ANORECTAL MALFORMATION INCIDENCE AND SIGNIFICANCE OF ASSOCIATED ANOMALIES

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ABSTRACT

Objective: To determine the incidence and types of associated anomalies in children with anorectal malformations.

Methods: One hundred and sixteen patients, with anorectal malformations seen at King Hussein Hospital between 1982-1996, were reviewed for associated genitourinary and other systems anomalies. Assessment of these anomalies was done by clinical, radiological, sonography and isotope scan.

Results: Out of 116 patients with anorectal malformation, high imperforate anus was found in 44.8% (male: 42, female: 10), and low anorectal anomaly in 55.2% (male: 26, female: 38). Genitourinary abnormalities were encountered in 38 patients. Associated congenital defects were seen in 45 patients: Urinary tract (24%), Cardiac (4.3%), Sacrum and vertebral deficit (8.6%), Genital (8.6%), Limbs (9.4%), Cloaca (5%), Esophageal Artesia (5%), Down's syndrome (3.4%), Hydrocephalus (1.7%) , Potter syndrome (0.8%), and others (10.3%). We observed that the incidence of associated anomalies was highest in males with high anorectal malformations (56%), and lowest (10%) in those females with low anorectal anomaly. Of the hundred and sixteen patients, 68 were males and 48 were females.

Conclusion: The genitourinary tract is the most serious and the commonest site to have associated defects in patients with anorectal malformations. The higher the malformation the more frequent is the associated anomalies with severe urologic problem. Early diagnosis and treatment should be carried out to prevent sequale of fatal associations.

Key words: Anorectal malformations, Urogenital, Additional association.

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Introduction

Anorectal malformations (ARMs) have been a topic of concern for the pediatric surgeons all over the world. Most studies in the literature have either concentrated on onset of associated anomalies or a subset of them. Others studies have given an overview of spectrum of anomalies but have not gone into detailed analysis.[1]

Reported associated congenital anomalies including renal and bladder agenesis (Potter syndrome),[2] ectopic bilateral vasa differentia into the posterior urethra (unpublished), mature sacral teratoma, gastroesophageal reflux and pyloric stenosis. It is well known that children with ARM have a high incidence of associated structural genitourinary anomalies and other systems.[3] Many of the anomalies can be explained by an event in early fetal life.[4] During weeks 5 to 7 of gestation there is almost simultaneous division of the cloaca into the bladder and anorectal canal, development of the mullerian, wolffian and urethral structures and formation of the lumber spine. Any arrest or insult to fetal development at this stage may result in a combination of anorectal, urinary tract, genital and lower spinal anomalies.[5] Shortly after the first month of embryologic development, the urorectal septum grows caudally to divide the cloaca into the anterior urogenital sinus and the rectum posteriorly.

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Abnormalities of embryogenesis occurring at this time may result in imperforate anus.[6] Associated anomalies have been well characterized, and they occur more frequently with high anorectal lesions.[5] Many large series were published that revealed a high incidence of urologic anomalies in infants with imperforate anus.[6] Urinary tract problems are common in patients with imperforate anus with a reported incidence of 26-50.[5] Herein, we discuss 116 children with anorectal malformations (ARM). The purpose of this study was to evaluate the frequency of coexisting urinary and other malformation of other systems that coexists in patients with ARM eg, vertebral, neurologic, cardiac, and genital.

Further, we also tried to find out the pattern of coexistence of these anomalies and compare our observations with those existing in the literature.[1] The VACTERL association encompasses defects in multiple organ systems, including vertebral anomalies, imperforate anus, cardiac lesions, tracheoesophageal, anorectal; and genitourinary malformations (V=vertebral; A=anorectal; C=cardiac; TE=tracheoesophageal fistula with esophageal atresia; and R= radial limb or renal) .Genitourinary anomalies are frequently associated with imperforated anus even in the absence of other features of VACTERL syndrome.[7] These anomalies not only lead to the overall mortality but also contribute to significant morbidity.[1] Comparing their distribution among subgroups based on the sex and nature of ARM (High/ Low), an aspect that had been deeply highlighted in this study.

The associations of imperforate anus and structural genitourinary anomalies, including recto-urinary fistulas, renal agenesis, ureteopelvic junction obstruction, vesicoureteric reflux, cryptorchidism and hypospadias, are well known. After repair of imperforated anus, we must attend to the symptoms caused by these genitourinary abnormalities.

Death from renal failure is reported in up to 6% of children with high anorectal anomaly. In recent year, advances in renal transplantation and the management of end-stage renal disease (ESRD) have extended these therapies to infants in the first 2 years of life.[7-9]

Methods

Records of 116 patients were available for study. There were 68 males and 48 females, their age at presentation was between one day and 13 years**,** medical and radiological reports of these patients were reviewed. Data regarding patient gender, anorectal lesions, the presence of urinary tract, genital, spinal and other systems anomalies were recorded. The results of clinical examination, invertograms, echocardiogram, abdominal sonography were included. Some investigations were performed selectively on few patients who had specific indications; eg, micturation cystourethrography (MCUG), in patients with abnormal ultrasonography of the abdomen. MRI spine when clinical stigma of spina bifida, sacral, or vertebral deformity were present. Children with low imperforated anus "infralevator" in which p-p distance was less than 1 cm on invertogram was seen in covered anus or cutaneous perineal fistula.

All other children were classified to have a supralevator (high) lesion. All patients with high type imperforate anus had temporary colostomy shortly after birth (within 24-48 hours) as an initial operation. Urinary tract anomalies were defined as any renal, ureteral, or bladder malformation, detected by radiographic evaluation. Eighteen categories of anomalies were classified in our patients: Urologic, cardiac, vertebral, limbs, GIT, spine and spinal cord related, genital, and miscellaneous. Data according to the type of anomalies and sex of the patients were enrolled in 4 subgroups (Table I).

The girls with cloacal abnormality (no=6) were classified as high ARM because all these subjects had a Trans or supralevator pouch. The data were analyzed to see if significant difference in the incidence of associated anomalies was present within the above-stated subgroups. We also compaired our findings with those of other reports in the literature. Genital malformations were defined as any anomaly of the penis, or scrotum in boys and of introitus or vagina, in girls. Specific imaging for uterine lesions was not performed in girls, but all patients underwent adequate clinical examination of the external genitalia and therefore, genitalia were noted to be normal or abnormal. Patients were considered to have a spinal anomaly whether the anomaly involves the bony spine or spinal cord. Sacral anomaly was defined as any defect of the sacral segments. Follow up of all children ranged from 2 to13 years.

Results

Out of 116 children with anorectal malformations, there were 58.6% males (n=68) and 41.4% females (n=48). The majority had presented in the neonatal period. High imperforate anus was found in 52 patients, including 6 cases of persistent cloacae. Low malformation were found more frequently among females (n=38) than males (n=26). Female to male ratio 1.4:1.

Concomitant urogenital anomalies were found in 38 patients (32.7%). Of these (n=21) occurred in males with high type anomaly, and (n=6) with low type. While in females (n=5) with high type, and in 6 of low type. (Table II).

Table I. Incidence of congenital anomalies in children with anorectal malformations

Type of Anomaly	High Types		Low Types			
	Male	Female	Male	Female	Total	Mortality
Urogenital	23	3	8	4	38	1 death
Sacrum defects	6	1			7	
Vertebral		1		1	2	
Cardiac	3	1		1	5	
Limbs	8	2	1		11	
CDH			1	1	2	
Pectus			1		1	
Ribs			1		1	
Hemangioma trunk			1		1	
Sacrum (teratoma)	1				1	
Sacrum (meningocele)	1				1	
Down's syndrome	2		2		4	1 death
Potter syndrome	1				1	1 death
Gastroesophageal reflux				1	1	
High arch or cleft palate	1		1		2	
Microcephaly + Micropthalmia	1				1	1 death
Hydrocephalus	2				2	1 death
Esophageal atresia	3	1	1	1	6	1 death
Total %	52 (60%)	9 (10%)	17 (20%)	9 (10%)	87	

Table II. Incidence of urogenital anomalies in children with anorectal malformation

Type of anomaly	High type	Low type	0utcome		
	Male	Female	Male	Female	
Non functioning kidney (lt) Hydronephrotic (Rt)		1			
Cystic dysplastic kidney (It)	1				nephrectomy
Rudimentery kidney and ureter (Rt)		1			nephrectomy
Ectopic herseshoe kidney (Rt)	1				
Single kidney (Vesicoureteric reflux) (VUR)	1				chronic renal failure
Single hydronephrotic kidney	1				death
Single ectopic kidney	1				
Single orthotopic kidney	3				
Dilated P.C system and ureter	2	1	2	1	
Vesicoureteric reflux	1			1	
Ectopic kidney (It)+ PUJ (Rt)				1	
Pelviureteric junction obstruction (PUJ)	1		1		
Hypoplastic kidney		1	1		
Ureteropelvic junction stenosis	1				
Hypoplastic kidney bilateral				1	
Ectopic kidney (Rt)+ hydronephrotic (It)+ bilateral (VUR)		1			chronic renal failure
Renal agenesis poter's syndrome	1				death
Hydrocele (Rt)	1				
Hypospadias + scrotal deformities	2			2	
Cryptorchidism	4		2		
Total %	21 (55%)	5 (13%)	6 (16%)	6 (16%)	

Structural genitourinary anomalies**:** Hydronephrosis, single kidney with or without ectopia were the most common associated anomaly in our study with preponderance in high male anomalies. The primary VUR was found in only (n=3)), Ureteropelvic junction obstruction (PUJ) (n=3), Ureteovesical junction stenosis (n=1), Cryptorchidism (n=6), Hypospadias (n=4), Potter's syndrome (n=1) (Table II).

Additional associated congenital defects were seen in 45 patients; the incidence was 3 times more in males than in females, and six times more in males of high type anomaly than those of low (6:1) (Table I).

Down syndrome (n=4**),** Extremities (limbs)

malformations had an overall incidence of 9.4%. Cardiac defects, esophageal atresia and cloacal in nearly 5% in each. (Table I).

Sacral related: deficit, meningocele, and teratoma in 7.7%. Here, it is worth mentioning that we have seen pyloric stenosis in two of our patients who presented late at age of 4 and 6 week respectively, and ectopic severely dilated bilateral vasa deferens to the posterior urethra in one patient, the diagnosis was made by cystourethrography, cystoscopy and by surgical exploration (unpublished), which may represent a new association.

On the other hand, patients without associated anomalies significantly appear in those with low ARMs, particularly females (n=34) and males (n=15).

Males with high anorectal anomalies (ARA) had significantly higher incidence of all types of anomalies (Table I).

Mortality was seen only in those with high ARA (4 male and 1 female). Additionally, when the patients with multiple anomalies were studied, simultaneous occurrence of associated anomalies affecting 3 systems were found in 22 patients (18.9%), 4 systems in 8 patients (6.8%), and 5 systems in 7 patients (6%).

All components of VACTERL were present only in one male patient, who was managed abroad. Nephrectomies were done in 2 patients either due to dysplastic or rudimentary kidneys .Chronic renal failure developed in two of our patients: due to a single refluxing kidney in one, and bilateral vesicoureteric reflux in the other (Table II).

Mortality of (5.1%) was related to associated anomalies (Renal, pulmonary failure, and malignancy), and not to the imperforate anus. We observed that the incidence of associated anomalies was highest in males with anorectal malformations (56%), and lowest (10%) in those females with low anorectal anomaly. From data of the 4 subgroups, interesting conclusion was drawn (Table I),

Discussion

Anorectal malformations (ARMs) are one of the common congenital anomalies encountered in the newborn period. Congenital anomalies of the genitourinary tract are the leading associated anomalies in infants with ARM.[1] In general an incidence of associated genitourinary anomalies ranging from 26% to 50% has been reported, which is the most serious and frequent site of associated defect in ARM. The higher the malformation the more frequently is associated with severe urologic problem.[4] Their frequency and variety influence prognosis and survival rate. All kinds of associated anomalies are possible but there are some typical combinations.[10] Lesions coexisting with imperforate anus increase the challenge of managing such cases, not only can these anomalies be a source of morbidity, but they can be a source of life threatening complications.[5] Although, the incidence of associated anomalies was found to vary widely in different studies.[1] The risk for both sets of problems increased with the level of the anorectal lesion.[9] In our study, there was a higher prevalence of ARM among boys especially who had high type of anorectal anomaly (ARA). Boys were much more likely to have a genital or upper urinary tract problem than girls (10 or 0 versus 29 or 9).

While in the literature, reported children with congenital ARA, 25 to 55 percent have urogenital tract abnormalities.[4] In our series the incidence of urogenital anomalies was 55.2% in high types males and 15, 7% in low type males, Versus 13% in high type females and 15.7 % in low types. (Table II) These results show the need for evaluation of urinary tract during the neonatal and early infantile period in all children even in low type of lesion. So, the different functional and anatomical abnormalities can be examined and treated depending on the nature and severity of the lesion.Thus, urologic investigation in cases of high defects may represent a higher priority than the colostomy itself.[3] Hydronephrosis, urosepsis, and metabolic acidosis from poor renal function represent the main source of mortality and morbidity in newborns with ARM.

The urological malformations associated with ARM are not only anatomical ,but also functional, the later being related to congenital neurovesical dysfunction (NVD) (5.7% to 32%) encountered in the literature.[7] Urinary and fecal incontinence often are the only presenting symptoms, not only related to the heights of anorectal defect, but also to the state of the spinal cord , however because urinary incontinence is frequently accepted as normal for the pediatric population, the diagnosis of neurogenic dysfunction will be delayed in many children which can result in advanced urinary tract disease and upper tract deterioration.[3] Sacral anomalies were found in 9 children (7.7%) most of them had partial sacral agenesis. NVD may however occur even in the absence of sacral anomalies.[11,12] The extraordinary high incidence of unsuspected lesions known to cause progressive bowel, bladder, and musculoskeletal dysfunction.MRI or US tomographies of the spine as an essential tool in the diagnostic work-up in patients with imperforate anus is recommended.[5,12] Children with sacral agenesis should receive urodynamic investigation within the first weeks of life to begin appropriate urologic treatment.[3,11,13]

In addition authors also recommend postoperative urodynamic assessment in boys with rectouretral fistula and girls with persistent cloacae to detect dysfunction caused by iatrogenic denervation of the pelvic nerves and nerve plexus injury.[8]

In children and myelodysplasia, long term follow up is mandatory especially those who have lower urinary tract dysfunction so that upper tract deterioration can be prevented.[13-15]

Although the incidence of primary VUR which is considered functional rather anatomical, ranges from 0.5% to 18% in asymptomatic patients; it is quoted to be around 35% among children and ARM, on average the literature reveals an overall incidence reflux of 20% with 39% in high disorders and 20% in low defects.[1] In our study, VUR observed in 3 patients (2.5%) of whom two were, males developed chronic renal failure. We admit that this low incidence was a result of our attempt to perform definitive investigations such as MCUG in which either a clinical or radiological indication existed. Authors recommend a renal and bladder u/s and voiding cystourethrogram in the early neonatal period in all patients with ARM.[9] Nuclear medicine renography is useful in selective circumstances. Because reflux nephropathy and neurovesical dysfunction represent preventable causes of renal deterioration, consideration should be given for a completely diverting colostomy especially in the presence of reflux or already compromised upper urinary tract.

Among the upper urinary tract anomalies hydronephrosis, single kidney and ectopia were most common and documented in 15/38 (39.4%) followed by dysplastic, rudimentary or hyperplasia in 5/38 (13%), pelviureteric junction obstruction (PUJ) in 3/38 (7.8%). So, renal dysfunction, nephrectomies or even death were encountered in six patients (5.1%), mainly due to single hydronephrotic, dysplastic or hypoplastic kidneys with or without VUR. These results show the need for early evaluation of urinary tract. Otherwise, these associated anomalies may go undetected and can be the cause of significant clinical morbidity and permanent renal damage in later life if not managed properly initially.

Regarding the association of the lower urogenital abnormalities in girls, Fleming *et al* reported 14% incidence of vaginal anomalies in 162 female patients, with imperforate anus, longitudinal vaginal septum was the most common anomaly. In our series this may be underestimated and may be partially explained by less thorough examination or documentation.[1,5] Leavitt *et al* recommend a good endoscopic examination, dye study, inspection at the time of surgery, MRI and ultrasonography of the female genital tract to detect and treat these anomalies before they get symptomatic.[1]

Whereas in boys the genital anomalies are more readily apparent, in our series this observation was documented in 11 patients (Table II). Six cryptorchidism, four hypospadias and one hydrocele.

In our series there were two mortalities in males with ARM from renal insufficiency the first from renal failure due to single hydronephrotic kidney and the second from agenesis of both kidneys bladders and ureters together with cleft palate and bilateral pnumothorax (Potter's syndrome).

In the English literature there are few patients reported to have bladder agenesis with ARM. Living patients with bladder agenesis have been described,[2,17] however, infants with imperforated anus and (ESRD) may successfully be managed by renal dialysis and renal transplantation co-ordinate with staged bowel reconstruction.[7]

Down syndrome as reported in literature ranges from 2.6% to 8%. In our series 4 cases (3.4%) were encountered, of these one died one year later because of AML and the other died at home for unknown cause.[10]

Knowledge of VACTERL association is important in evaluating the newborn with congenital abnormality. The VACTERL association is a tendency of concurrence rather than as absolute interrelationship, no specific genetic pattern for this association has been proven, while vertebral anomalies were reported to be a prominent one, they often cause severe intractable scoliosis in the growing child and they may be associated with neurological abnormalities, Limb anomalies being the prominent part of associated abnormalities in our series. (Table I)

The infant's morbidity from congenital anomalies is greatly increased by the number of lesions; early recognition of occult defects is therefore extremely important.[18]

Regarding the cardiovascular lesion, ventricular septal defect is the most common cardiac lesion.[18] No specific anomaly predominates in our 4 cases including both cyanotic and acyanotic heart disease.

Esophageal atresia is the 4th most frequent associated anomaly in our study (Table I). Other miscellaneous congenital abnormalities including cleft palate, pectus excavatum, suprnumery ribs, hemagioma of the trunk, pyloric stenosis, and gastro-esophageal reflux have been seen in our series. Lately we had a male patient found to have bilateral ectopia of vasa deferentia to the posterior urethra in association with high imperforate anus, postoperatively presented with recurrent epididemo-orchitis (new association).

As in the earlier studies, boys were found to have higher incidence of urologic anomalies.[1] Various factors could be responsible for this observation, such as the overall higher incidence of high ARM among boys with increased incidence of associated anomalies, the higher incidence of communicating fistula of the rectal pouch with urologic tract in boys could be another reason. Thus, among patients with ARA, male sex is certainly a disadvantage.[1]

Lesions coexisting with imperforated anus increase the challenge of managing these cases. Not only can these anomalies be a source of morbidity, but they can be a source of life threatening complications. A five percent mortality rate with the cause of death directly related to conditions other than imperforated anus in our series (n=6) were due to coexisting lesions (Table I).

Conclusion

The genitourinary tract is the most serious and the commonest site to have associated defects in patients with anorectal malformations. The higher the malformation the more frequent is the associated anomalies with severe urologic problem. Early diagnosis and treatment should be carried out to prevent sequale of fatal associations.

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