Renal Cystic Disease

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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

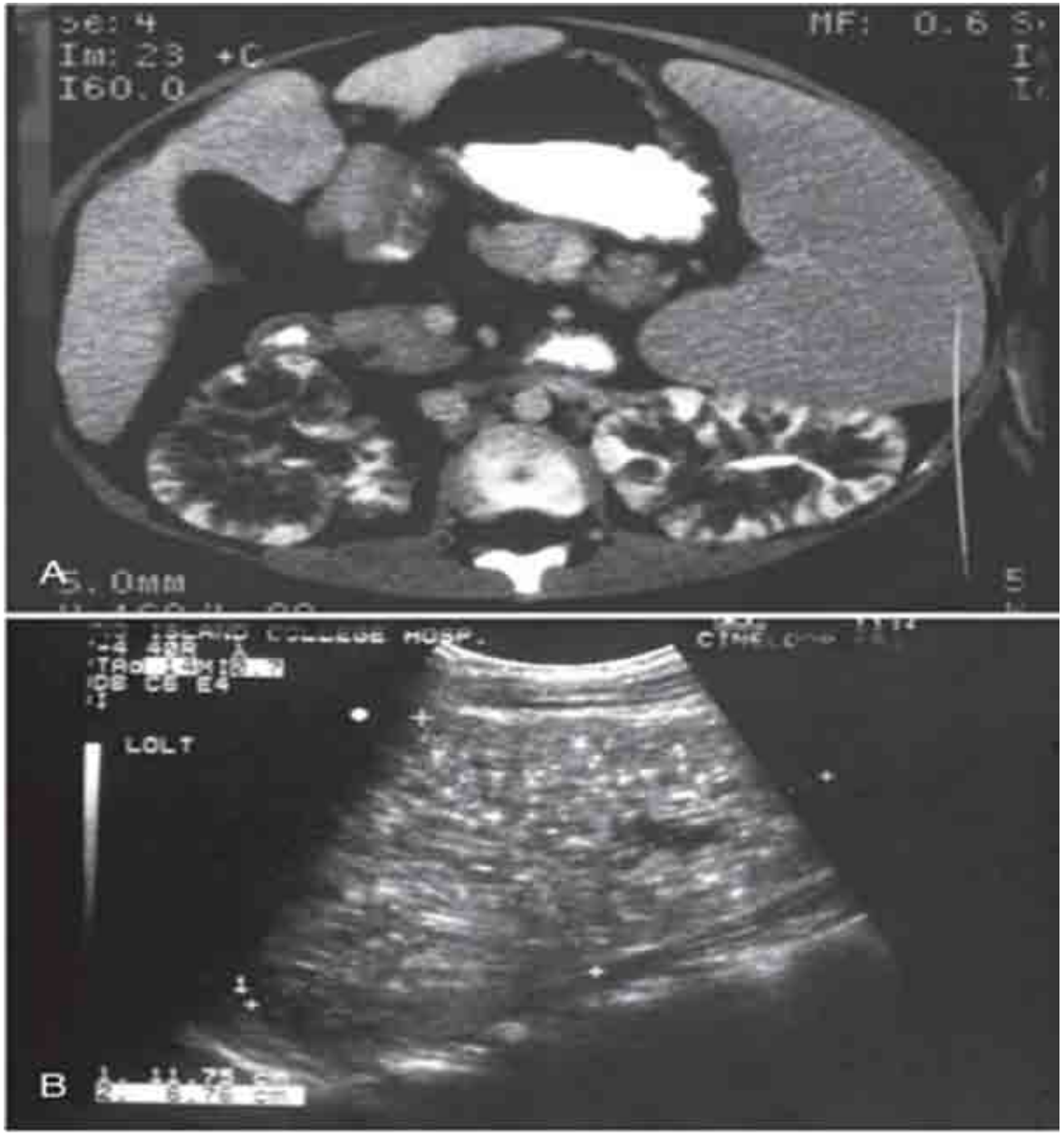
Non-Genetic Renal Cystic Disease

- 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

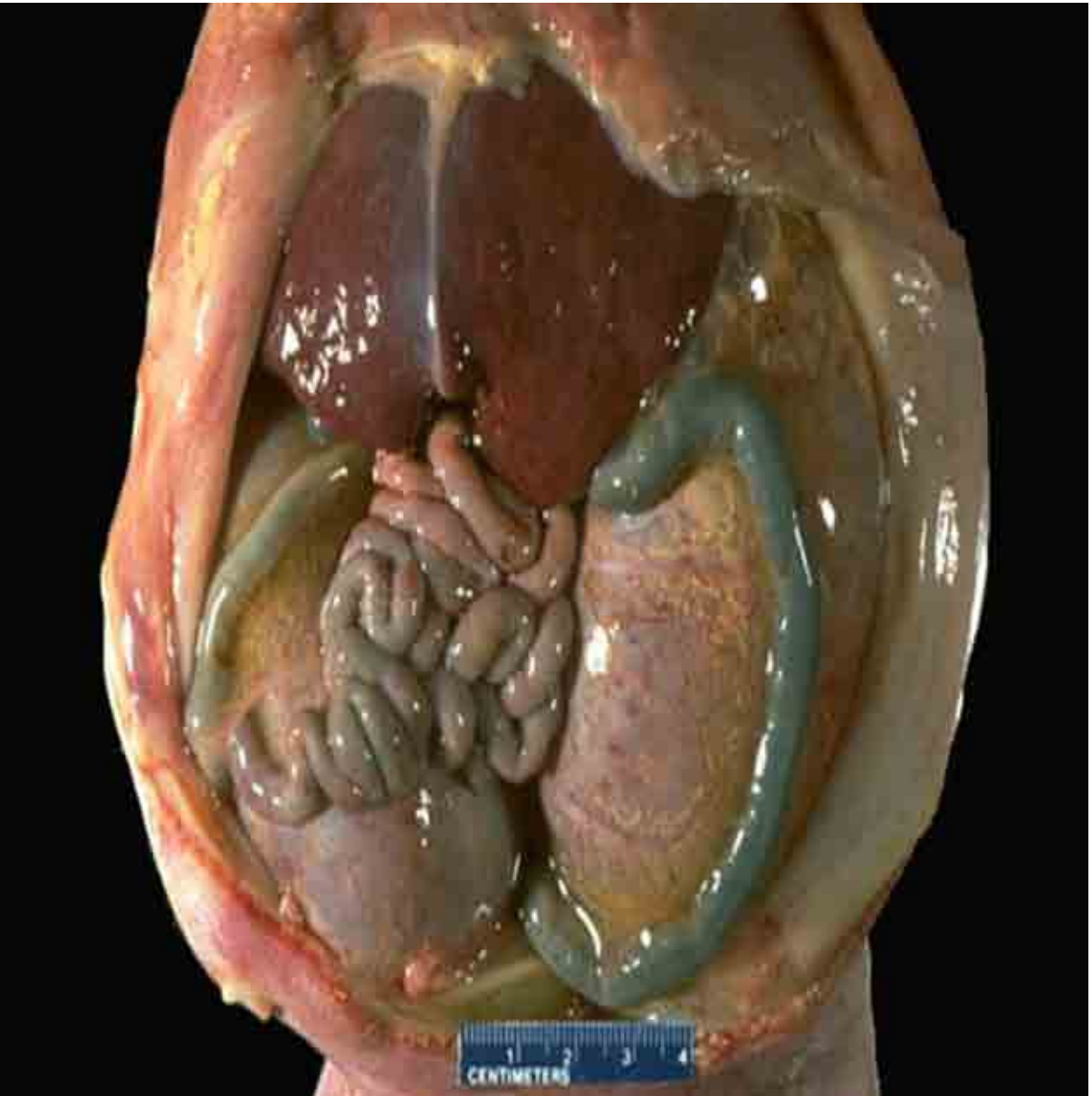
Autosomal Recessive Polycystic Kidney Disease

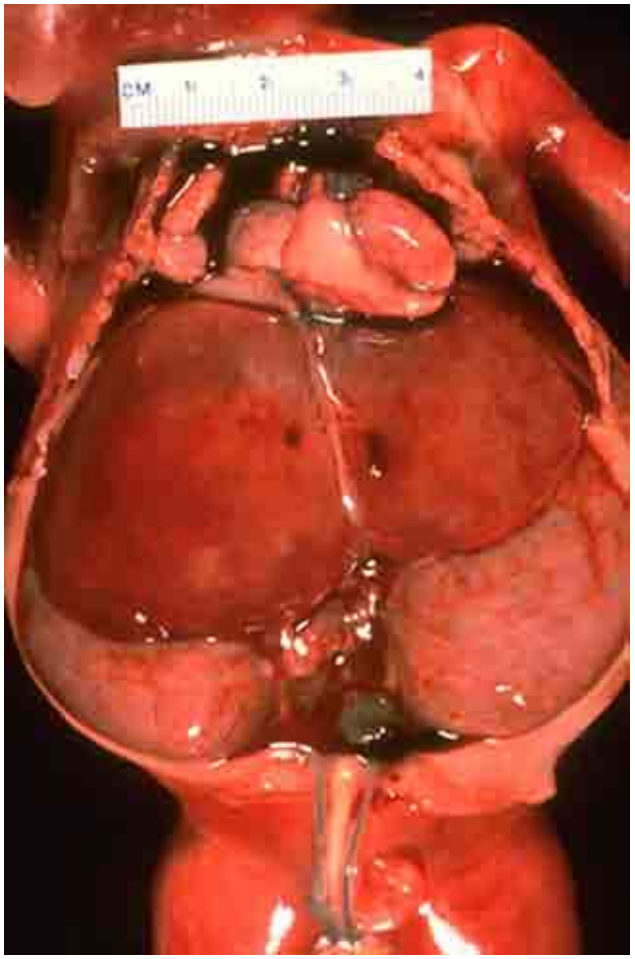
- Appears early (antenatal diagnosis)
- Echogenic but homogenous kidneys on U/S due to small cysts throughout parenchyma
- Usually fatal within months
- Associated with hepatic fibrosis - biliary ectasia, periportal fibrosis
- Mutation of PKHD1 gene on Cr 6
- Large kidneys, sponge

CT - ARPKD

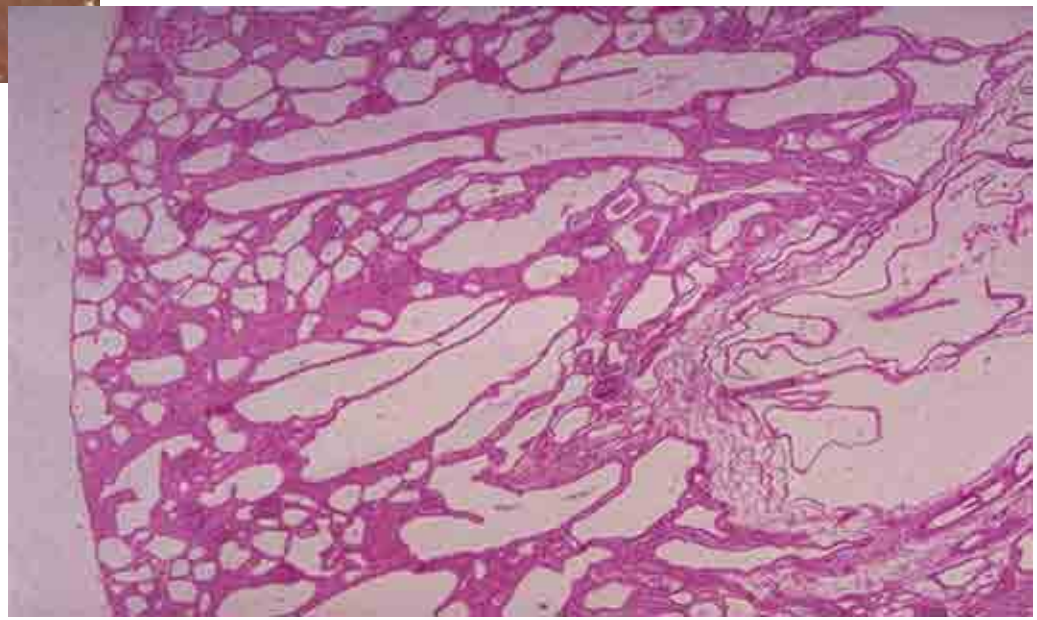


Fetus with ARPKD





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

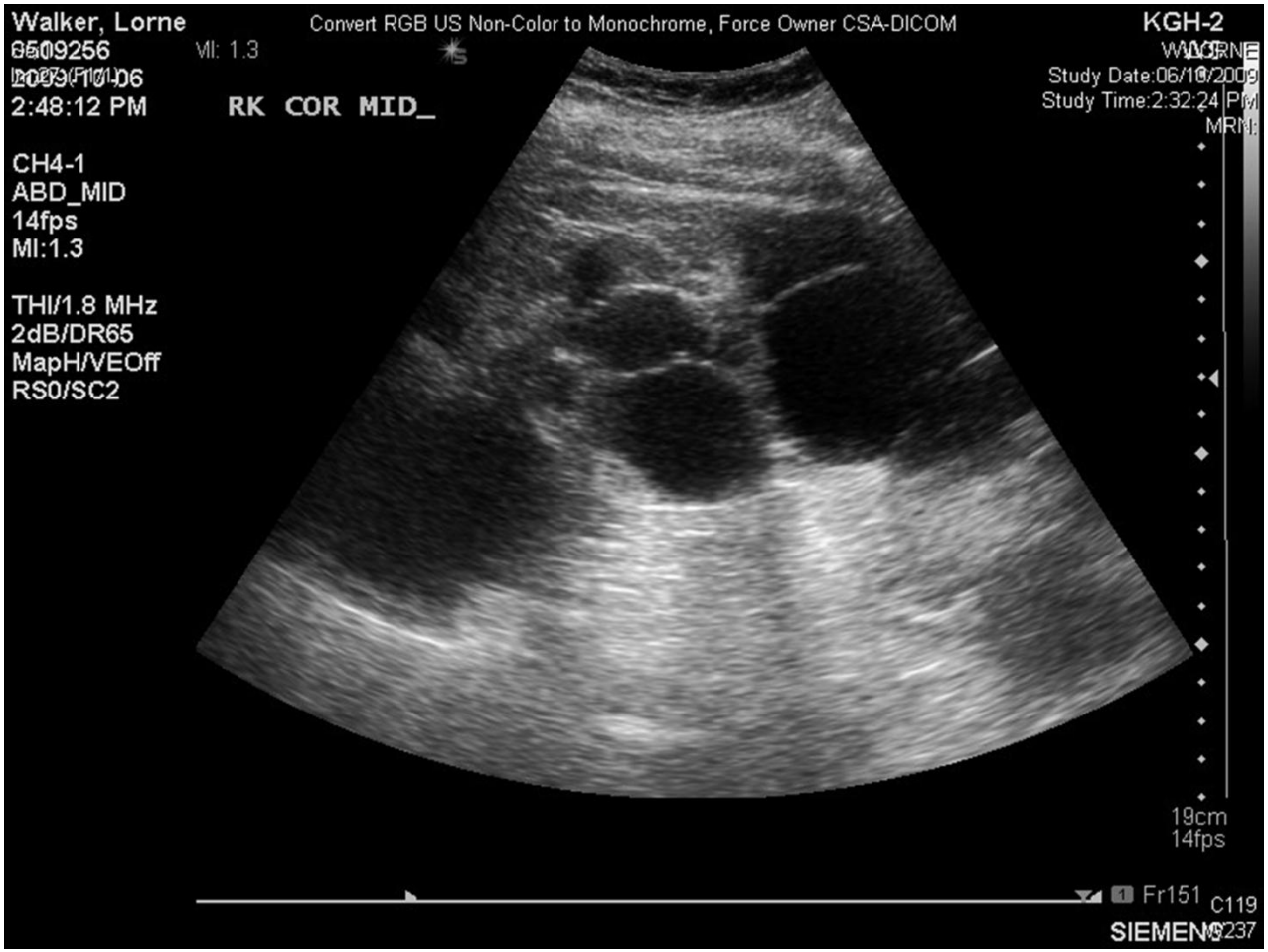
- Loss of polarity of epithelial cells anywhere in nephron
- Cell proliferation – outpouching, collection of tubular fluid
- Histology
- Cysts variable in size – from mm to several cm.
- Association with RCC
- No increased risk

ADPKD – Evaluation

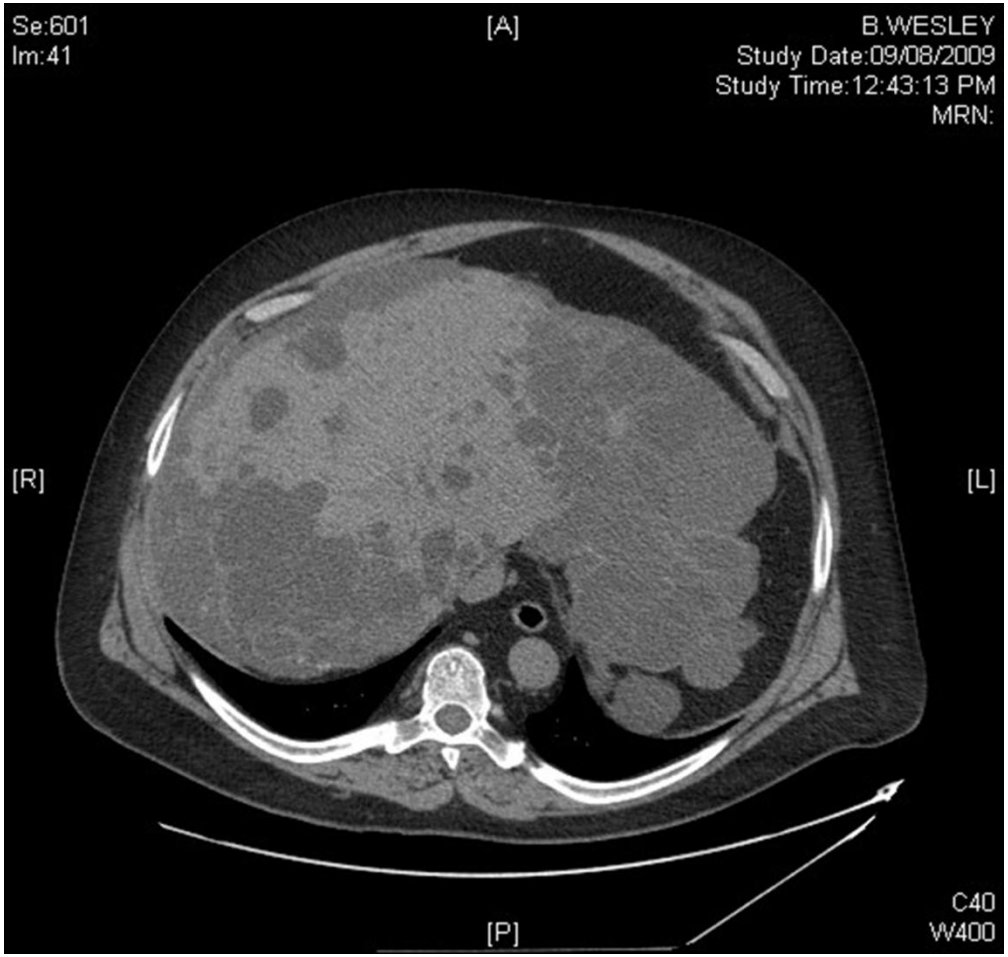
- Diagnosis (in absence of positive family history)

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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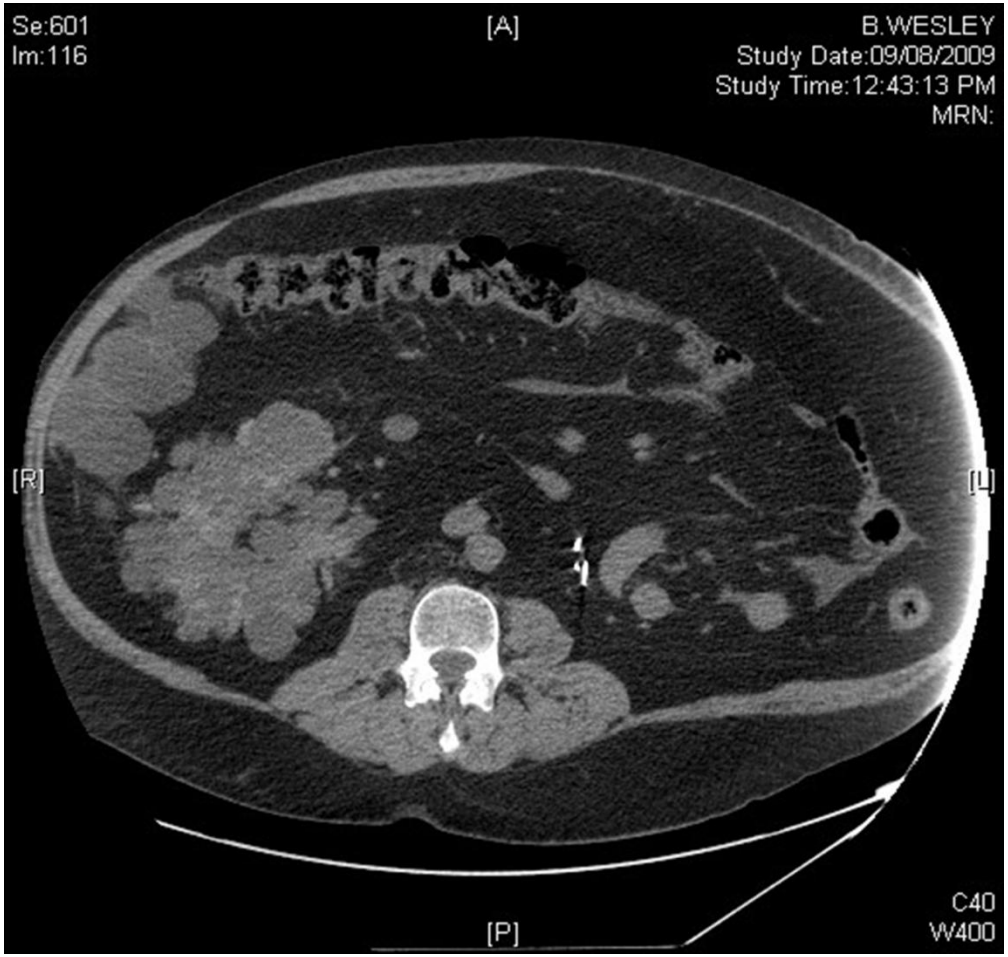
CT – ADPKD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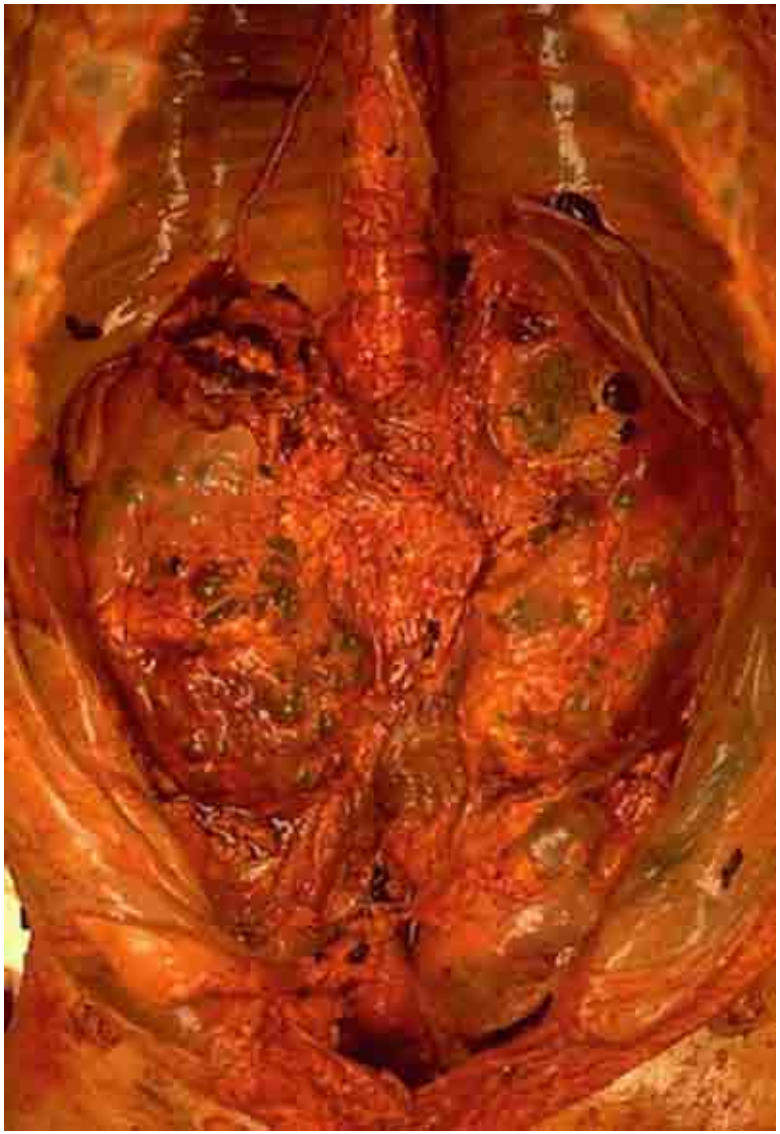
ADPKD



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 meganephrosis

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%

Cerebal 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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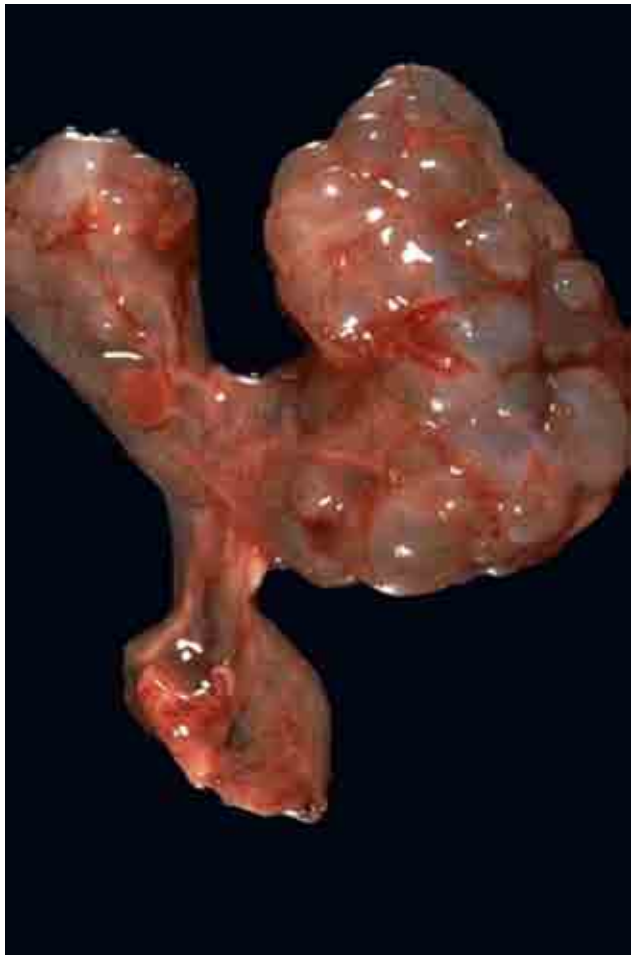
Multicystic Dysplasia - Kidney (MCDK)

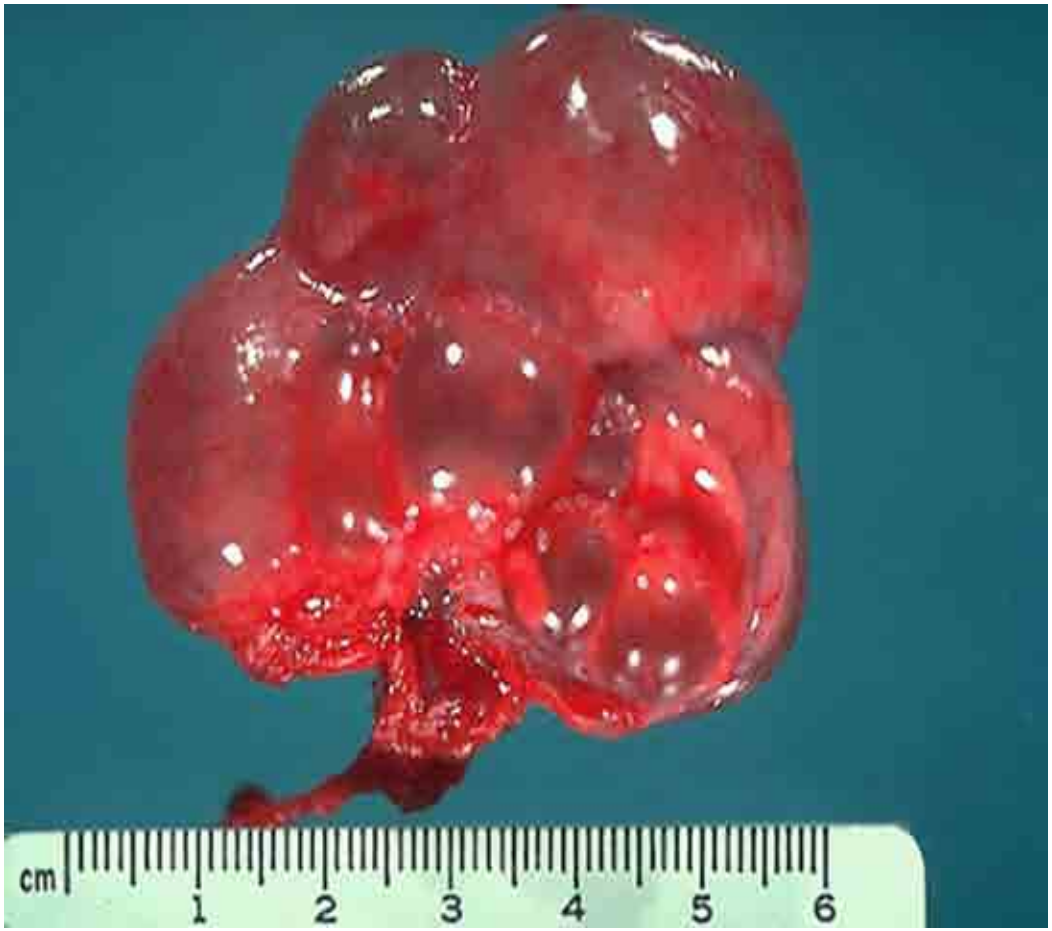
- Nongenetic dysplasia presenting at birth or shortly after
- Active process – so may involute, remain the same, or get larger
- Monitor for 5 years
- Intervention

- Increase in solid tissue
- Compromise of vital function

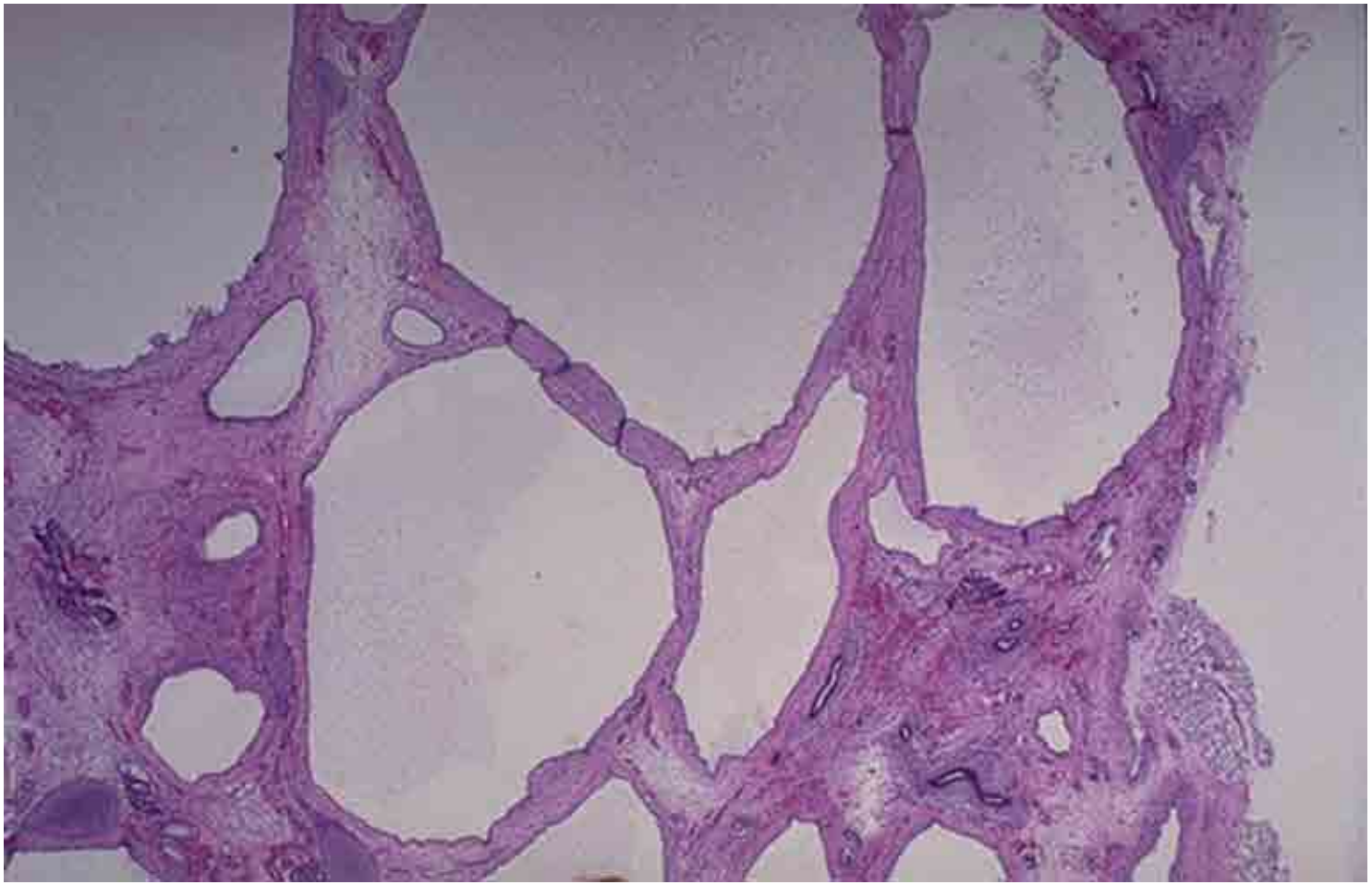
- Risk of contralateral reflux - 15%

MCDK





MCDK – micro

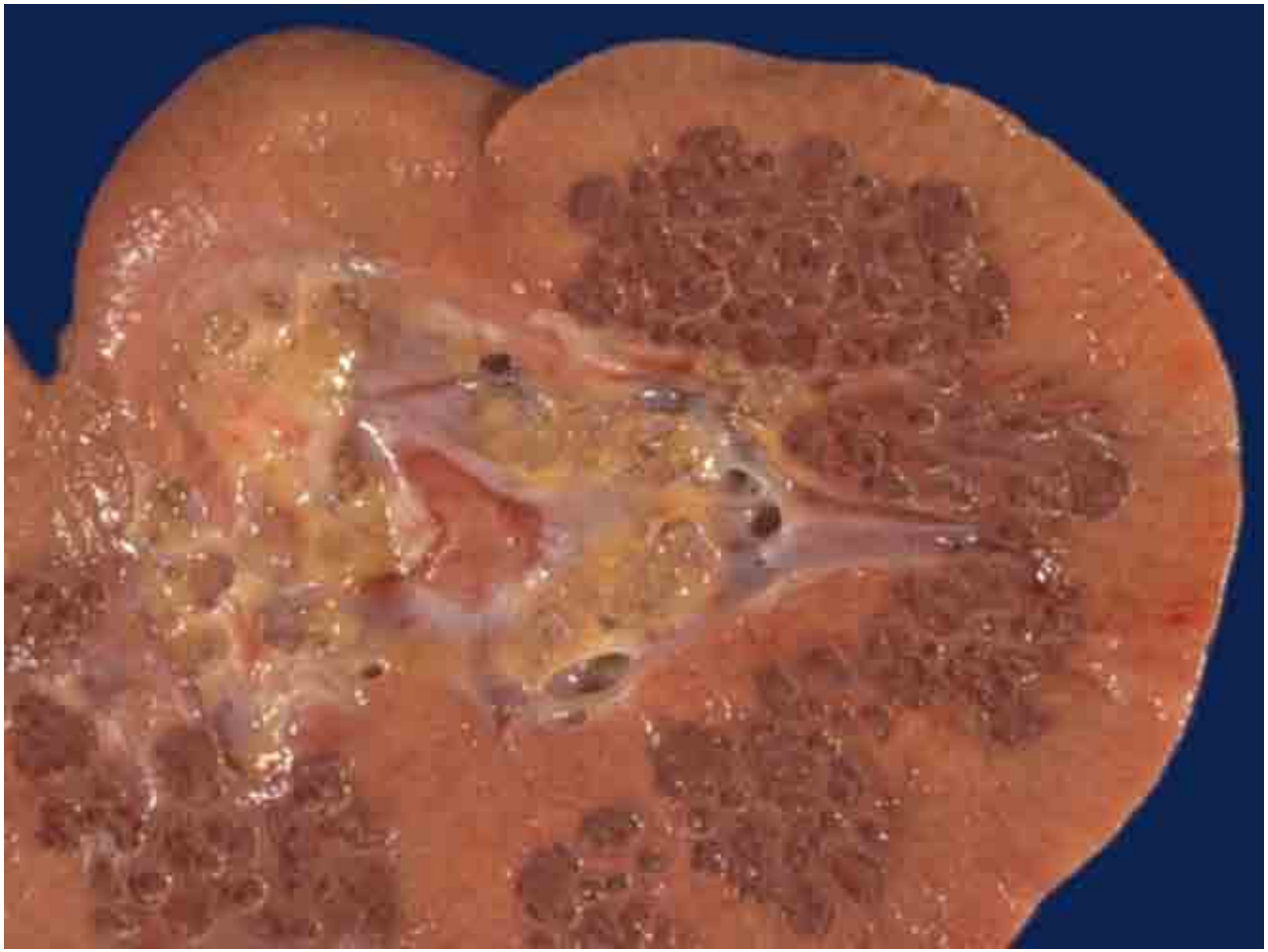


Medullary Sponge Kidney

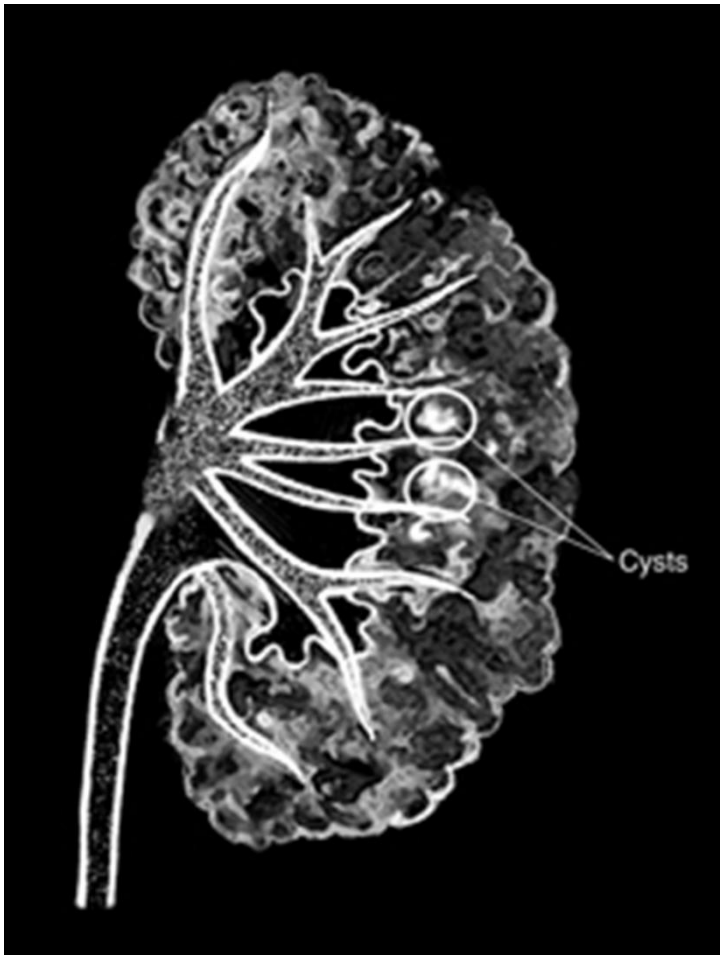
- Noninheritable condition – usually incidental finding
- Due to dilated collecting ducts – “blush” in papillae on IV contrast studies
- Increased risk of

1. Nephrolithiasis (50-60%)
2. Hypercalciuria (at least 33%)
3. Urinary tract Infection (20-33%)
4. Hematuria (0-18%)

MSL – Microscopic



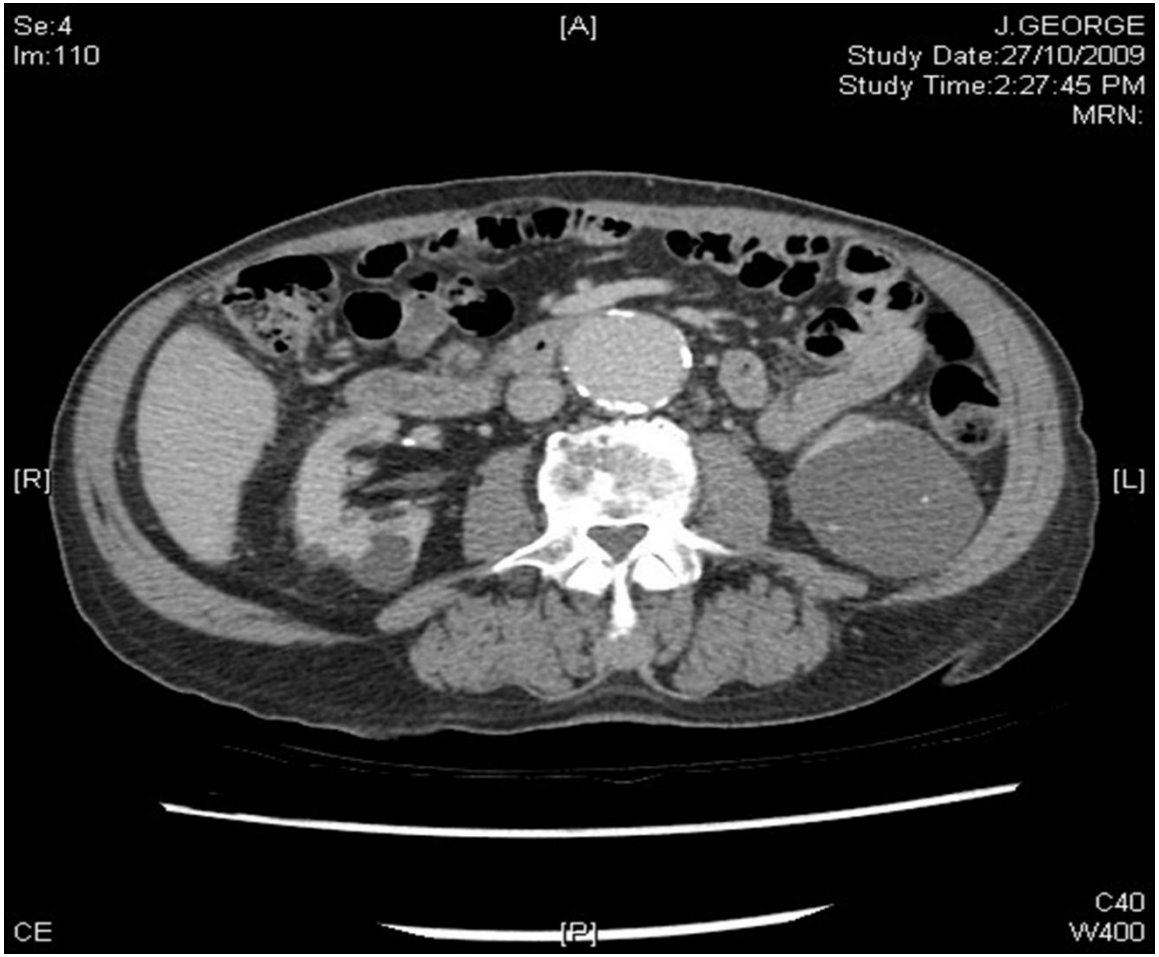
Cast of MSK Kidney



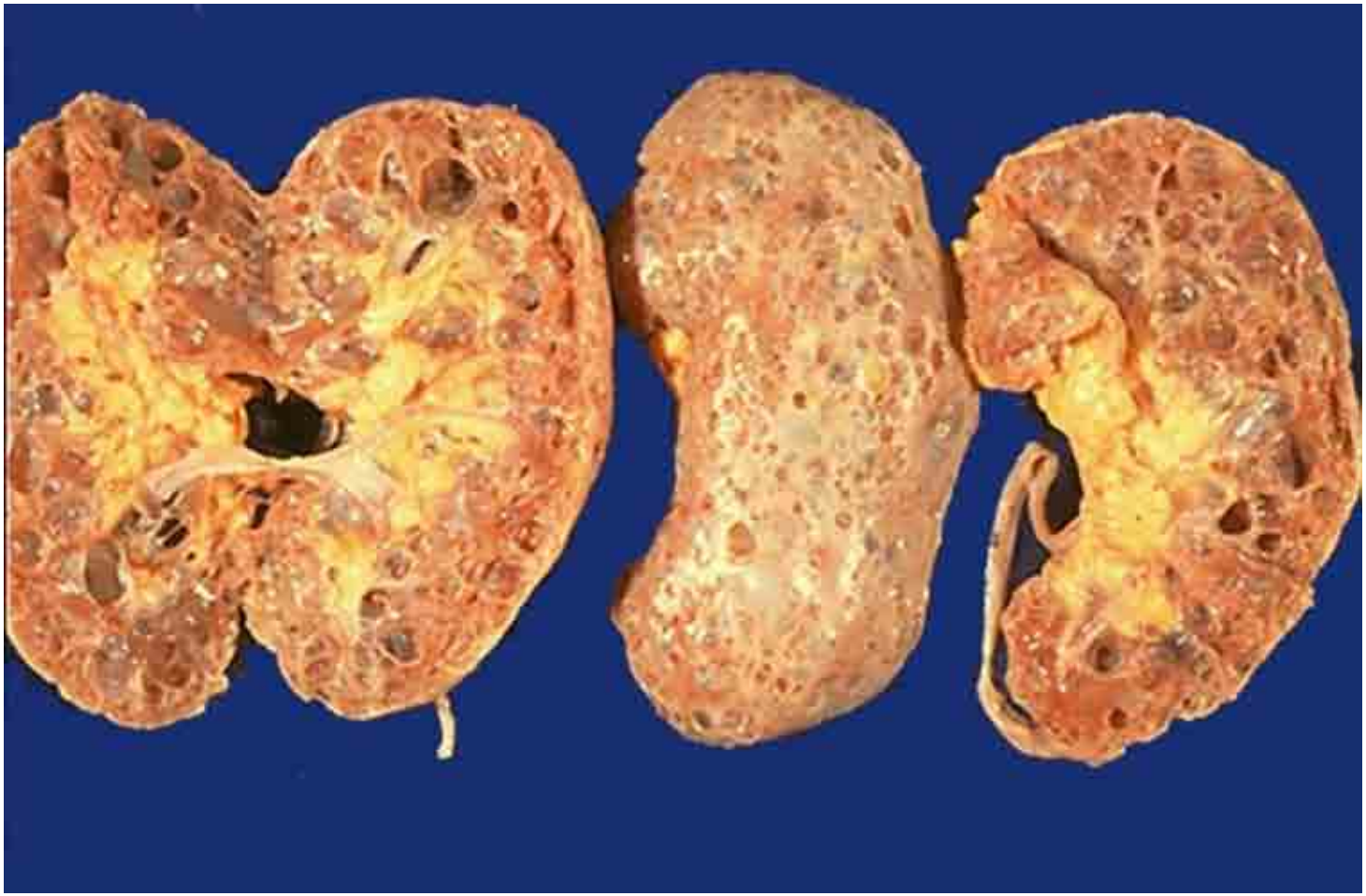
Acquired Renal Cystic Disease

- 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



Bosniak Classification of Incidental Renal Cysts

Category I	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.
Category II	Looks benign with some radiologic concerns including septation, minimal calcification, and high density; requires no surgery.
Category II F	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.
Category III	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.
Category IV	Clearly a malignant lesion with large cystic components, irregular margins; solid vascular elements; requires surgical removal.

Bosniak III –non contrast