Rare renal conditions cause structural and functional abnormalities of the kidneys, and have varying effects on patient morbidity.

Understanding rare renal conditions

In this article...

- Overview of rare renal diseases
- How they affect the structure and function of the kidney
- The effect of these conditions on patients

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Abstract

Rare renal conditions can lead to structural and functional problems in the kidney resulting in long-term health problems and possible chronic kidney disease. This article provides an overview of some of these rare conditions.

There are thought to be more than 100 rare kidney diseases (Shayman, 2006), all of which can cause structural and functional abnormalities, ranging from congenital abnormalities to conditions that cause kidney stone formation (nephrolithiasis). Their effects vary, with patients being unaware of their condition, for example when they have only one kidney, while others develop long-term conditions such as chronic kidney disease.

Congenital
The kidney is the most common site for congenital abnormalities, many of which result in impaired renal function (Newson, 2013). In some cases, a kidney may be absent (renal agenesis), and this is often found incidentally.

Renal hypoplasia (small kidney) may cause hypertension and sometimes a nephrectomy (removal of the kidney) is required. Dysplastic and multicystic kidneys may fail to function and are frequently associated with other developmental abnormalities of the genitourinary system (Box 1).

Glomerular abnormalities
The glomeruli in the kidney filter out soluble waste products while retaining proteins and cells. Diseases of the glomerulus usually result in waste products accumulating in the blood and can cause blood and protein to leak into the urine.

“Glomerulonephritis” describes a range of conditions that often have an autoimmune aetiology. Some of these can be rapidly progressive, such as Goodpasture’s syndrome, in which the glomeruli are attacked by abnormal antibodies. Others progress slowly over decades, such as IgA nephropathy in which 20% of patients progress to end-stage renal failure within 20 years (Galla, 1995).

Glomerular disease can be genetic, such as Alport’s syndrome, in which the gene for a form of collagen in the kidney (collagen type IV) is mutated. Abnormalities in the collagen in the kidney basement membrane cause gradual scarring of the kidneys and can progress to end-stage renal failure.

Glomerular disease can occur alone or be associated with an underlying medical problem such as infection, systemic lupus erythematosus or diabetes.

Renal tubular disease
Rare diseases that affect the renal tubules can have significant implications for fluid management, mineral homoeostasis and electrolyte balance (Chadha and Alon, 2009).

Keywords: Kidney/Genetics/Chronic kidney disease

●This article has been double-blind peer reviewed

5 key points

1 There are more than 100 rare kidney diseases
2 The kidney is the most common site for congenital abnormalities
3 Diseases of the glomerulus usually result in waste products accumulating in the blood
4 Rare diseases affecting the renal tubules can have significant implications on fluid management, mineral homoeostasis and electrolyte balance
5 Stone-forming conditions can increase the risk of chronic kidney disease

BOX 1. EXAMPLES OF RARE CONGENITAL DISORDERS

- Branchio-oto-renal syndrome (BOR)
- Renal coloboma syndrome
- Renal cysts and diabetes syndrome (HNF1B)
- Hypoparathyroidism, deafness and renal syndrome (HDR)
If a patient displays failure to thrive, polyuria and polydipsia, recurrent renal calculi, nephrocalcinosis, or unexplained hypertension, then tubular disorders should be considered. Common biochemical findings are listed in Box 2.

Patients with a confirmed diagnosis should have their biochemical results monitored closely during times of illness as levels can change and they may need their treatment to be adjusted temporarily.

Kidney stone-forming conditions
When the kidney functions normally, it is able to excrete metabolic waste, such as calcium and oxalate, and prevent kidney stones from forming. Patients with a rare kidney stone-forming condition have a high excretion rate of the minerals needed to make a kidney stone.

Stone-forming conditions, such as cystinuria, can also increase the risk of chronic kidney disease due to the damage caused by recurrent infections, surgery and hydronephrosis. Primary hyperoxaluria is associated with the overproduction of oxalate, resulting in calcium oxalate stones forming. This can eventually cause end-stage renal failure and intensive dialysis is needed to remove the oxalate and prevent calcium oxalate from being deposited in body tissue. Oxalate is the salt form of oxalic acid, and is a natural end product of metabolism.

Early treatment and appropriate management are vital to prevent adverse outcomes and delay chronic kidney disease progression.

Inborn errors of metabolism
The kidney can be affected by multisystemic inborn errors of metabolism. Cystinosis is a rare lysosomal-storage disease that causes cystine, an amino acid, to build up in the tissues. This can result in end-stage renal failure, diabetes, hypothyroidism, myopathy and central nervous system deterioration (Cherqui, 2012). Patients usually present in childhood with Fanconi syndrome and often need a kidney transplant.

In this condition, damage to the tubules causes important electrolytes such as sodium, potassium, phosphorus and bicarbonate to be wasted into the urine. Symptoms include:

- Poor growth;
- Excessive thirst;
- Excessive urination;
- Dehydration;
- Rickets.

Fabry disease is another lysosomal storage disease that is multisystemic and presents with a variety of symptoms including severe and progressive renal failure (Feriozzi et al, 2009).

Ciliopathies
Cilia are slender, microscopic, hair-like structures that extend from the surface of all cells and are broadly divided into two groups – motile and non-motile primary cilia.

Defective and dysfunctional non-motile cilia are now understood to be the cause of many rare genetic conditions called ciliopathies (Lee and Gleeson, 2011). The cilia in the kidney point into the urine and sense the flow of urine; they alert their cells that there is urine flow and produce signals that control urine concentration. If cilia are defective, there is no signalling and excessive cell division leads to uncontrolled cyst formation in the kidneys. Ciliopathies result in developmental delay, cystic kidney, retinal defects and polydactyly (an abnormal number of digits).

Conclusion
Although the diseases themselves are rare, having an understanding physiology of rare conditions can help nurses to manage and understand more common renal diseases. NT

References

Further information can be found at www.rarerenal.org

### BOX 2. TUBULAR DISEASE BIOCHEMISTRY

The following common biochemical findings are associated with renal tubular diseases:

- Hyperchloraemia (high level of chloride within the blood)
- Metabolic acidosis
- Metabolic alkalosis with or without hypokalaemia (primary increase in serum bicarbonate, with or without low serum potassium)
- Hypoatraemia with hyperkalaemia (low serum sodium with high serum potassium)
- Hypocalciuria with normal serum calcium (high urine calcium levels with normal levels of calcium in the blood)

Source: Bagga et al (2005)