النادي الطلابي
كلية الطب

Pathology GUS

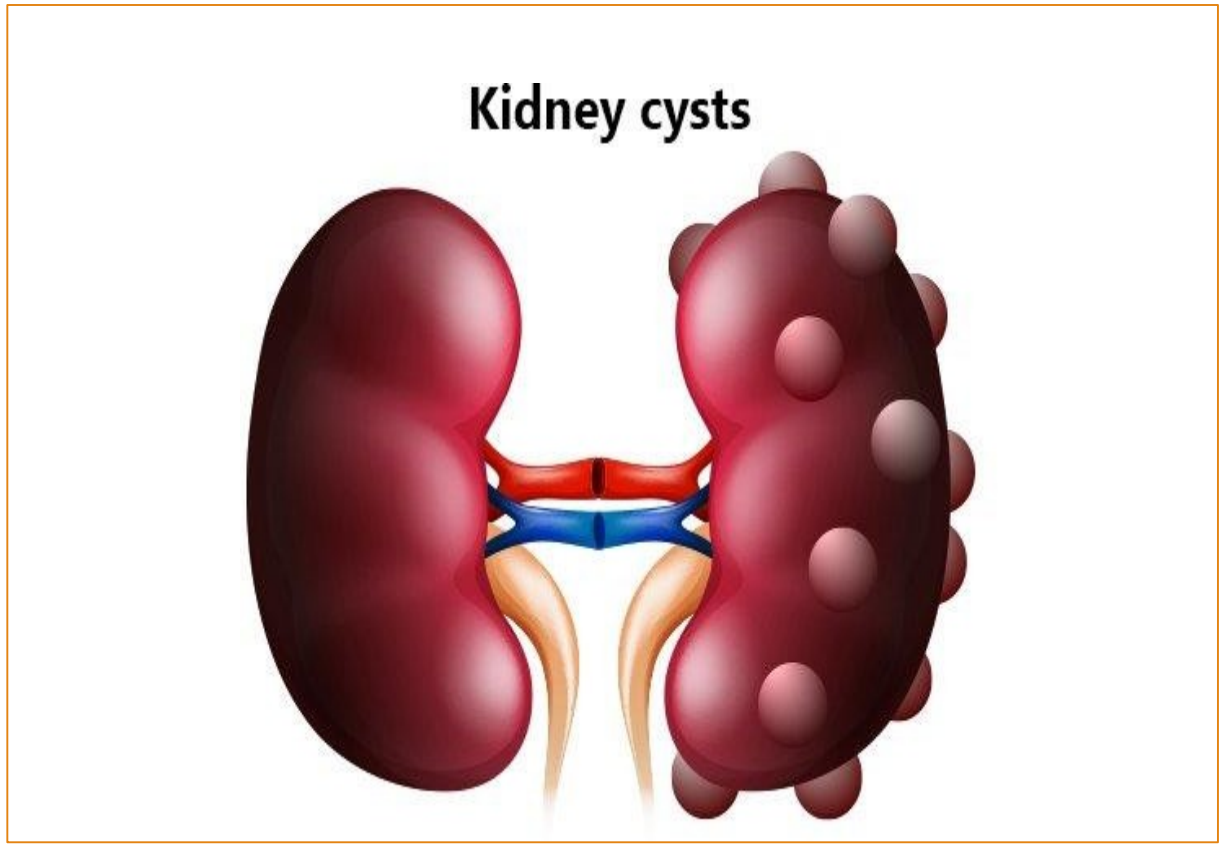
Done By Dana Alkhatib

Corrected By Leen Musleh

THE UNIVERSITY OF JORDAN

CYSTIC DISEASES of THE KIDNEY

Dr. Nisreen Abu Shahin



Types of cysts

The importance of the cysts and the characteristics differ between the different types.
All of them share that we need to differentiate them from other serious conditions like tumors

1 Simple Cysts

2 Dialysis-associated acquired cysts

3Autosomal Dominant (Adult) Polycystic Kidney Disease

4Autosomal Recessive (Childhood) Polycystic Kidney Disease

5 Medullary Cystic Disease

All of these diseases are tubular diseases

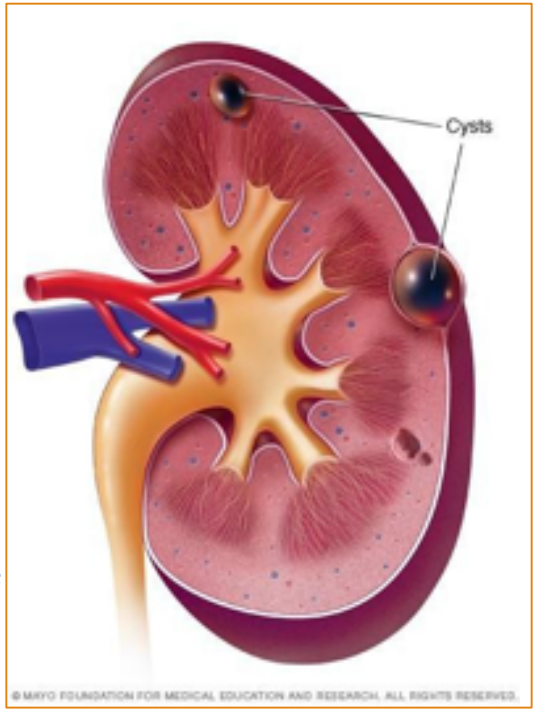
1- Simple Renal Cysts

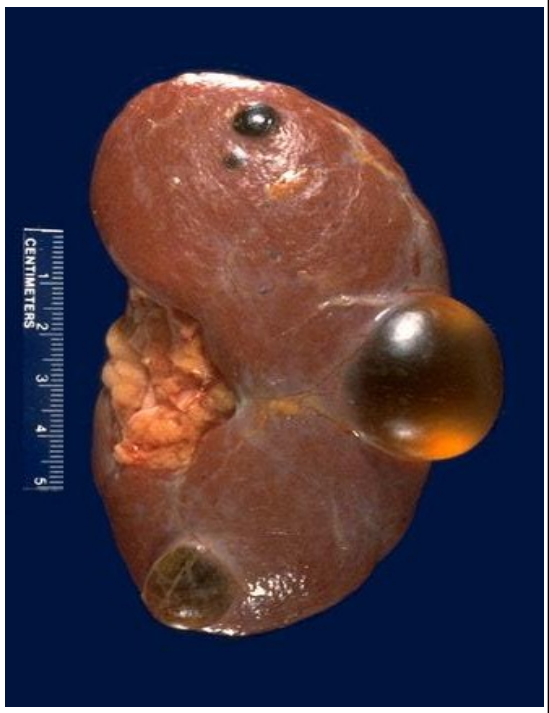
- **Multiple or single**
- **1-5 cm in diameter**
- **filled with clear fluid.**
- **confined to the cortex.**
- **no clinical significance.** Asymptomatic
- **Usually discovered incidentally** (for example when the patient does a radiological image for something else) **or because of hemorrhage** (when the cyst is large, it can lead to hematuria) **and pain**

Bleeding into a cyst occurs due to ruptured blood vessel near the cyst or direct trauma to the cyst & rupture or infection or stone formation

- **Importance: to differentiate from kidney tumors**

By radiological evaluation or histopathological confirmation





You can identify different sized smooth cysts in the cortex of the kidney. Theses cysts have a single locule (lumen) with thin walls without septations

The management of the cyst if it's asymptomatic is just to follow up with the patient, but if there are symptoms, we should perform surgical removal of the cysts

2- Cysts Associated With Chronic Dialysis

- **patients with renal failure who have prolonged dialysis*.**

Twice or 3 times a week for decades

Acquired disease because of chronic dialysis

- **both cortex and medulla**
- **Complications: hematuria; pain**

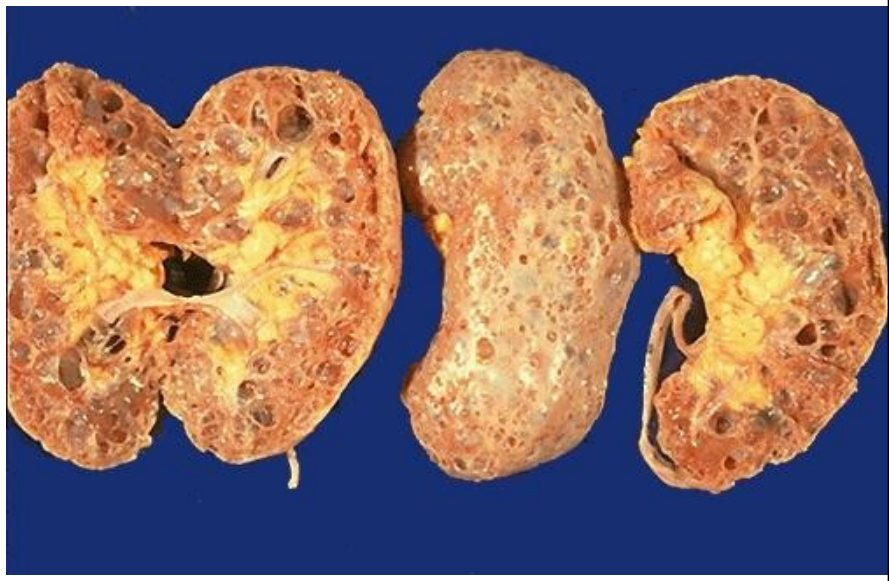
Because of rupture or trauma

Could be asymptomatic and sometimes increases the chance of UTIs & stone formation

- **Increased risk of renal carcinomas**

(100 times greater than in the general population)**

The best management is renal transplantation and removal of the diseased kidney



Large number of cysts overall the non-functioning kidney

Mechanism of cyst formation:
the non-functioning kidney is still a viable tissue and is receiving blood, but because of its decreased function, inflammation, irritation & fibrosis occur which may stimulate some growth factors & chemokines, leading to cysts.

3- Autosomal Dominant (Adult) Polycystic Kidney Disease

The manifestations appear during adulthood

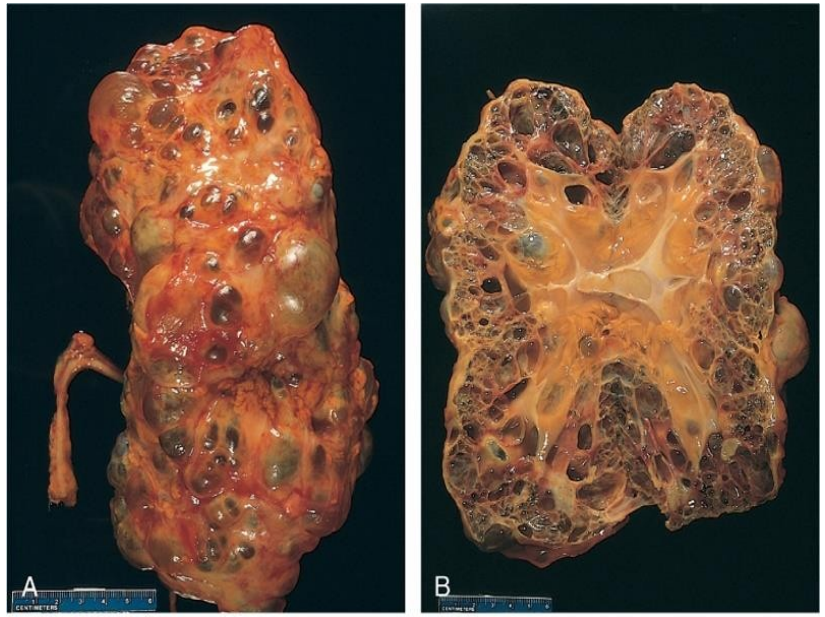
- **multiple bilateral** (both kidneys) **cysts**

Begins as numerous cysts in the cortex then replaces all the parenchyma

- **eventually destroy the renal parenchyma.**

By compression and ischemia, leading to loss of renal function and the start of clinical symptoms

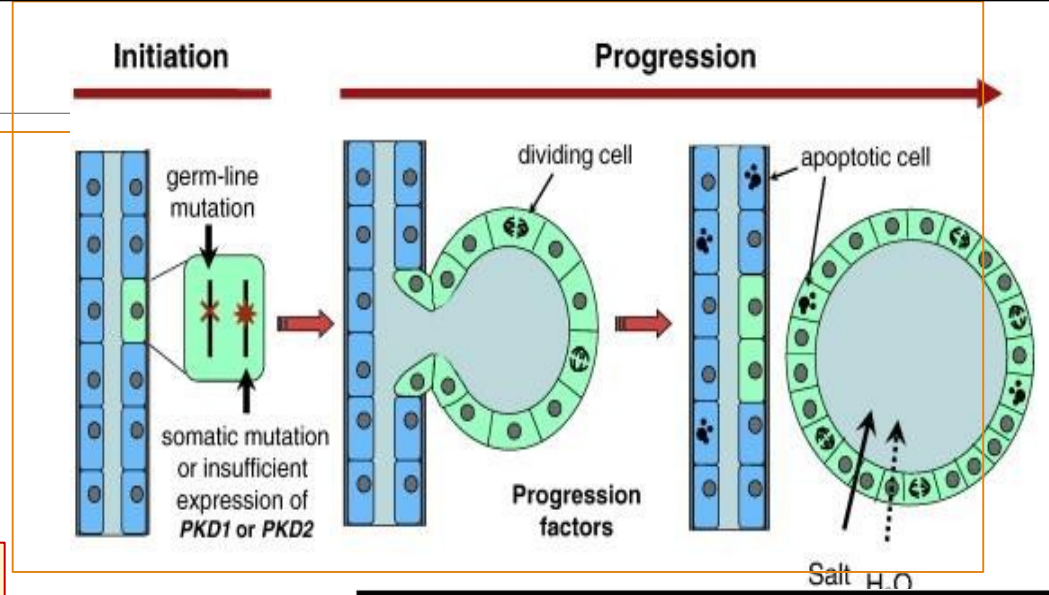
- **Incidence (1: 500-2000) persons** Not too rare
- **10% of chronic renal failure.**
- **inheritance of one of 2 autosomal dominant genes:**
- **(1)- *PKD1:* 85-90% (encodes polycystin-1)**
- **(2)- *PKD2* :10-15% (encodes *polycystin- 2).***



The weight of a normal single kidney is around 150g, but the weight of the diseased kidney could reach 2-4 kg and the disease affects both kidneys

These genes are not similar in structure and present on different chromosomes, PKD1 on chromosome 16
And PKD2 on chromosome 4; both are highly expressed in kidney tubules (primary cilia), endothelial cells, and smooth muscle cells of blood vessels. The loss of the expression of these genes leads to cysts formation.

(Adult) Polycystic Kidney Disease



Clinical presentation :

- ***asymptomatic*** ***until the 4th decade***
- **Symptoms:** ***flank pain*** **, heavy dragging sensation, abdominal mass,**

  hemorrhage, obstruction, ***Intermittent gross hematuria***

Higher chance to develop recurrent UTIs or stone formation

Complications

1 ***hypertension*** **(75%)***

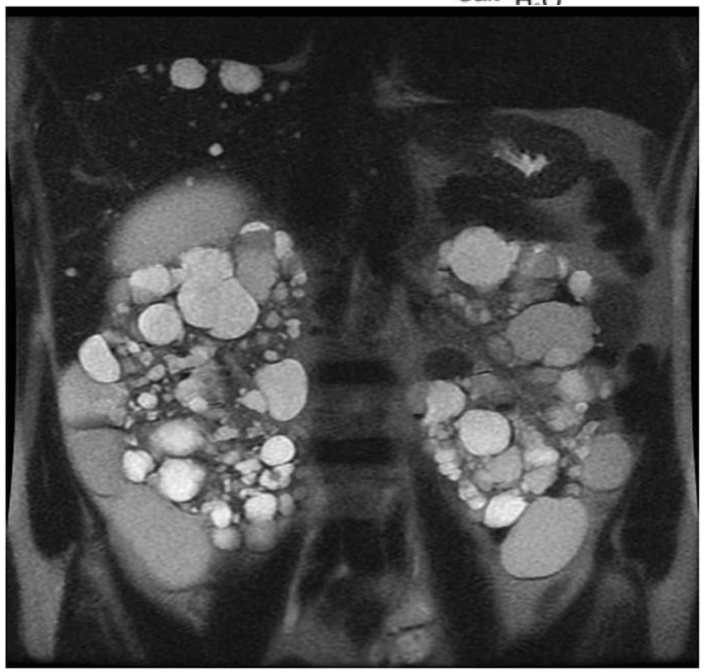
2 ***urinary infection***

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

4- renal failure at age 50

Theses complications are related to the loss of expression of PKD genes in blood vessels

The management:
Removal of kidneys and renal transplantation

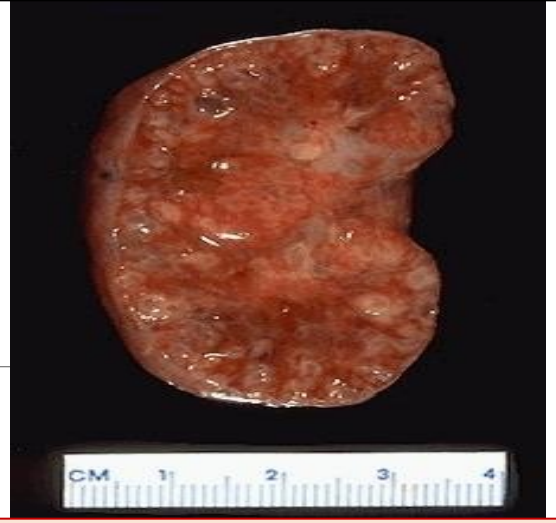


Coronal T2-weighted image shows symmetric enlargement of the kidneys, which contai n multiple cysts with variable size

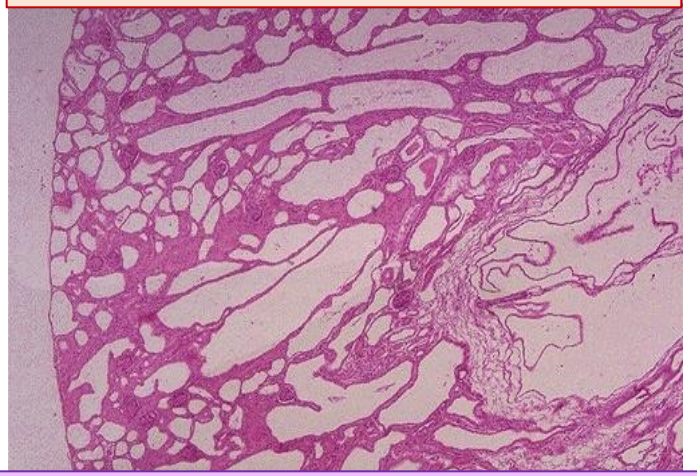
4-Autosomal Recessive (Childhood) Polycystic Kidney Disease

The manifestations appear during childhood

- **autosomal recessive**
- **1:20,000 live births.** Less common than the adult type
- **Types: perinatal, neonatal, infantile, and juvenile.** According to the onset of manifestations
- **Presents early in life** Renal impairment
- **Associated with liver cysts**
- **Mutations in *PKHD1* gene coding for *fibrocystin*.**
- **Fibrocystin may be involved in the function of cilia in tubular epithelial cells .**



The kidney is enlarged and contains multiple small cysts

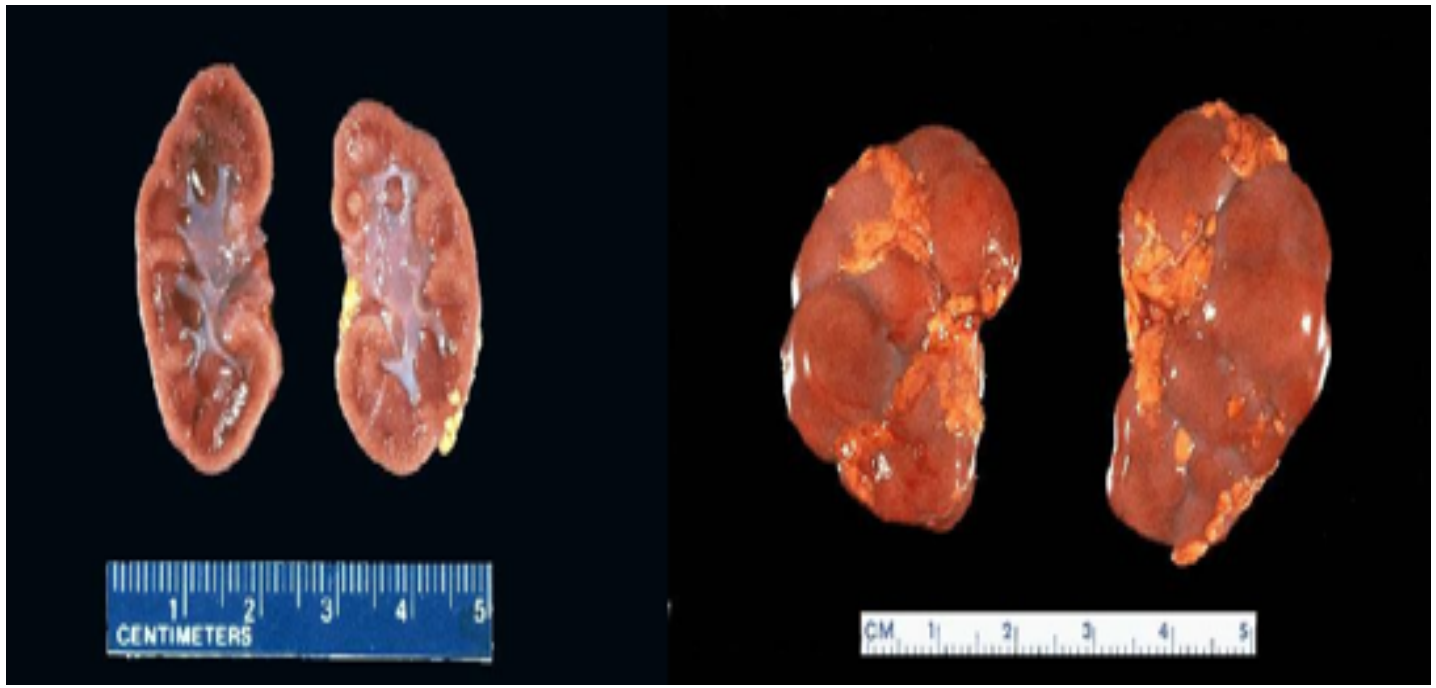


Microscopic: the whitish space represents the location of the cyst; the pink is the intervening renal parenchyma. With time, the whitish color increases because of the increasing number & size of cysts, finally resulting in chronic renal failure.

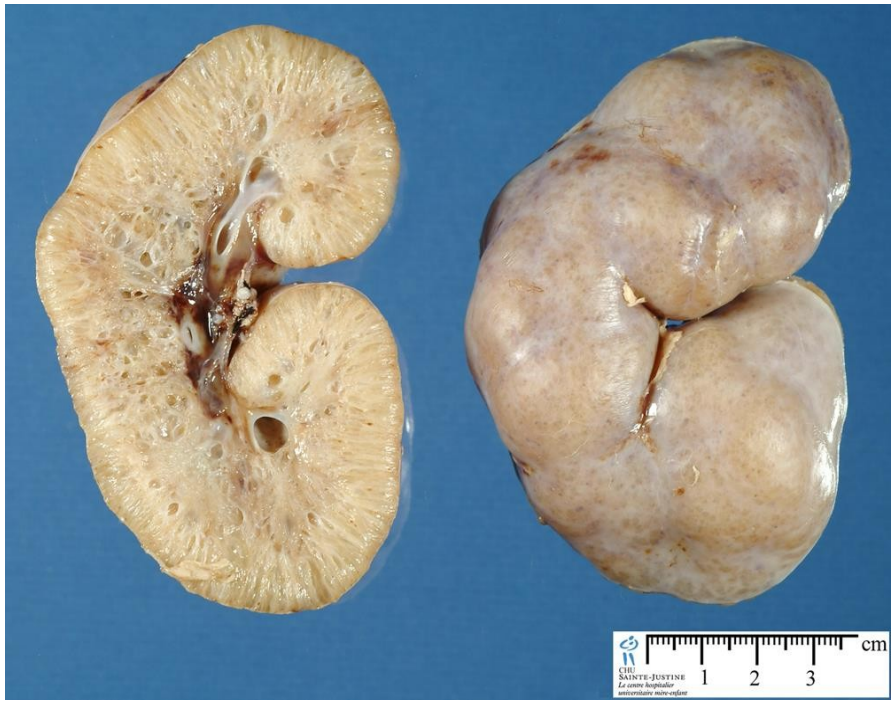
The best management is renal transplantation

Normal vs childhood polycystic kidneys

NORMAL TERM INFANT KIDNEYS



CHILDHOOD POLYCYSTIC KIDNEYS



The kidney is larger than normal, the outside is smooth, but the inside is abnormal and there is loss of corticomedullary differentiation & large number of small cysts

Adult vs childhood polycystic kidney disease

Variable sized cysts

Lobulated and abnormal outer surface

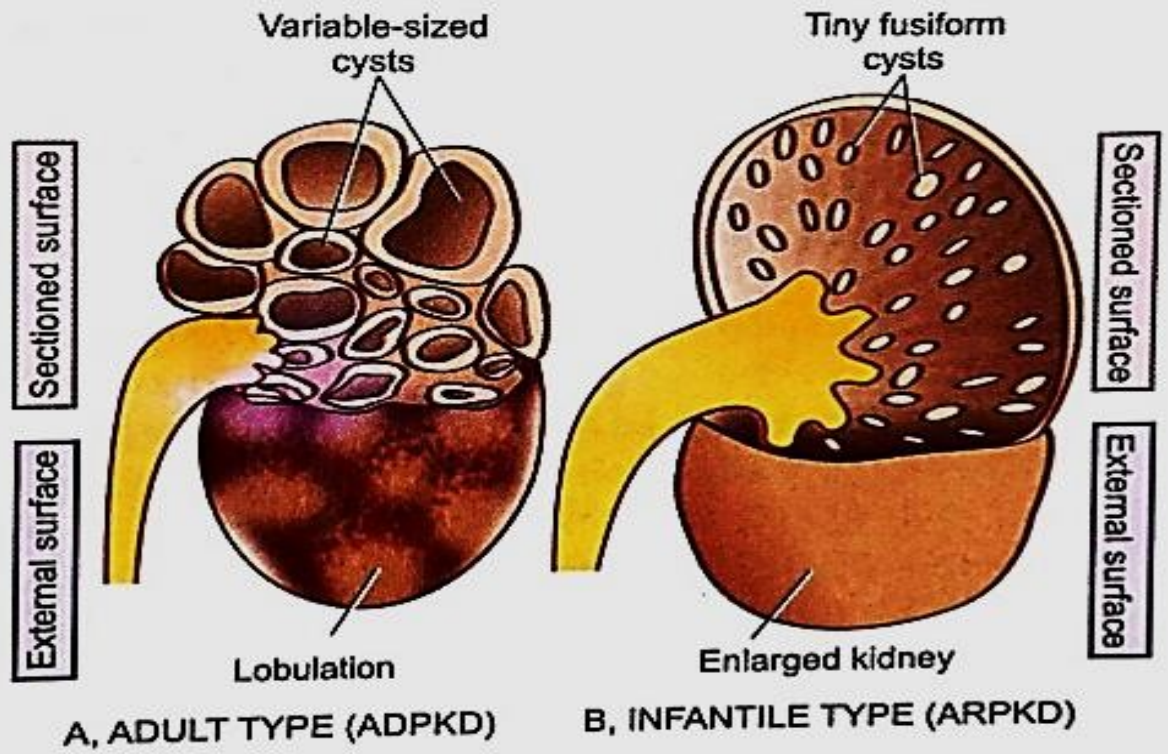


Figure 22.8 Polycystic kidney disease. Diagrammatic representation of comparison of gross appearance of the two main forms.

tiny fusiform cysts

The outside is smooth

5- Medullary Cystic Disease

➢2 major types:

In both of them, the cysts are located in the medulla of the kidney (what a shocker!)

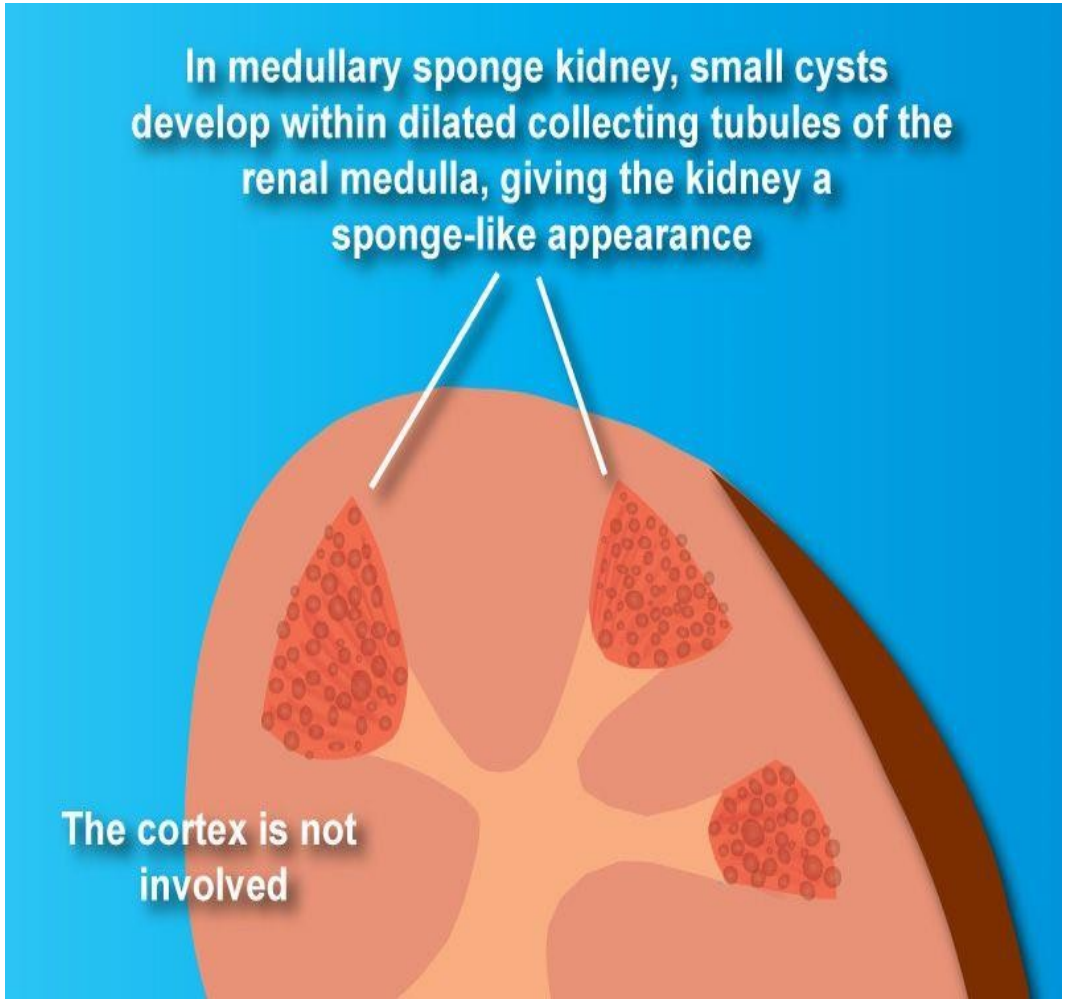
1-Medullary sponge kidney

- **Rare and nonthreatening condition.**
- **Rare, developmental abnormality characterized by ectatic (dilatory) or cystic malformations in the medullary collecting ducts of the kidney resulting in medullary cysts.**
- **Most patients are asymptomatic and the condition may be diagnosed based on incidental findings following radiologic investigation for other reasons.**

(Some patients may develop mild symptoms related to stone formation (flank pain and painful hematuria)

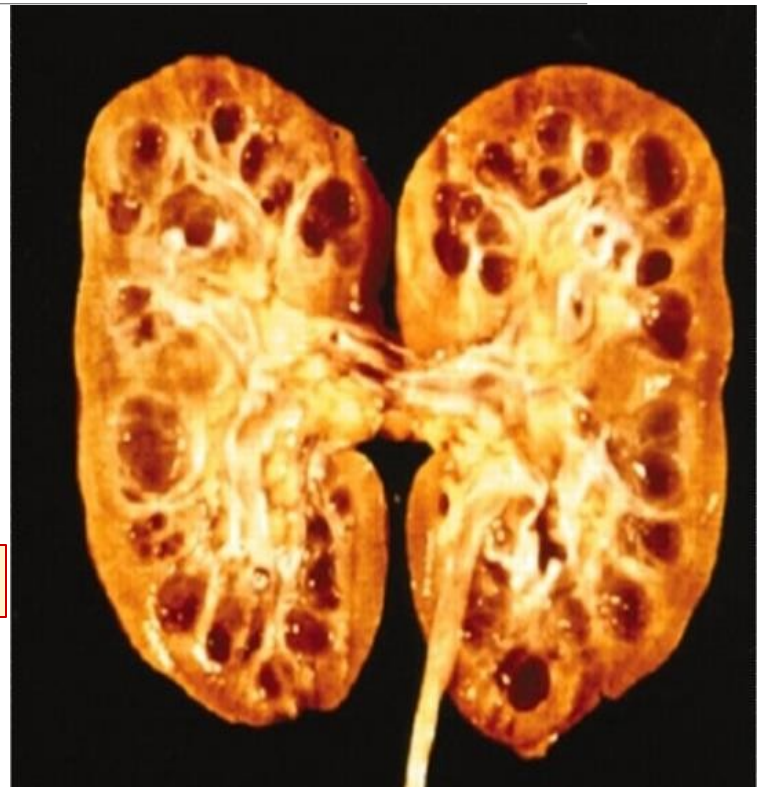
- Favorable and good prognosis

Spongy appearance of medulla



2-N*ephronophthisis-medullary cystic disease complex*

- **- almost always associated with renal dysfunction.**
  - Eventually leads to chronic renal failure
- **- usually begins in childhood.**
- **- Cysts are at cortico-medullary junction**
- **More than 9 gene mutations are described**
  - Most of them are autosomal recessive
  - It's an inherited disease
- **All share in common renal histologic triad of tubular basement membrane disintegration, tubular atrophy with cyst development, and interstitial cell infiltration with fibrosis**
  - Bad & not favorable prognosis



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

- **Clinical features:**
- **polyuria** (increased urine) **and polydipsia** (increased thirst)

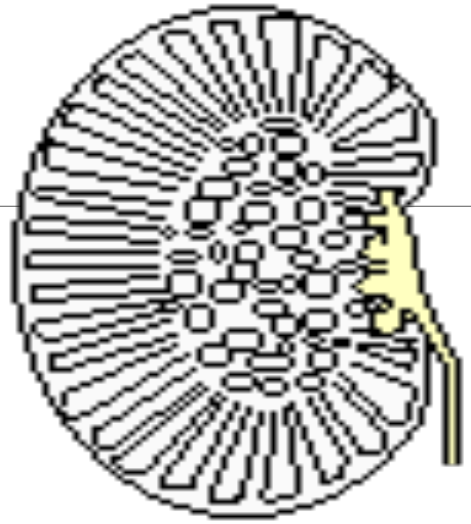
(↓tubular function).

- **renal failure over 5-10-year**
- **A positive family history and unexplained chronic renal failure in young patients should lead to suspicion of medullary cystic disease.**

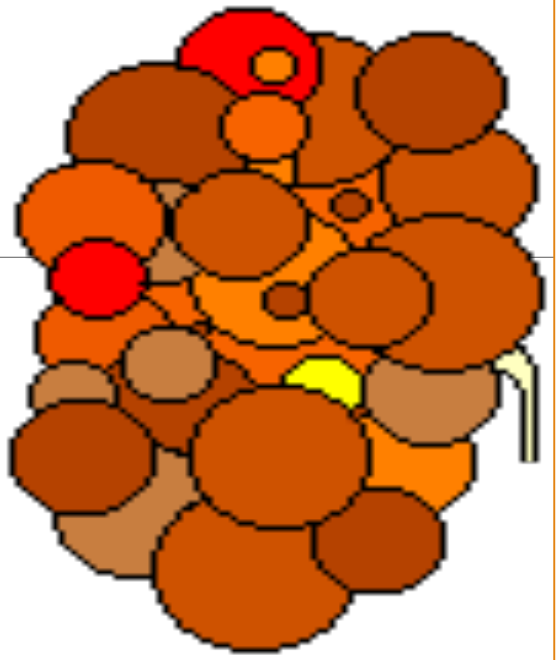
Kidney Cysts

No cysts

Simple cysts



Recessive polycystic



Dominant polycystic

Hydronephrosis is not cysts

"Dysplasia"

Medullary sponge

Medullary uremic

Dialysis cystic