

## Case 110 Congenital disease of both kidneys



Figure 110.1

The specimen shown in Fig. 110.1 of both kidneys, ureters, bladder and adjacent aorta was obtained at postmortem on a 40-year-old man in the days before much could be done for patients with this congenital condition.

### What is this abnormality called, and what is its embryological explanation?

Polycystic disease of the kidneys. It is believed to be due to failure of many of the tubules of the metanephros, which later develops into the kidney, to join with the metanephric duct, which gives rise to the calyces, the pelvis of the ureter and the ureter itself (Fig. 110.2).

There may be associated cysts in other viscera, particularly the liver (30%), lungs, spleen and pancreas. In addition, there is a strong association with intracranial

berry aneurysms and the danger of subarachnoid haemorrhage.

### What is its inheritance?

The condition is inherited as an autosomal dominant form, which presents usually in middle age. A less common autosomal recessive type presents in childhood, with renal failure. Genetically, the dominant form may result from a number of different gene mutations, the commonest being in the *PKD1* gene (chromosome 16p13.3).

### How may this condition present clinically?

To some extent, this may be classified according to the age of the patient. It is surprising that patients with this gross bilateral renal disease may sometimes live on, untreated, into their sixties or more:

- *Antenatal*: The renal masses may be detected on routine ultrasound.
- *At birth*: There may be obstructed labour due to the abdominal masses.

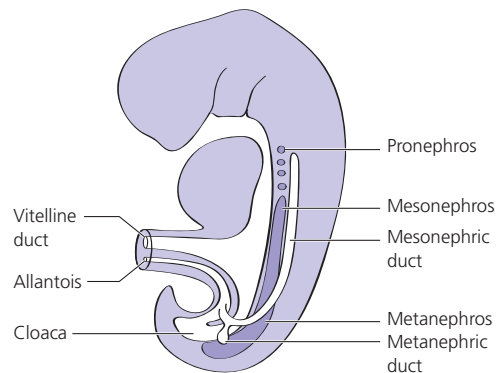


Figure 110.2 Development of the pro-, meso- and metanephric systems.

- *Infancy:* The baby may die from multiple congenital anomalies and renal failure.
- *Young adults:* Large bilateral symptomless masses may be found on a routine medical examination.
- *Middle-age and older adults:* Hypertension, loin pain, haematuria, cyst infection, renal failure.

**What are the two common forms of death from this condition, if untreated?**

The patient either dies from renal failure or from the complications of hypertension – cardiac failure or a cerebrovascular catastrophe (cerebral haemorrhage or

rupture of an associated berry aneurysm).

**How is this condition treated?**

Dialysis is commenced when the patient develops renal failure, with by kidney transplantation for those who are fit enough.

Occasionally bilateral nephrectomy may be required, either to make room for the transplant, to treat the otherwise uncontrolled hypertension, or for recurrent pain, infection or haematuria.