Genomics Education Programme

100,000 Genomes Project Eligibility wheels A tool for clinicians

Rare Diseases: Renal and urinary tract disorders – Syndromes with prominent renal

abnormalities #1

(1)
Recruitment should NOT include unaffected relatives (normal screening) unless part of a TRIO/DUO. Multiple affected relatives are preferable to singleton cases.
(2)

- Family history of renal failure or proteinuria, consanguinity or syndromic features
- Kidney biopsy in ≥1 affected relative showing: diffuse mesangial sclerosis, focal segmental or likely genetic glomerular pathology
- Steroid sensitive nephrotic syndrome (diagnosed <5 yrs)</li>
- Steroid resistant nephrotic syndrome (SRNS)

