Alpha-Galactosidase A

Description	Activity of α-galactosidase in white blood cells
Indication	Screening for Fabry disease
Additional Info	Deficiency of α-galactosidase due to *GLA* gene mutation causes Fabry disease, an X-linked recessive lysosomal storage disease in which globotriaosylceramide, a glycosphingolipid, accumulates within the body cells, leading to impairment of normal organ and tissue function. Patients with Fabry disease can present with a number of signs or symptoms of variable degree. • Peripheral nerves: burning/tingling/numbness in the extremities, Reynaud's-like symptoms • Skin: angiokeratomas, telangietasia, anhidrosis (lack of sweating), hyperhidrosis (excessive sweating), oedema • Gastrointestinal tract: abdominal pain, diarrhoea, nausea, vomiting • Eyes: corneal clouding and other ocular manifestations • Ears: progressive hearing loss, tinnitus, vertigo • Heart: hypertension, cardiomyopathy • Brain: transient ischaemic attack, stroke • Kidneys: proteinuria, haematuria, lipiduria, chronic kidney disease In addition to symptomatic treatment, enzyme replacement therapies have been licensed for Fabry disease.
Concurrent Tests	Lysosomal enzyme screen
Dietary Requirements	N/A
Interpretation	Males: Low or absent α-galactosidase A is consistent with Fabry disease. Genetic confirmation is recommended. Females: Low or absent α-galactosidase A is consistent with Fabry disease. However, α-galactosidase activity may be misleading in female carriers due to the random nature of X-inactivation. Thus, a normal α-galactosidase activity does not exclude Fabry disease in females. Genetic analysis is recommended to provide definitive confirmation.
Collection Conditions	No restrictions
Frequency of testing	Repeated measurement inappropriate