

# Polycystic kidney disease

## MCQs

- Autosomal dominant polycystic kidney disease type 1 is associated with a genedefect in:

- A. Chromosome 4
- B. Chromosome 8
- C. Chromosome 12
- D. Chromosome 16**
- E. Chromosome 20

ADPKD type 1 = chromosome 16 = 85% of cases

- A 26-year-old man with loin pain and haematuria is found to have autosomal dominant polycystic kidney disease. A defect in which one of the following genes is likely to be responsible?

- A. Fibrillin-2 gene
- B. Polycystin gene
- C. Fibrillin-1 gene
- D. Von Hippel-Lindau gene
- E. PKD1 gene**

- 24-year-old man who has a sister with polycystic kidney disease asks his GP if he could be screened for the disease. What is the most appropriate screening test?

- A. PKD1 gene testing
- B. CT abdomen
- C. Urine microscopy
- D. Ultrasound abdomen**
- E. Anti-polycystin 1 antibodies levels

Ultrasound is the screening test for adult polycystic kidney disease

- Autosomal dominant polycystic kidney disease type 2 is associated with a genedefect in:

- A. Chromosome 4**
- B. Chromosome 8
- C. Chromosome 12
- D. Chromosome 16
- E. Chromosome 20

ADPKD type 2 = chromosome 4 = 15% of cases

- Causes of nephrotic syndrome include all the following Except.

- a) SLE
- b) DM
- c) Amyloidosis
- d) Membranous glomerulonephritis
- e) **Autosomal-dominant polycystic kidney disease**

- what is the most common site for extra-renal cysts in a patient with autosomal dominant polycystic kidney disease (ADPKD)?

- A. Pancreas
- B. Brain
- C. Liver**
- D. Spleen
- E. Thyroid

**Most common location of extra-renal cysts in ADPKD is liver**

**Liver cysts are present in 70% of patients with ADPKD. Around 8% of patients have berry aneurysms**

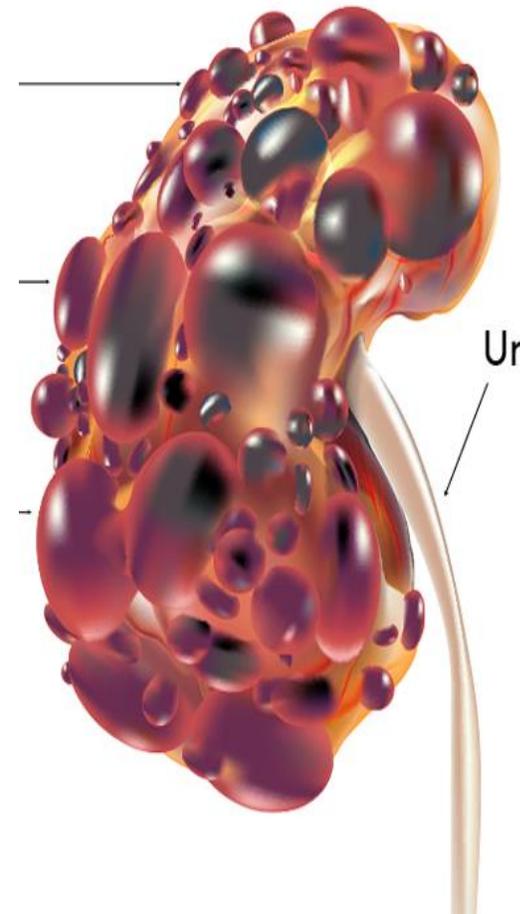
- one of the following statements is true regarding autosomal recessive polycystic kidney disease?

- A. Onset is typically in the third decade
- B. Liver involvement is rare
- C. Is due to a defect on chromosome 16
- D. More common than autosomal dominant polycystic kidney disease
- E. May be diagnosed on prenatal ultrasound**

# Mini-OSCE

Q5: which one of the following is NOT among the signs of this condition:

- Hematuria
- **Nephrotic syndrome**
- Loin pain
- Hypertension
- Renal failure



# Q1

Q1:whats your Dx?

Polycystic kidney disease

Q2: investigation :

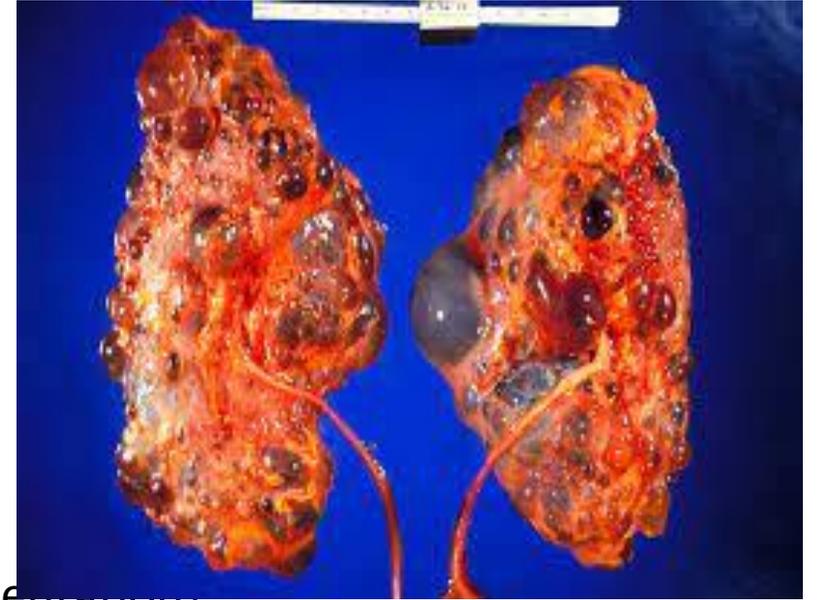
Ultrasound

Q3:pattern of inheritance :

Autosomal Dominant



**Station 4:** A patient presented to the ER with hematuria and ..... he died. This was found on autopsy.



1) What is your diagnosis?  
Polycystic kidney disease

2) What is the inheritance trait of this disease?  
Autosomal dominant

3) What is the investigation that should have been done on presentation?  
Renal ultrasound