CLINICAL CHARACTERIZATION OF RENAL CYSTIC DISEASES IN CHILDREN: A SINGLE CENTER EXPERIENCE

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OBJECTIVES

•Renal cystic disease (RCD) constitutes an important and leading cause of end-stage renal disease (ESRD) in children. RCD can be acquired or inherited; isolated or associated with extra-renal manifestations. The precise diagnosis represents a difficult clinical challenge [1,2].
•Aim of this work was to:
  –study the clinical phenotypic features of RCD.
  –define the etiology (clinical – imaging – histopathology).
•To our knowledge, this is the most comprehensive analysis of pediatric RCD reported from a regional single center.

METHODS

•Cross sectional study.
•All patients (up to 18 years of age) with RCD presenting to CPNT, Cairo University (major national referral center) through a period of 2 years.
•Clinical records, imaging studies and pathology reports of all patients with a diagnosis of RCD were reviewed.
•Patients were subjected to:
  –two generation family pedigree.
  –clinical and laboratory update including CKD staging.
  -abdominal ultrasonographic examination [3].
  -additional imaging studies and renal biopsy when indicated.

RESULTS

–105 patients belonging to 100 families.
–59 males and 46 females.
– Age: from 1 day to 17 yrs (median 2.7 yrs).
–21% were diagnosed antenatally.
– 5% were diagnosed at birth.
–The remaining 74% presented at a median age of 4.2 years (range 3 days – 10 years).
– Presumably inherited RCD represented 70.5%.
–NPHP (and related ciliopathies) represented the largest group (36%).
–The most common clinical features included polyuria (60%), UTI (31.4%) and hypertension (27%).
–49 cases reached ESRD.
–Extra-renal manifestations were present in 45 cases.
–Ultrasonography could determine the pattern of RCD in 95 patients (90.5%).
–10 patients required additional renal imaging.
–Renal biopsy was performed in 12 patients with a presumable diagnosis of NPHP.
–Two patients with ARPKD were biopsied; one because abdominal CT suspected malignant infiltration and the other was an excision biopsy of a large kidney at the time of transplantation.
–8 unclassified syndromic cases;4 of them with unclassified renal phenotype.
–They had various combinations of renal, neurological and dysmorphic features.
– Interestingly, 7 out of the 8 patients had early presentation in the first year of life. Fig 2 and 3 describe two of them.

Clinical classification of the studied cases

Group		Frequency	
PKD	ARPKD	31	25
	ADPKD		5
	Hypoplastic GCKD		1
NPHP	Juvenile	30	21
	Infantile		9
NPHP related ciliopathies	JSRD	8	5
	BBS		3
MCDK	Isolated MCDK	19	15
	MCDK with VUR		3
	MCDK with PUJO		1
Cystic dysplasia	Isolated	2	1
	Syndromic (McCune Albright)		1
Simple renal cysts		3	
Dialysis associated cysts		3	
PUV with calyceal cysts		1	
Syndromic; unclassified		8	

GCKD: glomerulocystic kidney disease, NPHP: nephronophthisis, JSRD: Joubert syndrome related disorders, BBS: Bardet-Biedl syndrome, MCDK: multicystic dysplastic kidneys, LAD: leukocyte adhesion defect

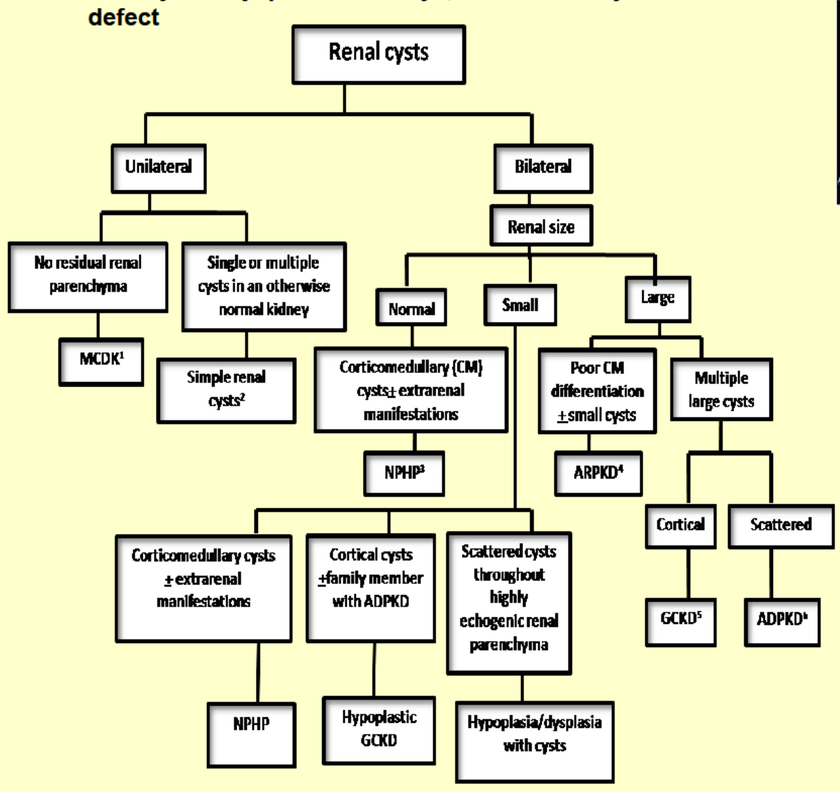

Fig. 4 Algorithm for diagnosis of renal cystic diseases.

1 DMSA scan should be done to confirm non-function.
2 Unilateral ADPKD may start as a solitary cyst and then cysts increase in number, so follow-up is necessary.
3 If no cysts could be detected by high resolution USS, renal biopsy should be done if molecular genetic analysis is not available.
4 The presence of evident congenital hepatic fibrosis supports the diagnosis.
5 Renal biopsy may be indicated to confirm the diagnosis of GCKD in doubtful cases. For GCKD, a parent may be also affected.

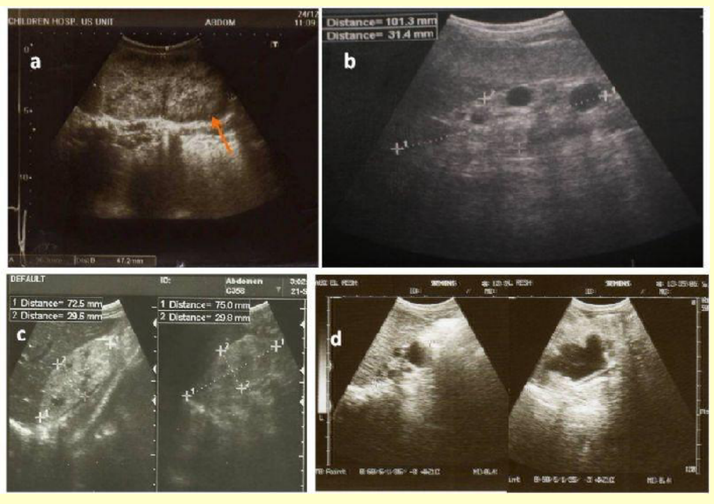

Fig. 1 a Renal USS of a 10 mo old boy with solitary ARPKD.
b Solitary ADPKD in a 9 yr old boy.
c Six yr old girl with juvenile NPHP.
d Right involuted MCDK and left PUJO in a 32 mo old boy.

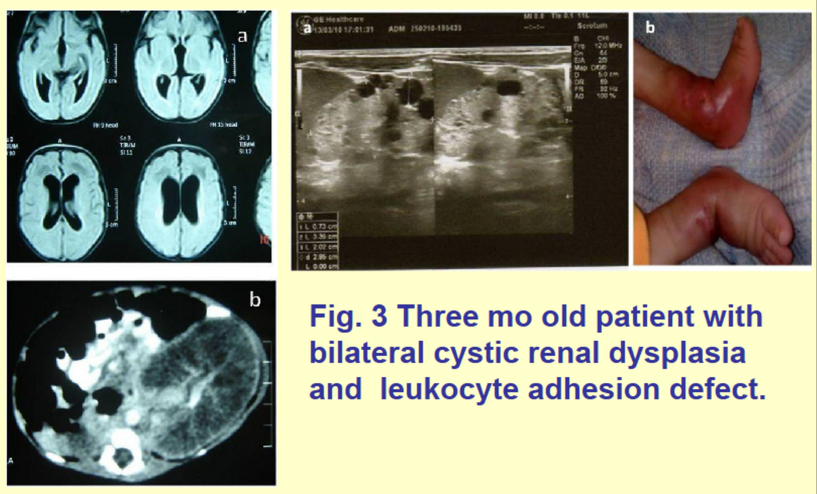

Fig. 2 a Brain MRI of a syndromic unclassified case showing ventriculomegaly.
b Abdominal CT of the same case showing solitary ARPKD.

Fig. 3 Three mo old patient with bilateral cystic renal dysplasia and leukocyte adhesion defect.

CONCLUSION

-While inherited disorders were the most common in this series, it might not reflect their prevalence in the community since the study was based in a major referral center.
-Extra-renal manifestations were common in the context of well defined syndromes, except in 8 patients possibly representing newly described constellations.
-Ultrasonography is a useful screening and initial diagnostic tool with a role for additional imaging, genetic studies and possibly biopsy in selected cases.
-We conclude an algorithm (fig. 4) as a helpful tool for categorization of RCD, modified from Greenbaum et al., 2008 [5].

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