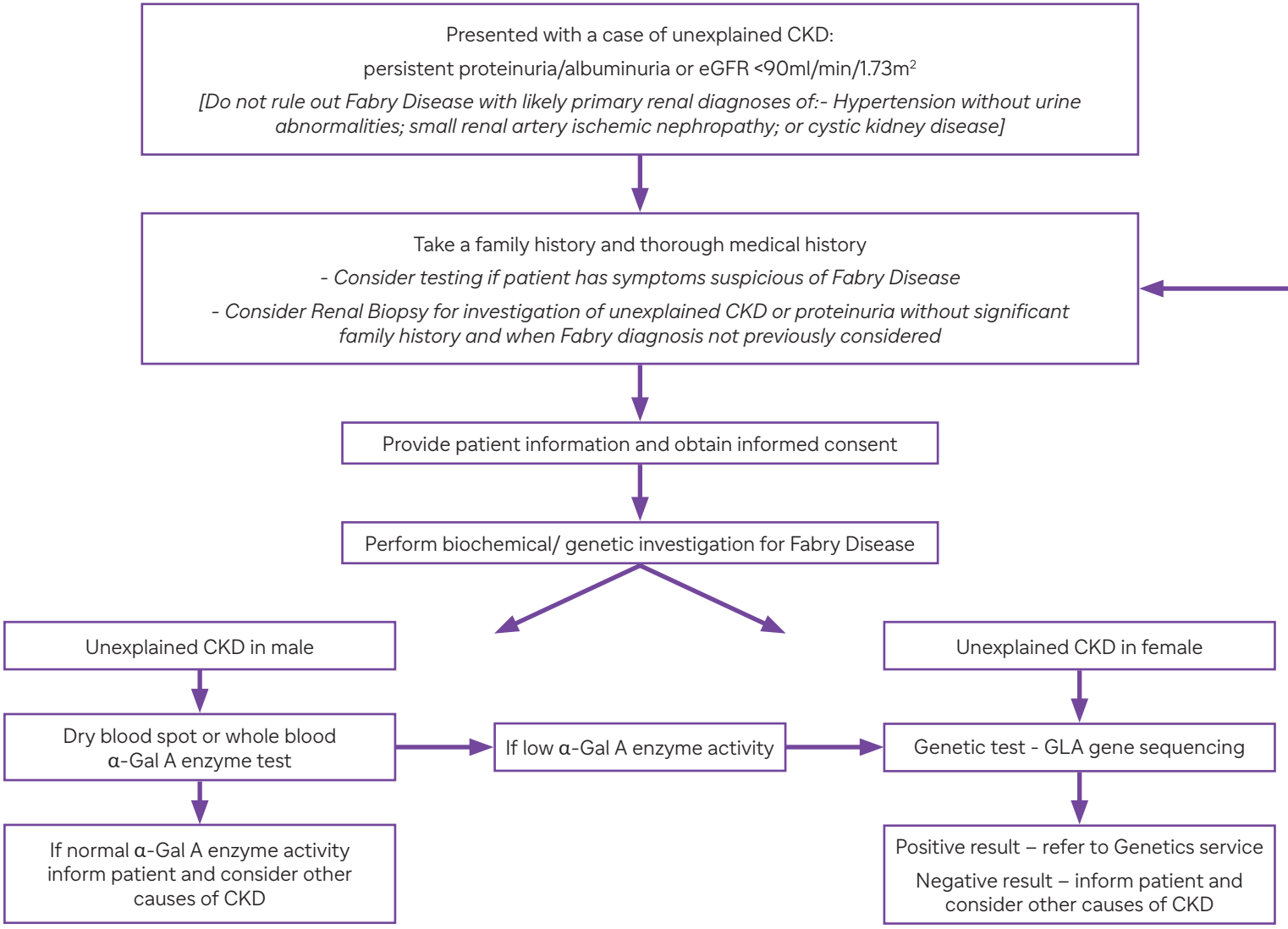
Diagnosis of Fabry Disease through Renal Services[1]

sanofi

- Fabry Disease is a life-limiting, inherited condition resulting from deficient α-galactosidase A (α-GAL A) activity[2,3]
- This causes pathogenic accumulation of globotriaosylceramide (Gb3 or GL-3) and other metabolites that can lead to cardiac hypertrophy, fibrosis and conduction abnormalities[3]



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 – birefrigent 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

Adapted from the UK Kidney Association's 'Diagnosis of Fabry Disease through Renal Services' algorithm

References : **1.** UKKA (2022) Diagnosis of Fabry Disease through Renal Services. Available at : https://ukkidney.org/sites/renal.org/files/radar/pictures/RA-RareRenal-Fabry-diagnosis-algorithm.pdf. Accessed February 2024. **2.** MacDermot KD, et al. J Med Genet. 2001;38 (11):769–775. **3.** Germain DP. Orphanet J Rare 2010;5(30):1-49.

MAT-XU-2300274 (v3.0) Date of preparation: February 2024