Objectives

• Be able to diagnose renal cystic disease
• Genetic / non-genetic
• Be able to describe patterns of various renal cystic disease on routine imaging studies
• Be able to counsel patients with renal cystic disease regarding
  • Prognosis
  • Required follow-up investigations
• Associated findings
Genetic Renal Cystic Disease

- ARPKD (Autosomal Recessive PKD)
- ADPKD (Autosomal Dominant PKD)
- Juvenile Nephronophthisis – Medullary CD
- Juvenile Nephronophthisis (autosomal recessive)
- Medullary Cystic Disease (autosomal dominant)
- Congenital Nephrosis (autosomal recessive)
- Familial Hypoplastic Glomerulocystic Disease (autosomal dominant)
- Others – e.g. Cystic Fibrosis, VHL
Non-Genetic Renal Cystic Disease

- Multicystic Dysplastic Kidney
- Benign Multilocular Cyst (Cystic Nephroma)
- Simple Cysts – Bosniak Classification
- Medullary Sponge Kidney
- Sporadic Glomerulocystic Kidney Disease
- Acquired Renal Cystic Disease
- Calyceal Diverticulum
- Cystic Renal Cell Carcinoma
Autosomal Recessive Polycystic Kidney Disease

- Appears early (antenatal diagnosis)
- Echogenic but homogenous kidneys on U/S due to small cysts throughout parenchyma
- Usually fatal within months
- Associated with hepatic fibrosis - biliary ectasia, periportal fibrosis
- Mutation of PKHD1 gene on Cr 6
- Large kidneys, sponge
Fetus with ARPKD
ARPKD
Autosomal Dominant Polycystic Kidney Disease

• Common cause of ESRD (7-15%)
• May present in newborn but most common presentation 30-50 years
• Two (?3) genes identified – PKD1, PKD2
• PKD1(Cr 16) – more hypertension, infections – younger age at presentation, onset of renal failure
• PKD2 (Cr 4) – older at presentation
ADPKD - Presentation

- Age 30-50
- Hypertension
- Renin mediated
- Microscopic/ Gross hematuria
- Flank pain
- Stones in 20-30 %
- GI symptoms
- Incidental finding of liver/kidney cysts on U/S
- Berry aneurysm (10 –40%)
ADPKD - Etiology

• Loss of polarity of epithelial cells anywhere in nephron
• Cell proliferation – outpouching, collection of tubular fluid
• Histology
• Cysts variable in size – from mm to several cm.
• Association with RCC
• No increased risk
ADPKD – Evaluation

• Diagnosis (in absence of positive family history)
  1. Presence of bilateral cysts with at least 2 of:
  2. Bilateral renal enlargement
  3. 3 or more hepatic cysts
  4. Cerebral artery aneurysm
  5. Cysts of arachnoid, pineal, pancreas, spleen
U/S – Right Kidney
CT – ADPKD
CT – ADPKD
ADPKD
ADPKD - Treatment

• Role of genetic counselling
• Role of hypertension management
• Risk of infection
• Avoid nephrotoxins
• Management of pain – medical vs surgical
Role for unroofing cysts
• Management of mecanephrosis
Juvenile Nephronophthisis/ Medullary Cystic Disease

• **Juvenile Nephronophthisis**
  - Usually autosomal recessive
  - 3 types – juvenile (NPH1), adolescent (NPH2), infertile (NPH3) genes
  - Presentation mean age 13 – renal failure

• **Medullary Cystic Disease**
  - Usually autosomal dominant
  - Older age at presentation (20-40)
Juvenile Nephronophthisis/ Medullary Cystic Disease

Presentation

• Polydipsia / polyuria in more than 80% (not to the degree of patients with DI) resistant to vasopressin

• Polyuria due to inability to conserve sodium – so salt restriction not indicated in these patients

• Salt losing nephropathy

• Associated with retinal disorders (retinitis pigmentosa), skeletal abnormalities, hepatic fibrosis

• Various syndromes associated with JN (Bardet-Beidl, Senior-Loken, Alstroms)
JN/MCD Complex

Histology

- Interstitial nephritis leading to atrophy
- Cysts – 85% with MCD vs 40% with JN
- Cysts at cortico-medullary junction
- Cysts small 0.5 cm;

Treatment

- Salt replacement
- Supportive therapy
Congenital Nephrosis

- Finnish Type – recessive - Cr 19
- Diffuse Mesangial Sclerosis – 1/3 familial

Clinical Features
- Large placenta / large kidneys
- Early onset severe proteinuria leading to edema, renal failure - death
Multiple Malformation Syndromes with Renal Cysts

Autosomal dominant-
• von Hippel Lindau – VHL gene on Cr 3
• Tuberous Sclerosis – TSC1 on Cr 9 or TSC2 on Cr 16

Autosomal recessive
• Meckel’s Syndrome
• Jeune’s Asphyxiating Thoracic Dystrophy
• Zellweger’s Cerebrohepatorenal Syndrome
• Ivemark’s Syndrome (renal-hepatic-pancreatic dysplasia)
Multiple Malformation Syndromes with Renal Cysts

X-linked Dominant
- Orofaciodigital Syndrome I

Chromosomal Disorders
- Trisomy 13 (Patau)
- Trisomy 18 (Edward)
- Trisomy 21 (Down)
Tuberous Sclerosis

Presentation
• Epilepsy 80%
• Mental retardation 60%
• Adenoma sebaceum 75%

Cerebral hamartoma hallmark

Renal involvement
• Cysts (lined with hypertrophic hyperplastic eosinophilic cells)
• Angiomyolipoma (40-80%)
• Renal cell carcinoma (2%)
von Hippel Lindau Disease

Cerebral and retinal hemangioblastoma – major cause of morbidity and mortality

Cysts
• Pancreas
• Kidney – 76%
• Epididymis

Epididymal cystadenoma
Pheochromocytoma – 10-17%
Renal cell carcinoma - in 50%
von Hippel Lindau Disease

Management

• Surveillance with U/S – CT q 1-2 years
• Conservative approach to renal lesions – segmental resection
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Multicystic Dysplasia - Kidney (MCDK)

- Nongenetic dysplasia presenting at birth or shortly after
- Active process – so may involute, remain the same, or get larger
- Monitor for 5 years
- Intervention
  - Increase in solid tissue
  - Compromise of vital function
- Risk of contralateral reflux - 15%
MCDK
MCDK – micro
Medullary Sponge Kidney

- Noninheritable condition – usually incidental finding
- Due to dilated collecting ducts – “blush” in papillae on IV contrast studies
- Increased risk of
  1. Nephrolithiasis (50-60%)
  2. Hypercalciuria (at least 33%)
  3. Urinary tract Infection (20-33%)
  4. Hematuria (0-18%)
MSL – Microscopic
Cast of MSK Kidney
Acquired Renal Cystic Disease

• Associated with ESRD – patients on Hemodialysis

• Hyperplastic renal cysts – increased risk of progression to RCC within 10 years of starting dialysis

• Cysts may regress with transplantation
Acquired Cystic Disease
Patient on Hemodialysis
Acquired Renal Cystic Disease
<table>
<thead>
<tr>
<th>Category</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Category I</td>
<td>Simple benign cyst with (1) good through-transmission (i.e., acoustic enhancement), (2) no echoes within the cyst, (3) sharply, marginated smooth wall; requires no surgery.</td>
</tr>
<tr>
<td>Category II</td>
<td>Looks benign with some radiologic concerns including septation, minimal calcification, and high density; requires no surgery.</td>
</tr>
<tr>
<td>Category II F</td>
<td>Although calcification in wall of cyst may even be thicker and more nodular than in category II, the septa have minimal enhancement, especially those with calcium; requires no surgery.</td>
</tr>
<tr>
<td>Category III</td>
<td>More complicated lesion that cannot confidently be distinguished from malignancy, having more calcification, more prominent septation of a thicker wall than a category II lesion; more likely to be benign than malignant; requires surgical exploration and/or removal.</td>
</tr>
<tr>
<td>Category IV</td>
<td>Clearly a malignant lesion with large cystic components, irregular margins; solid vascular elements; requires surgical removal.</td>
</tr>
</tbody>
</table>
Bosniak III – non contrast