Genetic nephrolithiasis

Including: Cystinuria; hyperoxaluria; Dent's disease

Cystinuria

- AR
- Encodes a family of solute carrying tubular proteins
- Defective uptake of cystine from urine leads to hyper-saturation in filtrate and stone formation
- Presents with nephrolithiasis
- Investigate: Urine microscopy shows hexagonal crystals, raised cystine on 24 hour urine collection, radio-opaque stones on imaging
- Treatment: increase fluid intake, can add in penicillamine or others to increase solubility in urine

Hyperoxaluria

- AR
- Defective proteins associated with alanineglyoxylate aminotransferase.
- Normally Glyoxalate converted to glycine
- In hyperoxaluria compensatory pathway leads glyoxalate to be converted to oxalate
- Systemic hyperoxalaemia and hyperoxaluria
- Presents with calcium oxalate nephrolithiasis as well as systemic oxalate deposition affecting joints, heart and blood vessels
- Investigations: raised urinary oxalate excretion, calcium oxalate renal stones
- Treatment:
    - Liver-kidney transplant (the oxalate metabolic pathway occurs in the liver)
    - Pyridoxine (vitamin B-6) is a cofactor in this chemical pathway and be given as a holding measure

Dent's disease

- XR
- Defect in CLCN5, renal chloride channel
- Failure of transport of proximal tubule proteins from urine to blood
- Presentations: Nephrolithiasis, nephrocalcinosis, Fanconi's syndrome
- Treatment with renal transplant once end-stage renal failure sets in.