SNS COLLEGE OF ALLIED HEALTH SCIENCES

SNS Kalvi Nagar, Coimbatore - 35

Affiliated to Dr MGR Medical University, Chennai

DEPARTMENT OF CARDIOPULMONARY PERFUSION CARE TECHNOLOGY

COURSE NAME: NEPHROLOGY

III YEAR

UNIT : PATHOLOGY OF KIDNEY

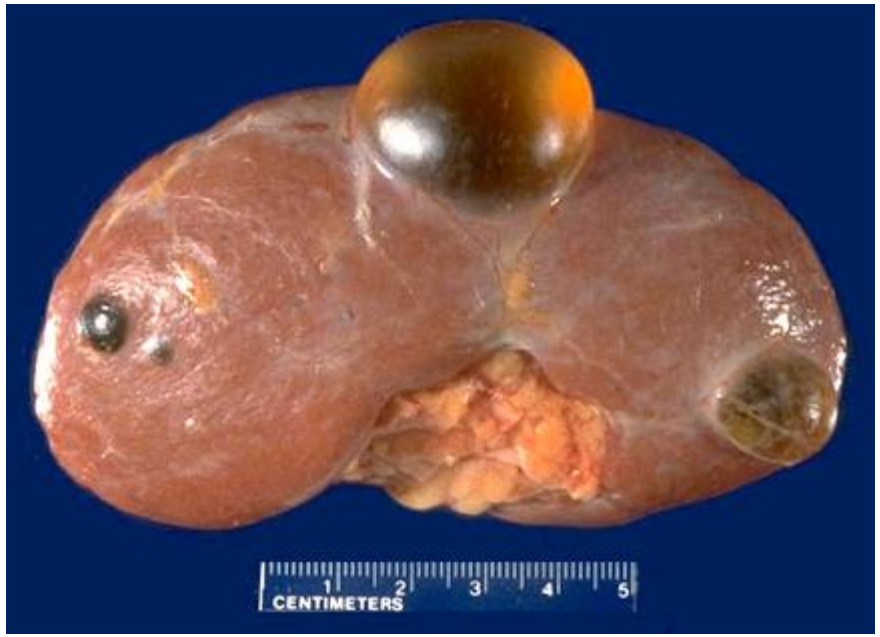
TOPIC : CONGENITAL KIDNEY DISEASE

Congenital Kidney Disease

- Congenital kidney disease has been characterized into different types based on the differences in shape, size, location, cysts and absents of kidney.

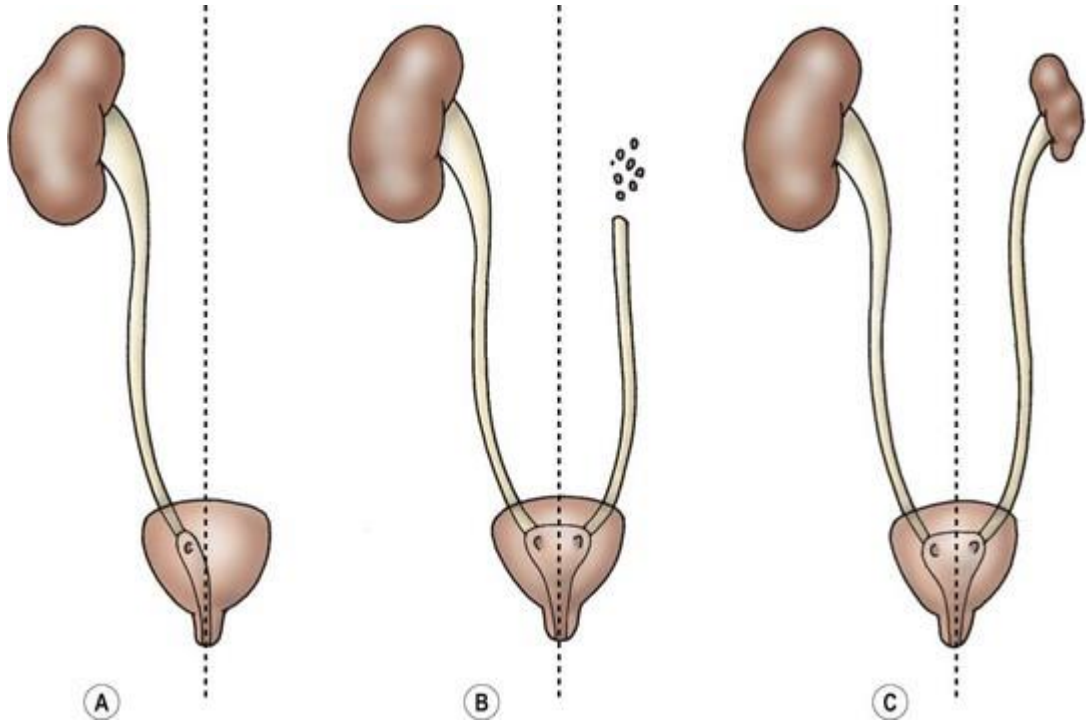
They different forms are,

- **Agenesis**
- **Hypoplasia**
- **Ectopic**
- **Horseshoe**



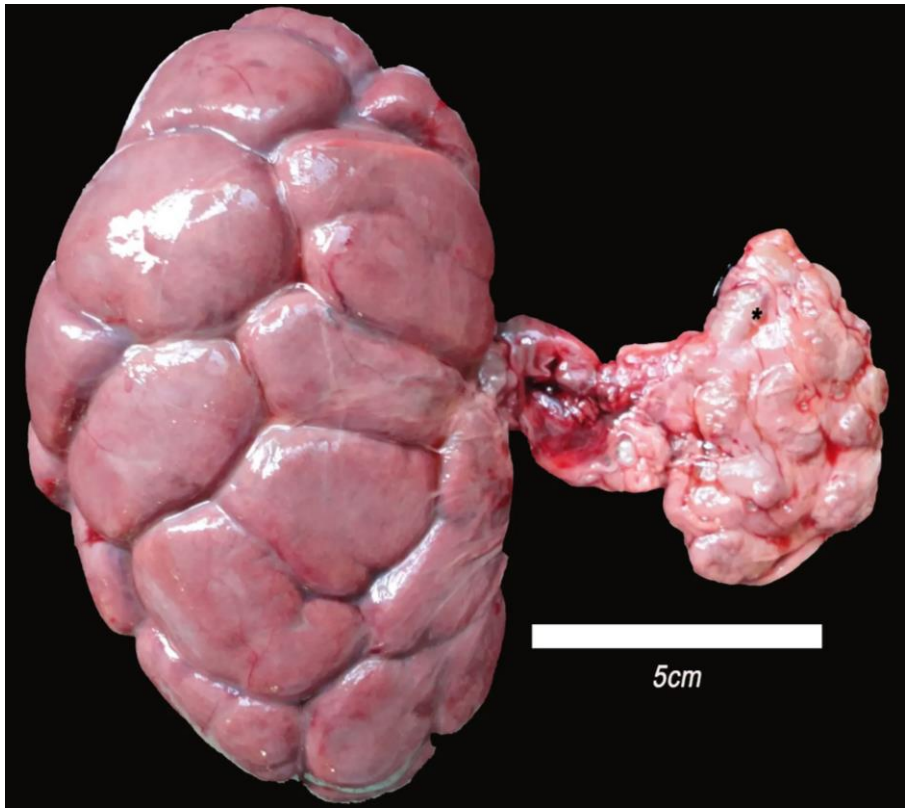
Agenesis of the kidney(absence)

- Bilateral agenesis is incompatible with life, seen in stillborn.
- Unilateral type is uncommon.
- The opposite kidney is usually enlarged as a result of compensatory hypertrophy



Hypoplasia(small size kidney)

- Is **failure of the kidney to develop** to a normal size.
- This anomaly may occur bilaterally, resulting in renal failure and early childhood death.
- Unilateral cases are more common.
- The opposite kidney is also enlarged due to compensatory hypertrophy.

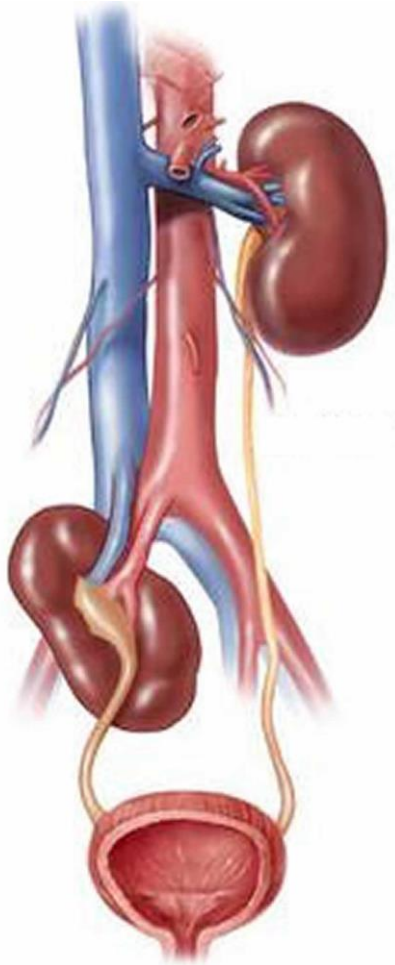


Ectopic Kidneys

- These kidneys **lie within the pelvis.**
- They are usually normal or slightly small in size.

Because of their abnormal location can cause :

- Kinking or tortuosity of the ureters may cause some obstruction to urinary outflow.
- Difficulty in labor in females.
- Misdiagnosis as pelvic tumors & abscesses.
- Have long renal artery that may have many complications like damage during surgery.

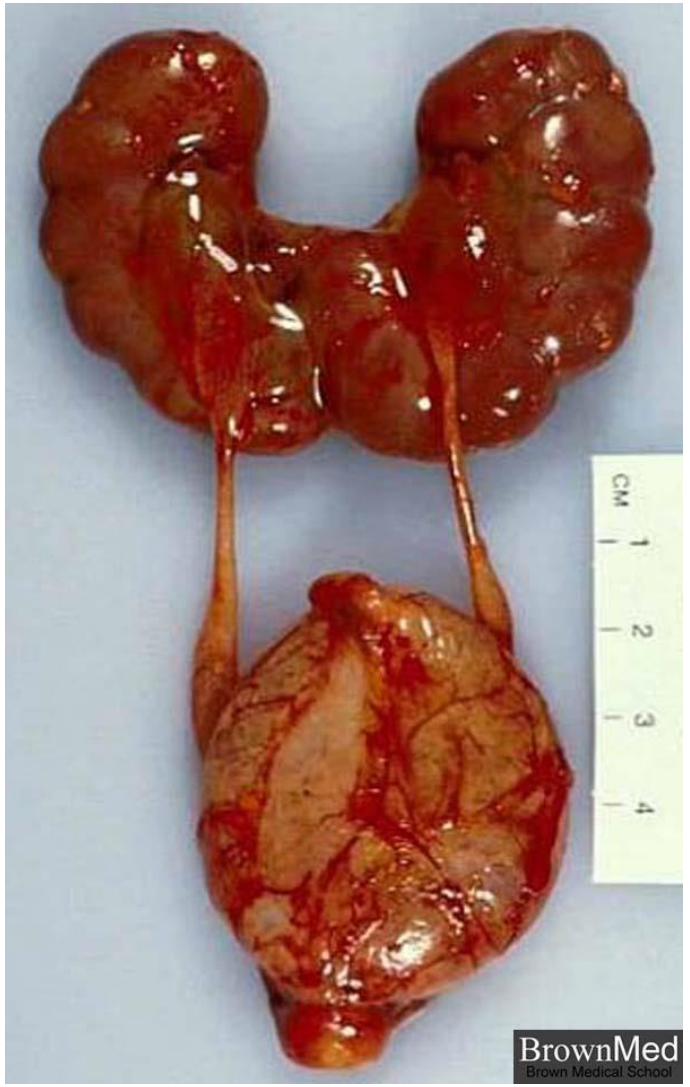


Horseshoe kidneys

Fusion of lower poles of the kidneys that is continuous across the midline anterior to the great vessels.

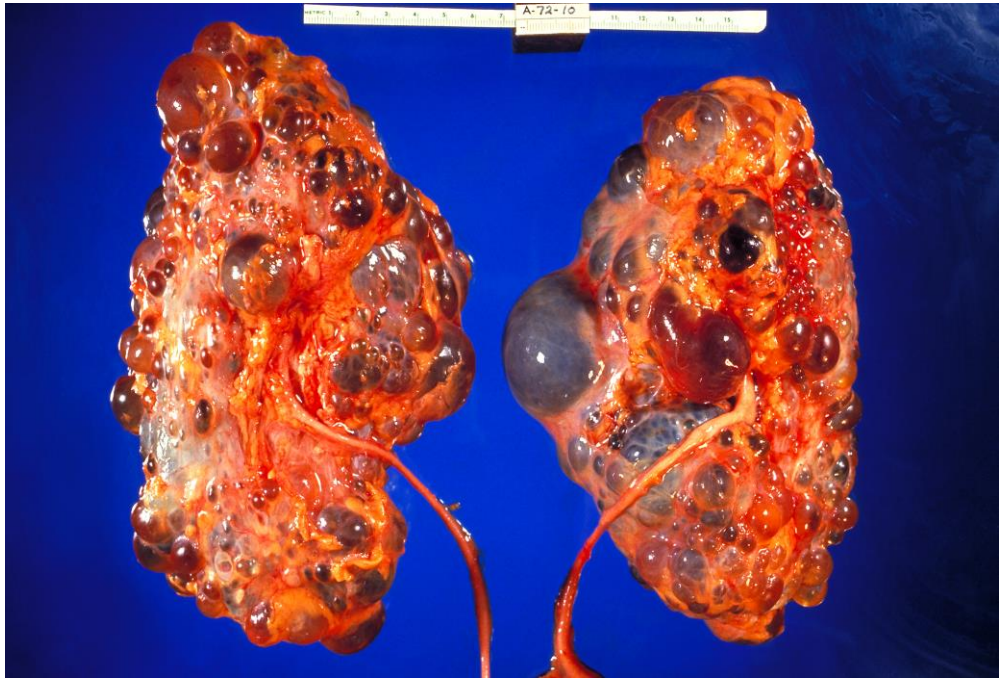
This may cause many complications:

- Partially obstructed the ureters, which result in hydronephrosis.
- Recurrent UTI.
- Stone formation.



Cystic disease of kidney

- They are **heterogeneous group comprising hereditary, developmental and acquired disorders.**



- They are important for several reasons
- They are reasonably common and often represent diagnostic problems for clinicians, radiologists and pathologists.
- Some forms are major causes of chronic renal failure.
- They can occasionally be confused with malignant tumors.

Classification of Cystic diseases

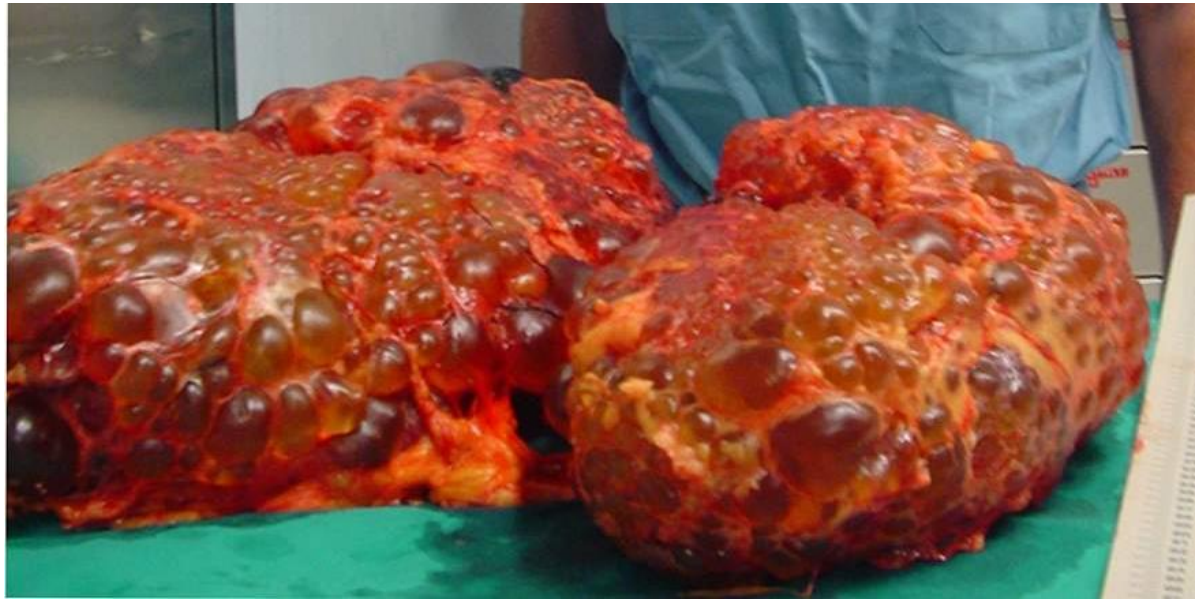
- **POLYCYSTIC KIDNEY**
  Autosomal DOMINANT (ADULTS)
  Autosomal RECESSIVE (CHILDREN)

- **ACQUIRED KIDNEY DISEASE**

- **SIMPLE KIDNEY DISEASE**

- **DIALYSIS-ASSOCIATED CYSTIC DISEASE**

- **PARASITIC CYSTS** (e.g. hydatid cyst).



Autosomal Dominant Polycystic Kidney Disease

- Multiple expanding cysts of both kidneys that ultimately destroy the intervening parenchyma.
- It affects roughly 1 of every 400 to 1000 live births

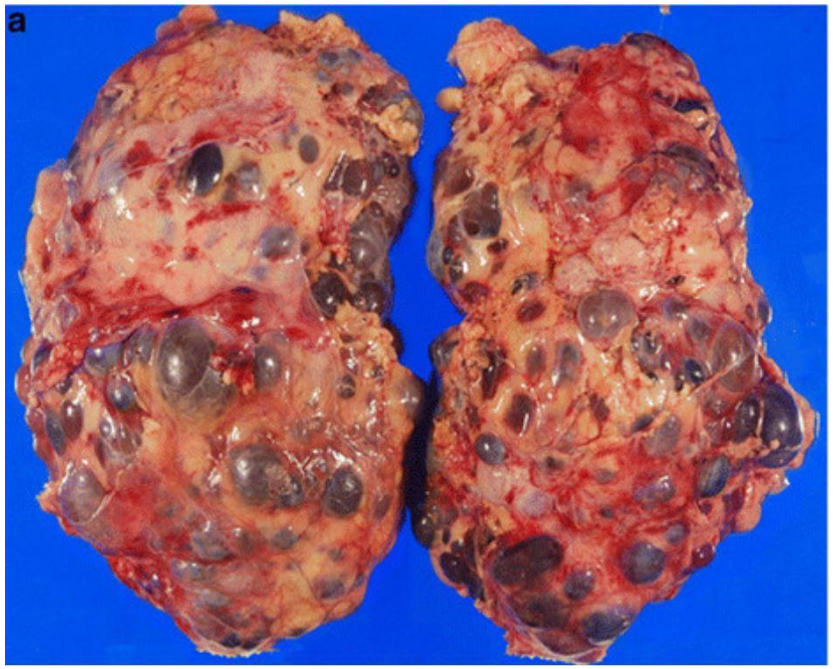
It can be caused by inheritance of at least two Autosomal dominant genes of high penetrance:

- PKD 1, present on chromosome 16, mutant in 90% of cases.
- PKD 2, present on chromosome 4, mutant in10%of cases

Autosomal Dominant Polycystic Kidney Disease

Gross:

- Large kidney (4 kg) of each kidney, & present as abdominal mass.
- Mass of cysts of varying sizes up to 3 to 4 cm in diameter without intervening parenchyma.
- Cysts are filled with fluid (clear, hemorrhagic).



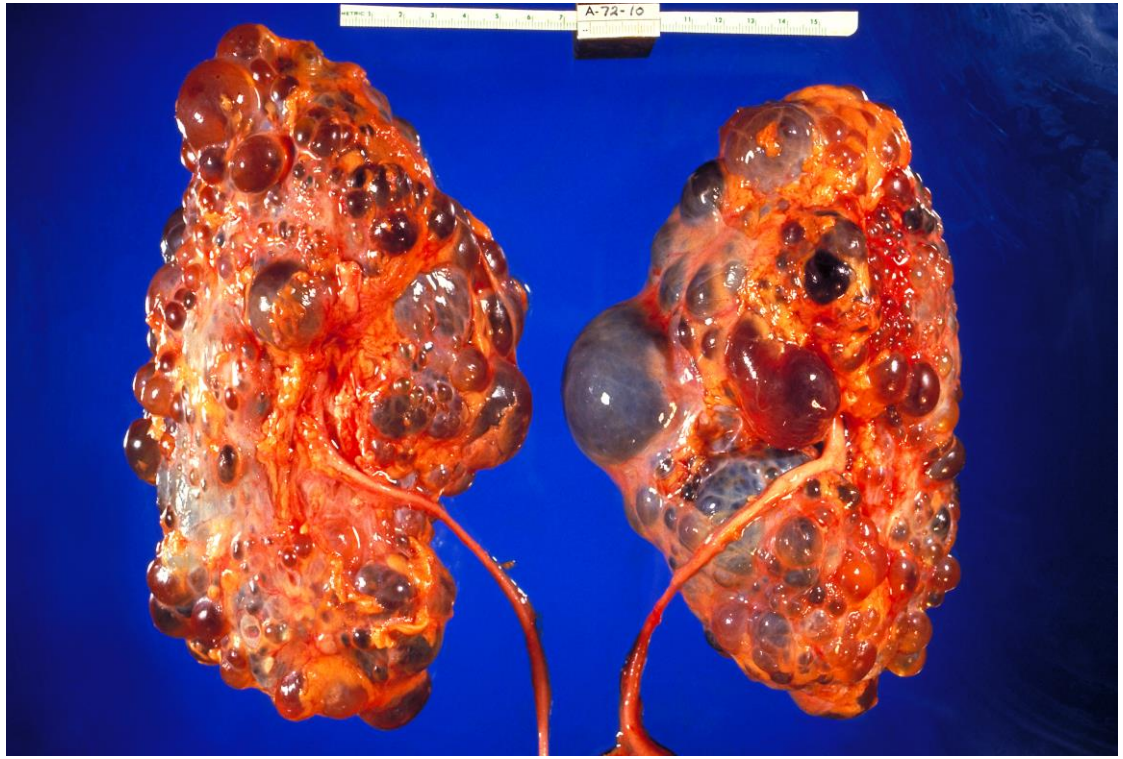
Microscope:

- Cysts arise from tubules or collecting ducts.
- Often have atrophic lining.
- Superimposed infection is common.

Autosomal Dominant Polycystic Kidney Disease

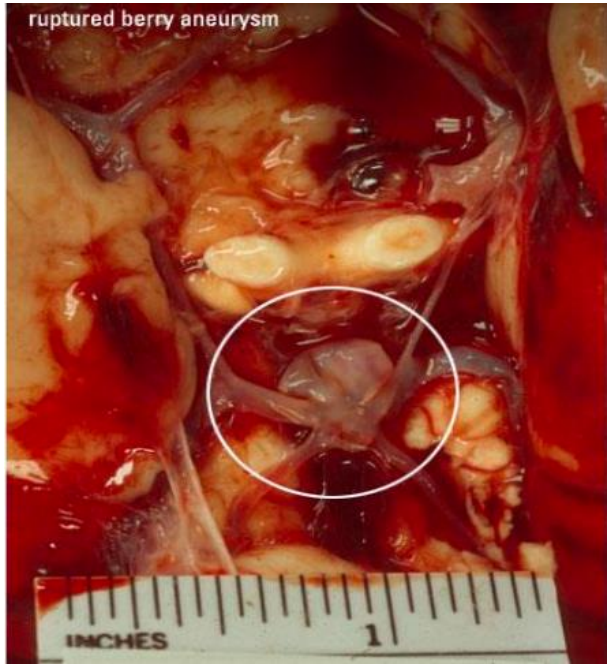
Clinical:

Many of these patients remain **asymptomatic** until about the 4th or 5th decade of life when renal insufficiency occurs because the cysts initially involve only portions of the nephrons, so renal function is retained



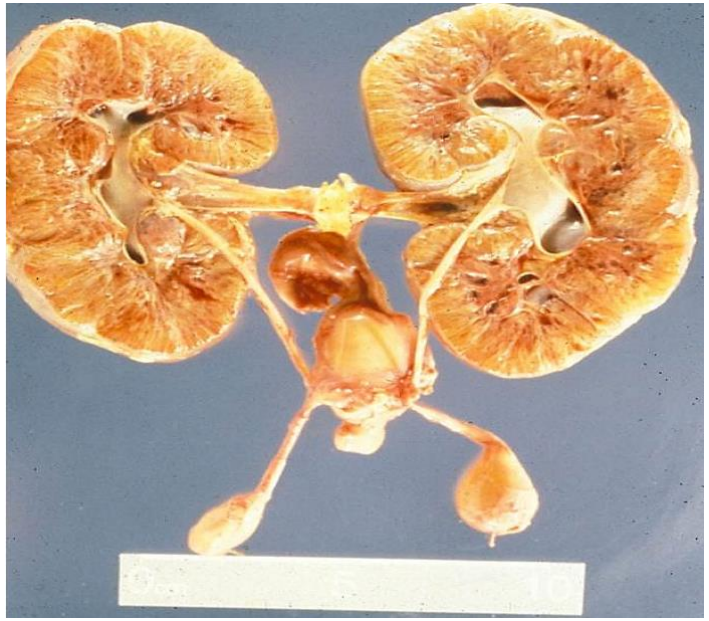
Autosomal Dominant Polycystic Kidney Disease

- Flank pain, hypertension, haematuria
- Progressive renal failure
- Large lesions are palpable
- 40% have cystic disease of the liver (most common), spleen, pancreas, brain
- **Berry aneurysms in circle of Willis** and can cause death
- 10% of patients due to subarachnoid haemorrhage.
- Death due to uraemia or hypertension
- 40% of patients die of coronary or hypertensive heart disease
- 25% of infection
- 15% of ruptured berry aneurysm or hypertensive intracranial haemorrhage.



Autosomal Recessive (Childhood) Polycystic Kidney Disease

- Is rare anomaly due to mutation of gene PKHD1 on chromosome 6
- Perinatal, neonatal, infantile and juvenile types are present
- The first 2 are the most common
- In all cases are associated with liver cysts (congenital liver cirrhosis).

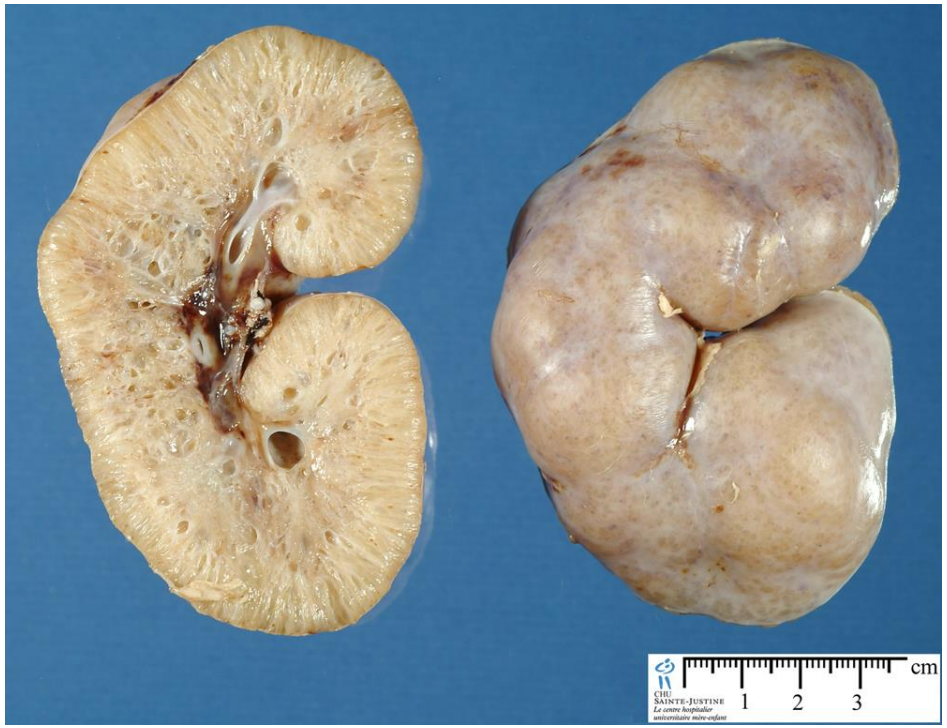


- **Gross:** bilateral Small cysts in the cortex & medulla (sponge like appearance).
- Elongated channels at right angles to the cortical surface.
- **Microscopic:** Uniform lining of cuboidal cells (Originate from collecting ducts).

Autosomal Recessive (Childhood) Polycystic Kidney Disease

Morphology:

- The kidneys are enlarged (bilateral) and have a smooth external surface.
- On cut section, numerous small cysts in the cortex and medulla give the kidney a sponge-like appearance.
- **Smooth, Small, Sponge kidneys**
- The cysts are dilated channels perpendicular to the corticomedullary junction.
- Cysts originate from collective tubules and are lined by uniform cuboidal cells
- In almost all cases, the liver has cysts with portal fibrosis as well as proliferation of portal bile ducts.

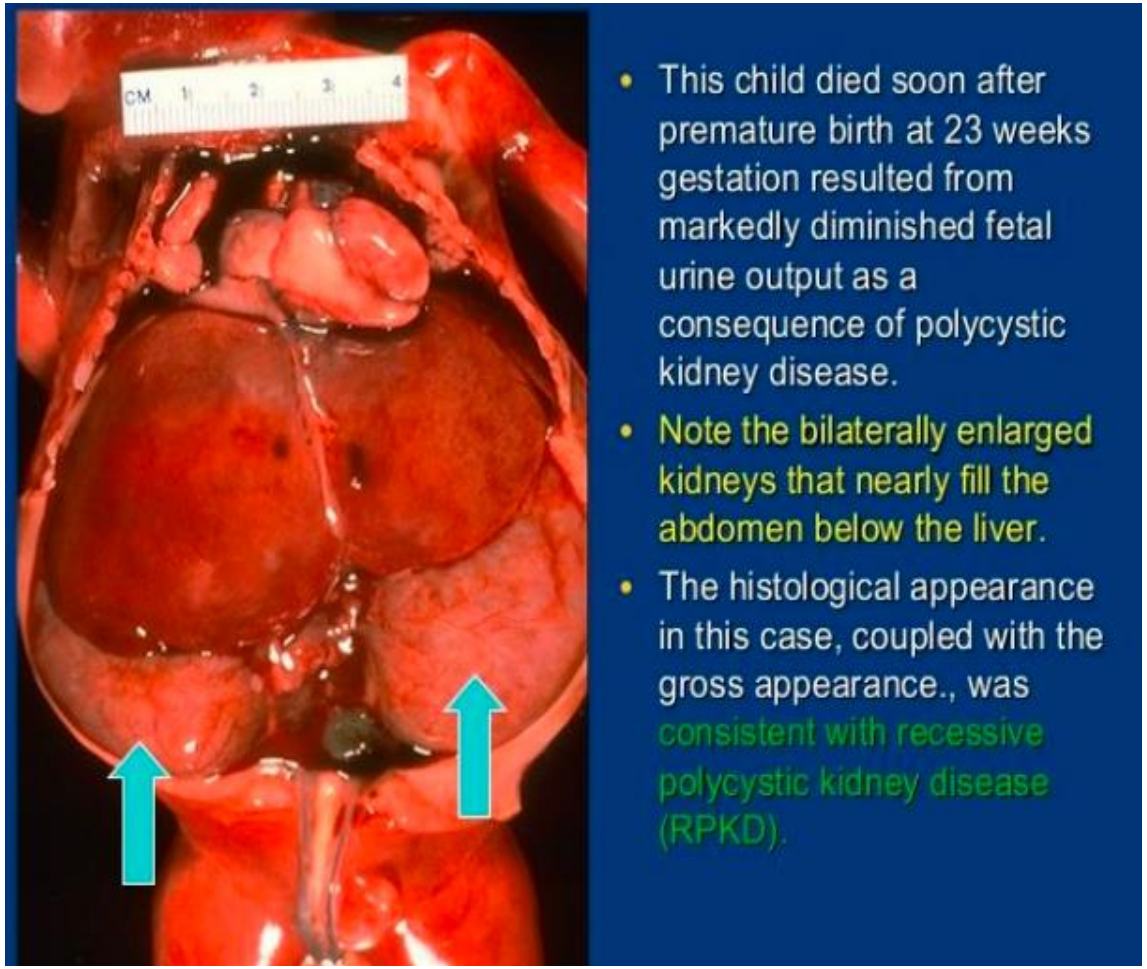


Autosomal Recessive (Childhood) Polycystic Kidney Disease

Clinical Features

- serious manifestations are usually present at birth, and the young infant might succumb rapidly to renal failure. Patients, who survive infancy, may develop congenital hepatic fibrosis.

- Liver: epithelium lined cysts and proliferation of bile ducts

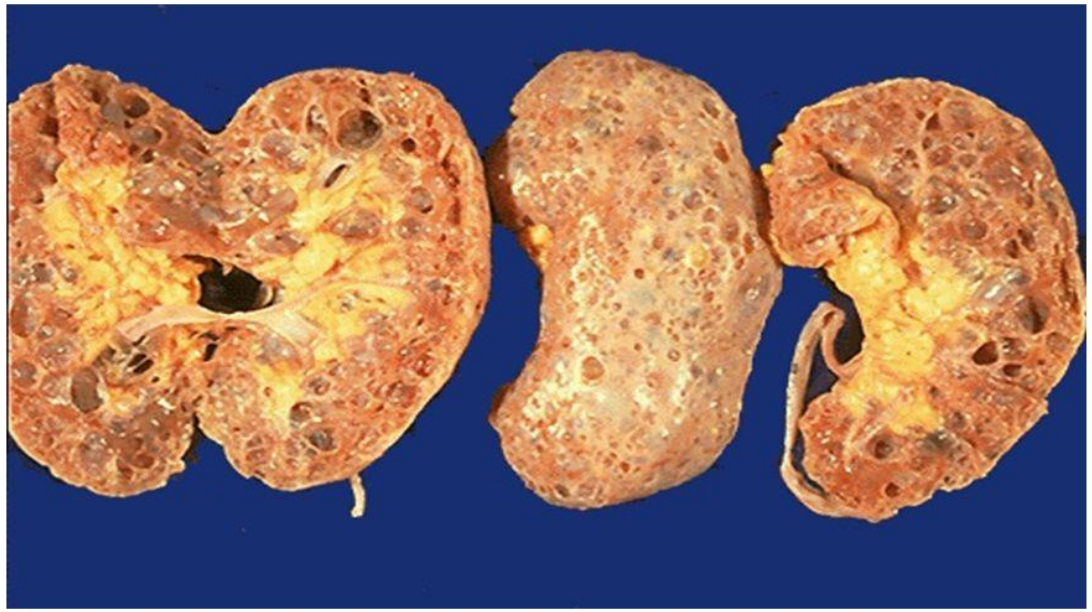


Acquired (Dialysis-Associated) Cystic Disease

- The kidneys of patients on chronic dialysis, sometimes exhibit numerous cortical and medullary cysts.

- **The cysts measure 0.5-2cm**, contain clear fluid, are lined by either hyperplastic or flattened tubular epithelium and often contain **calcium oxalate crystals.**

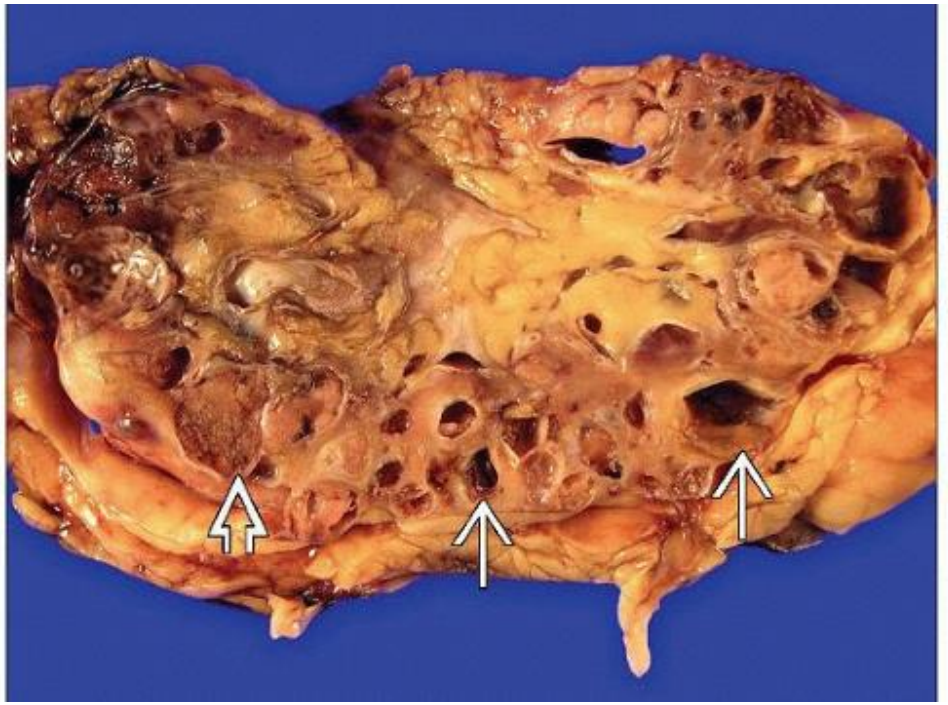
Cystic change associated with chronic renal dialysis.



Acquired (Dialysis-Associated) Cystic Disease

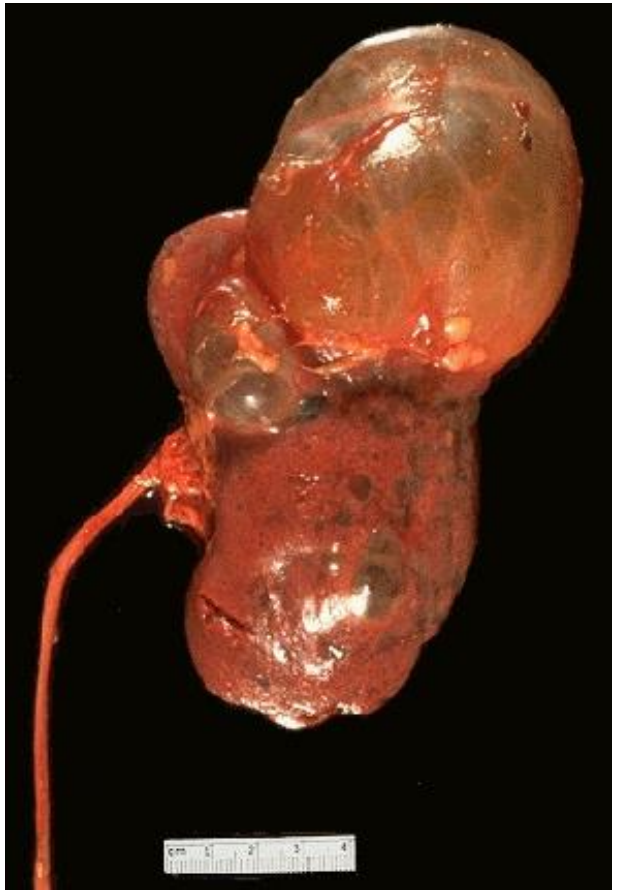
- **Cause:** They probably form as a result of tubular obstruction due to interstitial fibrosis or by oxalate crystals.

- Most are **asymptomatic**, but sometimes the bleeding inside the cysts cause **hematuria**. The most important complication is the development of **renal cell carcinoma** in the walls of these cysts in about 7% of patients during 10 years period.



Simple Cysts

- **Acquired,** Incidental, very common
- These occur as **single or multiple**, usually cortical.
- The size range from **1-10 cm** or more.
- They are translucent and filled with clear fluid.
- They are lined by a **single layer of cuboidal or flattened epithelium.**
- They are common postmortem findings. On occasion, hemorrhage into them and they may cause **sudden pain, and calcification** may be visible radiologically.
- The main importance of these cysts is in their differentiation from **kidney tumors.**



THANK YOU

REFERENCES:

- Text book of Pathology Harsh Mohan

- Textbook of Pathology for Allied Health Sciences, Ramadas Nayak