DISEASE:
Fabry disease

NAME:	Fabry disease
DESCRIPTION:	A rare genetic, multisystemic lysosomal disease characterized by specific cutaneous (angiokeratoma), neurological (pain), renal (proteinuria, chronic kidney failure), cardiovascular (cardiomyopathy, arrhythmia), cochleo-vestibular and cerebrovascular manifestations (transient ischemic attacks, strokes). The phenotypic expression depends on age of onset and, in females, the level of X-inactivation.
ORPHACODE:	324
SYNONYMS:	Alpha-galactosidase A deficiency Anderson-Fabry disease Angiokeratoma corporis diffusum Diffuse angiokeratoma FD
XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM
ANALYTE(S):	GLA
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/94

RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Cardiopathies, hereditary (gene panel)
- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Enzymatic dosage Fabry disease
- Fabry disease
- Fabry disease
- Hypertrophic cardiomyopathy (gene panel)
- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- galactosidase alpha

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL
- Hypertrophic cardiomyopathy (75 genes) - IPG
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG
