Cysts in the kidney: What do they mean?

Dr Mohan Shenoy
Consultant Paediatric Nephrologist
Royal Manchester Children’s Hospital
Outline

• Introduction

• 6 cases

• Clinical assessment

• Summary
Cystic kidney disease

- Inherited cystic kidney disease
  - ADPKD, ARPKD, TS, Renal Cysts and Diabetes Syndrome

- Cystic dysplasia including multicystic dysplastic kidney

- Ciliopathies
  - Nephronophthisis, Bardet-Biedl Syndrome

- Simple cyst
Cyst position

Renal dysplasia

ARPKD

ADPKD

Complicated cyst

Medullary sponge kidney

Exophytic cyst

Calyceal diverticulum

Simple renal cyst
Case History 1

- 13 year, girl
- Rheumatology – joint pains
- US abdomen – part of investigations
ADPKD

- Most common hereditary kidney disorder
  - 1 in 500 births
- Progressive enlargement of cysts leading to CKD in 50% by 40-50yrs
- Accounts for 5% of ESRD
- Mutations in PKD1 (85%) and PKD2 (15%)
- 90% will have an affected parent
ADPKD – US Diagnostic criteria

- 3 or more cysts – unilateral or bilateral age 15-39y
- 2 or more cysts in each kidney age 40-59y
- Fewer than 2 cysts at ≥40y excludes the disease

ADPKD

- Mild disease in childhood
- Screening family members
- Annual BP and urine for protein
- Routine cerebral aneurysm screen not required unless strong family history
- Ongoing trials
Case history 2

- Term male infant
- Respiratory distress soon after birth
- Abdominal distension and vomiting ?NEC
Diagnosis?
ARPKD

- Mutations of PKHD1
- 1 in 20,000 live births
- Antenatally large bright kidneys, oligohydramnios
- Severe respiratory distress at birth ~ 30% mortality
- Severe hypertension, large palpable kidneys, CKD
- Hepatic fibrosis, portal hypertension
ARPKD
Case history 3

- 9yr Asian boy
- Presents with frank haematuria
- BP 121/72
- No oedema
- Urine M,C&S - >100 rbc, no growth
- FBC, creatinine, coagulation - normal
- No proteinuria
US kidneys

Polycystic kidney disease

Parental US kidneys normal
US 12 months later…
Cortical tubers and periventricular calcification
## Tuberous Sclerosis

<table>
<thead>
<tr>
<th>Major features</th>
<th>Minor features</th>
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<tbody>
<tr>
<td>Facial angiofibroma ($\geq 3$) or fibrous cephalic plaque</td>
<td>Confetti’ skin lesions</td>
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<tr>
<td>Non-traumatic ungual or periungual fibroma ($\geq 2$)</td>
<td>Multiple, randomly distributed pits in dental enamel ($\geq 3$)</td>
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<tr>
<td>Hypomelanotic macules ($\geq 3$, at least 5mm diameter)</td>
<td>Gingival fibromas ($\geq 2$)</td>
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<tr>
<td>Shagreen patch</td>
<td>Retinal achromic patch</td>
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<tr>
<td>Multiple retinal hamartomas</td>
<td>Multiple renal cysts</td>
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<tr>
<td>Subependymal nodule</td>
<td>Non-renal hamartoma</td>
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<tr>
<td>SEGA</td>
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<tr>
<td>Cortical tuber</td>
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<tr>
<td>Cortical dysplasia</td>
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<tr>
<td>Cardiac rhabdomyoma</td>
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<tr>
<td>Lymphangiomyomatosis</td>
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<tr>
<td>Renal angiomyolipoma ($\geq 2$)</td>
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Case history 4

- Antenatal scan – R kidney multiple cysts, L kidney normal

- Postnatal US confirms findings
DMSA
The multicystic dysplastic kidney

• Developmental abnormality where renal parenchyma is completely replaced by multiple non-communicating cysts
• Kidney non-functioning
• Proximal ureter is atretic or non-patent

• 1 in 2400 to 4300 Males 2:1 Females
MCDK: Investigation

- Postnatal USS
- DMSA to confirm non-function and normality of contralateral kidney
- ?MCUG
  - has been performed historically because VUR detected in around 25%
  - Aslam and Watson Arch Dis Child 2006;91:820
    - 27/143 (19%) had contralateral VUR
    - I-II 74%, III 22%, V 1 child
    - UTI incidence similar VUR vs no VUR
    - No new scar formation (USS only)
Conservative vs Surgery

- Conservative management advocated on basis that most kidneys involute with time and the risk of complications is low
Complications of the MCDK

• Mass effect
  – Rare problem in antenatal or newborn period
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- Mass effect
  - Rare problem in antenatal or newborn period
- Hypertension
  - Systematic review of 29 studies revealed 29 cases in 1115 patients

Narchi H. Arch Dis Child 2005;90:921
Complications of the MCDK

• Mass effect
  – Rare problem in antenatal or newborn period

• Hypertension
  – Recent systematic review of 29 studies revealed 29 cases in 1115 patients
    Narchi H. Arch Dis Child 2005;90:921

• Malignancy
  – 9 cases reported (none in systematic review); very difficult to know whether MCDK a true risk factor
MCDK; long term outlook

- Medium term prognosis excellent provided contralateral kidney is normal
- Nottingham group advocate USS at 0, 2, 5 and 10 years

- Lifelong annual follow up
  - BP and proteinuria

Mansoor O et al Ped Neph 2011
Case history 5

• Male child

• Antenatal scan bilateral bright cystic kidneys

• No family history of kidney disease

• Postnatal scan
Case history 5
Case history 5 – Week 1

- Creatinine 180umol/l
- BP 110/75mmHg
- Normal examination
- Voiding good volumes
DD of antenatal bright kidneys

- ADPKD
- ARPKD
- Primary hyperoxaluria
- Cystic dysplasia
- Syndromes
  - Bardet-Biedl, Meckel Gruber
Case history 5

- Age 4y
- Well
- Creatinine 130s
- eGFR 30ml/min/1.73m²

- Diagnosis: Cystic dysplasia

- HNF 1beta mutation pos
Spectrum of \textit{HNF1B} Mutations in a Large Cohort of Patients Who Harbor Renal Diseases

Laurence Heidet,\textsuperscript{*} Stéphane Decramer,\textsuperscript{†} Audrey Pawtowski,\textsuperscript{†} Vincent Morinière,\textsuperscript{*\dagger} Flavio Bandin,\textsuperscript{†} Bertrand Knebelmann,\textsuperscript{β} Anne-Sophie Lebre,\textsuperscript{†} Stanislas Faguere,\textsuperscript{β} Vincent Guigonis,\textsuperscript{γ} Corinne Antignac,\textsuperscript{α\dagger††} and Rémi Salomon\textsuperscript{*\dagger††}

\begin{figure}
\centering
\includegraphics[width=0.4\textwidth]{figure2}
\caption{Type of \textit{HNF1B} mutation (\square, deletion of the entire gene; \blacktriangleleft, missense mutations; \blacktriangle, truncating mutations) according to the renal phenotype in patients and affected relatives. 1, prenatal hyperchogenic kidneys; 2, hyperchogenic kidney diagnosed after birth; 3, MCD; 4, unilateral renal agenesis; 5, cystic disease; 6, renal hypoplasia; 7, tubulointerstitial nephritis; 8, pyeloureteral junction; 9, pelvic kidney; 10, lack of renal anomaly.}
\end{figure}

\begin{figure}
\centering
\includegraphics[width=0.4\textwidth]{figure3}
\caption{Type of \textit{HNF1B} mutation (deletion, missense mutations, truncating mutations) according to the GFR at last follow-up (\blacktriangle, GFR >80 ml/min per 1.73 m\textsuperscript{2}; \blacktriangleleft, GFR <80 ml/min per 1.73 m\textsuperscript{2}) and age at last follow-up (\blacktriangleleft).}
\end{figure}

Conclusions: This large series showed that the severity of the renal disease associated with \textit{HNF1B} mutations was extremely variable (from prenatal renal failure to normal renal function in adulthood) and was not correlated with the genotype.

Renal dysplasia and PUV
Simple cyst

- Very rare
- Diagnosis of exclusion
  - Often seen in a duplex kidney with non/poorly functioning half
  - May be early ADPKD
- Needs follow up
Take home messages

• Simple cysts are extremely rare in children

• Family history and parental US

• Absence of cysts in childhood does not exclude ADPKD

• Clinical examination
  – TS, Bardet Biedl syndrome

• Renal Genetic clinic