G.U.S.
Pathology

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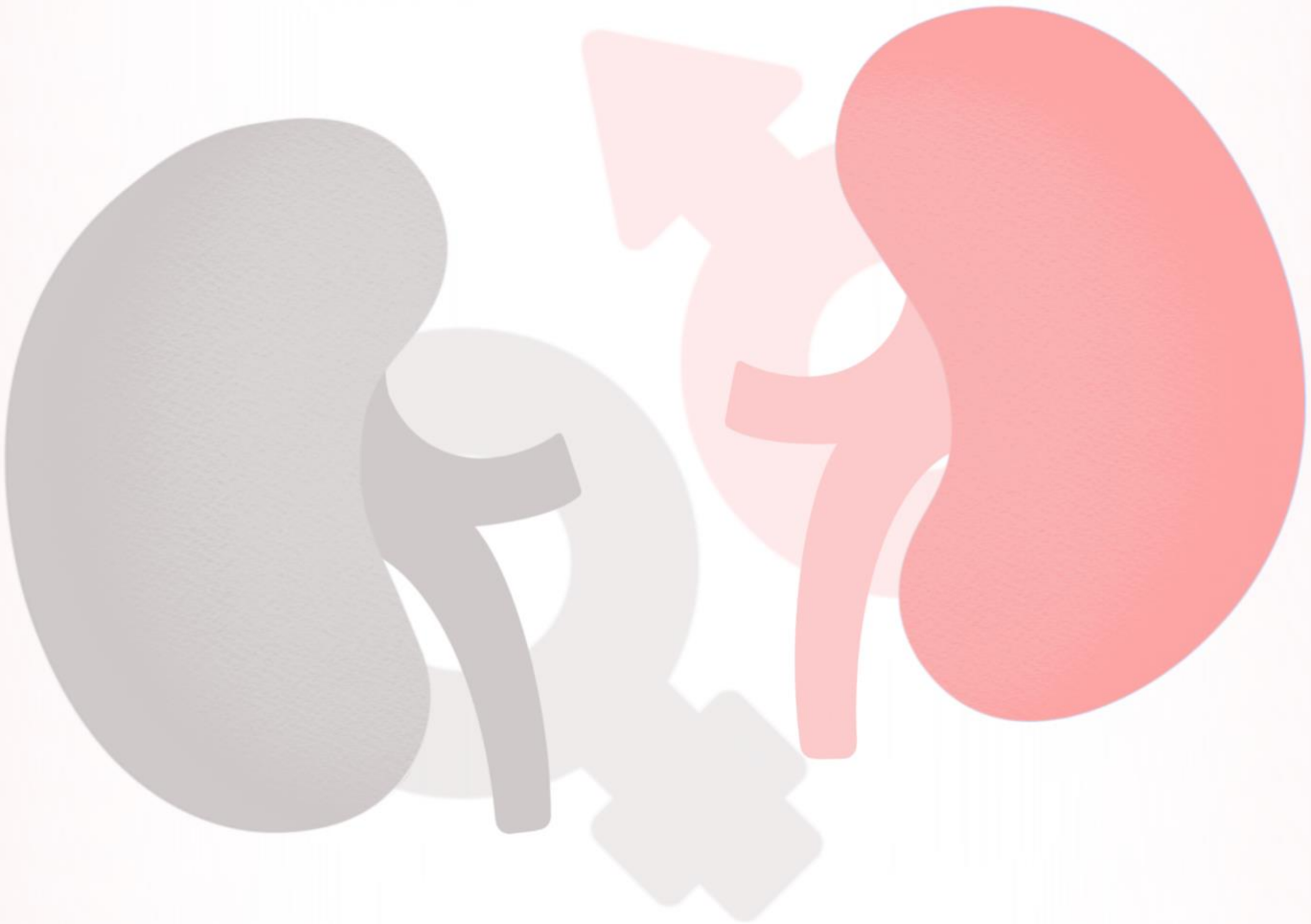

Sheet: Cystic diseases of the kidney

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In this lecture we are going to talk about cystic diseases of the kidney .

A cyst is a fluid-filled space and we have different types of cysts that may involve the kidneys , they range from completely innocent cysts with no clinical significance to others that are inherited and might lead to renal failure ; thus threatening the patient's life

So we have 5 major types of cysts are :-

1-Simple Cysts

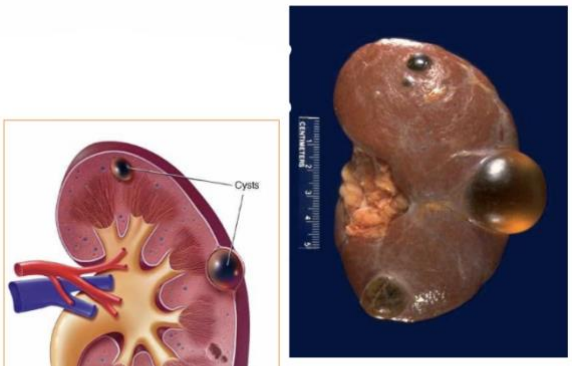
2-Dialysis-associated acquired cysts

3-Autosomal Dominant (Adult) Polycystic Kidney Disease

4-Autosomal Recessive (Childhood) Polycystic Kidney Disease

5-Medullary Cystic Disease

Simple Renal Cysts



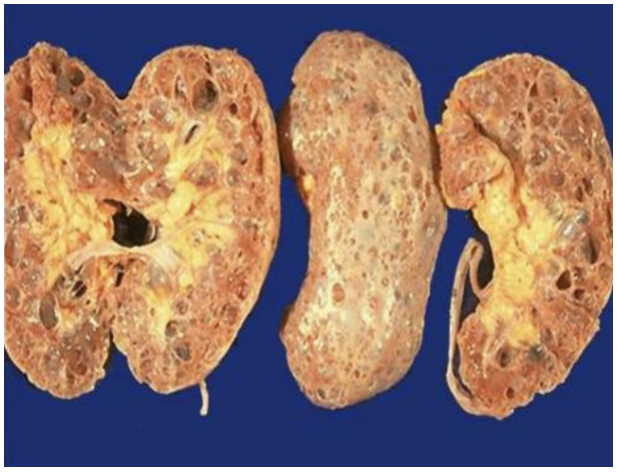
Multiple or single lesions , 1-5 cm in diameter , filled with clear fluid , confined to the **cortex.**

No clinical significance (hence given the name simple) , usually discovered incidentally or because of hemorrhage and pain

Importance: to differentiate from kidney tumors which are more serious.

Cysts Associated With Chronic Dialysis

Patients with renal failure who have prolonged dialysis , so they are acquired cysts.



They are numerous in number (as you can see from the picture) and involve **both cortex and medulla** .

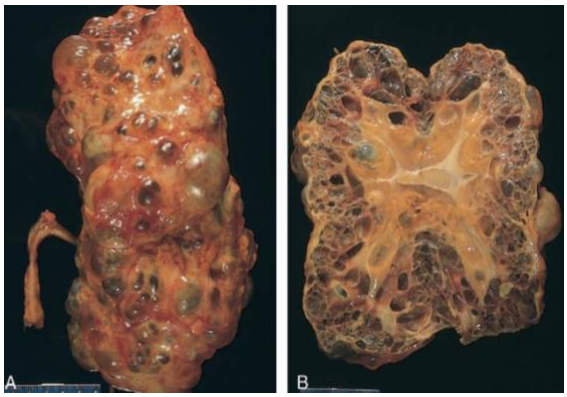
Complications: hematuria (due to hemorrhage inside of these cysts) ; flank pain , increased risk of renal carcinomas (100 times greater than in the general population).

How these cysts develop ??

This is the result of chronic inflammation and irritation that develops within the atrophic renal parenchyma (or degenerated renal parenchyma) ; this irritation and chronic inflammation might lead to cell division and with cell division cells might acquire additional mutations and later on they might go on or transform into malignant cells.

Autosomal Dominant (Adult) Polycystic Kidney Disease

Multiple bilateral cysts, these cysts replace the renal parenchyma and eventually destroy all of renal function. The size as well as the weight (it may reach to 1000-1500 gm) of the kidney are larger than normal. Cysts can be found both in the **renal cortex and renal medulla.**



The name adult comes from the fact that the symptoms and manifestations of this condition do not appear to be evident or obvious until the patient reaches adulthood.

The Incidence is around (1: 500-2000) persons .

This condition occur in 10% of chronic renal failure patients .

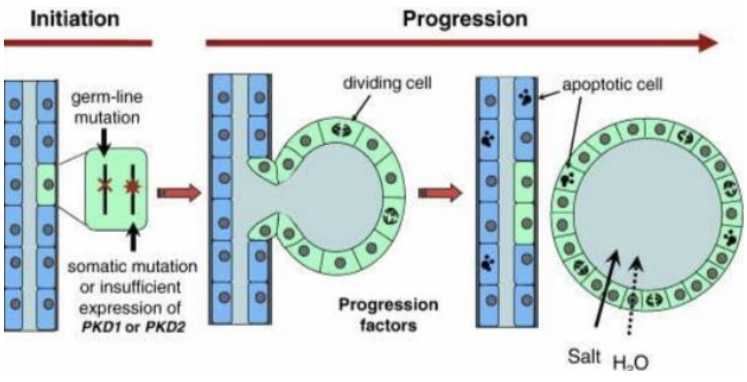
Inheritance of one of 2 autosomal dominant genes:

(1)- PKD1 (Polycystic kidney disease 1): 85-90% (encodes polycystin-1) (VIP)

(2)- PKD2 (Polycystic kidney disease 2): 10-15% (encodes polycystin- 2).

What happens in this disease is that the mutation that is present in either PKD1 or PKD2 leads to abnormal and progressive cell division within the cells of the renal tubules , with this continuous abnormal division there will be the formation of these multiple cysts which will eventually leave the original renal tubules and form these cystic spaces. This will also happen in the remaining renal tubules as well.

(Look at the figure)



Clinical presentation : (asymptomatic until the 4th decade)

Symptoms: flank pain, heavy dragging sensation (because of the large weight of the kidney) , abdominal mass, hemorrhage (due to rupture of these cysts) , obstruction (due to either of hemorrhage within the cysts or obstruction by stones) , Intermittent gross hematuria

Complications

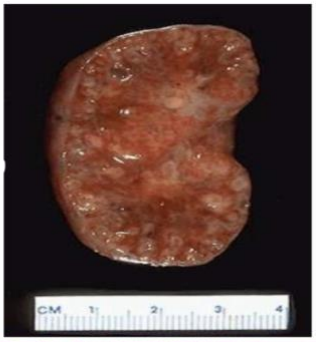
1- hypertension (75%)

2- urinary infection , frequent complication ; because inside these cysts we can have infections .

3- vascular aneurysms of circle of Willis (10% -30%) —> (rupture of these aneurysms can result in subarachnoid hemorrhage).

4- renal failure at age of 50 , MOST FREQUENT COMPLICATION (develops around 10 years following the initiation of symptoms at the age of 50)

Autosomal Recessive (Childhood) Polycystic Kidney Disease



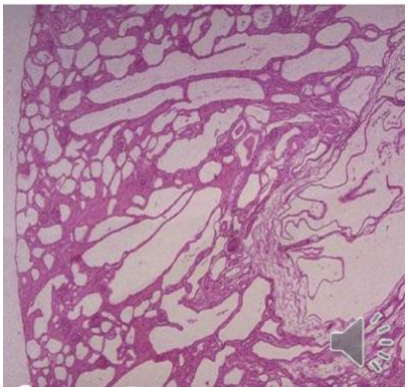
Autosomal recessive

1:20,000 live births , Presents early in life

Types: perinatal, neonatal, infantile, and juvenile.

Associated with liver cysts

Mutations in PKHD1 gene coding for fibrocystin (important in the development of renal tubules) , fibrocystin may be involved in the function of cilia in tubular epithelial cells .



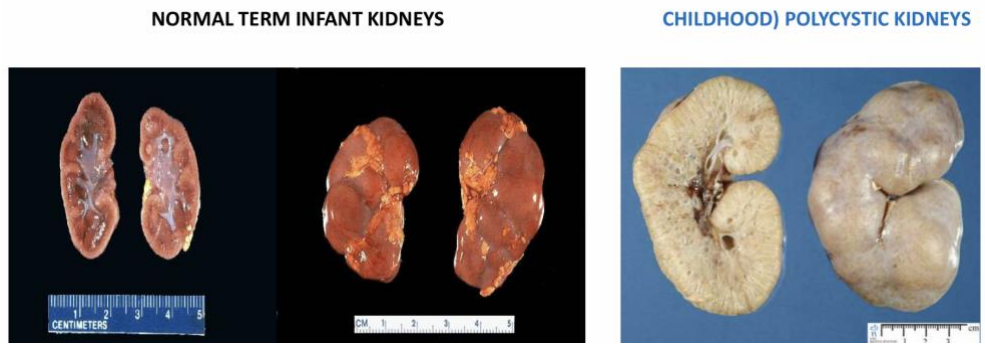
In the picture above , we can see a kidney that is involved by the childhood polycystic kidney disease which involved both the cortex and the medulla .

Also the diseased kidney under the microscope looks like this as → we can see the renal parenchyma is replaced by white spaces , these white spaces are the cystic structures that are developing from the abnormal renal tubules .

Normal vs childhood polycystic kidneys

Note :-

- The size of the kidneys is larger than normal
- The renal parenchyma of the diseased kidney is full of these cysts

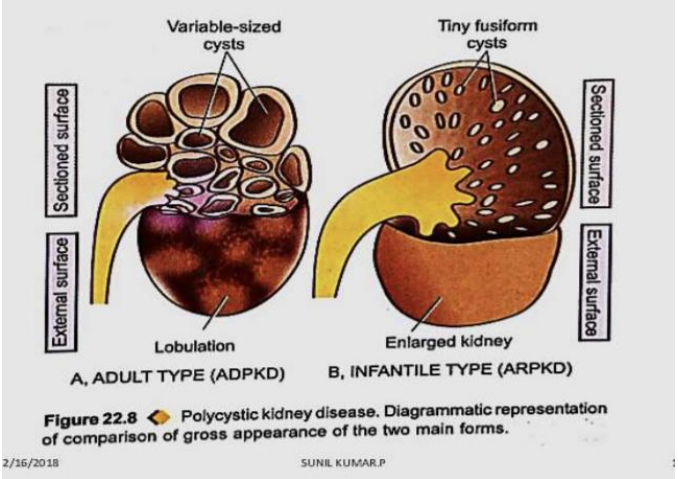


Adult vs childhood polycystic kidney disease

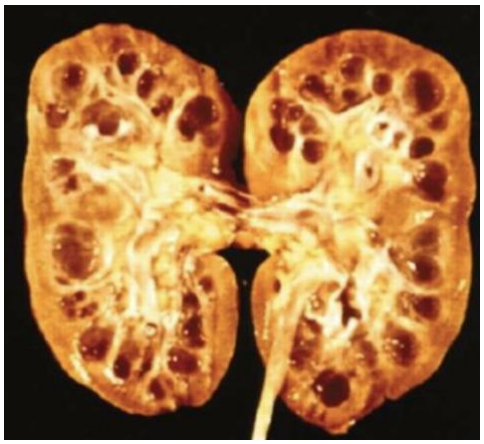
Adult type of kidney disease: variable-sized cysts, large cysts, Autosomal dominant

Infantile type of kidney disease: autosomal recessive, tiny fusiform cysts

<u>Both of these types can lead to progressive and chronic renal failure .</u>



Medullary Cystic Disease



2 major types:

1- Medullary sponge kidney: common and innocent condition.

2- Nephronophthisis-medullary cystic disease complex (or medullary uremic type): they are the least common and worst variant because they are almost always associated with renal dysfunction and progression towards renal failure.

Both types usually begin in childhood. And the Cysts are present at the cortico-medullary junction .

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

o Clinical features:-

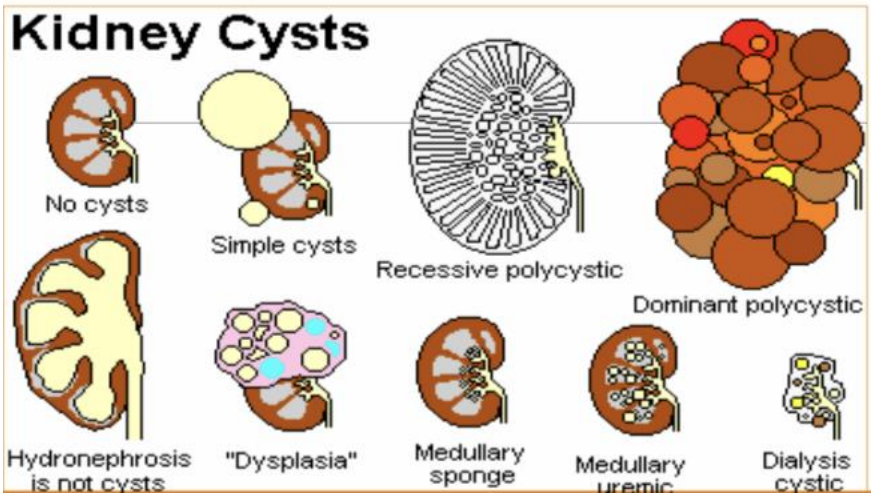
- Polyuria and polydipsia in a child who has some kind of renal impairment (↓tubular function).

- Renal failure over 5-10-year

- A positive family history and unexplained chronic renal failure in young patients should lead to a high clinical suspicion of medullary cystic disease.

- It's important to be recognized because it usually leads to a progressive renal failure during childhood or early adulthood.

In summary


Kidney Cysts
No cysts
Simple cysts
Recessive polycystic
Dominant polycystic
Hydronephrosis is not cysts
"Dysplasia"
Medullary sponge
Medullary uremic
Dialysis cystic