

# Lab Update no.72

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## GENETIC TESTING FOR HAEMOPHILIA FROM ROYAL BLOODLINES TO GENOMIC MEDICINE

Haemophilia A and haemophilia B are X-linked recessive bleeding disorders caused by deficiency of coagulation factor VIII (encoded by the *F8* gene) and factor IX (encoded by the *F9* gene), respectively.

According to the *World Federation of Hemophilia*, haemophilia A is one of the most common inherited bleeding disorders, affecting 1 in 5 000 male live births worldwide. Although typically seen in males, females may also be affected, either as asymptomatic carriers or, more rarely, with clinically significant disease due to factors such as skewed X-inactivation or homozygosity.

Ampath Laboratories now offers genetic testing for both haemophilia A and haemophilia B, in collaboration with the University of the Free State.

### UTILITY OF GENETIC TESTING

The initial laboratory diagnosis of haemophilia follows a stepwise approach using various coagulation studies and factor assays. Once the type of haemophilia is confirmed, genetic testing can be requested to confirm the severity of the disorder, predict response to replacement therapy and risk of inhibitor formation, guide reproductive decisions and for carrier detection. Targeted therapies used for specific genetic variants have been studied and might drive future haemophilia management.

**TABLE 1: MOST COMMON GENETIC ABNORMALITIES IN HAEMOPHILIA**

	SEVERE HAEMOPHILIA A	MILD OR MODERATE HAEMOPHILIA A	HAEMOPHILIA B
GENETIC FINDINGS	Intron 22 inversions (45%)	DNA sequence variants (76-99%)	Mostly DNA sequence variants
	Intron 1 inversions (2-5%)	Intron 22 inversions (1-4%)	
	DNA sequence variants (43-51%)	Intron 1 inversions (1-4%)	Large deletions (rare)
	Large deletions or duplications (<2%)	Large deletions or duplications (<1%)	

**TABLE 2: GENETIC TESTING AVAILABLE AT AMPATH**

	HAEMOPHILIA A (FACTOR VIII DEFICIENCY)	HAEMOPHILIA B (FACTOR IX DEFICIENCY)
TEST MNEMONIC	F8NGS	F9NGS
SPECIMEN TYPE	Peripheral blood (EDTA)	Peripheral blood (EDTA)
TEST METHODOLOGY USED	Fluorescent PCR, NGS and MLPA	NGS and MLPA
TURNAROUND TIME	6 weeks	6 weeks

PCR: polymerase chain reaction; NGS: next generation sequencing; MLPA: multiplex ligation-dependent probe amplification

Genetic counselling services are available for affected patients and their families. Bookings can be made at [geneticsclinic@ampath.co.za](mailto:geneticsclinic@ampath.co.za).

For more information, contact us at [ngs@ampath.co.za](mailto:ngs@ampath.co.za).

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