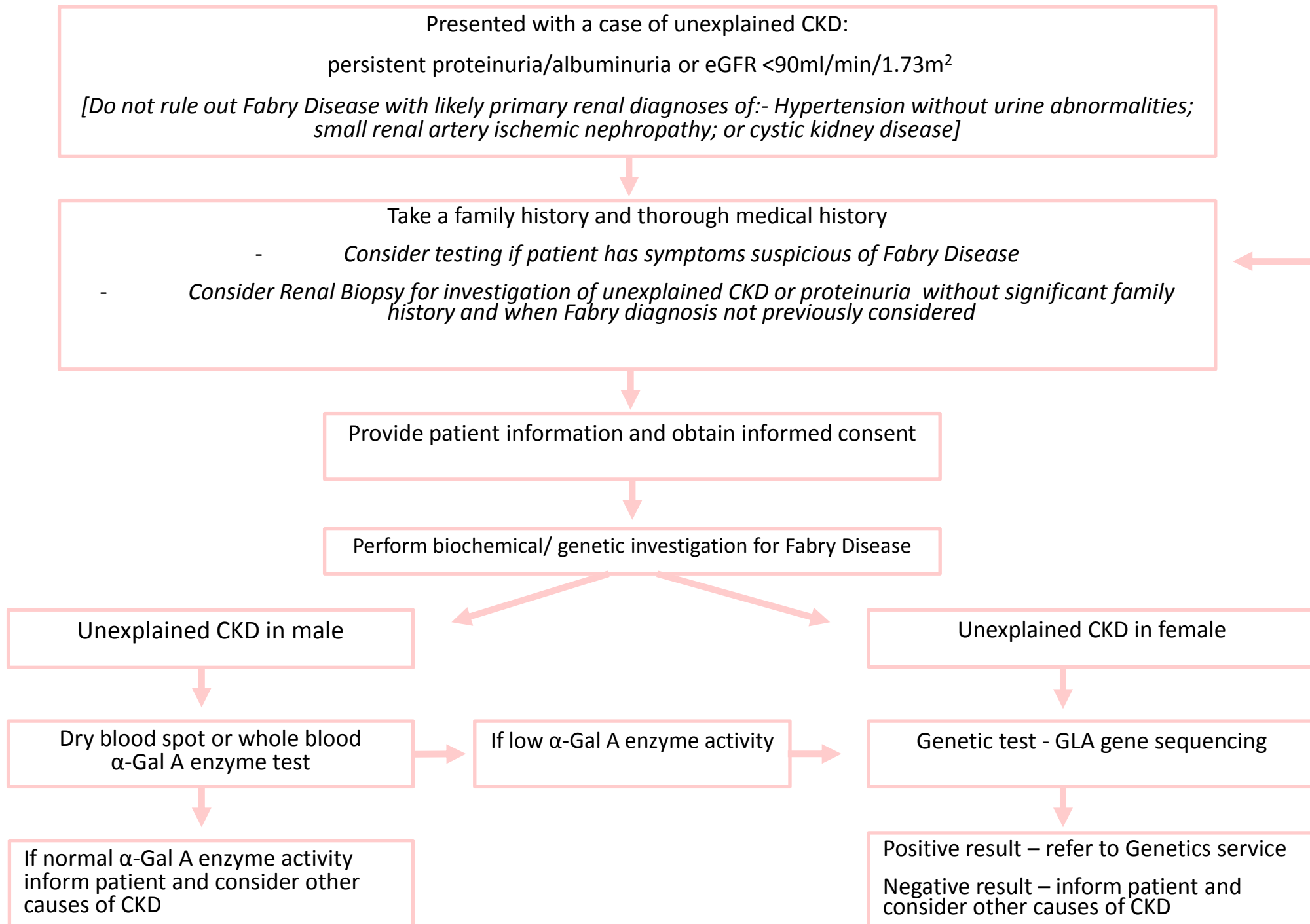


Diagnosis of Fabry Disease through Renal Services



Ancillary Information to support a suspicion of Fabry Disease

- Take family history and look for patterns compatible with X-linked inheritance – history of CKD, heart disease or stroke in family, especially at young age
- History and examination – look for angiokeratoma; lack of sweating; temperature intolerance; acroparesthesia (abnormal sensation or pain in hands/feet)
- Beware of pre-existing diagnostic labels (esp. chronic pain; fibromyalgia; IBS; growing pains; cardiomyopathy)
- Absence of proteinuria does not rule out Fabry Disease
- Fabry Disease may present with single organ involvement
- Parapelvic cysts (renal sinus cysts) on renal ultrasound
- Urinary sediment – birefringent lipid droplets – maltese crosses
- ‘Zebra bodies’ intra-cytoplasmic inclusion bodies seen on electron microscopy of renal biopsy within podocytes, mesangial cells and renal tubular cells