



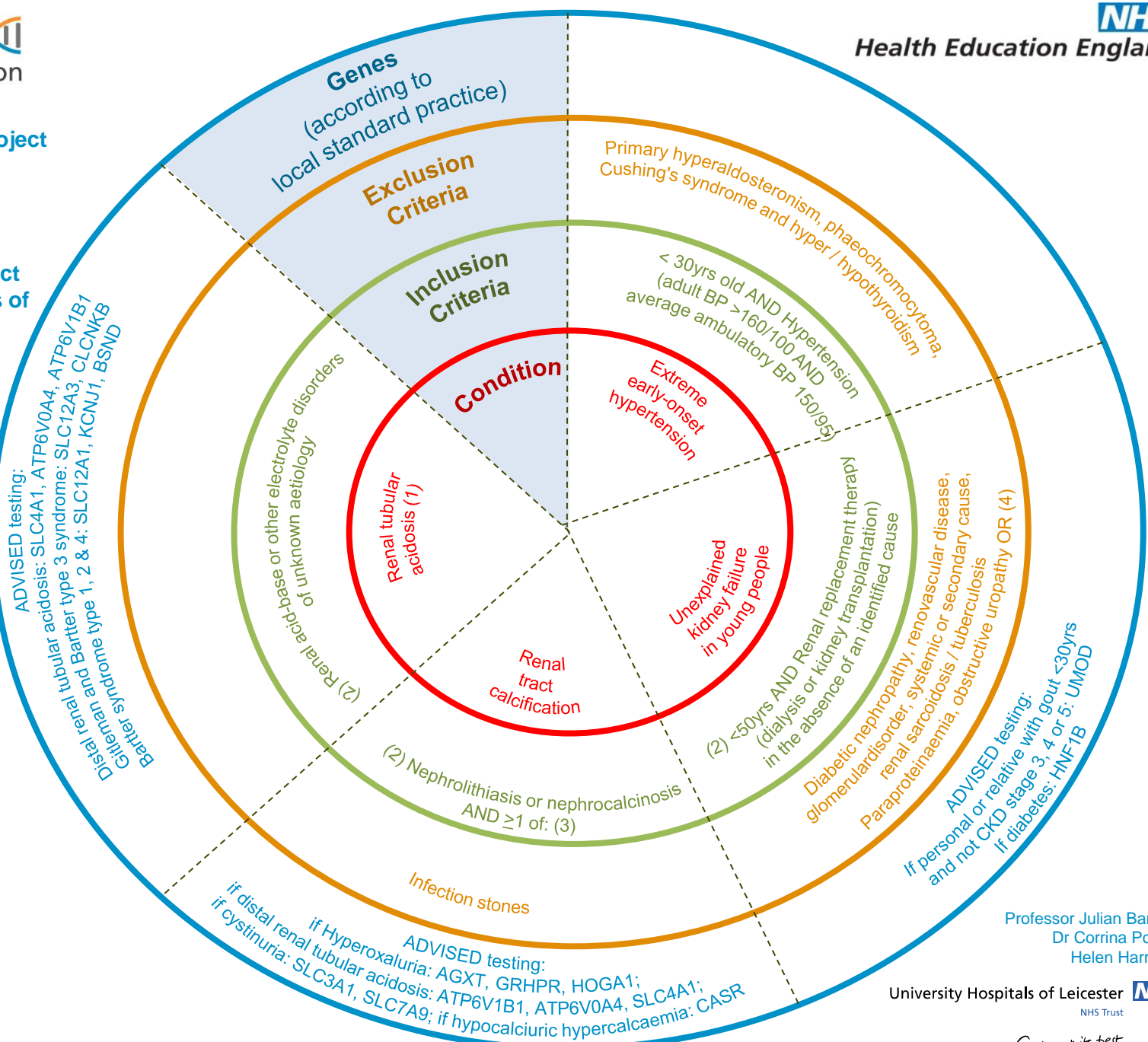
Rare Diseases: Renal and urinary tract disorders - Disorders of function

- (1)
- Distal renal tubular acidosis
  - Renal Fanconi syndrome
  - Gitelman and Bartter type 3 syndrome
  - Bartter syndrome type 1, 2, & 4,
  - Liddle syndrome
  - Gordon syndrome
  - Glucocorticoid remediable hypertension
  - Apparent mineralocorticoid excess
  - Pseudohypoaldosteronism type 1

(2)  
Recruitment should NOT include unaffected relatives (normal screening) unless part of a TRIO/DUO. Multiple affected relatives are preferable to singleton cases.

- (3)
- 1<sup>st</sup> episode <21yrs
  - FDR / SDR with stones /nephrocalcinosis
  - Hyper / hypocalcaemia,
  - Abnormal PTH with normal eGFR
  - Metabolic acidosis / alkalosis,
  - Hypercalciuria
  - Hyperphosphaturia
  - Hyperoxaluria
  - Aminoaciduria
  - Hypocitraturia
  - Hypermagnesuria

(4)  
Meets other renal condition criteria



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