Multicystic Renal Dysplasia, Bilateral

Alternative Names
MRD
Pelviureteric Junction Obstruction
PUJO
Hydronephrosis due to PUJO
Hydronephrosis Congenital

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations of the urinary system

Incidence per 100,000 Live Births
11-50

OMIM Number
143400

Mode of Inheritance
Autosomal dominant

Gene Map Locus
6p

Description
Multicystic renal dysplasia is a congenital dysplasia of the kidney characterized by large nonhomogeneous dilations of the collecting tubules. The dysplasias are usually unilateral, but it can be bilateral, segmental or focal, and the incidence is about 1 out of 5,000 to 10,000 births. There is a male predominance (ratio M2.4/F1) but females are twice as likely to have bilateral multicystic dysplastic kidney disease and associated non-renal abnormalities. Severe bilateral defects are lethal. Partial dysplastic involvement of both kidneys eventually leads to renal function impairment.

Molecular Genetics
Genetic studies have assigned one of the loci for multicystic renal dysplasia to the short arm of chromosome 6. A chromosome 19 breakpoint has also been observed in some patients with bilateral multicystic renal dysplasia, appearing to have occurred in intron 7 of the USF2 gene.

Epidemiology in the Arab World
Bahrain
Amin and Hameed (1984) reviewed 23 patients (18 males, 5 females) who had undergone surgery for congenital hydronephrosis. Most of the patients were adults, and the most common clinical presentation in them was loin pain. The younger patients presented with occasional loin pain, pyrexia and hematuria. Of these patients, 11 had a PUJO on the right side, whereas the remaining were obstructed on the left side. Only one patient showed bilateral obstruction. Amin and Hameed (1984) considered the Anderson Hynes pyeloplasty to be an effective surgical method, and performed the procedure on 18 of the patients. The remaining patients were managed using Culp-Deweerd-Scardino plasty, Y-V Foley plasty, or uretrolysis. At the time of the review, 22 patients continued to be symptom free.

Oman
Fernandes et al. (1998) presented a six week-old-boy with bilateral emphysematous pyelonephritis and cystitis secondary to bilateral peviureteric junction obstruction. He presented with fever, failure to thrive, abdominal distension, and loose stools with non bilious vomiting. He was found to be dehydrated and febrile with the left kidney palpable, and mild hepatomegaly was felt. Investigations revealed hyponatremia (125mmol/l), hyperkalemia (6.5mmol/l), urea of 9.4mmol/l, creatinine of 53mmol/l, and bicarbonate of 15mmol/l. On ultrasound scan, bilateral renal pelvises were dilated with highly reflective echoes in both kidneys (persistent with gas). IVU showed bilateral dilatation of the pelvicalyceal systems with gas bubbles in the

renal collecting system and the bladder wall which was also noticed on a plain abdominal X-ray. Posterior urethral valves and vesico-ureteric reflux were excluded by a cystourethrogram, and a DTPA renogram detected features of bilateral pelviureteric junction obstruction. Anderson-Hynes pyeloplasty was done on the right side five weeks after admission, and then four weeks later, on the left side. On follow up, he was thriving well, with no further attacks of urinary infection, and a repeat IVU after one year showed mild hydronephrosis with no signs of obstruction.

Qatar

Akl and Zayyoud (1983) reviewed the medical records of 83 children who presented with renal disorders in Qatar during a single year period. Three of these patients were diagnosed with uretero-pelvic junction obstruction.

Ehlayel and Akl (1992) reviewed the records of all patients in Qatar diagnosed with chronic renal failure between the years 1982 and 1990. Of the total of 30 such cases, four patients were found to have been diagnosed with PUJO.

United Arab Emirates

Abou-Chaaban et al. (1997) studied the pattern of pediatric renal diseases among children in the Dubai Emirate during the period from 1991 to 1996. In this period, a total of 712 pediatric patients, including 230 nationals of the United Arab Emirates, were seen with various renal problems. Among the patients with congenital renal anomalies, Abou-Chaaban et al. (1997) observed 12 subjects with pelviureteric junction obstruction.

Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January 1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Sporadic conditions accounted for 26% of the cases. In their study, Al Talabani et al. (1998) observed 15 cases of uni- or bilateral hydronephrosis born to non-related parents from the United Arab Emirates. No recurrence was reported in the family. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities overall in the United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi, the capital of United Arab Emirates.

References

Abou-Chaaban M, Al Murbatty B, Abdul Majid M. Spectrum of pediatric renal diseases in Dubai. Saudi J Kidney Dis Transplant. 1997; 8(3):310-3.

Akl K, Zayyoud M. The spectrum of childhood kidney disease in Qatar. Qatar Med J. 1983; 4(2):95-7.

Al Talabani J, Shubbar AI, Mustafa KE. Major congenital malformations in United Arab Emirates (UAE): need for genetic counselling. Ann Hum Genet. 1998; 62 (Pt 5):411-8. PMID: 10088038

Amin E, Hameed T. Retrospective study on 23 cases of congenital hydronephrosis. Bahrain Med Bull. 1984; 6(3):105-8.

Ehlayel MS, Akl KF. Childhood chronic renal failure in Qatar. Br J Clin Pract. 1992; 46(1):19-20. PMID: 1419547

Fernandes LP, Sajwany MJ, Derweesh A. Emphysematous pyelonephritis and cystitis associated with bilateral pelviureteric junction obstruction: a case report. J Pediatr Surg. 1998; 33(5):739-40. PMID: 9607484.

Related CTGA Records

N/A

External Links

http://www.amershamhealth.com/medcyclopaedia/volume%20IV%202/MULTICYSTIC%20DYSPLASTIC%20KIDNEY.ASP

http://www.emedicine.com/ped/topic1493.htm

http://www.humpath.com/article.php3?id_article=2418

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