Genito-Urinary Abnormalities in Children with Anorectal Malformations

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Between 30-60% of children with an anorectal malformation (ARM) have a genito-urinary abnormality (GUA) [1-4]. In general, GUA's are more common in males with the exception of vesico-ureteric reflux (VUR) which is twice as common in affected females [5]. In addition, high ARM's or complex malformations such as cloaca or "pouch colon" have a very high incidence of GUA [6]. Two percent of anocutaneous fistulae and as many as 90% of cloacal anomalies have a urological abnormality [7]. The frequency of GUA is also directly proportional to the diligence with which they are investigated. The practice of carrying out routine MCU's even in children with a low ARM has revealed a significant incidence of VUR's [8].

Involvement of both the GU and the GI systems probably results from an embryological accident around the twenty-eighth somite stage at 4-5 weeks of gestation [9]. Failure of development of the urorectal septum results in a recto-urinary fistula in the male and a recto-genital fistula in the female [10]. A defect of the cranial component of the septum (Torneux's fold) results in a fistula to the bladder or posterior urethra while recto-bulbar fistulae result from a defect of the caudal component (Rathke's plicae). Spinal abnormalities of ARM probably result from disordered neurulation at the caudal end, an event in the fourth week of embryogenesis [9].

The various GUA encountered and their relative frequencies in ARM's is tabulated in Table 1. We have also tabulated the relative frequencies of GUA encountered in 154 children with imperforate anus treated at the Hospital for Sick Children, Great Ormond Street, London between 1976 and 1990. The commonest anomaly encountered is VUR [11]. Vertebral-spinal abnormalities are included in this discussion because of their association with neuropathic bladder. The unexpected high incidence of VATER syndrome in our series reflects the tertiary referral nature of our hospital.

Vesico-ureteric reflux (VUR)

VUR occurs in 35-40% of children with ARM [2,3,11-12]. The more complex the anomaly the higher is the incidence of reflux, e.g. 68% of cloaca and 60% of "pouch colons" have reflux [6,7]. Until recently, most reflux was presumed to be primary, however, an increasing number of children with neurovesical dysfunction are being discovered [13-15]. In the presence of a rectourinary fistula, it is conceivable that bacterial spillover with oedema of the VUJ could cause a borderline valve to reflux. Indeed, cessation of low-grade VUR after definitive surgery and closure of fistula is common [3,12].

Early detection of VUR is essential to preserve renal function, and some advocate that MCU's should be mandatory in all cases, even low ARM's [8]. Most VUR's including a few high-grade reflux [17], resolve with time. Persistent VUR necessitates investigations for a neuropathic bladder, where as many as two-thirds will ultimately require surgery [16]. Ureteric reimplantation, which is usually performed **after** a pull-through operation, can sometimes be difficult because of scarring at the bladder base [18]. Submucosal teflon injection (STING) is an attractive option and a 78 per cent success rate has been reported in the difficult setting of neuropathic bladder, in normal bladders the success rate can exceed 85-90%[19].

Spinal abnormalities

Malformations of the lumbosacral spine are re-

Table 1. Incidence of various genitourinary anomalies in children with anorectal malformations

Anomaly	Incidence	References	GOS experience #
1. VUR	35-40 %	2,3,11,12	25.3 %
2. Vertebral and spinal	13-45 %	20-24	14 %
3. Neuropathic bladder	7-18 %	12-15	8.7 %
4. Rectourinary fistula*			
Males	86 %	10	
Females	78 %		
5. Renal/ureteric anomalies (hydronephrosis, agenesis, dysplasia, duplications, ectopic ureter, ureteroceles)	15-25 %	1-4	27.3 %
6. Müllerian abnormalities	31-45 %	30,31	2 %
7. Renal failure	< 1 %	11	2 %
8. Genital anomalies (hypospadias, undescended testes, ambiguous genitalia)	3-5 %	10	8.7 %
9. Stigma from operative damage - vas, urethra, ureter	< 1 %	10,33	4 %
10. VATER syndrome	3-6 %	34-36	14 %
11. Recurrent epididymitis	Rare	6,24	2.8 %
12. Bladder exstrophy	Rare	37	0 %
13. Rectal enterolithiasis	Very rare	38	0 %

** High or intermediate anomalies,*
 # GOS experience: genito urinary abnormalities seen in 154 children with imperforate anus treated at Hospital for Sick Children, Great Ormond Street, London, between 1976-1990.

ported to occur in 13-45% of ARM's [20-24]. The commonest malformations are sacral agenesis and hemivertebrae. Their importance lies in the fact that they are often associated with neurovesical dysfunction (NVD) and VUR. Thus, in a series of children treated with PSARP (posterior sagittal anorectoplasty), 60 per cent of those with abnormal sacrums had urinary incontinence [18]. In another series, 72 per cent of ARM's with an abnormal spine had GUA [20].

The vertebral anomaly may be an indication of an underlying correctable lesion of the spinal cord, which may otherwise present later with NVD. The symptoms often become manifest at the time of a growth spurt when traction on the cord occurs. Because of failure of ossification of the posterior arches in the first year of life, an ultrasound can detect these anomalies in infancy. Ultrasound scans of the spine have recently been advocated as a screening investigation in all cases of high ARM's or those with an abnormal sacrum [25-28]. An NMR scan is required for final anatomic delineation of the lesion.

Karrer reported preservation of function with early neurosurgical release, however, absence of urodynamic evaluation both before and after the release limits the usefulness of his conclusions [21]. Rarely, a patient may deteriorate following prophylactic detethering [28].

Neurovesical dysfunction (NVD)

Overt NVD has been reported in 7-18 per cent of ARM's [13-15]. The risk of NVD is highest in patients with a cloaca or with a vertebral anomaly. In a study of 90 patients with ARM, 16 were found to have NVD; 13 of these had vertebral abnormalities [29].

The fact that NVD is often diagnosed several months folowing a pull-through operation seems to imply that operative damage to the pelvic nerves is an aetiological factor. However, urodynamic studies on 32 patients with ARM's and NVD revealed that the majority were hyperreflexic, indicating an upper motor neuron lesion [30]. Secondly, although NVD may exist at birth, it is often diagnosed later when toilet training is being attempted. Thirdly, NVD may manifest later as a result of spinal traction at the time of a growth spurt [31].

Clinically, NVD presents as failure to empty the bladder or as incontinence. In a long-term study by Holschneider, 20 per cent of children with ARM were incontinent and VUR was present in half of these bladders [16]. Urodynamic studies show hyperreflexia; the sphincters are either dyssynergic or weak in 40 per cent [25,30,32]. The management of these patients requires either pharmacological manipulation (mainly oxybutinin) or clean intermittent catheterization (CIC). In a long-term study of 32 children, 19 ultimately needed bladder augmentation, only 16 were fully continent [30]. The im-

portance of ruling out spinal lesions cannot be overemphasised; of 16 patients with NVD, 6 were found to have a tethered cord [29].

Some authors have suggested performing screening urodynamics in all patients with a high ARM. In one study, 4 out of 14 screened were shown to have detrusor sphincter dyssynergia (DSD), all four had VUR or hydronephrosis [33]. Since all patients with a high ARM undergo a MCU, if facilities are available, a urodynamic study should be carried out at the same time.

Rectourinary fistula (RUF)

In males with a high or intermediate anomaly, 80 per cent have a fistula to the urethra and 6.6 per cent to the bladder [10]. Females with a similar anomaly have a rectogenital fistula in 78 per cent. A rectourinary fistula can have infective or biochemical complications.

Colonization of the urinary tract with fecal bacteria occurs in 21-48 per cent of all cases with recurrent UTI in a few [2,18,34]. Sepsis in a patient with ARM is deemed to be due to a RUF unless proved otherwise. Bacterial spillover with resulting VUR in a borderline valve has been referred to previously. Antibiotic prophylaxis is mandatory in all RUF's until definitive surgery has been carried out. Although a colostomy is supposed to be protective, UTI due to silent fecal spillover into the distal loop is known to occur [7].

Hyperchloremic acidosis is a known complication of RUF [35,36]. Dehydration, distal urinary obstruction and a proximal colostomy (e.g. right transverse), all increase the risk of absoption of chloride ions [37]. The occasional infant may be very sick with accompanying shock. If the fistula closure cannot be achieved earlier, oral bicarbonate should be prescribed. In more serious cases, bladder drainage with a suprapubic catheter or vesicostomy may be required.

Genital and renal anomalies

The commonest renal anomaly is a unilateral renal agenesis which should be confirmed by a renal scan. This serves not only as a prognostication but may also localise a small ectopic kidney which may present later in life with incontinence due to an ectopic ureter [18]. In the male, undescended testis and hypospadias occur with greater frequency [2]; a severe hypospadias with an empty scrotum can result in ambiguous genitalia. A full karyotypic, biochemical, hormonal and radiological workup is required. Ultimately it is the size of the phallus which determines the type of genitoplasty [38].

Cloaca and Müllerian abnormalities

Müllerian abnormalities are very common in cloacal anomalies and occur in 32-45 per cent of all female ARM's [39-41]. Uterine duplications are most common, and can result in premature labour or abortions. A Caesarian section at 33-34 weeks gestation would appear to be appropriate in the event of pregnancy in later life. Obstruction to uterine outflow may occur either in early infancy or at menarche. GUA's are particularly common in patients with cloacal anomalies and include deformed labia, neuropathic bladder, hypoplastic urinary sphincters, VUR, vaginal atresia and renal anomalies [7]. Surgical repair should only be attempted by an experienced surgeon, a posterior sagittal approach is favoured [10]. Reconstruction of the urethra from the urogenital sinus should be given top priority, vaginal reconstruction is best delayed till puberty.

Renal failure

Sepsis and infravesical obstruction are the main causes of renal failure. The latter can result from a urethral stricture or a neuropathic bladder; or from the pressure from a hugely distended hydrocolpos or "pouch colon" [6,7]. Urethral or suprapubic catheterization is both diagnostic and therapeutic.

Operative injury

Iatrogenic injury to the vas or ureter may occur even with the PSARP approach [42]. Urethral injury may occur from an inappropriate Foley's catheter or due to the surgery itself. It may manifest later as a urethral stricture or diverticulum [10].

VATER Syndrome

The non-random association of multiple malformations was first described by Burhan Say and

PS Gerald in 1968 [43], the term was it self first coined by Quan and Smith [44] in 1972. Later it was expanded to VACTERL, to include vertebral, anorectal, cardiac, tracheo-esophageal, renal and limb anomalies. Two or more components are required for diagnosis, the most common component is vertebral. Once the diagnosis is made, a dilligent search should be made for the other components by appropriate investigations. In selected centers, survival of these babies now approaches 80-90 per cent [5,45].

Recurrent epididymitis

The cause of recurrent epididymitis is not known. In some cases an ectopic ureter or a urethral abnormality has been found. Others have reported the unusual finding of reflux of dye into the ejaculatory ducts at the time of an MCU, and this may play a role in the recurrent epididymitis [5,18]. In frequent relapses, continuous antibiotic prophylaxis is indicated. Very occasionally vasectomy may be the only solution to the problem.

Bladder exstrophy

Bladder exstrophy may rarely coexist with ARM. Approximately 1 in 400,000 have the full blown cloacal exstrophy syndrome; severe renal and Müllerian abnormalities are the rule here [46]. Surgical management consists of an ileostomy, separation of bladder halves from the bowel and their approximation; appropriate surgery for the exomphalos and meningomyelocoele is also required. In genetic males, a feminizing genitoplasty is the only option [38].

Rectal enterolithiasis

In the presence of a rectourinary fistula, urine mixes with meconium in the rectum which then may calcify. Rarely, the calcified enterolith can be diagnosed antenatally [47].

Conclusions and recommendations

Every infant with an anorectal malformation should undergo careful physical examination and investigations to rule out associated genito-urinary abnormalities. Early detection and appropriate management can result in prevention of recurrent UTI and preservation of renal function, particularly in the setting of VUR. An x-ray of the lumbosacral spine and an abdominal ultrasound should be performed at birth. Those with a high ARM or with an abnormal vertebral radiograph should undergo a screening ultrasound of the spine to eliminate cord tethering. An MCU should be performed in all ARM's, including low anomalies. It can often be combined with a distal loopogram if a colostomy has been performed. The role of urodynamic studies without evidence of spinal abnormalities or neurogenic bladder is debatable, but a high proportion of neuropathic bladders will be identified. Where indicated, it can be combined with the MCU study.

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