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REVIEW ARTICLE

Congenital urological anomalies diagnosed in adulthood — Management considerations

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Abstract *Objective:* Despite worldwide availability of prenatal ultrasound, many patients are diagnosed in adult life with congenital anomalies such as ureteropelvic junction obstruction (UPJO), undescended testicle (UDT), ureterocele, hypospadias, vesicoureteral reflux (VUR) and primary obstructing megaureter (POM). The aim of this review was to describe these clinical conditions and their suggested management based on the available medical literature. *Review:* Adult UPJO is not a rare condition; symptomatic patients should be treated rather than observed. Treatment options are nephrectomy for non-functioning kidneys and reconstructive surgery for functioning renal units. The adult UDT has low fertility potential and increased cancer risk; hence most of the data in the literature indicate performing an orchiectomy. Adult ureteroceles are usually related to single systems and they are intravesical and less obstructive. For symptomatic patients endoscopic incision showed high efficacy for symptom elimination with minimal side effects. Primary hypospadias correction in the adult patient is feasible, but success rates are low compared to the pediatric age group. Secondary correction, whether primary correction was performed in childhood or adulthood, is a challenging task with a high complication rate. Treatment decisions regarding adult patients with VUR are difficult to make as the available data are inconsistent; there is no strict evidence that reflux in an adult is directly related to renal growth impairment, ascending pyelonephritis, and/or embryo loss in a pregnant woman. In contrast to the pediatric age group, adult POM is usually a symptomatic condition and related to a high complication rate including infections, stone formation and renal failure. Spontaneous resolution is rare and hence active surgical management is advocated.

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Conclusion: Congenital urological anomalies identified in adulthood are not rare and pose a management challenge to the urologist. For most of the reviewed diseases, evidence-based management direction is difficult due to a lack of randomized trials and long-term follow up.

Introduction

The urology system is easily demonstrated in prenatal sonography [1]. Prenatal identification of congenital urological anomalies has tremendous impact on the postnatal baby's life. Prenatal parental consultation enables explanation of the predicted anomalies and planning a postnatal follow-up regimen. Preparations for a high-risk baby with urologically related anomalies may reduce antenatal mortality and morbidity [2]. Congenital anomalies diagnosed in adulthood may require a different perspective than in children regarding management and surgical correction. Here we present descriptions of some of the anomalies for which surgical management is an option in children. We propose management options according to the current medical literature.

Adult Ureteropelvic Junction Obstruction (UPJO)

The incidence of UPJO in the pediatric age group is well defined, affecting around 60% of all newborns with hydronephrosis. The exact incidence of primary congenital UPJO in adults is unknown, although estimated as not uncommon [3]. In contrast to prenatal screening, adult UPJO may be discovered in several ways: (a) evaluation following symptoms such as chronic back pain, acute renal colic, especially after fluid overload, hematuria, UTI and pyelonephritis; and (b) incidentally during diagnostic imaging of the abdomen or spinal area for other problems such as abdominal or back pain. Diagnosis of adult UPJO does not differ from that of the pediatric age group, using nuclear medicine static and dynamic renal scans, intravenous pyelography, or CT urography. Unfortunately, there is no consensus regarding which is the optimal imaging modality to determine obstruction. Moreover, there are no uniform standards for how to perform the above-mentioned diagnostic tests, and various protocols exist. Management decisions in the adult are quite different. In children, the main goal is to eliminate the obstruction in order to restore maximal renal function and to allow maximal growth of the kidney. In adults, the ability to restore renal function or to allow compensatory renal growth following surgery is limited, as chronic obstruction may have caused irreversible changes in the renal parenchyma and vasculature, while maximal growth potential has already been achieved. Factors that should guide the physician regarding the management of adult UPJO are: symptoms, function, age and co-morbidities.

Symptomatic patients with UPJO should be treated when symptoms endanger the patient: recurrent UTI, pyelonephritis or hypertension can be life threatening, especially in elderly patients with other co-morbidities. Non-life-threatening symptoms such as chronic pain that interfere with a patient's quality of life may also be an indication for definitive treatment. Definitive treatment includes reconstructive surgery to eliminate the obstruction or nephrectomy. In order to choose the most suitable arm of treatment for the patient, several factors should be taken into consideration: the function of the obstructed kidney, the function of the other kidney, the patient's age and co-morbidities. A non-functioning symptomatic kidney in the presence of a normal functioning contralateral kidney should be removed. In patients with a single kidney or those with overall poor renal function, reconstructive surgery should be offered.

Nephrectomy in adults is a well established operation. The decision between open and laparoscopic routes should be made by the patient and the urologist based on individual circumstances. Both can achieve the surgical goal successfully, with the advantage of a shorter recovery period with less analgesic consumption in patients who undergo laparoscopic nephrectomy [4].

Reconstructive surgery in adults can be performed by a wider range of surgical techniques than in children. The open surgical approach — open pyelopasty — is still considered the gold standard for UPJO in adults, with a 91% success rate overall and 100% success rate for grade 1—3/4 hydronephrosis [5]. Laparoscopic and laparoscopic robotic pyeloplasty already show similar success rates to open surgery. Bauer et al. [6] compared 42 laparoscopic pyeloplasties to 35 patients who underwent open surgery, and the overall success rates were comparable (98% and 94%, respectively). Robot-assisted laparoscopic pyelopasty also showed comparable success rates with minimal complications and short recovery time [7,8]. In contrast to the pediatric age group, adult patient may choose to undergo endoscopic correction of the UPJO. Endoscipc endopyelotomy has the advantage of being a minimally invasive procedure that may be performed as ambulatory or day-care surgery, with short recovery time and rapid (within 24 h) return to normal activity. Due to its minimally invasive nature and the ability to perform the procedure under regional anesthesia, endoscopic endopyelotomy is more suitable to patients with co-morbidities. The idea was originally described by Wickham and Kellet [9] who, using an endoscopic urethrotome via an antegrade endoscopic approach, performed a full-thickness incision of the upper ureter and PUJ. Among the three initially treated patients, two achieved improved drainage. Danuser et al. assessed the results of 212 consecutive antegrade endopyelotomies over a period of 8 years and showed overall 85% success. With the introduction of smaller endoscopes with better visual imaging and the use of laser energy antegrade and later on retrograde, endopyelotomies became even easier to perform. Today, endopyelotomy for UPJO in adults is considered as first-line treatment in various urological centers [10]; however, more recent reviews with longer-term follow up show that success rates are worse than previously reported. Dimarco et al. [11] assessed the long-term results

of antegrade endopyelotomy (182 patients) and open pyeloplasty (175 patients). The estimated 3-, 5-, and 10-year recurrence-free survival rates for the endopyelotomy group were only 63%, 55% and 41%, respectively, compared to 85%, 80% and 75% for the pyeloplasty group ($p < 0.001$). Additional crucial information arising from this study is the fact that failures continue to appear after 5 and 10 years, and patients should be followed accordingly. Rassweiler et al. [12] showed similar results comparing the success rates of laser endopyelotomy (113 patients) and laparoscopic pyelopasty (143 patients). The laparoscopic procedure showed a superior overall success rate of 94.4% versus 72.6%.

In summary, for the adult patient with UPJO various optional treatment modalities are available: nephrectomy or a reconstructive procedure. Although minimally invasive endoscopic techniques enable almost any patient to have definitive treatment, recent reports show disappointing results and late appearance failure. Patients should be followed beyond 10 years, and given advice regarding the higher success rate of 'formal' open and laparoscopic pyeloplasty.

Adult cryptorchidism (undescended testis, UDT)

In the pediatric age group, orchidopexy at an early age is guided by the need to preserve functions being damaged by the inappropriate anatomic position of the UDT, i.e. sperm production and testosterone secretion. Unfortunately, spermatogenesis in the UDT decays with time and after the age of 2 years the rate of germ-cell aplasia irreversibly accelerates. Rogers et al. [13] analyzed the histology of 52 resected UDTs. Patients' mean age at surgery was 26 years, and among the 52 specimens only one (1.9%) testis showed normal spermatogenesis. In the remaining testicles the histology slices showed Sertoli cells only in 30 (58%) patients, maturation arrest in 15 (28.5%), and testicular agenesis in six (11.5%). The authors concluded that the majority of UDTs cannot contribute to fertility. Grasso et al. [14] produced similar results by performing testicular biopsies in 22 patients who had postpubertal orchidopexy for cryptorchidism. More than 83% of the biopsies showed azoospermia and severe oligospermia. Although Leydig cells are less vulnerable to damage, endocrine function is also impaired in the adult UDT [15].

Concerning malignancy it has already been shown that orchidopexy does not prevent or change the rate of testicular cancer [16]; hence orchidopexy may only improve the ability to palpate the testicle. Due to its abnormal location, UDT may cause discomfort and undergo torsion. Zilberman et al. [17] showed that the rate of salvaging UDT torsion is lower than for normally positioned testes. Among 11 patients with torsion of UDT only two testicles (18%) remained viable; five had massive necrosis during surgery and had to be resected and four vanished post orchidopexy.

In summary, the majority of adult UDTs have very low fertility potential, impairment of endocrine function and increased risk of testicular cancer. Hence, in patients with a normal contralateral testicle, UDT orchiectomy should be offered. In patients with a single testis or bilateral UDT, preservative management may be considered mandating careful follow up and awareness of the patient and physician of the possible complications.

Adult ureterocele

In the pediatric age group ureterocele may be related to a wide variety of complex anomalies, such as duplex kidney, ectopic ureter, bladder outlet obstruction, incontinence and reflux. In adults most ureteroceles are related to a single system. They are intravesicaly located and the degree of obstruction is less severe. Presentation in an adult could be either by flank/back pain and recurrent UTI, or asymptomatic hydronephresis detected incidentally. Stasis at the ureteral meatus due to the partial obstruction of the ureterocele may induce stone formation inside the ureterocele, causing renal colic and/or UTI. A single case report in the English medical literature described a patient who deteriorated to renal failure due to bilateral ureterocele [18]. Diagnosis can be established with ultrasound or intravenous pyelography in patients with normal renal function. Ultrasound demonstrates a cystic mass within the bladder; Doppler mode can demonstrate a urine jet at the meatus of the ureterocele with volume changes during urine expulsion from the meatus. With evaluation of renal parenchyma, degree of hydronephrosis and jet sign with the advantage of no radiation energy exposure, ultrasound may be a sufficient tool to diagnose and follow up adult patients with ureterocele [19]. Intravenous pyelography may demonstrate the classic picture of the 'cobra head' intravesically. In addition, renal function, excretion delay and degree of hydronephrosis can be assessed. Management should be based on symptoms and renal function. Symptomatic patients should be treated. In the case of no or poorly functioning kidney with normal contralateral kidney and normal renal function nephrectomy should be offered. For symptomatic functioning kidney endoscopic incision of the ureterocele with stone fragmentation will lead to symptom elimination with minimal morbidity. Chtourou et al. [20] described 20 patients with ureterocele diagnosed due to chronic back pain; the mean age was 48 years, 16 had a single system and four duplex. All had an endoscopic incision and, in the presence of stones, fragmentation. Elimination of pain was successfully achieved in all patients. In a single patient the procedure was complicated with sepsis and one patient developed transient VUR. Vasu et al. [18] described a rare case of bilateral ureterocele causing progressive renal failure that reversed following bilateral incision. Although these are single case reports, such complications should be born in mind.

In summary, adult ureteroceles are usually intravesical and only mildly obstructing. Although it seems logical to treat symptomatic patients with endoscopic incision, physicians should be aware of the lack of data in the medical literature; hence our recommendations for adults are based on scanty reports.

Adult hypospadias

Distal hypospadias in itself poses only a cosmetic problem, and sometimes may cause deviation of the urinary stream. Erectile function is fully preserved and is not related to the anomaly. Ejaculation and fertility are not impaired. There are very little data regarding hypospadias correction in adults. Adayener and Akyol [21] reviewed the results of primary distal hypospadias repair in 80 adults and secondary

repair in additional 17. The location of the meatus was glanular in six, coronal in 35 and subcoronal in 56. Operative technique used was meatal advancement in 42, Mathieu in 41 and tubularized incised plate in 14. Overall success rate was 91.3%. However, the meatal position-related success rate was slightly different with 91% success for glanular and coronal hypospadias and only 85% success rate for subcoronal hypospadias. This rate is low compared to the pediatric age group. Senkul et al. [22] achieved a 89.9% success rate operating on 59 adults with a mean age of 22 years, among whom 48 had distal, nine mid-shaft and two proximal hypospadias. Operating on secondary hypospadias yields much lower success rates, with no differences in complications and failure rates noted whether the failed primary correction was performed in childhood or in adulthood. Barbagli et al. [23] assessed 60 adults with complications following pediatric hypospadias surgery; 36% of the patients had one complication and 64% had two or more. Complications included stricture 34, residual hypospadias 26, fistula 18, meatal stenosis 11, penile curvature nine, hair four, diverticula two and stone in one. Twenty-nine patients had one-stage repair with buccal or skin grafts or direct repair, and 31 underwent multistage repairs with buccal or skin grafts. Forty-five (75%) patients had a final successful outcome, 15 (25%) failed. One-stage repair provided 24 (82.7%) successes and five (17.3%) failures. Multistage repair provided 21 (67.7%) successes and 10 (32.3%) failures. The authors concluded that adults with complications following childhood hypospadias repair are still a difficult population to treat with a high failure rate for reoperative surgery. Senkul et al. showed a 27% complication rate for second attempt hypospadias correction in patients operated in adulthood.

For comparison we review results from the experience in the pediatric age group. Snodgrass and Yucel [24] reported about 30 pediatric patients who had tubularized incised plate repair for mid-shaft hypospadias. Fistulae were noticed in 3/30 (10%) and an additional two patients (6%) had one stricture and one glans dehiscence. Cheng et al. [25] reported about 514 pediatric patients operated for distal (414) and mid-shaft (100) hypospadias. For the distal repair no fistulae were reported and only a single case (0.2%) of stenosis, in the mid-shaft group three (3%) fistulae were noticed and one (1%) case of urethral stenosis. Combining both groups overall complications were less than 1%. Hammouda et al. [26] with at least 6 months' follow up showed that 4% (2/48) of patients who had tubularized incised plate repair developed fistulae and 10% (5/48) meatal stenosis. Duncan and Snodgrass [27] reviewing 26 articles comprising around 2035 pediatric patients operated for distal hypospadias revealed an average complication rate of only 9%.

In summary, primary and secondary hypospadias correction in adults is feasible. Surgical success rates are lower and there is a high complication rate compared to the pediatric age group. Patients should be informed regarding these data.

Adult reflux

The treatment of pediatric reflux underwent tremendous changes in the past decade. Open surgery was replaced with conservative medical treatment and minimally invasive endoscopic procedures. Although a high percentage of patients with reflux will resolve spontaneously, in 10—40% reflux will persist (depending mainly on grade and patient age). Symptomatic reflux, causing new renal scars, and deterioration of renal function should be treated; however, the proper management for asymptomatic persistent reflux in the adult is yet to be determined.

Consideration of increased risk in the adult patient with reflux may arise for several reasons. In females, adulthood may be related to an increased rate of bacterial UTI during sexual activity and at menopause. Pregnancy is related to overall urinary tract dysfunction, bacteriuria and UTI. Above all the chance of spontaneous resolution of the reflux in adulthood is low. On the other hand, the adult kidney is more resistant to infection, is less susceptible to developing new scars following infection, and the kidney growth potential has already been achieved. Based on the available medical literature it is not possible yet to determine the exact effect of asymptomatic reflux on the adult patient. Choi et al. [28] assessed 86 adult females with pyelonephritis for the significance of VUR. All patients underwent VCUG, 31 of them at the 3rd treatment day and 55 at the 7th day. Only two patients (2.3%) had low-grade reflux. Hence the authors concluded that, unlike in children, VUR is not a significant factor causing ascending infection leading to the development of acute pyelonephritis in adult women. el-Khatib et al. [29] assessed the influence of reflux on the course of renal failure in adult patients with reflux nephropathy. Among several factors studied the presence of persistent reflux had no independent influence on the course of renal failure; the most important factor that was associated with renal function deterioration was proteinuria.

Olbing et al. [30] showed in a 10-year follow-up study that even in the presence of high-grade reflux the appearance of new scars is rare after the age of 5; moreover there was no difference in new scar formation between patients treated conservatively and patients who were treated surgically. Olbing et al. [31] in a different study assessed renal growth in patients with severe reflux. The study compared renal growth in two groups of patients with high-grade reflux, one treated conservatively and the other surgically. There was no significant difference in renal growth during 10 years between surgical and medical treatment in patients with severe reflux. The question whether reflux can impair pregnancy outcome was studied in several works: el-Khatib et al. [32] studied the outcome of 345 pregnancies in 137 women with reflux nephropathy. Overall fetal loss was 48 (14%) of which only six (2%) were therapeutic abortions. Fifty-two pregnancies took place in women with plasma creatinine >0.11 mmol/l prior to conception. Fetal loss after 12 weeks' gestation (excluding therapeutic abortions) was 18% compared to 8% in the 104 pregnancies where maternal plasma creatinine was lower than 0.11 mmol/l ($p < 0.05$). Maternal complications were also more common in the impaired renal function group ($p < 0.001$). Comparison of pregnancies in women with unilateral versus bilateral renal scarring revealed no significant difference in fetal loss but an increased incidence of maternal complications in the bilateral renal scar group ($p < 0.01$). Persistent VUR was not associated

with increased fetal loss or maternal risk; however, impaired renal function prior to conception is associated with increased fetal loss and maternal complications in pregnancy.

In summary, currently there is no firm evidence that reflux in an adult is directly related to renal growth impairment, ascending pyelonephritis and/or embryo loss in pregnant woman with reflux.

Adult primary obstructing megaureter (POM)

In the pediatric age group a high percentage of patients with megaureters may be manage conservatively as long as renal function is good, there are no UTIs and hydronephrosis is stable. Although adult POM is uncommon patients are usually symptomatic and need intervention. Hemal et al. [33] described a series of 55 adult patients treated over a period of 12 years: 52/55 (94%) were symptomatic presenting with flank pain, 20/55 (36%) had calculi, and one (1.8%) had obstructive jaundice due to a huge hydronephrotic kidney. Tatlisen and Ekmekcioglu [34] described five patients with POM who all suffered from flank pain. Dorairajan et al. [35] reported a series of 37 adult patients with POM: 26 (70%) had flank pain, 15 (40%) UTI, 17 (46%) urinary calculi and five (13.5%) presented with azotemia.

Diagnostic imaging included ultrasound, intravenous pyelography, diuretic renogram and VCUG as in the pediatric age group. Ghersin et al. [36] used antegrade injection of contrast material and multi-detector CT to diagnose urinary tract obstruction including megaureters without the need for intravenous contrast material injection, so that patients with impaired renal function could also be evaluated.

In contrast to the pediatric age group, uniformly, the medical literature recommends active treatment in adults with POM [33,35,37,38]. Surgical management for functioning renal units includes ureteral neocystostomy (open or laparoscopic) and endoscopic endoureterotomy (electrocoutry or laser), and nephrectomy for non-functioning units. Hemal et al. [33] operated 41 patients for POM: 38 patients underwent reimplantation (21 with tailoring and 17 without) and three endoscopic ureteral meatotomy. Forty three of the 47 (%) showed improvement in collecting system dilatation, and the mean follow-up time was 7 years. In this series five (12%) patients with bilateral POM suffered already from renal failure on diagnosis, despite surgical treatment only one patient improved, and two died of renal failure. Tatlisen and Ekmekcioglu [34] performed direct nipple ureteroneocystostomy in five adult patients in order to avoid ureteral tailoring. Ansari et al. [39] successfully performed three laparoscopic ureteroneocystostomies for POM; ureteral tailoring was done extracorporeally followed by Lich-Gregoir reimplantation. Biyani and Powell [40] performed four endoscopic laser ureterotomies for adult POM; a full-thickness 2.5 cm long incision was performed on the intramural and juxtavesical part of the ureter followed by internal stent diversion. In a 24-month follow-up period all patients improved; a single female patient developed reflux. Bapat et al. [37] used a cutting current to treat six POM. Follow up of 1–4 years showed reduction of hydronephrosis and elimination of symptoms.

In summary, adult POM in contrast to children is usually symptomatic. Due to symptoms complications such as infections, stones and reduced renal function, and low spontaneous resolution active management is advocated. For functioning units, reconstructive surgery is advised; for poorly functioning units nephrectomy would be a better option. Ureteral reimplantation with or without tailoring is an established procedure. Endoscopic endoureterotomy showed good results, but should be evaluated for a longer term with higher numbers of patients.

Conclusions

Diagnosis of a congenital urological anomaly in an adult is not a rare event. Management considerations in the adult patient are sometimes different compared to the pediatric age group. In adults, more emphasis is given to symptoms, patient age and additional co-morbidities. The adult age group may benefit from a wider choice of surgical techniques that cannot be applied in children such as endoscopic procedures. Despite utilizing similar operational skills and techniques, surgical outcome in adults may be different from that in children, as is clear in hypospadias cases. In general, there is a relatively large arsenal of available therapeutic solutions to offer to adult patients with congenital urological anomalies.

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