Single gene glomerular disease

Including: Alport's syndrome; Steroid resistant nephrotic syndrome; Nail-patella syndrome

Alport's syndrome

- Second most common cause of inherited renal failure
- Genetics of Alport's syndrome
  - Most commonly deletion on X Chr on COL4A5 – encodes collagen type IV (alpha5)
  - X-linked dominant
  - Less frequently can be autosomal dominant or autosomal recessive
- Presentation of Alport's syndrome
  - Sensorineural deafness
  - Haematuria (common), proteinuria and hypertension
  - Nephrotic syndrome in 30-40% of young adults
  - Can get blindness due to retinitis pigmentosa
- Associations with Alport's syndrome
  - Retinitis pigmentosa
  - Most common retinopathy is a "dot-and-fleck" pattern, anterior lenticonus is pathognomonic
  - Anti-GBM
  - Leiomyomatosis
- Work-up of Alport's syndrome
  - Renal biopsy, genetic testing, audiometry, ophthalmic review and renal USS
- Treatment of Alport's syndrome
  - Screening
  - Hearing aids
  - ACE-I or ARBs if proteinuria
  - Renal transplant – anti-GBM disease can occur in small number post-transplant

Steroid resistant nephrotic syndrome

- Autosomal recessive
- Congenital nephrotic syndrome usually presenting as focal-segmental glomerulosclerosis (FSGS) on biopsy
- Treated with alkylating agents, calcinuerin inhibitors, diuretic and antihypertensives.

Nail-patella syndrome

- Autosomal dominant – Chr 9 long arm 9q34
- Nail dysplasia, absent patella and steroid resistant nephrotic syndrome
- LMX1B gene defect causing problems with LIM homeodomain protein