Congenital abnormalities of the kidney and urinary tract (CAKUT)

Please note there are a multitude of CAKUT diseases and associated conditions. Two have been picked for illustrative purposes but the entire area is beyond the scope of this page: Vesicoureteral reflux (VUR) and Kallman's syndrome.

Vesicoureteral reflux

- Mixture of genetic phenotypes
- Graded 1-5 radiologically (1= reflux into ureter with no dilatation, 5 = gross dilatations of ureter, renal pelvis and calyces)
- Presentations: Childhood UTIs, adults: CKD with renal scarring or sequelae of CKD e.g. hypertension
- Investigations: DMSA or MAG-3 scan- renal scarring, gold standard is a micturition cystogram
- Treatment:
    - In children long term antibiotics are an option, surgical options are available but may not be better than other more conservative options
    - In adults managing CKD is the priority

Kallman's syndrome

- AD
- Defect in KAL1 encoding anosmia
- Primary defect is with gonadotropin releasing hormone deficiency with associated anosmia
- Apart from primary infertility and other reproductive failures renal agenesis may occur
- Renal agenesis Anosmia, hypogenitalism