DEFINITIONS

1. **Polycystic kidney disease** is characterised by the development and growth of cysts in the kidneys. Cysts may also occur in other organs, particularly the liver, but also in the pancreas, ovaries, gastrointestinal tract, spleen, lungs and the vascular tree.

2. There are two main forms -

   2.1 the commoner form is known as **adult polycystic kidney disease** or **autosomal dominant polycystic kidney disease**

   2.2 the other form is known as **infantile (or childhood) polycystic kidney disease** or **autosomal recessive polycystic kidney disease**.

ADULT POLYCYSTIC KIDNEY DISEASE

CLINICAL MANIFESTATIONS

3. This form of polycystic kidney disease may present at any time during life but, most frequently, it becomes symptomatic during the fourth to fifth decade with a gradual onset of renal failure.

4. The commonest presenting symptoms are flank pain and haematuria.

5. Other presenting symptoms may be headaches, gastrointestinal complaints, increase in abdominal girth, nocturia, frequency of micturition and polyuria. Some patients present with renal complications such as urinary tract infections, hypertension, renal calculi, or acute or chronic renal insufficiency.

6. Normal renal function is usually retained until middle life. The condition progresses steadily as the cysts increase in size. End-stage renal failure rarely occurs before 40 years of age. Approximately 50 per cent of patients have well preserved renal function at 70 years of age.

AETIOLOGY

7. **Adult polycystic kidney disease** is a disease inherited in an autosomal dominant pattern. The gene is carried on chromosome 16 and it has been estimated that complete penetrance of the gene will occur by the time the individual is aged 90. It is extremely common, occurring at a rate of 1 to 2 per 1,000 population.

INFANTILE (OR CHILDHOOD) POLYCYSTIC KIDNEY DISEASE

CLINICAL MANIFESTATIONS

8. **Infantile (or childhood) polycystic kidney disease** usually presents in the neonatal period and is rarely seen in older children and adults. It usually progresses to end-stage chronic renal failure before adolescence.

AETIOLOGY

9. This form of polycystic kidney disease is a rare disorder which is inherited in an autosomal recessive pattern.

CONCLUSION

10. **Polycystic kidney disease** exists in two forms, an infantile and an adult form. Both are inherited, being genetically determined.

REFERENCES

Gabow P A. Cystic disease of the Kidney. In: (Eds) Wyngaarden J B, Smith L H and Bennett J C. Cecil Textbook of Medicine. Philadelphia. W B Saunders Company. 19th Ed. 1992. p608-612.

Evans D B. Cystic and Congenital Abnormalities of the Kidneys. In: (Eds) Weatherall D J, Ledingham J G G and Warrell D A. Oxford Textbook of Medicine. 2nd Ed. 1987. Oxford. Oxford University Press. p18.164-18.165.

Rosai Juan. Ackerman's Surgical Pathology. 7th Ed. 1989. St Louis. C V Mosby Company. p857-858.

December 1992