# UKGTN Testing Criteria

| **Test name: Fabry Disease** | |
|---|---|
| | |
| **Approved name and symbol of disease/condition(s):**<br>**Fabry Disease** | **OMIM number(s):**<br>**#301500** |
| **Approved name and symbol of gene(s):**<br>galactosidase, alpha; GLA | **OMIM number(s):**<br>***300644** |

| | |
|---|---|
| **Patient name:** | **Date of birth:** |
| **Patient postcode:** | **NHS number:** |
| **Name of referrer:** | |
| **Title/Position:** | **Lab ID:** |

| **Referrals will only be accepted from one of the following:** | |
|---|---|
| **Referrer** | **Tick if this refers to you.** |
| Consultant Clinical Geneticist | |
| Consultant Metabolic Disease Specialist | |
| Consultant Cardiologist | |
| Consultant Nephrologist | |
| Consultant Dermatologist | |
| Consultant Ophthalmologist | |
| Consultant Neurologist<br>Consultant Physician | |

| **Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:** | |
|---|---|
| **Criteria** | **Tick if this patient meets criteria** |
| Individuals with deficient alpha-galactosidase enzyme activity in plasma or leucocytes | |
| | |
| **OR** patients with one or more of the following clinical symptoms:<br>• Angiokeratoma<br>• Acroparasthesia<br>• Anhidrosis<br>• Relevant ocular symptoms (e.g. corneal opacity, lenticular changes)<br>• Relevant cardiovascular/cerebrovascular disease e.g. LVH<br>• Relevant renal symptoms e.g. proteinuria | |
| **OR** at risk family members where the familial mutation is known | |

**If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.**