Other full case

Multicystic renal dysplasia: a diagnostic dilemma

Kiran Alam,¹ Manoranjan Varshney,¹ Mehar Aziz,¹ Veena Maheshwari,¹ Mahfooz Basha,¹ Kavita Gaur,¹ Imran Ghani²

¹Department of Pathology, J.N. Medical College, Aligarh, India;

²Department of Surgery, J.N. Medical College, Aligarh, India

Correspondence to Dr Kavita Gaur, kavgaur@gmail.com

Summary

A 3-year-old male child had left-sided abdominal lump for 4 months. Radiological examination revealed a cystic mass in the left kidney. A tentative diagnosis of multicystic nephroma was made on clinical and radiological examination. Cytology was inconclusive. Nephrectomy was done. A final diagnosis of multicystic dysplasia was made on histological examination.

BACKGROUND

The patient presented with symptoms suggestive of multicystic nephroma and the cytological examination was inconclusive. However, finally it was diagnosed as a case of renal dysplasia after histopathological examination.

CASE PRESENTATION

A 3-year-old male child presented to surgery outpatient department with complaints of lump on the left side of abdomen for 4 months. General and systemic examination were unremarkable. Blood pressure was 140/90 mm of Hg. Local examination revealed a ballotable mass in left lumbar region. It was non-tender and overlying skin was normal. CT scan revealed a homogenous cystic mass lesion with enhanced internal septae arising from left kidney. Right kidney was unremarkable. CT-guided fine needle aspiration cytology showed paucicellular smears composed of few spindle cells and occasional ill-formed tubule-like structure. So, no conclusive diagnosis was

made. Left-sided nephrectomy was done, which on gross examination showed a distorted specimen of kidney with attached ureter; kidney measuring $8 \times 7 \times 3.5$ cm and ureter 7×0.5 cm. Outer surface was irregular with areas of congestion and attached fatty tissue (figure 1A). On cut section, markedly dilated pelvis along with multiple cysts was present on the kidney (figure 1B). No corticomedullary differentiation was poor. Microscopic examination showed elongated cysts lined by epithelium which were disorganised. Primitive ducts surrounded by fibromuscular collars (figure 2A,B) and a focus of dysplastic cartilage was seen in the cortex (figure 3). Inflammatory cell infiltrate with features of chronic pyelonephritis (figure 4A) and haemosiderin laden macrophages were noticed (figure 4B). Markedly dilated pelvis with underlying hemorrhage and mononuclear cell infiltrate (figure 5A) along with normal looking areas were also present (figure 5B). Areas of hemorrhage and thick-walled blood vessels were seen. The lumen of ureter was patent and histologically

Figure 1 (A) Distorted specimen of kidney with attached ureter and markedly dilated pelvis. (B) Cut section showing dilated pelvis and multiple cysts.

unremarkable. On the basis of these findings, a final diagnosis of multicystic renal dysplasia was rendered.

In patients of renal dysplasia, an obstructive cause lower down should be looked for as early diagnosis and treatment to prevent the damage to other kidney.

DIFFERENTIAL DIAGNOSIS

- Ureteropelvic junction obstruction
- Multicystic nephroma
- Renal dysplasia secondary to other obstructive disorders of urinary tract.

Figure 2 (A) Section showing glomeruli and tubules lined by undifferentiated cells (H&E $\times$ 500). (B) Section showing tubules lined by undifferentiated cells (H&E $\times$ 500).

Figure 3 Section showing metaplastic cartilage (H&E $\times$ 125).

Figure 4 (A) Section showing features of chronic pyelonephritis (H&E $\times 500$). (B) Section showing haemosiderin laden macrophages (H&E $\times 125$).

Figure 5 (A) Section showing markedly dilated pelvis (H&E $\times 500$). (B) Section showing normal looking kidney (H&E $\times 500$).

TREATMENT

Left-sided nephrectomy was done.

OUTCOME AND FOLLOW-UP

The patient was discharged in good conditions.

DISCUSSION

The term renal dysplasia refers to abnormally differentiated kidneys presented as abnormal structural organisation with ill-developed metanephric elements. It may have different presentations that include hypoplasia, aplasia and multicystic dysplasia.

Multicystic dysplastic kidneys are usually non-functional and they contain multiple, variable sized and non-communicating cysts separated by dysplastic renal parenchyma and abnormal pelvicaliceal system. It usually manifests as a sporadic disease; however, few familial cases have also been reported.¹ Mutation in SIX 1 and PAX 2 genes are reported to be associated with renal dysplasia.^{2–4} Various teratogens and in utero exposure to various virus-like cytomegalovirus, enterovirus and adenovirus are assumed to cause renal dysplasia. It is more common in males with male to female ratio of 1.4:1.⁵ Diagnosis can be prenatal.⁶ The patients usually present with flank mass as in our case and may have hypertension.⁷ Contralateral vesicoureteric reflux is seen in 4–19% of cases.^{8–9} Several other renal anomalies like ureteropelvic junction obstruction, horse shoe kidney, segmental ureteral disparity, glomerulonephritis and pyelonephritis in contralateral kidney may be associated with multicystic renal dysplasia. It may be associated with other malformation like congenital heart diseases and intestinal atresia. This condition is usually unilateral; however, cases of bilateral renal dysplasia are also reported in literature, which usually results in stillbirth. Bilateral disease may be associated with pulmonary hypoplasia, amnion nodosum and oligohydramnios. Number of cysts varies, however, more than five cysts are seen in two-thirds of cases.¹⁰ Histologically, it is characterised by lobar disorganisation, primitive ducts and metaplastic cartilage.^{11–12} Metaplastic cartilage should be closely looked for in multiple sections, as it may be present in small foci like in our case and may be missed. Tubules and collecting duct are lined by undifferentiated cells which mimic like blastemal cells of Wilms' tumour.^{13–16} Features of chronic pyelonephritis may be present if the ureter is patent, and an ascending infection may result in chronic pyelonephritis as in our case. Differential diagnosis include ureteropelvic junction obstruction, multicystic nephroma and renal dysplasia secondary to other obstructive disorders of urinary tract. Nephrectomy is the treatment of choice especially when associated with hypertension which disappears after surgery.^{17–18} Laproscopic nephrectomy is also a treatment option but still controversial.¹⁹ Nephrectomy is the mainstay of treatment when renal malignancies are associated with this. Chances of development of renal malignancies in later life is still not clear as few authors reported no increased incidence of renal malignancies. Antibiotic prophylaxis should be given in patients with contralateral vesicoureteric reflux to prevent the chances of pyelonephritis. A lifelong follow-up is needed even after nephrectomy because of chances of hypertension, proteinuria and renal insufficiency in later life. In patients with one functional kidney, dietary modifications like low-fat and low-salt diet may be advised.

Learning points

- Renal dysplasia closely mimics Wilms' tumour clinically.
- Patient may have hypertension which disappears after nephrectomy.
- Metaplastic cartilage should be closely looked for in multiple sections.
- Early diagnosis may prevent damage to the other kidney.
- Lifelong follow-up is needed.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. **Belk RA**, Thomas DF, Mueller RF, *et al.* A family study and the natural history of prenatally detected unilateral multicystic dysplastic kidney. *J Urol* 2002;**167**(2 Pt 1):666–9.
2. **Buller C**, Xu X, Marquis V, *et al.* Molecular effects of Eya1 domain mutations causing organ defects in BOR syndrome. *Hum Mol Genet* 2001;**10**:2775–81.
3. **Ruf RG**, Xu PX, Silvius D, *et al.* SIX1 mutations cause branchio-oto-renal syndrome by disruption of EYA1-SIX1-DNA complexes. *Proc Natl Acad Sci USA* 2004;**101**:8090–5.
4. **Fletcher J**, Hu M, Berman Y, *et al.* Multicystic dysplastic kidney and variable phenotype in a family with a novel deletion mutation of PAX2. *J Am Soc Nephrol* 2005;**16**:2754–61.
5. **Robson WL**, Leung AK, Thomason MA. Multicystic dysplasia of the kidney. *Clin Pediatr (Phila)* 1995;**34**:32–40.
6. **Cunniff C**. Prenatal screening and diagnosis for pediatricians. *Pediatrics* 2004;**114**:889–94.
7. **Narchi H**. Risk of hypertension with multicystic kidney disease: a systematic review. *Arch Dis Child* 2005;**90**:921–4.
8. **Aslam M**, Watson AR. Unilateral multicystic dysplastic kidney: long term outcomes. *Arch Dis Child* 2006;**91**:820–3.
9. **Kuvertz-Broeking E**, Brinkmann OA, Von Lengerke HJ, *et al.* Unilateral multicystic dysplastic kidney: experience in children. *BJU Int* 2004;**93**:388–92.
10. **Strife JL**, Souza AS, Kirks DR, *et al.* Multicystic dysplastic kidney in children: US follow-up. *Radiology* 1993;**186**:785–8.
11. **Bernstein J**. Developmental abnormalities of the renal parenchyma-hypoplasia and dysplasia. *Pathol Annu* 1968;**3**:213–47.
12. **Bernstein J**. The morphogenesis of renal parenchymal maldevelopment (renal dysplasia). *Pediatr Clin North Am* 1971;**18**:395–407.
13. **Vellios F**, Garrett RA. Congenital unilateral multicystic disease of the kidney. A clinical and anatomic study of seven cases. *Am J Clin Pathol* 1961;**35**:244–54.
14. **Gaddy CD**, Gibbons MD, Gonzales ET Jr. Obstructive uropathy, renal dysplasia and nodular renal blastema: is there a relationship to Wilms tumor? *J Urol* 1985;**134**:330–3.
15. **Dimmick JE**, Johnson HW, Coleman GU, *et al.* Wilms tumorlet, nodular renal blastema and multicystic renal dysplasia. *J Urol* 1989;**142**(2 Pt 2):484–5.
16. **Noe HN**, Marshall JH, Edwards OP. Nodular renal blastema in the multicystic kidney. *J Urol* 1989;**142**(2 Pt 2):486–8.
17. **Susskind MR**, Kim KS, King LR. Hypertension and multicystic kidney. *Urology* 1989;**34**:362–6.
18. **Kullendorff CM**. Surgery in unilateral multicystic kidney. *Z Kinderchir* 1990;**45**:235–7.
19. **Ionouchene S**, Mikhaylov N, Novozhilov V, *et al.* Laparoscopic nephrectomy: advantages of technique in infants and newborns. *J Laparoendosc Adv Surg Tech A* 2009;**19**:703–6.

This pdf has been created automatically from the final edited text and images.

Copyright 2011 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.

BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Alam K, Varshney M, Aziz M, Maheshwari V, Basha M, Gaur K, Ghani I. Multicystic renal dysplasia: a diagnostic dilemma. *BMJ Case Reports* 2011; 10.1136/bcr.03.2011.3989, date of publication

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow