

# GENETIC TESTING FOR CARDIOVASCULAR DISORDERS

## June 2023



### LOCAL TESTING

Test	Mnemonic
Familial hypercholesterolemia	FHNGS
Pharmacogenomics panel	PHARMA
Marfanoid syndromes/aortopathies (15 gene panel)	MARFAN

### OVERSEAS TESTING

For many genetic cardiac conditions (such as inherited cardiomyopathies and arrhythmias), local curated diagnostic NGS panels are not available. We are able to facilitate testing of appropriately selected individuals through Invitae Laboratories (USA). This involves:

- A **handling fee** (NGCF, R1372), billed by Ampath.
- A **testing cost (\$349)** which the patient pays directly, via an online link, to the overseas lab.
- Testing requires:
  - A completed Invitae consent form, signed by the patient.
  - A completed Ampath requisition form (select NGCF), **including clinical information** (phenotype, family history and suspected diagnosis) and the specific requested panel.
  - Two EDTA tubes of blood, drawn by Ampath.
- Turnaround time is ~4 weeks and the original report is emailed to the clinician.
- **These costs are NOT covered by medical aids.**



#### GENETIC COUNSELLING CLINIC

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Depending on the cardiovascular condition, genetic testing may be useful for diagnosis, prognosis, treatment, family screening, and reproductive planning. Genetic counselling plays an important role in assessment and care of patients undergoing genetic testing.

**Pre-test genetic counselling** includes obtaining a detailed family history. Counselling allows for the patient to understand the potential benefits, limitations and process of genetic testing, preparing them for the possible outcomes.

**Post-test counselling** is important in explaining the meaning of results, discussing risks to other family members and facilitating testing of other at risk individuals, in the case of positive results.

AmPath offers face to face and remote genetic counselling to referred patients.

**Appointments can be made telephonically at 012 678 0645 or via email at [geneticsclinic@ampath.co.za](mailto:geneticsclinic@ampath.co.za).**