Algorithm for the diagnosis and assessment of Fabry disease patients

Family member with Fabry disease

Symptoms suggestive of Fabry disease

(e.g. angiokeratomas, acroparesthesiae) see Table 1

Biochemical/genetic analysis: males

- Measurement of plasma or urinary Gb₃ or lyso-Gb₃
- Measurement of plasma α-galactosidase A activity
- Confirmation of Fabry disease by genetic analysis of the GALA gene

Genetic/biochemical analysis: females

- Genetic analysis of the GALA gene
- Measurement of plasma or urinary Gb₃ or lyso-Gb₃

Initial positive diagnosis of Fabry disease - referral to tertiary centre



Baseline valuation at tertiary centre



Pain/QOL

Structured assessment scales (e.g. BPI, VAS, SF-36)

Neurological system

- Brain MRI
- Temperature sensitivity tests
- Electromyography

Eyes/ears

- Slit -lamp examination
- Audiogram

Renal function

- Urine dipstick test (microalbuminuria/ proteinuria)
- 24-hour urine (protein)
- Biopsy (only if indicated)
- Ultrasound (vascular lesions)

Cardiography

- ECG
- Echo
- 24-hour Holter
- Assessment of cardiac function

GI system

- Diarrhoea
- Constipation
- Abdominal pain

Dermatology

- Angiokeratomas
- Dyshidrosis
- Telangiectasia



Follow-up

Enzyme replacement therapy Concomitant therapy