Spectrum of Congenital Anomalies of the Kidney and Urinary Tract in Children

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ABSTRACT:

BACKGROUND:

Congenital anomalies of the kidney and urinary tract comprise a wide range of structural malformations that result from defects in the morphogenesis of the kidney and/or urinary tract

OBJECTIVE :

Study the types of renal anomalies. Study their clinical presentations, age of presentation, way of diagnosis, and complications.

PATIENTS AND METHODS:

A descriptive study was conducted from 1st January 2015 till 1st January 2016. Patients were collected from 4 pediatric nephrology centers. Data included: age of diagnosis, type of renal anomaly, radiological methods of diagnosis, clinical presentation and associated complications, family history of congenital anomalies and consanguinity.

Thorough physical examination was done to all children. Investigations send were Blood urea, Serum creatinine , urinalysis, urine culture, imaging study were documented.

RESULTS:

In this study 160 patients were included. Males were more affected than females 1.54:1. Most of the patients diagnosed at >1- ≤ 5 age group, 78 patients (48.8%). Most of the patients had negative family history, 124 (77.5%), and most of their parents were not consanguine (58.1%). Vesicoureteral reflux (VUR) was the commonest anomaly detected in 67 patients (41.9%), followed by Renal agenesis in 24 patients (15.0%).

Abdominal Ultrasound was the commonest radiological method used for diagnosis in 152patients (95.0%), followed by voiding cystourethrogram (VCUG) in 80 patients (50.0%).

Urinary symptoms were most common presentation in 93patients (58.1%). A higher complication was UTI (62.5%).

CONCLUSION:

The commonest renal anomaly was VUR, followed by Renal agenesis, then PUJ, These renal anomalies were mostly diagnosed at >1-≤ 5 age group. Males exceeded the number of females, and the majority of patients were diagnosed initially by ultrasound

Most common presentation was urinary symptoms. The most prominent complication was UTI

KEY WORDS: renal, anomalies, children.

INTRODUCTION:

Congenital anomalies of the kidney and urinary tract [CAKUT] comprise a wide range of structural malformations that result from defects in the morphogenesis of the kidney and/or the urinary tract. (1) This wide range of renal system structural and functional malformations that occur at the level of the kidney, collecting system, bladder, or urethra account for about 40–50% of children with chronic kidney disease (CKD), and are the most common cause of end-stage renal disease (ESRD) in children. (2, 3).

Nephrogenesis continues at a nearly exponential rate and is complete by the 36th wk of gestation. At least 16 signals agents have been identified that regulate renal development. Defects in any of signals activities could cause a kidney not to form (renal agenesis), to differentiate abnormally (renal dysgenesis). (4)

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Bilateral and unilateral renal agenesis occurs as an isolated, nonsyndromal, sporadic abnormality detected during prenatal ultrasonography. While the bilateral renal agenesis have incidence about 1 in 3000 births, the unilateral renal agenesis have incidence about 1 in 500-800 births.[5]

Renal dysgenesis refers to maldevelopment of the kidney that affects its size, shape, or structure. The 3 principal types of dysgenesis are dysplastic, hypoplastic, and cystic. If the entire kidney is dysplastic with a preponderance of cysts, the kidney is referred to as a multicystic dysplastic kidney (MCDK), it have incidence of approximately 1/2000 [4]

Autosomal Recessive Polycystic Kidney Disease is an autosomal recessive disorder occurring with an incidence of 1 : 10000 to 1 : 40000 in which both kidneys are markedly enlarged and grossly show innumerable cysts throughout the cortex and medulla. [6]

Vesicoureteral reflux (VUR) is the retrograde flow of urine from the bladder up to the ureter or even up to the kidney.Most VUR results from congenital incompetence of the ureterovesical (UV) junction, a structure that matures through early childhood. VUR may be familial ; 30% to 40% of siblings of a child with VUR also have VUR. VUR may also be secondary to distal bladder obstruction or other urinary tract anomalies. VUR is most often identified during radiologic evaluation following a UTI. No clinical signs reliably differentiate children with UTI with and without VUR [4]

In pelvi-ureteric junction obstruction (PUJ), the proximal obstruction at the ureteric junction of the renal pelvis, resulting in reduced or absent urinary flow and a high-pressure renal pelvis. PUJ is increasingly diagnosed with maternal Ultrasonography (US) screening and it is the most common cause of antenatal hydronephrosis (7 in 1500 live births). Children may present with failure to thrive, flank pain, urinary tract infections, a palpable mass, hematuria, hypertension, or renal impairment.[7]

Posterior urethral valves (PUV) are a congenital urethral anomaly seen in boys due to an obstructing membrane. PUV accounts for 3–9% of all cases of hydronephrosis .Presence of PUV is often suspected antenatally, but is confirmed by a postnatal Voiding CystoUrethroGram (VCUG) [8, 9]

AIMS OF THE STUDY:

Study the types of renal anomalies. Study their clinical presentations, age of presentation, way of diagnosis, and complications.

PATIENTS AND METHODS:

This is a descriptive study, conducted from 1st January 2015 till 1st January 2016; patients were collected from Pediatric Nephrology Clinic and Pediatric ward from 4 pediatric hospitals in Iraq: Imamain Kadhimein Medical City, Central Child Teaching hospital, Children Welfare Teaching hospital and Karbala Pediatric Teaching hospital.

The study included 160 patients with already diagnosed renal anomalies, 23 patients of them had combined renal anomalies. Children with acquired anomalies, incomplete file records and incomplete investigations were excluded .

Data were collected from the patients file records for the following information: age of presentation, age of diagnosis of renal anomalies, family history of congenital anomalies, consanguinity between the father and mother, radiological methods of diagnosis, clinical presentation and associated complications. Missing data were taken from by direct interview with the patients themselves or their parents .

Thorough physical examination was done to all. Diagnosis of failure to thrive was considered if a child's weight is below the 5th percentile. [4]

Investigations included renal function test (blood urea, serum Creatinine), urinalysis, urine culture.

Cut values for blood Urea and Serum Creatinine were figured as reference values. [4, 9, 10]

Stages of CKD, Estimated GFR based on serum creatinine using Schwartz formula [4, 9, 10]

Imaging study were documented in the study are: US of abdomen, VCUG, Intravenous Urogram (IVU), CT-scan abdomen. Some patients have more than one image study. One patient had DMSA, and other one had retrograde Pyelography were done outside Iraq.

UTI was considered positive when patient had symptoms and/or findings on urinalysis, confirmed by a urine culture. [4, 11]

Presence of stone disease was confirmed by renal ultrasonography, plain abdominal radiograph, or computed tomography (CT) scan in some cases. [12]

Acute kidney injury is an abrupt loss of kidney function that develops within 7 days. [13]

Age of presentation was regarded as the age for early signs and symptoms developed. Age of diagnosis was regarded as time when confirmation by radiological study done for the diagnosis.

RESULTS:

One hundred sixty patients were studied with renal anomalies, were included in the study. Age at time of presentation was range between 1day to 16years, and the mean was 47.07±SD 49.89 months, while the age at time of diagnosis was range between prenatal diagnoses to 13 years old with a mean 31.36± 37.44 months. The most frequent age group at time of presentation was >1- ≤ 5 age group including 84 patients (52.5%). See table (1).

The most frequent age group at time of diagnosis also was >1-≤ 5 age group 78 patients (48.8%), followed by ≤1 year age group 61 patients (38.1%), Thirteen patients out of 160 were diagnosed prenatally (8.1%). See table (1). Regarding patients distribution according to gender, there were 97 males (60.6%) and 63 females (39.4%) with M/F ratio 1.54:1. See table (1)

Most of the patients had negative family history 124 (77.5%), and most of their parents were not consanguine (58.1%). See table (1)

Regarding types of Renal anomalies of the study group, VUR was the most frequently seen in 67 patients (41.9%), followed by Renal agenesis in 24 patients (15.0%).Fifteen patients with VUR were associated with Neurogenic bladder. Figure (1)

Urinary signs and symptoms were the most common clinical presentation seen in 93 patients (58.1%) that include ≥ 1 of the following: oliguria, dribbling, frequency, polyuria, gross hematuria, dysuria, turbid urine, and enuresis. Fever ranked second detected in 77patients (48.1%). See table (2)

Abdominal findings on examination included ≥ 1 of the following: abdominal mass, distention, pain. Urinary tract infection was the most frequent complication accounted for (62.5%), followed by hydronephrosis (39.4%). See table (3)

Abdominal Ultrasound was the most Radiological method used for diagnosis of these anomalies in 152patients (95.0%), followed by VCUG in 80 patients (50.0%). See table (4).

Table 1: Demographic characteristics of patients with renal anomaly.

		No.	%
Age (yr) Age of Presentation			
≤ 1		41	25.6
> 1 - ≤ 5		84	52.5
> 5 - ≤ 10		27	16.9
> 10		8	5.0
Age (yr) at diagnosis			
Prenatal Diagnosis		13	8.1
≤ 1		48	30.0
> 1 - ≤ 5		78	48.8
> 5 - ≤ 10		19	11.9
> 10		2	1.2
Gender			
Male		97	60.6
Female		63	39.4
Family history	Positive	36	22.5
	Negative	124	77.5
Consanguinity	Positive	67	41.9
	Negative	93	58.1

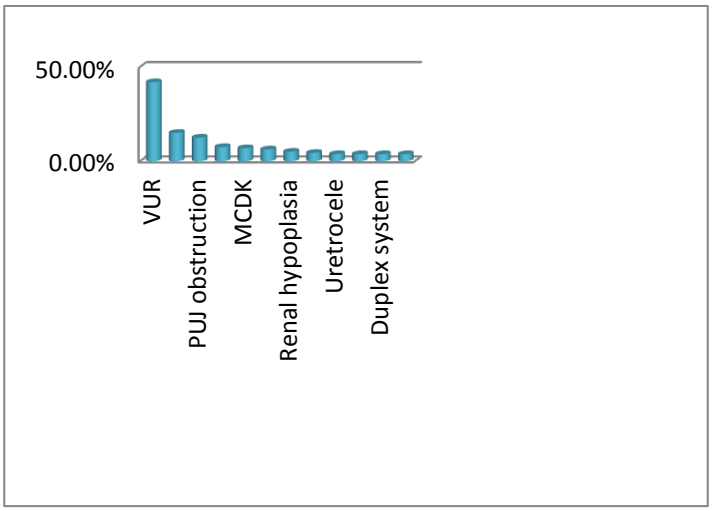

Figure 1: Patients distribution according to types of renal anomalies.

Table 2: Patients distribution according to clinical presentation.

Clinical presentation	No.160	%
Urinary symptoms	93	58.1
Fever	77	48.1
Abdominal symptoms	41	25.6
Acute kidney injury	15	11.9
Prenatal	13	9.4
Accidentally diagnosed	11	6.9
Improper Weight gain	10	6.3

*patients may have more than one clinical presentation

***Table 3: Patients distribution according to associated complications.**

Complications	No.160	%
Urinary tract infection	100	62.5
Renal impairment	55	34.4
FTT	38	23.8
Stones	15	9.4

*Patients may have more than one complication.

***Table 4: Patients distribution according to radiological methods of diagnosis of renal anomalies.**

Radiological method	No.160	%
Ultrasound	152	95.0
VCUG	80	50.0
IVP	41	25.6
CT scan	35	21.9
Retrograde Pyelography	1	0.6
DMSA	1	0.6

*Patients may have more than one radiological method of diagnosis.

DISCUSSION:

Age at time of presentation ranged between 1day to 16years, and the mean was 47.07 ± SD 49.89 months, while the age at time of diagnosis ranged between prenatal diagnoses to 13 years old with a mean 31.36 ± SD37.44 months. We notice that there was considerable delay between the age of presentation and that of diagnosis (about 1.5 year) which occurred due to fact that the study was done in tertiary centers and patients had seen by different medical centers before referral to these tertiary centers, or lack of the health awareness.

During comparison with Iraqi study by Faisal Ch. Hasoon,2009 [14] , which had age of presentation 15days- 11years with the mean 36 months and age of diagnosis from perinatal to 7 years with the mean 24 months, we notice there is nearly similar gap between age of presentation and age of diagnosis (about 1 year). In Turkey study by Bulum B *et la*, 2013 [15] , age range of presentation ranged from perinatal to 7 years with the mean 37.2±SD 44.4 months, and Egyptian study by Neveen A. *et la*,2015 [16] , the age of diagnosis ranged between 1day to 13years and the mean was 25months.

Prenatal diagnosis was done for 13 cases (8.1%) of the patients, which was less than Turkey study by Bulum B. *et la* (12.6)and Egyptian study by Neveen A. *et la* (36.6%) diagnosed prenatally[15,16]. This might be related to difference in antenatal care of pregnant women or the need for specialist of Diagnostic Radiologist.

Prenatal diagnosis was included with clinical presentation because it is the early indicator for the presence of renal anomaly which is confirmed after delivery.

Male predominance (60.6%) was evident with an approximate male/female ratio 1.54:1. In several studies from different regions of the world, male gender was predominant as well[14, 16, 17].

Positive family history was found in 36 patients (22.5%), this agree with the Hwang et la (USA) study [17] who detected 21.4% positive family history, and this confirms that renal anomalies are mostly sporadic [18,19] , however the Turkey study by Bulum B. et la [15] , found only 8.8% positive family history, that was due to selection of first degree relative.

Positive consanguinity was found in 67 patients (41.9%), which is nearly similar to Egyptian study [16] 49.5%, this reflects the high rate of consanguine marriage in Arab population.

In this study, VUR was the commonest abnormality which agree with other investigators [14, 15, 17, 18] . On the other hand Egyptian study [16] was disagree with our result in that the most common anomaly was PUV (36.4%), this might be due to collection of patients from Pediatric Urological Unit where PUV patients frequently visited.

We did not study the primary and the secondary VUR due to PUV or other causes separately .

Regarding clinical presentation of the study patients, urinary symptoms were the commonest (58.1%), followed by fever (48.1%), while Egyptian study reported abdominal symptoms (50.5%), and followed by urinary symptoms (37.3%). This difference might be due to the difference in the types of renal anomalies of the two studies which reflects different clinical presentation.

Similarly to Neveen study [16], UTI was the commonest complication.

Abdominal Ultrasound was the mostly used radiological study in 152 patients as an initial easy method of investigation. Confirmation of a lot of anomalies required another radiological method accordingly. High frequency of VUR and PUV in this study required VCUG as the method of diagnosis of these anomalies. In Iraqi study by Muneera Fadhil (2009), mentioned that (59.4%) of infants and children with symptomatic UTI had positive ultrasound findings, and (32.6%) of them had positive findings by VCUG. [19]

CONCLUSION:

The commonest renal anomaly was VUR, followed by Renal agenesis, then PUJ, These renal anomalies were mostly diagnosed at >1-≤ 5 age group. Males exceeded the number of females, and the majority of patients were diagnosed initially by ultrasound

Most common presentation was urinary symptoms. The most prominent complication was UTI

Recommendations : Development of a comprehensive treatment plan between primary care physicians, Urologist and pediatric nephrologists for children with renal anomalies in order to provide optimal care . With more efforts should be aimed at improving antenatal diagnosis.

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