Inherited Kidney Diseases

There are various kidney diseases, ranging from relatively common to rare disorders and from benign disorders to those with a high morbidity and mortality. Presentation may also vary - eg, renal mass, loin pain, failure to thrive, short stature, hypertension or renal dysfunction.

Advances in genetic techniques are providing novel insights into kidney diseases, especially diagnosis, classification, pathogenesis and therapy. Many congenital kidney diseases are due to single gene defects (eg, some cases of nephrotic syndrome resistant to steroids). It is also becoming clear that some adult-onset kidney diseases - which are far more common - are associated with risk alleles (genetic variants linked to an increased risk of developing certain diseases). An example is focal segmental glomerulosclerosis and chronic kidney disease in African-American patients.[1]

Congenital anomalies of the kidney and urinary tract anatomy represent approximately 30% of all prenatally diagnosed malformations.[2]

Main groups of inherited kidney diseases

- Cystic kidney diseases:
  - Autosomal dominant polycystic kidney disease.
  - Autosomal recessive polycystic kidney disease.
  - Nephronophthisis: juvenile and adult form.
  - Medullary sponge kidney.

- Alport's syndrome and variants.
- Bartter's syndrome.
- Inherited metabolic diseases with renal involvement:
  - With glomerular involvement - eg, diabetes mellitus, genetic amyloidosis, Anderson-Fabry disease.
  - With non-glomerular involvement - eg cystinosis (and other causes of inherited renal Fanconi syndrome), cystinuria (autosomal recessive disorder with the formation of cystine stones in the kidneys, ureter and bladder), hyperoxaluria.

- Other inherited diseases - eg, congenital nephrotic syndrome, nail-patella syndrome (autosomal dominant - results in small, poorly developed nails and kneecaps and may be associated with proteinuria, haematuria and end-stage kidney disease).
- Primary immune glomerulonephritis (occasionally familial - eg, IgA nephropathy).
- Various renal diseases with genetic influence - eg, reflux nephropathy, haemolytic uraemic syndrome.

Assessment

- Prenatal diagnosis may be possible.
- Full clinical assessment, including family history and assessment of family members where appropriate.
- Assessment of renal function.
- Imaging of the urinary tract - eg, ultrasound, CT, MRI scanning.
- Renal biopsy.
- Genome testing.[3]
- Full evaluation for associated defects.
Management

- The management will depend on the underlying disorder, degree of renal dysfunction and associated defects.
- Genetic counselling is useful in patients and relatives where there is a defined autosomal dominant condition. However, where there is familial aggregation of congenital renal malformations with no defined genetic abnormality, genetic counselling may be of less benefit. [4]
- The role of prenatal interventions and postnatal therapies in cases of congenital kidney and urinary tract anomalies requires further research. [2]

Further reading & references


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