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Unrepaired cloacal exstrophy in an adult: Medical and ethical considerations

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ABSTRACT

Cloacal exstrophy, also known as OEIS complex, is a rare condition, comprised of severe congenital anomalies. This case report describes a 24-year-old 46,XY individual who had repair of the omphalocele at birth, but has remained with an unrepaired bladder exstrophy. This case highlights the intersections between medical decision-making, individualized management of complex patients, and ethical considerations for adults lacking capacity.

1. Introduction

Cloacal exstrophy, also known as OEIS complex, is a rare syndrome affecting between 1 in 200,000 to 1 in 250,000 live births.[1] This complex includes omphalocele, bladder exstrophy, imperforate anus, and spinal defects. Some individuals may also present with limb defects, genital defects, and Müllerian duct anomalies.[1] Modern medical advancements have improved the survival rate; however, adults born with OEIS complex may encounter multiple quality of life (QOL) issues including urinary and fecal continence, sexual function, and psychosocial challenges.[2] This case highlights the intersections between safe medical decision-making, individualized management of complex patients, and ethical considerations for adults lacking capacity.

2. Case report

This patient is a 46,XY 24-year-old born with cloacal exstrophy. Given the paucity of genital tissue, she was gender assigned female at birth. During omphalocele closure, a colostomy was created, remnant hindgut was repurposed as a neovagina, and a right orchiectomy was performed. The bladder was not reconstructed, nor was a left orchiectomy performed as the testis could not be identified.

She has had numerous other procedures including revisions to the colostomy, complex abdominal closures with biologic dermal substitutes, spinal surgeries, repair of Chiari II malformation, and cholecystectomy. Her abdomen and pelvis are shown in Fig. 1. Additionally, her history is significant for severe developmental delay and autism. Her biological mother serves as her proxy decision-maker for medical treatment.

Prior to presentation at our institution, she had been voiding into a diaper from her open hemi-bladders, with no specific care to the bladder mucosa. Her bowels were managed with a colostomy. She has been maintained on hormone suppression with a gonadotropin-releasing hormone analogue given the remaining left testicle.

She initially presented with emesis and decreased ostomy output. There was concern for a small bowel obstruction (SBO) in a large hernia near the neovagina. She was managed conservatively with nasogastric (NG) decompression and total parenteral nutrition (TPN) given her hostile surgical abdomen. However, she failed this conservative trial and discussions began about operative management.

Prior to intervention, repeat imaging was obtained including an upper gastrointestinal (GI) series and stomagram. These were suggestive of severe gastroparesis or gastric outlet obstruction than SBO. Abdominal magnetic resonance imaging (MRI) similarly showed no concerns for SBO and identified the left testicle surrounded by loops of bowel near the colostomy site (Fig. 2).

Ultimately, open surgical management was deferred after reviewing the abdominal MRI and multidisciplinary discussion, involving the family. She underwent pyloric dilation with Botox injection to the esophageal sphincter and tolerated nasojejunal (NJ) feeds. Bladder

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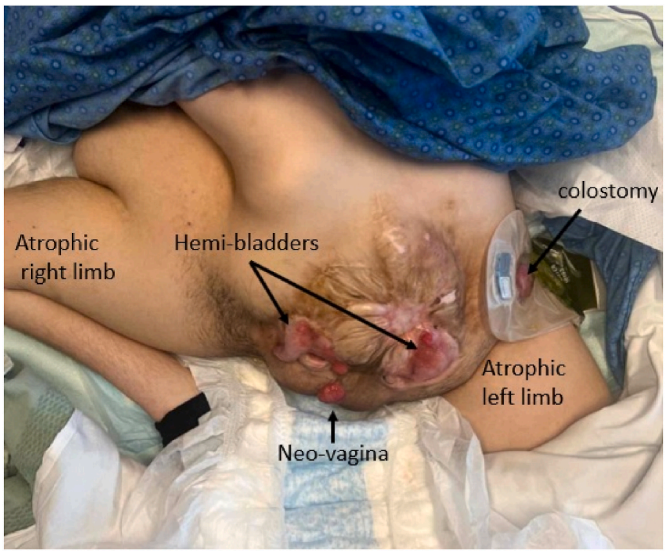

Fig. 1. Photograph showing exposed hemi-bladders, neovagina, and LLQ colostomy.

biopsies were performed and did not show any signs of malignancy. She was discharged home with local GI, colorectal, and urology follow-up. A shared decision was made with the patient's mother to continue annual surveillance for malignancy of the hemi-bladders with physical exam and the remaining testis with ultrasound.

3. Discussion

3.1. Sex assignment and neovagina management

Sex assignment in newborns with ambiguous genitalia has been a controversial topic within pediatric urology for decades. The current literature on the outcomes of gender converted 46,XY cloacal exstrophy patients is extremely limited. At the time these decisions were made, cloacal exstrophy would have been a condition with very poor survival. Newer techniques improved survival; however, phallic reconstruction was in its infancy and not widely available, which may have influenced some of these difficult decisions. Rearing as male is accepted in the contemporary era, as brain imprinting by androgens has already occurred *in utero* and maintaining the testes allows for preservation of future fertility potential. While creation of a functional phallus can be challenging, it is far from impossible with the further development of phalloplasty.[3]

In our practice, the recommendation is to incorporate all the gastrointestinal tissue into the fecal stream, creating a true end colostomy.[4] Only after deciding between stoma pull-through vs. permanent stoma, based on the results of bowel management via stoma, hindgut could then be considered for reconstruction.[5]

When initially discussing possible operative management, the possibility of removing the neovagina was raised. If involved near a large hernia, causing an SBO, the thought was that maintaining it might cause more harm than good. Given her significant delay, use of her neovagina for sexual purposes would be unlikely to be on a consensual basis. A permanent stoma was also better for QOL based on her immobility and care dependence, leading to the discussion of using this tissue for urologic reconstruction.

3.2. Quality of life

Progress in early surgery and supportive care means that management priorities have shifted from survival to QOL with a particular focus on continence and long-term psychological outcomes. Discussions were

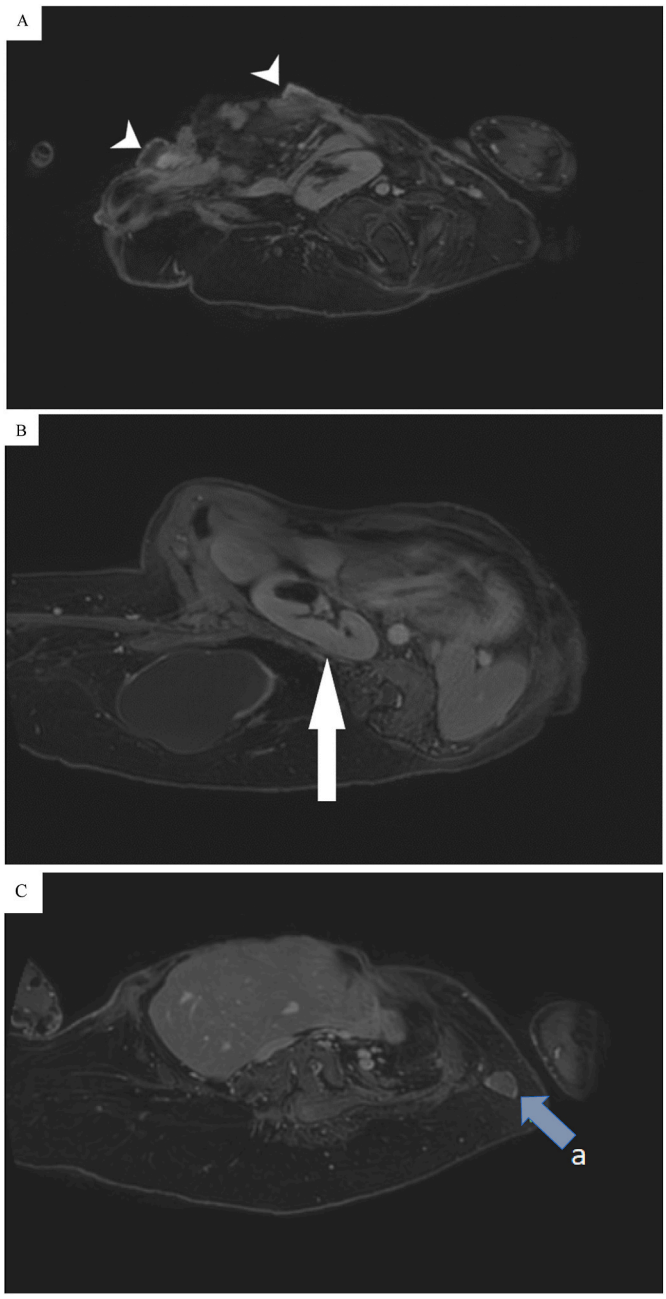

Fig. 2. T1 Magnetic Resonance Imaging without contrast, axial view. (**A**) Left pelvic kidney. Note the kidney is malrotated and posterior to the exposed hemi-bladders, making future percutaneous access unsafe(arrowheads). (**B**) Right bifid kidney malrotated in transverse lie, also in a location for unsafe percutaneous access (arrow). Note the large lumbosacral myelomeningocele. (**C**) Possible left testis in the soft tissue posterosuperior to the LLQ colostomy (arrow). Note the minimal margin of soft tissue overlying the liver.

had with her mother about how to manage her urinary system. We discussed possible exstrophy closure, with likely augmentation and continent channel creation. However, given the severity of her pubic diastasis, osteotomies would not be possible to ensure appropriate soft tissue coverage of the pelvis and avoid closure failure (Fig. 3). We discussed removing the bladder halves and performing a diversion with a continent pouch or an incontinent ileal conduit. Continence was not a priority for the patient and her mother, an ileal conduit was seriously considered. However, our patient currently has stable renal function, so the possibility of not only compromising this function but also limiting future access to her upper tracts, led to the shared decision to defer

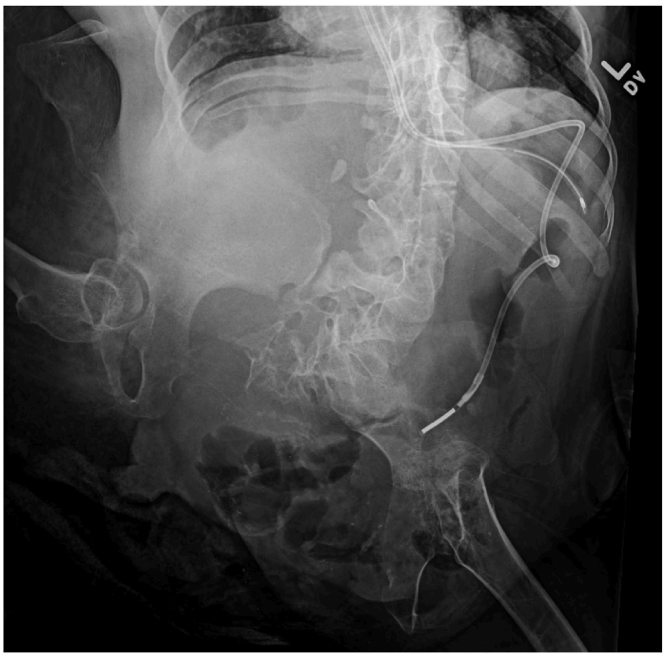

Fig. 3. Abdominal X-ray of the pelvic anatomy.

surgical management. Additional surgical risk was not in line with QOL goals, which were, in many ways, already met.

4. Conclusion

In summary, this rare case adds to the literature regarding considerations in managing an adult with unrepaired cloacal exstrophy. An interdisciplinary discussion with the patient's proxy decision-maker was needed to clarify the definition of QOL and treatment goals. Balancing the safety and effectiveness of treatment with the patient's welfare and family's needs were key to the care of this patient.

Consent

The authors received consent from the patient's proxy decisionmaker to publish this case report.

Declaration of competing interest

None.

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References

1. Bernardini Laura B, Dickie Belinda H, Estroff Judy A. OEIS syndrome: omphalocele, exstrophy of the Cloaca, imperforate anus, and spinal defects. *NeoReviews*. October 2021;22(10):e709–e718. https://doi.org/10.1542/neo.22-10-e709.
2. van Vliet R, Roelofs LA, Rassouli-Kirchmeier R, et al. Clinical outcome of cloacal exstrophy, current status, and a change in surgical management. *Eur J Pediatr Surg*. 2014:87–93.
3. Diamond DA, Burns JP, Mitchell C, Lamb K, Kartashov AI, Retik AB. Sex assignment for newborns with ambiguous genitalia and exposure to fetal testosterone: attitudes and practices of pediatric urologists. *J Pediatr*. 2006;148(4):445–449. https://doi.org/10.1016/j.jpeds.2005.10.043.
4. Soffer SZ, Rosen NG, Hong AR, Alexianu M, Peña A. Cloacal exstrophy: a unified management plan. *J Pediatr Surg*. 2000;35(6):932–937. https://doi.org/10.1053/jpsu.2000.6928.
5. Levitt MA, Mak GZ, Falcone Jr RA, Peña A. Cloacal exstrophy–pull-through or permanent stoma? A review of 53 patients. *J Pediatr Surg*. 2008;43(1):164–170. https://doi.org/10.1016/j.jpedsurg.2007.09.039.