

CYSTIC DISEASE OF KIDNEY:

It Represent a heterogeneous group of hereditary, development and Acquired disorders by the presence of cysts in one or both kidney.

CLASSIFICATION:

① POLYCYSTIC KIDNEY DISEASE: → Adult

- AUTOSOMAL DOMINANT POLYCYSTIC D

- AUTOSOMAL RECESSIVE POLYCYSTIC D

Child

② MEDULLARY CYSTIC DISEASE:

- Medullary sponge kidney

- Nephrocalcinosis

③ Medullary cystic disease.

④ Acquired cystic disease.

⑤ Lobar renal cysts.

⑥ Extraparenchymal renal cysts

AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY:

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

↓
Renal failure

PATHIOGENESIS: 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 cells

↓
Dysregulation of
signaling

↓
Altered cell
polarity

↑ cell proliferation

↑ secretion of
fluid

↓
cyst formation

↓
Damage to the
glomerulus & vessels

consequence

↓
Inflammation of
fibrosis in the
interstitium

PKD1 - Gene Mutation:

PKD1 gene encodes a cell membrane - Associated protein called polycystin 1

→ Germline mutation in the PKD1 gene:

PKD gene requires a second hit for the loss of 2nd allele of the gene to develop the disease.

→ Consequences 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, Bosellated

Cut section → numerous cysts of varying size

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

MICROSCOPY: cysts.

Number: Numerous cysts.

Origin: arise at Any level of the nephron, show variable morphology, lined by lining epithelium.

Lining: composed of columnar epithelium.

Renal parenchyma show foci of interstitial fibrosis & nephropathy

C.F

→ flank pain, bilateral Abdominal mass, hypertension

Extrarenal Associated congenital Anomalies.

⇒ polygenic liver

⇒ Biliary Aneurysms

⇒ Mitral valve prolapse

AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE

genetically different from Adult polycystic kidney disease.

→ perinatal

→ neonatal

→ infantile

→ juvenile

PATHOGENESIS:

(Polycystic kidney hereditary disease)

Mutation in PKHD1 gene cause ARPKD.

↳ codes for a protein called fibrocystin (FPC)

↓

seen in CD, TA loop of Herli.

seen in

Fibrocystin: primary cilia of epithelial cell of renal tubules & bile duct, it may be a cell surface

Receptor involved in collecting duct & biliary differentiation

MORPHOLOGY:

→ enlarged of smooth apparatus.

Cat section → Medulla of cortex replaced by numerous small cysts.

cyst tend to be linear & Radiate from the Medulla.
to the outer cortex - sponge like appearance to the kidney

MICRO

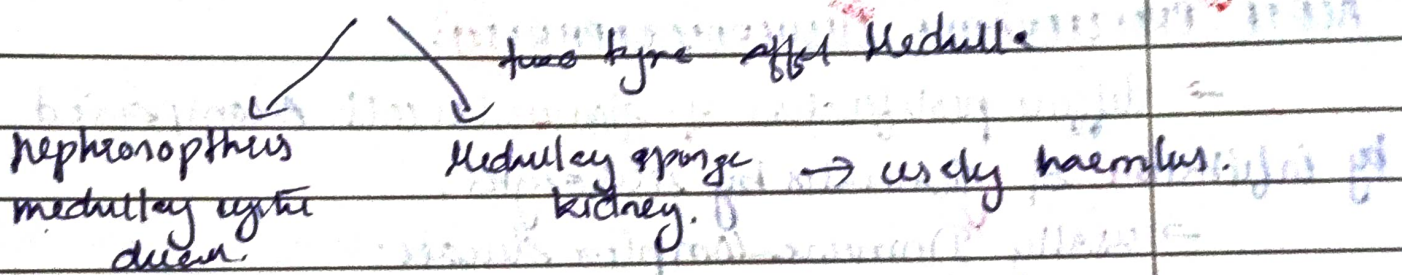
shape: cyst is cylindrical.

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

C.F → large, cystic kidney at Birth.

development → congenital hepatic fibrosis.

HEPATIC DISEASE WITH CYSTS:



↳ begins in childhood

Renal dysfunction → chronic kidney disease

MORPHOLOGY → small and contracted kidney.

MICRO → cyst are lined by flattened or cuboidal epithelium.
tubular atrophy & fibrosis.

C.F → polycystia & polydipsia