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Review Articles

Anorectal malformation and associated end-stage renal disease: Management from newborn to adult life

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Abstract

Background/Objective: Renal failure remains one of the most significant causes of morbidity in patients with anorectal malformations (ARM). In the modern era, an increasing number of children born with ARM and genito-urinary (GU) anomalies reach adulthood and require continued multidisciplinary care for the rest of their life. The aim of this study is to present our institutional experience in the management of pediatric chronic renal failure related to severe GU anomalies and anorectal malformations.

Methods and Results: Three hundred twenty-one patients with ARM have been followed at our institution since 1987. Six patients developed end-stage renal disease (ESRD) and received a kidney transplant at different ages. One patient is currently followed for mild, progressive chronic renal failure. These seven cases are reported along with a broad discussion concerning etiology of renal failure, neonatal surgical management, pediatric dialysis, urologic issues, and kidney transplantation.

Conclusion: Complex GU anomalies associated with ARM require a long-term approach by specialized pediatric and adult clinicians to optimize the care of this selected population of patients.

Anorectal malformation (ARM) is an abnormal termination of the distal anus and rectum that comprises a wide spectrum of diseases involving both the alimentary and genito-urinary tracts. The incidence is approximately 1 in 5000 births and slightly more common in males. Up to 25%–60% of children with this malformation have coexisting genito-urinary (GU) abnormalities and the occurrence changes with increasing complexity of the anorectal defect [1,2]. Most of the major GU anomalies are found in females with cloaca (90%) and males with bladder neck fistulae

Abbreviations: ARM, anorectal malformation; GU, genito-urinary; ESRD, end-stage renal disease; CAKUT, congenital anomalies of kidney and urinary tract; UTIs, urinary tract infections; PD, peritoneal dialysis; ES, exit-site; HD, hemodialysis; VUR, vesico-ureteral reflux; VCUG, voiding cysto-urethrogram; CIC, clean intermittent catheterization; PSARP, posterior sagittal anorectoplasty.

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Table 1 Major congenital genito-urinary anomalies present in children affected by anorectal malformation [1–4].

Etiology	Incidence
Vesico-ureteral reflux	30%–40%
Renal agenesis	10%–18%
Neurogenic bladder	6.5%
Hypoplasia–dysplasia	5%–6%
Uretero-pelvic junction obstruction	1%–2%
Multicystic kidney disease	1%–2%

(52%). In contrast, lower perineal and cutaneous fistulae are associated with a lower incidence of associated GU anomalies (14%) [2,3]. Table 1 shows the incidence of major GU anomalies associated with ARM.

The world incidence of chronic renal failure ranges from 12 to 14 new cases per million per year in the pediatric population [4–6]. While diabetes mellitus and hypertension are the leading causes of end-stage renal disease (ESRD) in adult patients, congenital anomalies of kidney and urinary tract (CAKUT) represent the primary cause of irreversible kidney failure in children [5]. Historically, the rate of death from ESRD in children affected by anorectal malformations has been reported between 2.5% and 6% [7].

The aim of this study is to present our experience with chronic renal failure in children with ARM and discuss etiology, management, and outcome in this particular group of children.

1. Case reports

1.1. Case 1

A 49-year-old male presented with history of high ARM and recto-urethral fistula corrected in the first year of life with an abdomino-perineal pull-through without recognition/ligation of the fistula. The fistula was eventually closed at the age of 26 years old after several episodes of urinary tract infections (UTIs) and progressive worsening of the kidney function. Genito-urinary anomalies included a right dysplastic kidney and left severe vesico-ureteral reflux (VUR). In the 1970s poor knowledge of the anorectal malformation anatomy together with the poor urologic management of the VUR in this patient resulted in progressive renal failure and ESRD at 41 years of age. No urodynamic studies were available. Cadaveric kidney transplant was performed at age 47 years with current normal kidney graft function.

1.2. Case 2

A 36-year-old female was born with a cloaca. Abdomino-perineal pull-through with defective closure of the left fistula along the bladder pouch was performed at age 2 years. Subsequent multiple pyelonephritic infections led to progressive bilateral scarring and further renal function deterioration. Bilateral ureteral reimplantation was performed at 6 years of age. Right nephrectomy, resection of the bladder pouch and vaginal reconstruction with ileum interposition were performed at the age of 12 years. Delayed intervention for the VUR together with an inappropriate repair of the cloaca was the main cause of early renal failure. No urodynamic studies were available. Cadaveric kidney transplantation was performed at the age of 18 years but, she had a progressive loss of graft function, over a 10-year period, due to chronic allograft injury.

1.3. Case 3

A 26-year-old female was born with a cloaca. Recurrent febrile UTIs with progressive bilateral scarring on DMSA scan were noted. An abdomino-perineal pull-through without vaginal reconstruction and bilateral ureteral reimplantation were performed at age 6 years. Delayed management of the bilateral severe VUR together with an incomplete and late repair of the cloaca resulted in chronic kidney failure. Hemodialysis was started at 12 years of age. No urodynamic studies were available. Cadaveric kidney transplant was followed by multiple reflux UTIs to the graft. Progressive worsening of the kidney function ensued during the next 10 years post-transplant with currently moderate chronic kidney disease (GFR 50 ml/min/1.73 m^2).

1.4. Case 4

A 21-year-old male with high ARM and recto-urethral fistula underwent neonatal colostomy and right cutaneous ureterostomy (left kidney agenesis). He was corrected at the age of 1 year with a posterior sagittal anorectoplasty (PSARP) and few months later had ureterostomy closure with right ureteral reimplantation that was followed by irretractable stenosis and recurrent UTIs. Then, the right cutaneous ureterostomy was re-established and kept in place for about 12 years. Progressive renal failure required creation of an arterio-venous fistula for hemodialysis at age 15 years. Pre-transplantation urodynamic study showed normal bladder pressures with low capacity. Cadaveric donor kidney transplant was completed at the age of 18 years and the patient continues to have normal graft function at 3-year follow-up.

1.5. Case 5

A 21-year-old male with history of high ARM and recto-urethral fistula underwent neonatal colostomy. He also had bilateral severe VUR, a dysplastic right kidney, and penoscrotal hypospadias. PSARP was performed at the age of 6 months and complicated by iatrogenic urethral stenosis and ischemic rectal stenosis. A vesicostomy was created at the age of 1 year and is still in place even after creation of a