

# INTERNATIONAL GENETIC TESTING REFERRALS

## PATHOLOGY SOLUTIONS ARE IN OUR DNA



Ampath Genetics offers a referral and clinician support service for constitutional genetic testing that is not available in South Africa. For patients, pre- and post-test genetic counselling are also offered.

Whilst the repertoire of locally available genetic tests is increasing, genetic testing for many rare disorders remains unavailable in South Africa at present. Based on the clinical features and family history, a next generation sequencing (NGS) panel, or broader genomic testing, such as whole exome sequencing (WES) may be indicated.

The majority of international NGS genetic panel testing is referred to INVITAE LABORATORIES in the United States, while some single gene tests, panels and WES are referred to CENTOGENE LABORATORIES in Germany. On rare occasions, a sample may be sent elsewhere when the specific testing required is not available at either of these laboratories. In general, medical aids do not cover the costs of international genetic testing referrals (see table 1 for more information). The referral and testing process is summarised on the next page.



### FAST FACTS

- Clinical phenotypic information from the referring doctor is essential when requesting international/ panel testing
- Informed consent is required
- Testing costs are paid by the patient, directly to the referral laboratory
- Clinical and genetic counselling support is offered by Ampath Genetics

**For more information, contact**  
geneticsclinic@ampath.co.za or  
012 678 0645

**TABLE 1: INTERNATIONAL TESTING**

Invitae Laboratories	Centogene Laboratories
<ul style="list-style-type: none"><li>• NGS panels in the clinical areas of inherited cancer syndromes, cardiology, neurology, immunology, metabolic, and paediatric and rare diseases.</li><li>• There is a standard panel cost of \$399*, payable directly by the patient.</li><li>• Family testing is available (at no additional cost, aside from additional courier charges) when pathogenic variants are identified, within a limited time period.</li></ul>	<ul style="list-style-type: none"><li>• Additional NGS panels and single gene tests requiring specific expertise/techniques, as well as whole exome and genome sequencing.</li><li>• Testing costs vary according to the test and are payable directly by the patient.</li><li>• Family testing is available at an additional cost to the family.</li></ul>

\* Price correct at time of publishing (September 2024) and is subject to change.

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## SUMMARY OF THE REFERRAL AND TESTING PROCESS

