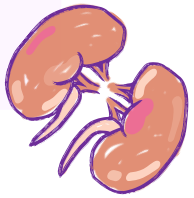
بسم الله الرحمن الرحيم

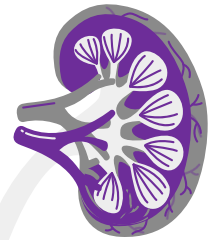
Renal Cysts

MID | Lecture 5

﴿قُلْ بِفَضْلِ ٱللَّهِ وَبِرَحْمَتِهِۦ فَبِذَٰلِكَ فَلْيَفْرَحُوا۟ هُوَ خَيْرٌ مِّمَّا يَجْمَعُونَ﴾

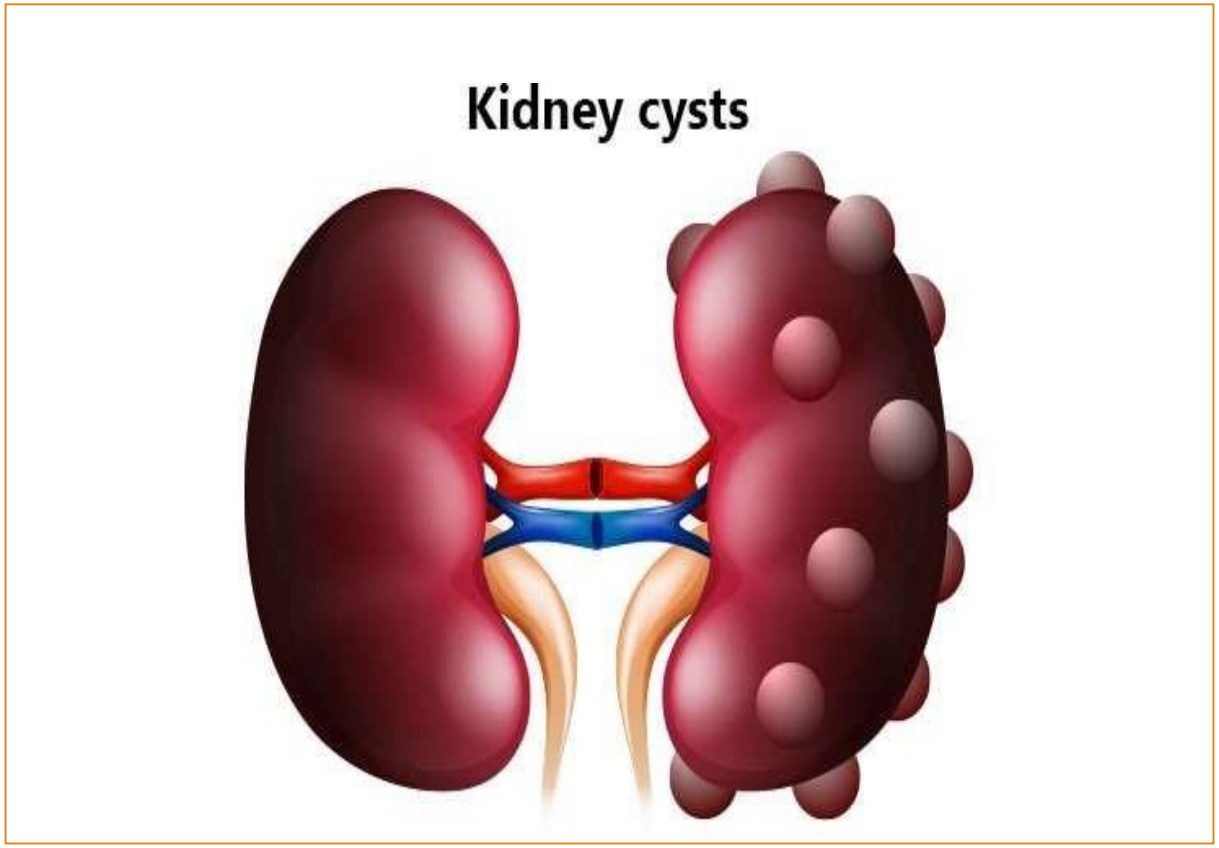
Written by: Layan Fawarseh
Aya Ghalayini





CYSTIC DISEASES of THE KIDNEY

Dr. Nisreen Abu Shahin



Types of cysts

As you know a cyst is a space that is filled with fluid. There are different types of cysts that can involve the kidney which range from completely innocent legions that have no clinical significance to others that are inherited and may lead to renal failure and can threaten the life of the patient. The major five types are:

1 Simple Cysts

2 Dialysis-associated acquired cysts

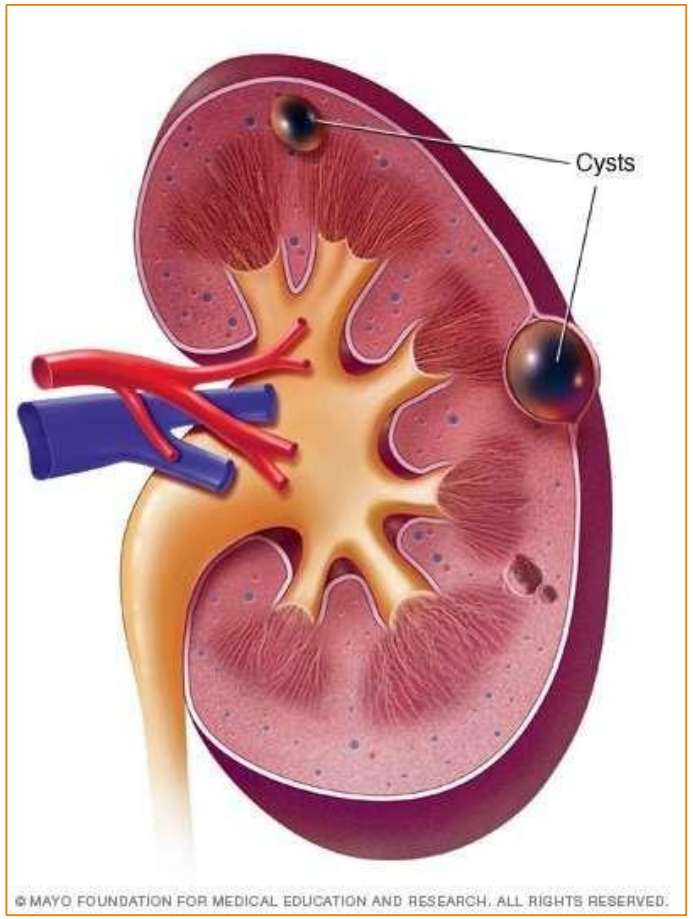
3 Autosomal Dominant (Adult) Polycystic Kidney Disease

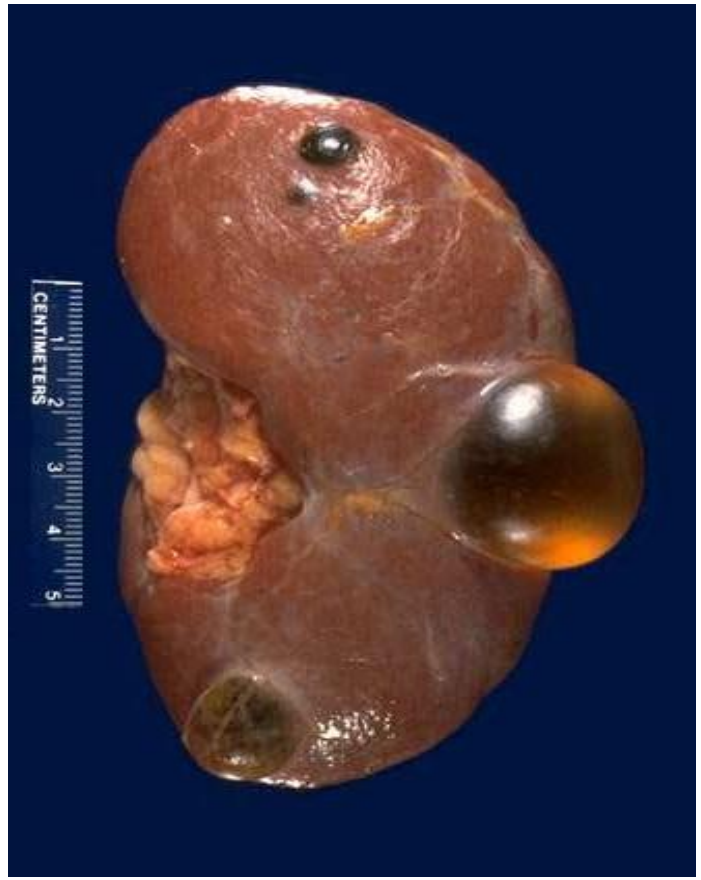
4 Autosomal Recessive (Childhood) Polycystic Kidney Disease

5 Medullary Cystic Disease

1- Simple Renal Cysts

- Have usually a favorable prognosis.
- **Multiple or single** lesions.
- **1-5 cm in diameter** or even larger**.**
- **filled with clear fluid.**
- **confined to the cortex.**
- **no clinical significance** hence the name "SIMPLE"**.**
- **Usually discovered incidentally or because of** mild symptoms such as **hemorrhage and** flank **pain**
- **Importance: to differentiate from kidney tumors.**

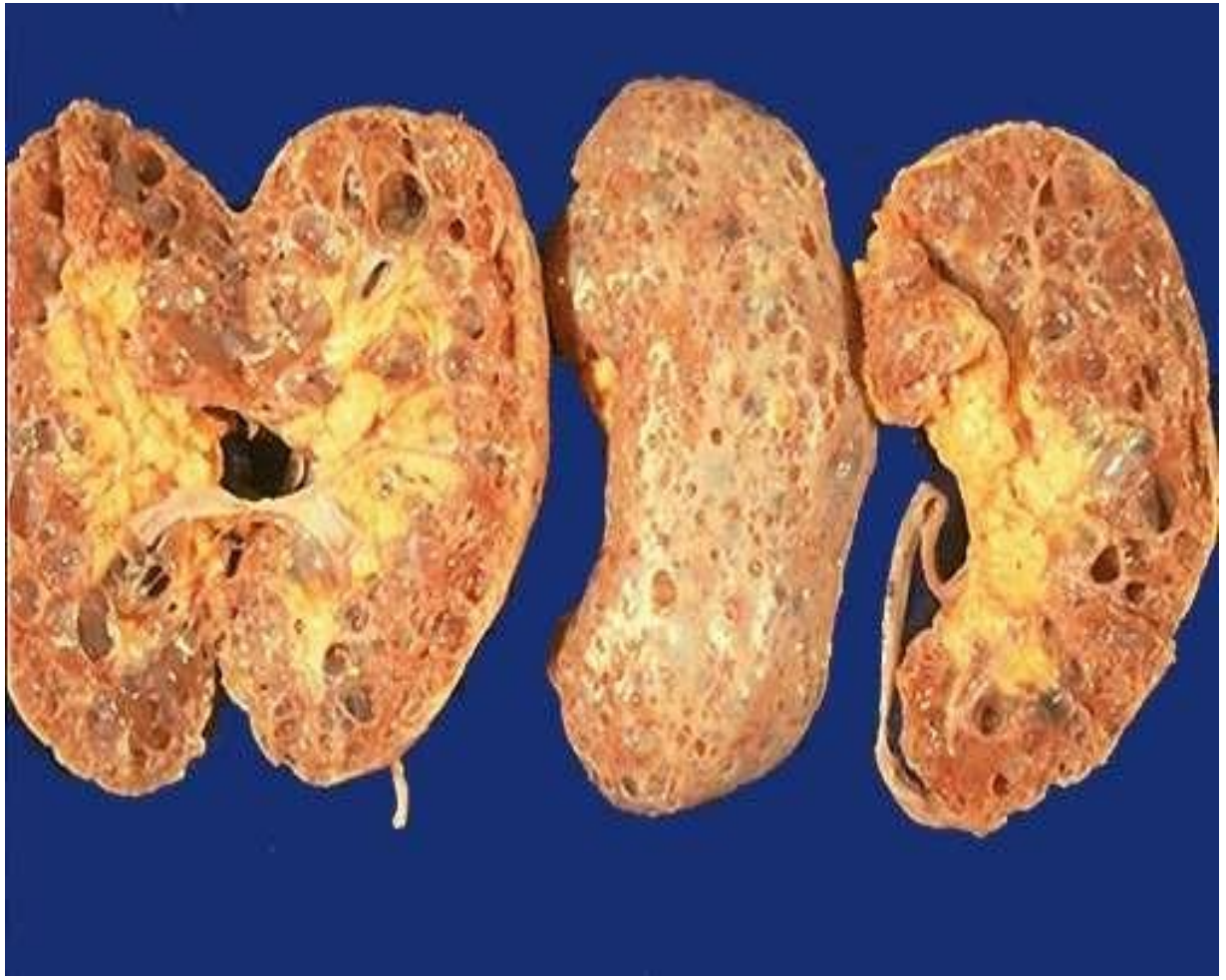




2- Cysts Associated with Chronic Dialysis

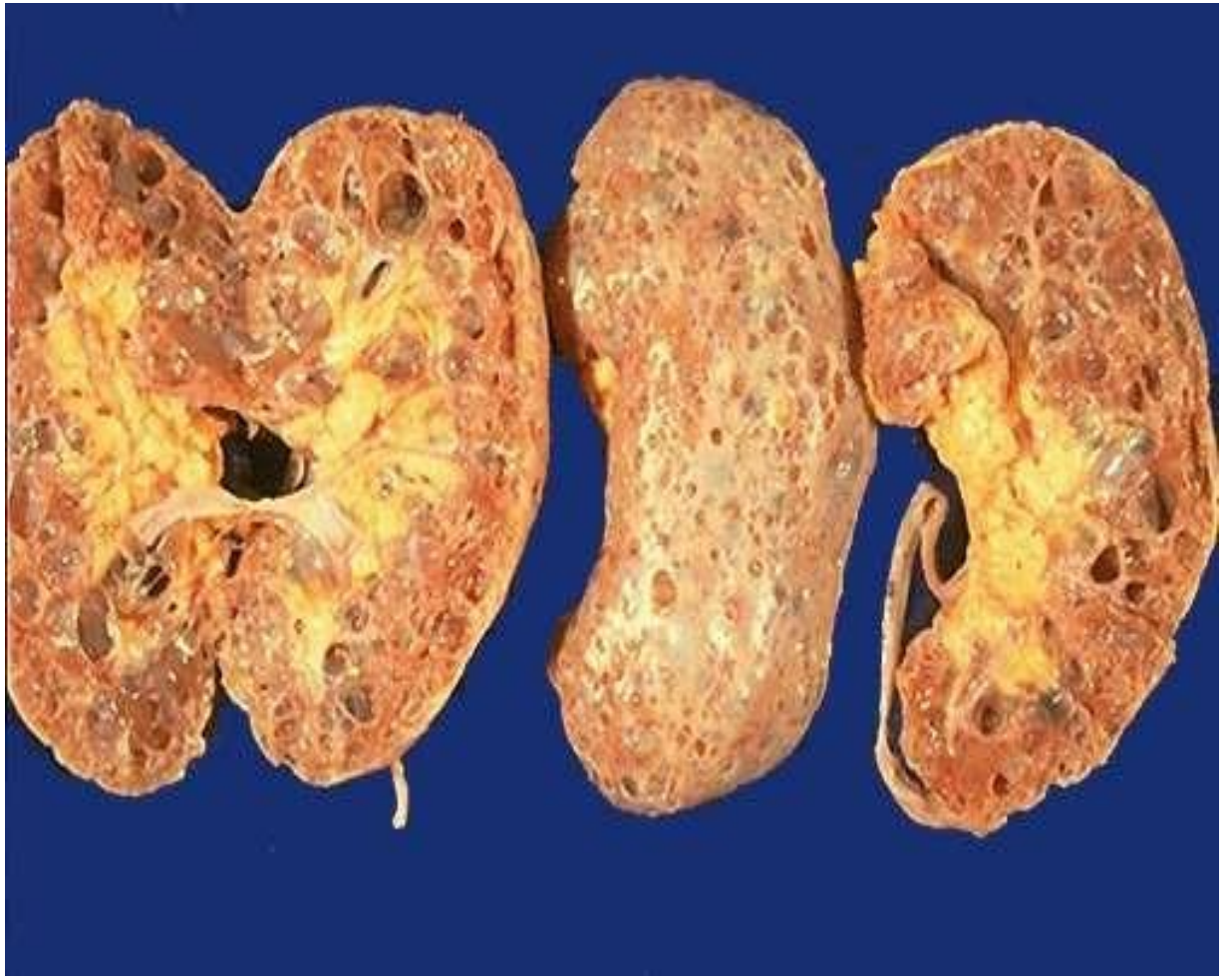
- Numerous Acquired Cysts that occur in patients with renal failure who have prolonged dialysis.
- Both cortex and medulla
- Complications: **hematuria** (due to hemorrhage inside one of these cysts); flank **pain**
- The most feared complication:

Increased risk of renal carcinomas (100 times greater than in the general population), *see next slide.*



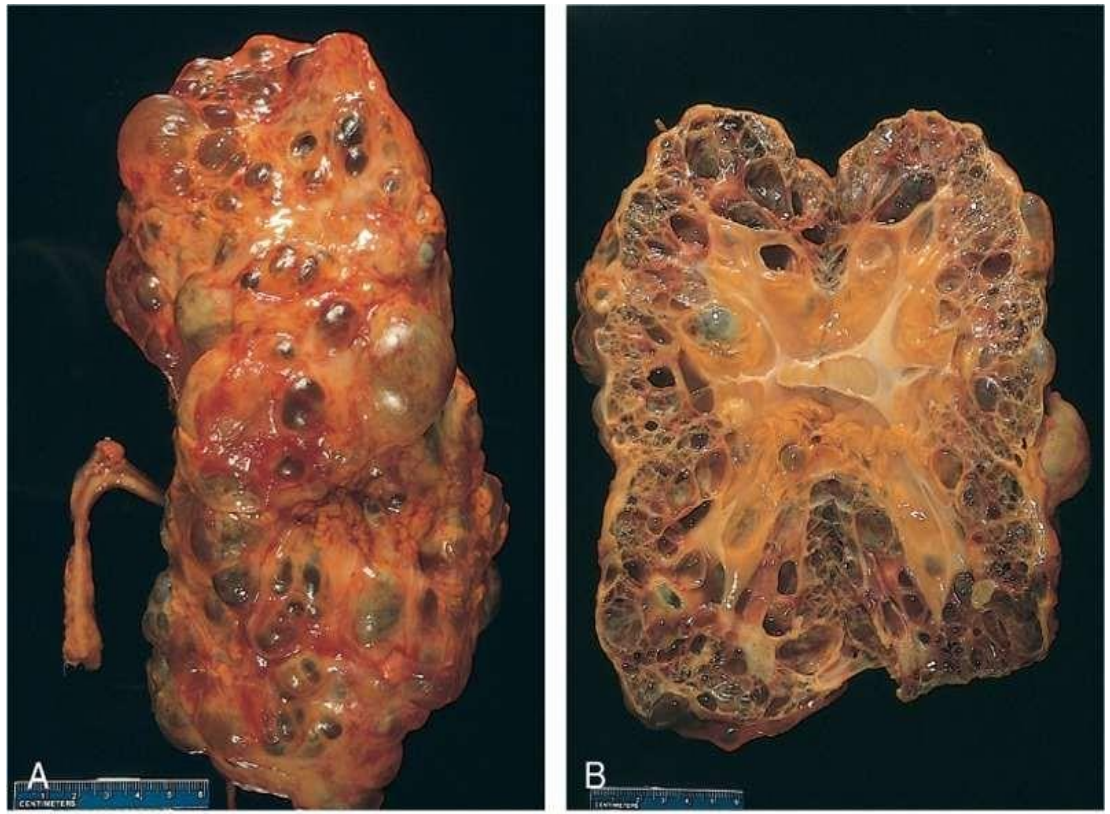
2- Cysts Associated with Chronic Dialysis – con.

- These cysts develop to malignant cells as a result of chronic inflammation and irritation that develops within the atrophic renal parenchyma (degenerated renal parenchyma), this irritation and chronic inflammation might lead to a cell division there. With cell division, cells might acquire additional mutations and later on they might go on or transform into malignant cells.



3- Autosomal Dominant (Adult) Polycystic Kidney Disease

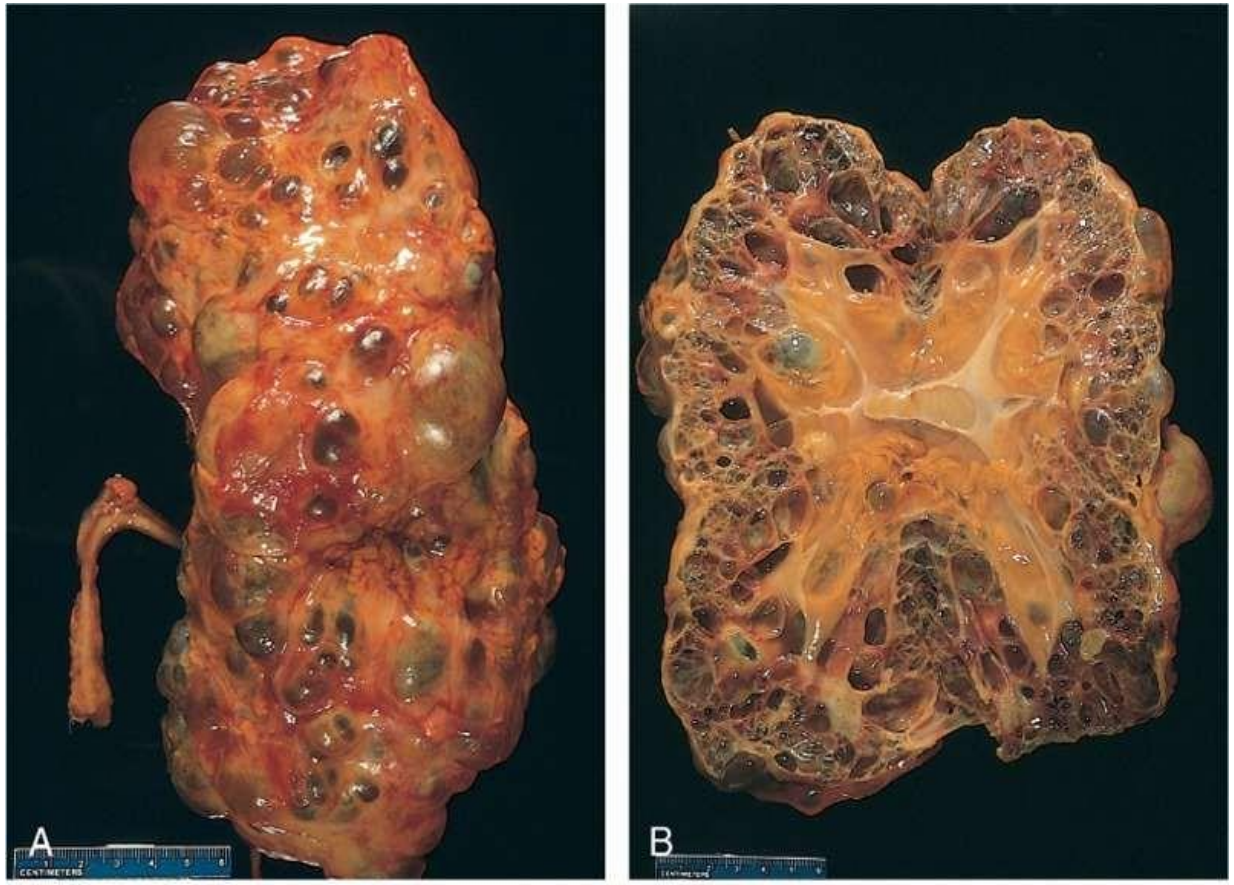
- "Autosomal dominant": Inherited
- "Adult": the fact that symptoms and manifestations of this condition does not appear to be evident or obvious until the patient reaches adulthood
- "Polycystic": **multiple bilateral cysts** involve both cortex and medulla.
- The size of the kidney: enlarged, and its weight may reach 1500 g – normal kidney weight: 250 g for both kidneys –.



3- Autosomal Dominant (Adult) Polycystic Kidney Disease – con.

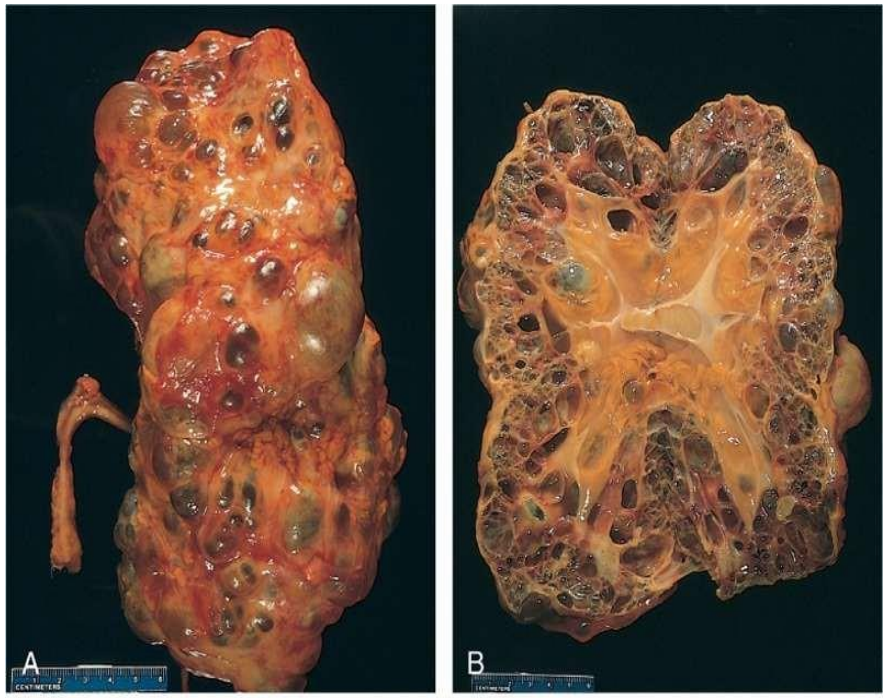
- eventually destroy the renal parenchyma **therefore destroy all the renal functions**.
- Incidence (1: 500-2000) persons
- 10% of chronic renal failure.
- inheritance of one of 2 autosomal dominant genes:
- (1)- *PKD1:* 85-90% (encodes polycystin-1)
- (2)- *PKD2* :10-15% (encodes *polycystin- 2).*

PKD: Polycystic Kidney Disease.



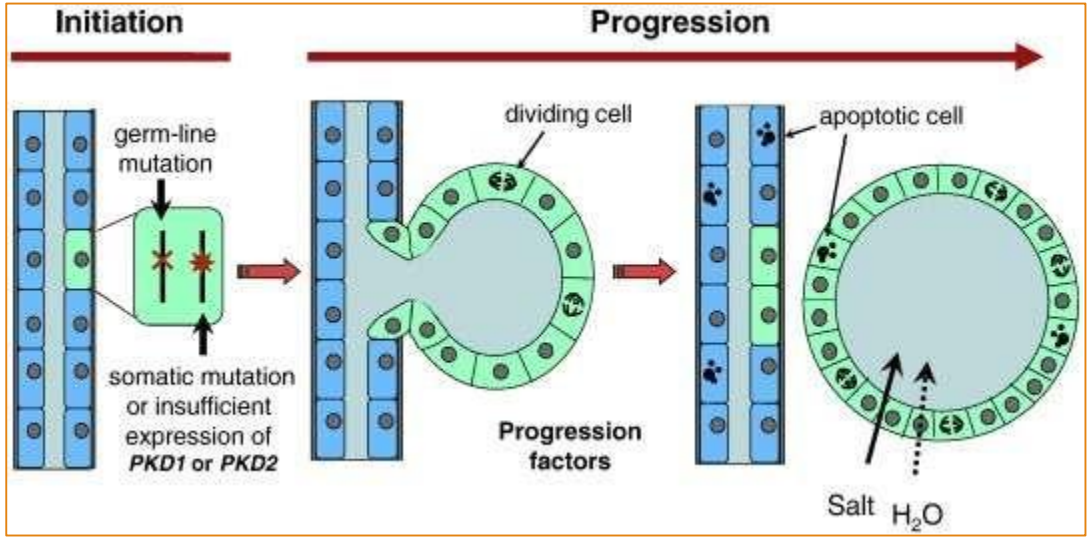
3- Autosomal Dominant (Adult) Polycystic Kidney Disease – Extra

- **polycystin-1:** PC1 is a membrane-bound protein 4303 amino acids in length expressed largely upon the primary cilium, as well as apical membranes, adherens junctions, and desmosomes. PC1: has been proposed to act as a G protein-coupled receptor.
- PKD1 consists of a voltage-gated ion channel fold that interacts with PKD2.
- **Polycystin-2:** is a transmembrane protein encoded by the human gene PKD2, associated with type 2 autosomal dominant polycystic kidney disease. It acts as an intracellular calcium release channel, impacting cellular functions and membrane potential.



(Adult) Polycystic Kidney Disease

a somatic mutation in either the PKD1 or PKD2 gene leads to abnormal and progressive cell division in the epithelial cells of the renal tubules. This division becomes discontinuous and autonomous, resulting in the formation of multiple cysts. These cysts eventually detach from the original tubules and form independent cystic spaces, and the same process continues in other renal tubules.



Clinical presentation :

- ***asymptomatic* until the 4th decade.** Patients are asymptomatic until the 3rd or 4th decade of life.
- **Symptoms: *flank pain* , heavy dragging sensation**(due to kidney enlargement)**, abdominal mass, hemorrhage** due to ruptured cysts, **obstruction** caused either due to hemorrhage within cysts or by kidney stones **and*Intermittent gross hematuria***

COMPLICATIONS

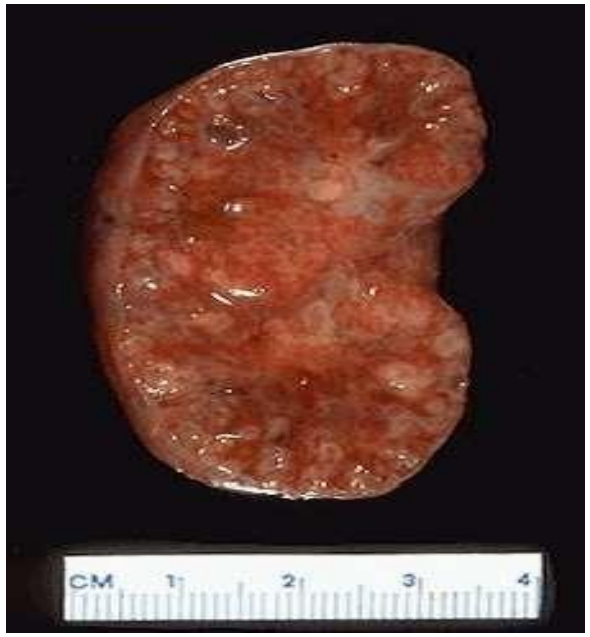
1 ***hypertension* (75%),** which is a major complication.

2 ***urinary tract infection*** (cysts can serve as sites for infection)

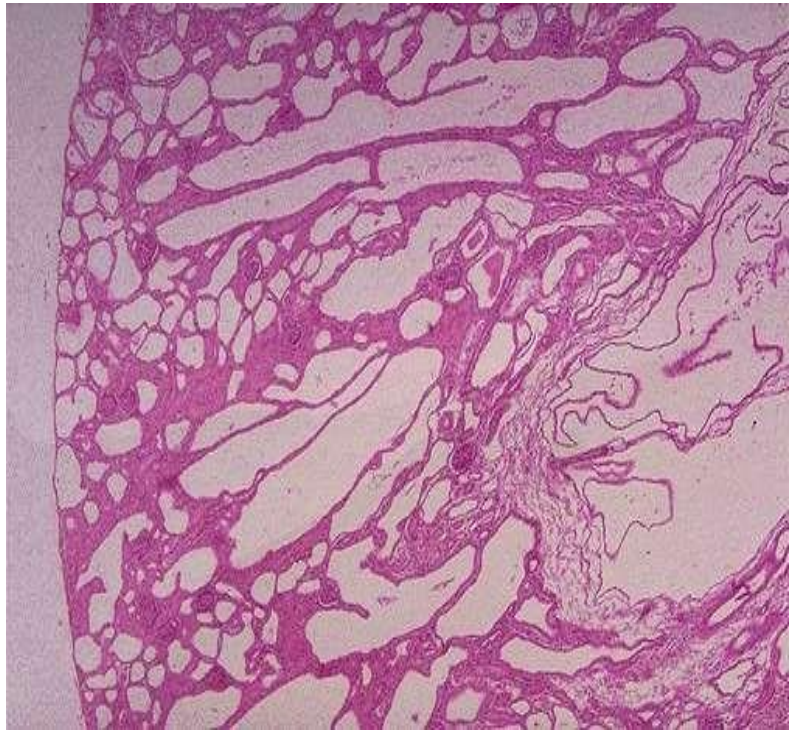
3 **vascular aneurysms of circle of Willis (10% -30%)→→ (subarachnoid hemorrhage)**

4 **chronic renal failure at age 50 (≈25% ; % increases with age)**

4-Autosomal Recessive (Childhood) Polycystic Kidney Disease

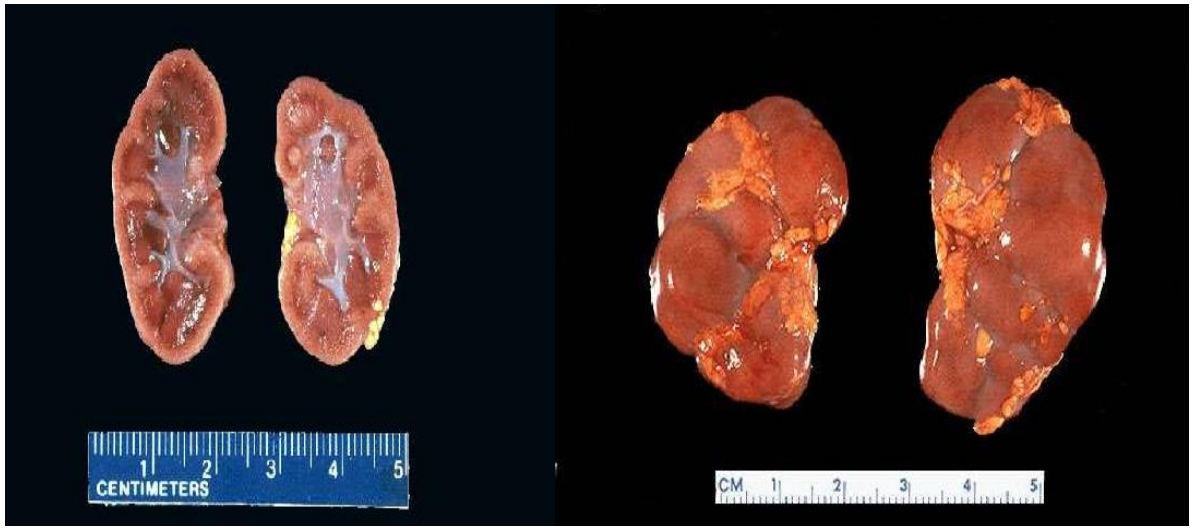


- **autosomal recessive**
- **1:20,000 live births.**
- **Types** based on the age of onset: **perinatal, neonatal, infantile, and juvenile.**
- **Presents early in life**
- **Associated with liver cysts and fibrosis**
- **Mutations in *PKHD1* gene coding for *fibrocystin*.**
- **Fibrocystin may be involved in the function of cilia in tubular epithelial cells .**
- Patients typically have bilateral renal cysts and may also have associated liver cysts.



Normal vs childhood polycystic kidneys

NORMAL TERM INFANT KIDNEYS



CHILDHOOD POLYCYSTIC KIDNEYS

- Morphologically, the kidneys show multiple cysts involving both the cortex and the medulla. Microscopically, the normal renal parenchyma is replaced by cystic spaces derived from abnormal renal tubules.
- Compared to a normal infant kidney, the affected kidney is enlarged, and its parenchyma is filled with numerous small cysts throughout both cortical and medullary regions.

Adult vs childhood polycystic kidney disease

Feature	Adult polycystic kidney disease	Childhood polycystic kidney disease
Cyst size	Large, variable-sized cysts	Small, fusiform cysts
Distribution	Distort and enlarge the kidney	Distributed throughout the renal parenchyma
Kidney appearance	Markedly enlarged and deformed	Enlarged with uniform small cysts
Outcome	Progressive chronic kidney failure	Progressive chronic kidney failure

Adult vs childhood polycystic kidney disease

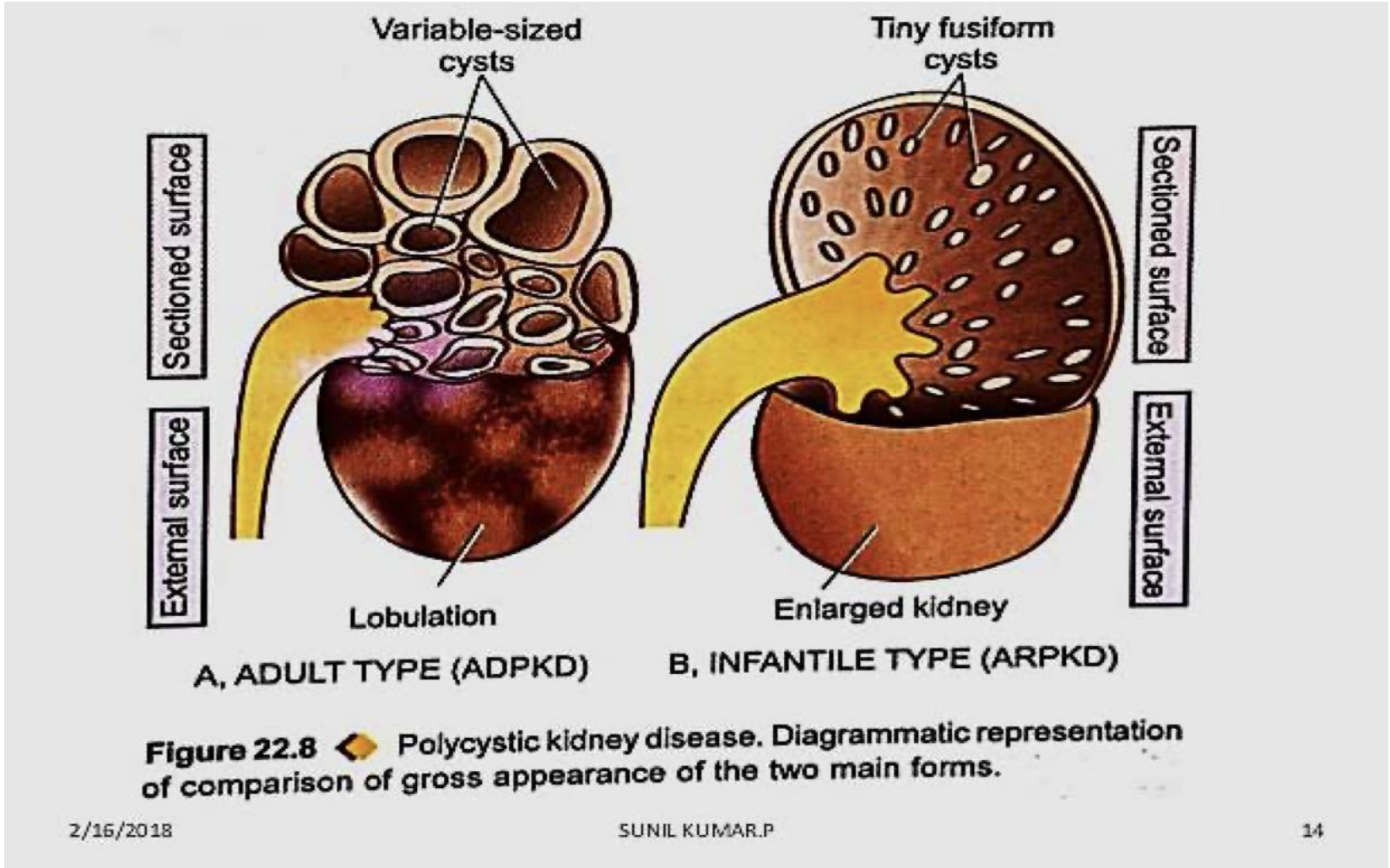


Figure 22.8 Polycystic kidney disease. Diagrammatic representation of comparison of gross appearance of the two main forms.

5- Medullary Cystic Disease

- **2 major types:** medullary sponge kidney and nephronophthisis.

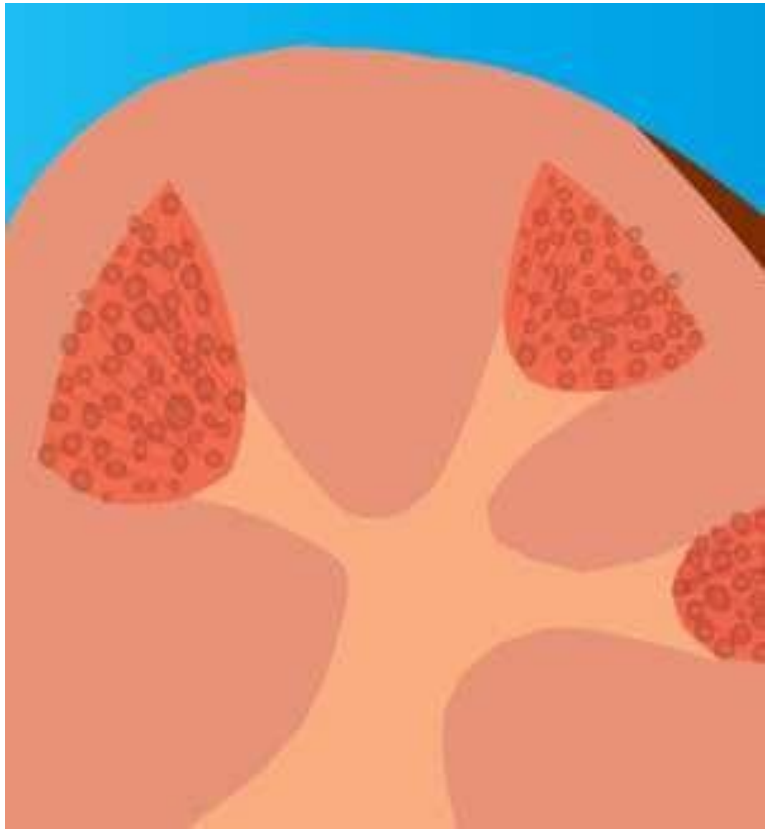
1 medullary sponge kidney

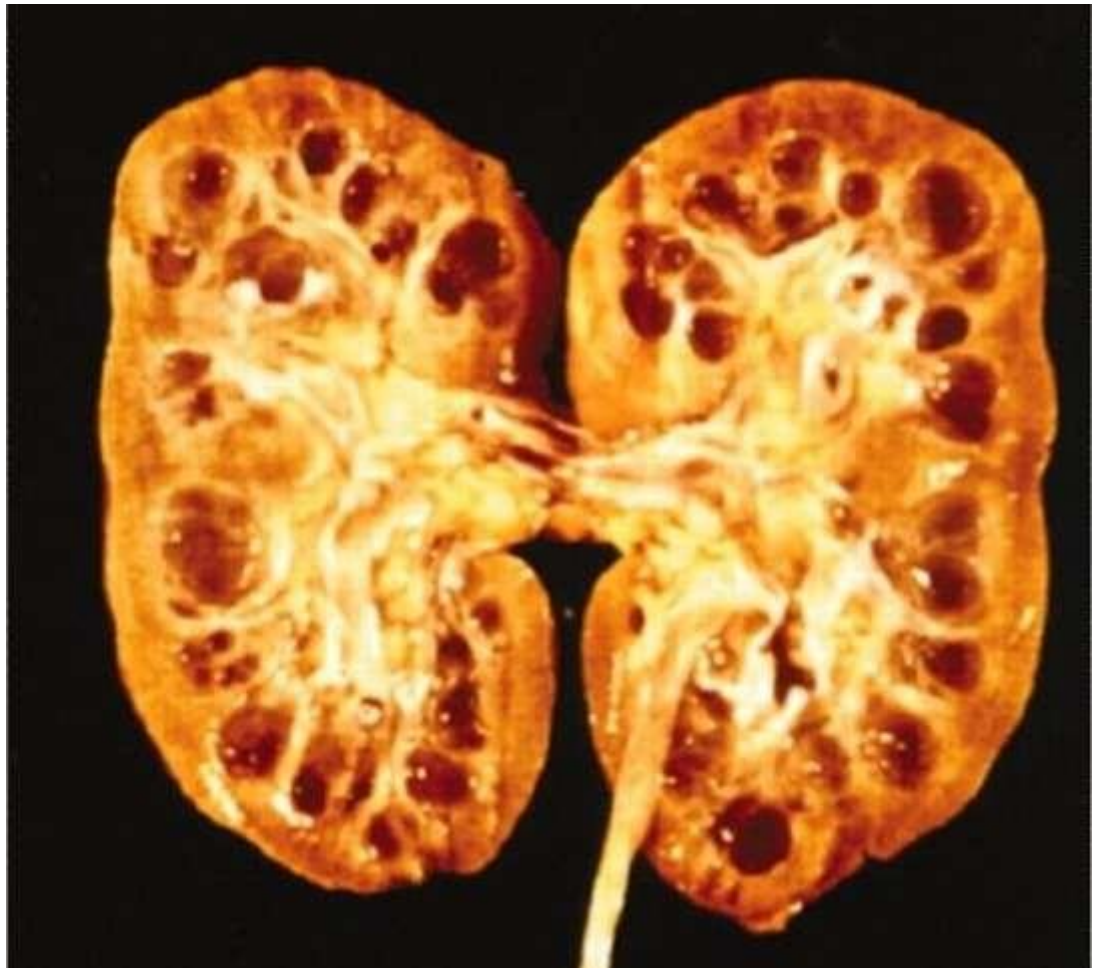
- **common and innocent condition (**more common and relatively benign form)

2-nephronophthisis-medullary cystic disease complex

- **almost always associated with renal dysfunction**
- So it's less common but more severe, often leading to renal dysfunction and progression to renal failure.
- **usually begins in childhood** (both types)
- **Cysts are at cortico-medullary junction** (both types)

Medullary Sponge Kidney





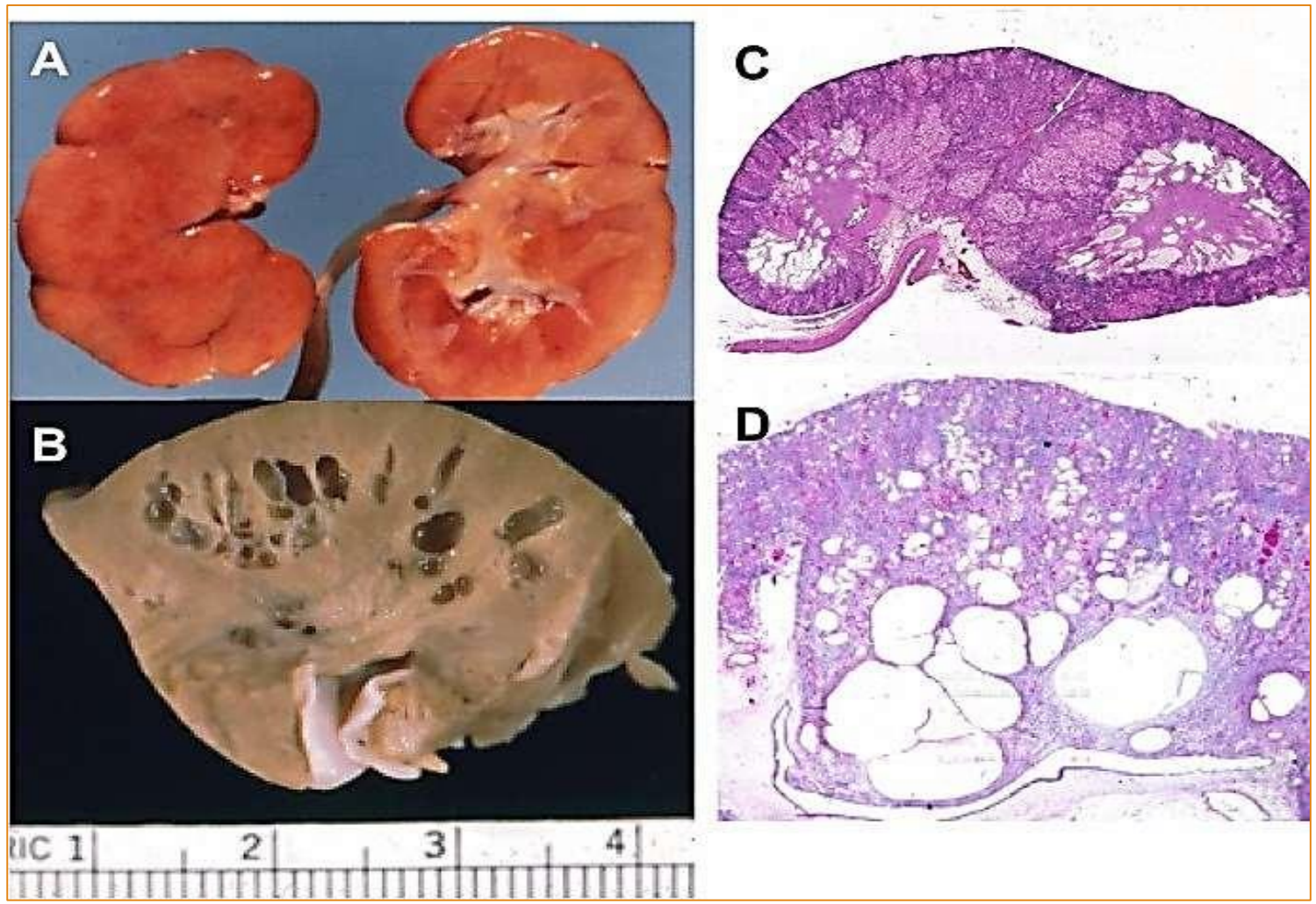
5- Medullary Cystic Disease

2- Nephronophthisis-medullary cystic disease complex (medullary- uremic type)

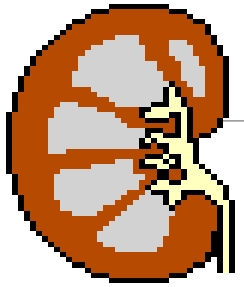
- A worse disease with progressive renal impairment
- polyuria and polydipsia (↓tubular function)
- renal failure over 5-10 years
- A positive family history and unexplained chronic renal failure in young patients should lead to suspicion of medullary cystic disease.

➢ Nephronophthisis typically presents in children with **polyuria, polydipsia, renal impairment and a family history of unexplained chronic renal failure**. This combination should raise strong suspicion, as the disease often progresses to renal failure in childhood or early adulthood.

Nephronophthisis-medullary cystic disease complex (medullary- uremic type)

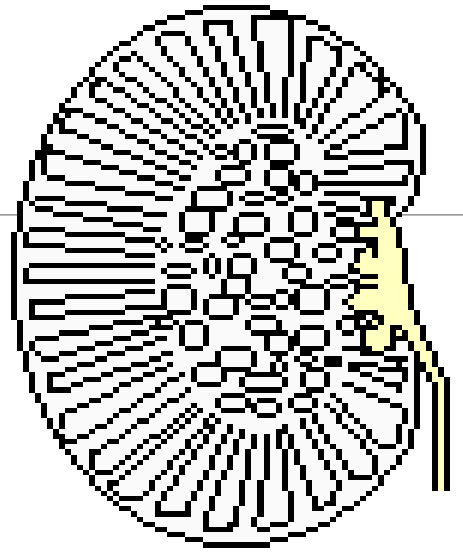


Kidney Cysts

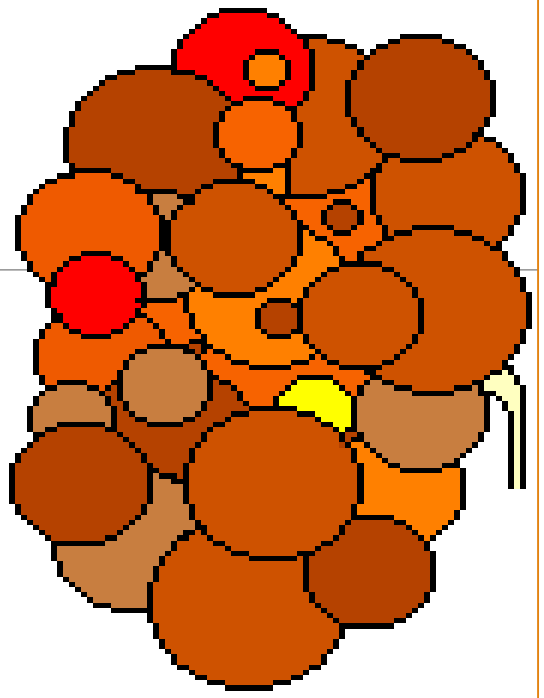


No cysts

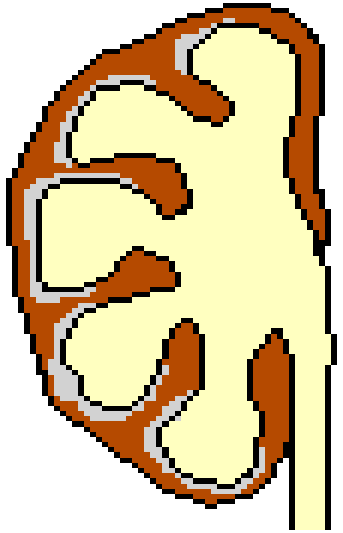
Simple cysts



Recessive polycystic



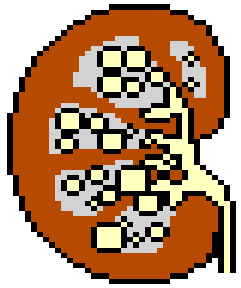
Dominant polycystic



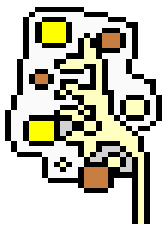
Hydronephrosis is not cysts

"Dysplasia"

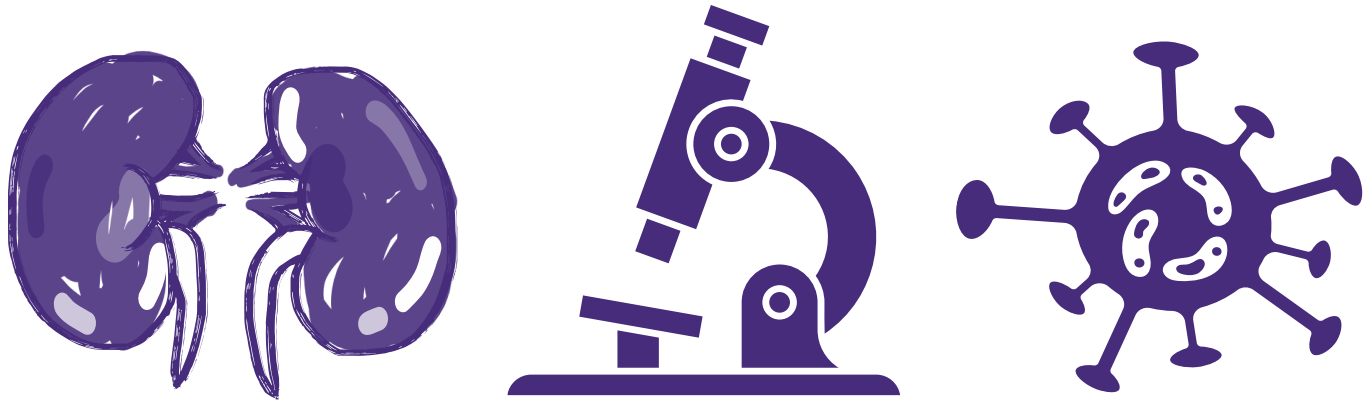
Medullary sponge



Medullary uremic



Dialysis cystic



PATHOLOGY
QUIZ
LECTURE 5

External Resources

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