# CYSTIC DISEASE OF KIDNEY:

It Represent a heterogeneous group of hereditary, development And Acquired disorder by the presence of cysts in one or both kidney

## CLASSIFICATION:

① POLYCYSTIC KIDNEY DISEASE: → Adult
- AUTOSOMAL DOMINANT POLYSTIC D
- AUTOSOMAL RESSESSIVE POLYSTIC D
Infantile

② MEDULLARY CYSTIC DISEASE:
- Medullary sponge kidney
- Nephronophthisis.

③ Medullary cystic diseas.
④ Acquired cystic diseas.
⑤ Localized Renal cysts
⑥ Extraparenchymal Renal cysts

## AUTOSOMAL DOMINANT POLYSTIC KIDNEY:

→ ADPKD is a hereditary disorder characterized by multiple cysts in both kidney. → These cyst expand & destroy the intervening kidney parenchyma
↓
Renal failure.

PATHOGENESIS: ADPKD is a genetically heterogeneous disorder.

Associated with mutation in PKD1 And PKD2 gene

↓

ADPKD occur due mutation in Any one.

PKD – tumor suppressor gene.

⇒ PKD1 gene encodes a protein called polycystin 1 & PKD2 gene encodes polycystin – 2

**MECHANISM OF CYSTS FORMATION:**

Mutation in PKD1 And PKD2 gene

↓

Reduced production of polycystin 1, 2

↓

Defect in mechanosensing in tubular cilia

↓

Altered cell polarity

Disrupted Ca signaling

↓

↑ secretion of fluid

↑ cell proliferation

↓

Cyst formation

↓

Consequence

↓

Damage to the glomerular & vessels

Inflammation & fibrosis in the interstitium.

## PKD1 - Gene Mutations:

PKD1 gene encodes a cell membrane - Associated protein called polycystin

→ Germline mutation in the PKD1 gene:

PKD gene Require a second hit for the loss of 2nd allel of the gene to develop the disease.

→ Consequence of mutation

↓

Reduced production of polycystin 1

↓

cause defect in mechanosensing by tubular epithelial cell

↓

disrupt $Ca^{2+}$ channel signaling (essential for maintenance of epithelial cell)

↓

↑ cell proliferation, ↑ secretion of fluid

Altered cell polarity. → cyst formation

## MORPHOLOGY:

GROSS: Involves both kidney.

Enlarged kidney, Bosselated.

Cut section → numerous cysts of varying size.

Content → clear, serous, turbid, Red to brown.

## MICROSCOPY: cysts:

Number: Numerous cysts

Origin: arise at Any level of the nephron, show variable lining epithelia.

Lining: absorbed columnar epithelium.

Renal parenchyma show foci of interstitial fibrosis & tubular atrophy

C.F

→ flank pain, bilateral Abdominal mass, hypertension

Extrarenal Associated congenital Anomalies.

⇒ polycystic liver
⇒ Berry Aneurysms
⇒ Mitral valve prolapse.

## AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE

genetically different from Adult polycystic kidney disease.

- → perinatal
- → neonatal
- → infantile
- → juvenile

**PATHOGENESIS:** (Polycystic kidney hepatic disease)

Mutation in PKHD1 gene. cause ARPKD.

↳ codes for a protein called fibrocystin (FPC)

↓

seen in CD, TA loop of Henle.

Fibrocystin: seen in primary cilia of epithelial cell of Renal tubes & bile duct. it may be a cell surface Receptor involved in collecting duct & biliary differentiation

## MORPHOLOGY:

→ enlarged & smooth kidneys.

Cut section → Medulla & cortex Replaced by numerous small cysts.

cyst lead to be linear of Radiate from the Medulla to the cortex - sponge like appearance to the kidney

**MICRO**

shape: cyst is cylindrical.

lining: All the cyst are lined by cuboidal epithelial cells Reflecting their origin from the collecting ducts.

C.F → large, cystic kidney at Birth.
develop → congenital hepatic fibrosis.

**MEDULLARY DISEASE WITH CYSTS:**

two type affect Medulla

Nephronophthisis medullary cystic disease.

↳ begins in childhood
Renal dysfunction → chronic kidney disease.

Medullary sponge kidney. → cystic haemolysis.

MORPHOLOGY → small And contracted kidney.

MICRO → cyst are lined by flattened or cuboidal epithelium.
tubular Atrophy of fibrosis.

C.F → polyuria & polydipsia