabilization of the patient's gastrointestinal malformations, lower extremity separation could be addressed. First, a thorough preoperative assessment was performed to assess feasibility and develop an approach for separation. [**See Video 1 (online),** which displays the preoperative visit before lower extremity separation.] CTA and 3D CT were performed to further investigate the extent of lower extremity fusion, to assess for additional musculoskeletal or neurovascular abnormalities, and determine if surgical separation was vascularly possible (**Supplemental Digital Content 1**, **http://links.lww.com/PRSGO/C767**). Imaging revealed a dysplastic pelvis with possible posterior dislocation of the hips, and type 1 sirenomelia (fusion of superficial connective tissues only), with no noted bony fusion or orthopedic malformation in the lower extremities. She had trifurcation of the vessels at the popliteal fossa as expected. In addition to imaging, a team of plastic and orthopedic surgeons assessed joint mobility at all levels of the lower extremity and determined this patient would be an excellent candidate for separation, given preservation of joint mobility despite congenital fusion between the lower extremities. This case was approached like a dorsal rectangular flap syndactyly release. [**See Video 2 (online),** which displays the preoperative marking, operative plan, and separation of lower extremities of a patient with sirenomelia.] Large Z-plasty flaps were raised, and the soft tissue within the skin bridge was dissected to provide adequate skin flaps to enable closure without the need for skin grafting (Fig. 1). A quadrangular flap was raised to reconstruct the perineum, and the corners were de-epithelialized and folded in to produce a neo-vulva. **[See figure, Supplemental Digital Content 2,** which displays the immediate postoperative (intraoperative) pictures of a 11-month-old female child with sirenomelia. **http://links.lww.com/PRSGO/C768**.] The introitus may be deepened at a later age in a future surgery, if desired. At 15 months old, 4 months postoperative, the incisions are well-healed, and the child continues to progress with physical activity and weight-bearing bilaterally (Fig. 2). **(See figure, Supplemental Digital Content 3,** which displays the postoperative image of a patient with sirenomelia 4 months after lower extremity separation. **http://links.lww.com/PRSGO/C769**.) [**See Video 3 (online),** which displays the postoperative video showing the patient's lower extremity function after surgical release.] She performs regular physical and occupational therapy for strength, mobility, and scar care.

Takeaways

Question: In surviving patients with sirenomelia, how should lower extremity surgical separation be approached?

Findings: We present a unique case of a surviving 11-month-old child with sirenomelia who underwent a novel procedure in which the fused lower extremities were separated and repaired. This case was approached much like a large dorsal rectangular flap syndactyly release. Large Z-plasty flaps provided adequate soft tissue coverage without the need for perineal skin grafting. A quadrangular flap was designed to reconstruct the perinium and produce a neo-vulva using de-epithelialization.

Meaning: We achieved successful lower extremity separation, repair, and neo-vulvar reconstruction in a case of type I sirenomelia.

DISCUSSION

Sirenomelia is rare and almost always lethal in utero or immediately after birth. As such, much of the research surrounding this disease focuses on prenatal diagnosis and case studies investigating etiology, comorbidities, and causes of mortality in sirenomelia patients.[1,6–15] Literature surrounding the surgical management of these patients, and their lower extremity separation, is incredibly scarce, and there exist only four described cases of this operation.[5,16–18] Guidera et al performed a staged separation in an infant, in which the infant underwent tissue-expander placement at age 3 months, followed by lower extremity

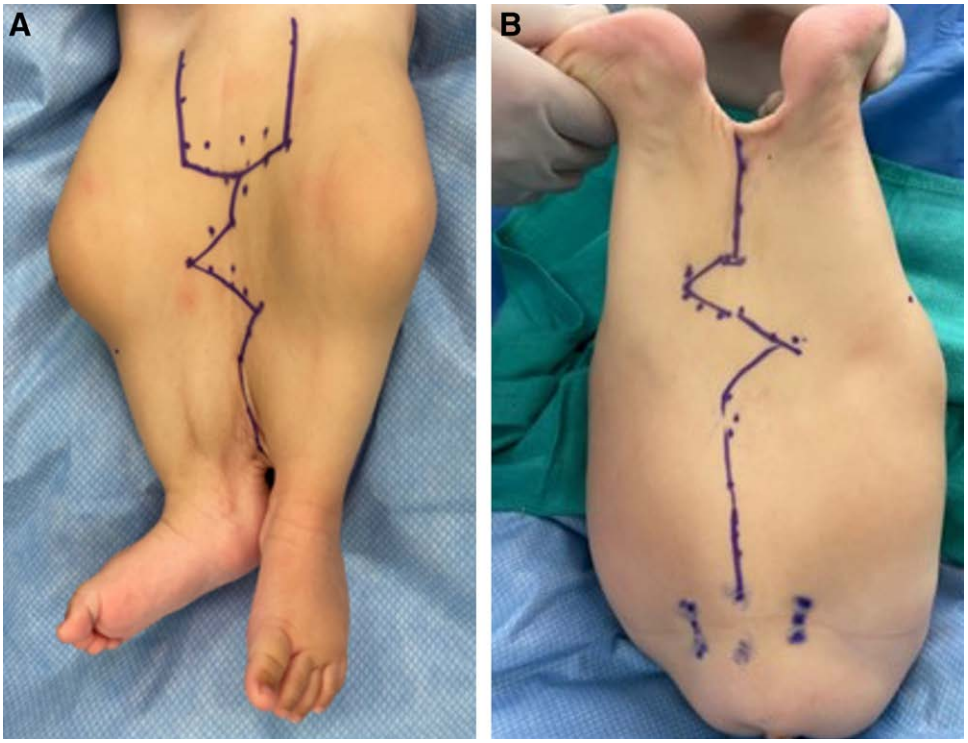

Fig. 1. Preoperative anterior (A) and posterior (B) markings in an 11-month old female child with sirenomelia.

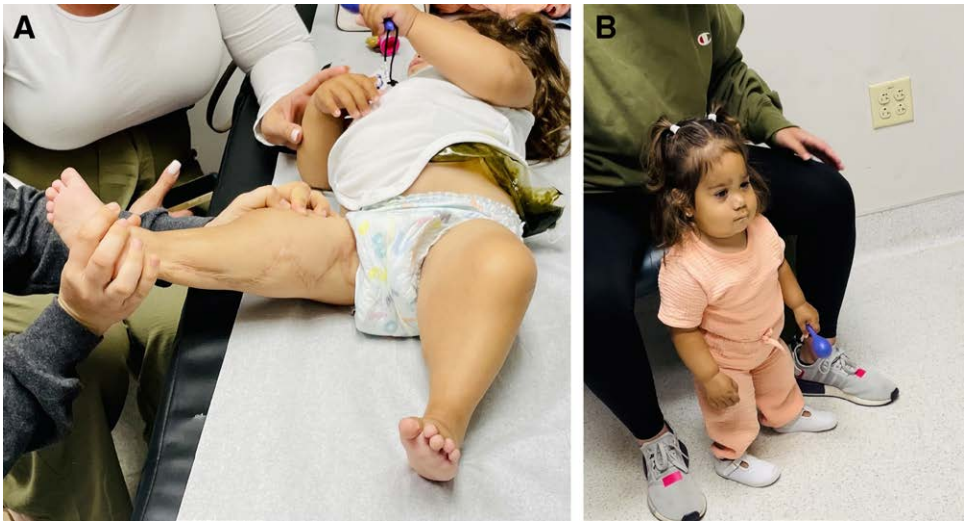

Fig. 2. Images of the patient at 25 months of age (14 months postoperative) demonstrating lower extremity range of motion (A) and independent ambulation (B) after surgical release.

separation at the age of 4 months, with skin grafting from the thighs and buttocks to cover residual defects.[16] A similar approach was used by Messineo and colleagues, who placed tissue expanders in a 5-month-old patient with sirenomelia, and expanded the lower extremity skin bridge for 2 months, then removed the expanders and used a multidisciplinary surgical team, consisting of a vascular surgeon, microsurgeon, plastic surgeon, and pediatric surgeon, to separate the lower limbs.[17] Recently, Pederson et al also performed a staged separation on a patient with sirenomelia. In the first stage, performed at 14 months of age, a tissue expander was placed in the anterior upper part of the legs to allow for closure of the thighs without need for skin grafting upon separation during the second stage. After 3 months of tissue expansion, the lower extremities were separated in a fashion similar to our approach, using large Z-plasty flaps to separate the thighs. A through-knee amputation was performed, given the

severity of deformity of the lower legs; however, a pedicled flap was salvaged from the lower legs and used for perineal reconstruction.[18]

In contrast to the cases above, in which a staged separation was performed, Stanton and colleagues completed their separation in a single stage, without use of tissue expanders. On day 15 of life, the patient underwent lower extremity separation with locally based skin flaps to close the defects. This was later followed by Z-plasties to elongate the scars. The initial procedure was complicated by wound breakdown which required debridement.[5]

Although the fused lower extremities were among the most distressing features to the parents of our patient, before management of this patient's lower extremity fusion, additional anomalies and comorbidities had to be diagnosed and managed. This included thorough gastrointestinal, renal, genitourinary, and cardiovascular examination and workup. Secondly, we also had to consider any anatomical constraints which might threaten potential repair options. Of note, given the location and presence of this patient's cloaca, there was consideration of flap placement that would not compromise the repair, but also would not result in urine contaminating the wound site, as this would be a great risk for infection and ultimate breakdown of the reconstruction. In this patient, anatomic diversion of the GI system was necessary, but urine was eliminated through a cloacal orifice located caudal to any incision sites. After medical stability of the patient was certain, plans were made for surgical intervention. A multidisciplinary team discussed optimal timing for lower extremity repair. We believed that at the age of 11 months, the patient was medically stable and a suitable candidate for the longer period under general anesthesia that would be required for this procedure. The patient's gastrointestinal issues were addressed early in life, allowing for 6 months of nutritional optimization via G-tube before the operation. Finally, we wanted to achieve separation before the age children typically begin ambulating independently. This would allow sufficient time for the patient to work with orthopedics and physical therapy to potentially achieve ambulation at an age similar to that of her peers. Throughout the perioperative process, discussion with the patient's parents were necessary to understand their goals of treatment and to establish realistic expectations. They were also counseled on risks of the procedure, including incomplete separation and need for surgical revisions in the future. Ultimately, the goals of this procedure are to introduce lower extremity function, most notably ambulation, and to improve appearance.

Given the similarity in anatomy to that of a complete digital syndactyly, the surgical approach of this case revolved around similar principles which guide syndactyly separation.[19] In particular, our approach mirrored the modified Flatt technique,[20,21] where a combination of a proximal/cephalad quadrangular flap was used in conjunction with large Z-plasty flaps to achieve separation and closure. This approach allowed separation and repair in a single stage, without need for multi-stage approach involving tissue-expander placement or skin grafting with additional donor sites, as was required in other instances of sirenomelia separation.

CONCLUSIONS

Sirenomelia is a rare congenital anomaly with a paucity of literature surrounding its management. We performed a single-stage surgical separation in a surviving sirenomelia patient, with good aesthetic and functional outcomes. Medical comorbidity stabilization, multidisciplinary care, and thorough preoperative imaging are vital aspects of successful repair in these patients.

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DISCLOSURE

The authors have no financial interest to declare in relation to the content of this article.

PATIENT CONSENT

The parents or the guardian provided written consent for the use of the patient's image.

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