FAST FACT: MELANOMA GENETIC TESTING OPTIONS

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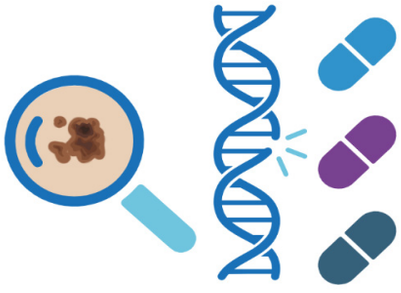
In South Africa, melanoma is the 4th most prevalent cancer in men, and the 5th most prevalent cancer among women. Genetic testing plays an integral part in the diagnosis, prognostication, and management of melanoma.

Various testing options available at Ampath are summarised below, as well as relevant biomarkers and their matched targeted therapies



Mnemonic	Details	Genes tested	Turnaround time	Sample requirements
GFISH	FISH	*RREB1* (6p25), *MYB* (6q23), *CCND1* (11q13)	4 working days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 4 normal slides (not charged) with 10 micron thick unstained recuts
BRAF	*BRAF* PCR	*BRAF V600 E/E2/D/D2/K/R*	4 working days	Formalin Fixed Paraffin Embedded Tissue (FFPE) 8-12 normal slides (not charged) with 10 micron thick unstained recuts or 10 micron thick recuts in an Eppendorf tube or Paraffin Embedded tissue block
EGFRASSEQ	DNA only NGS panel (no fusions)	*BRAF, CDKN2A, CTNNB1, GNA11, GNAQ, KIT, NRAS, PIK3CA, PTEN, TP53*	10 working days	
OCASUB	DNA and RNA panel	*AKT3, ALK, ARID2, BAP1, BRAF, CCND1, CDKN2A, CDK4, CYSLTR2, EIF1AX, ERBB4, EZH2, HRAS, GNAQ, GNA11, GRIN2A, KIT, KRAS, MAP2K1, MDM2, MET, MITF, MPA3KA, NF1, NRAS, NTRK, PIK3CA, PLCB4, PPP6C, PRKAR1A, PTEN, RAC1, RET, ROS1, RB1, SF3B1, TERT, TP53, TYR*	14 working days	
TMB	Tumour mutational burden measurement	90 genes	14 working days	

Biomarker	Therapeutic Implications
***BRAF* V600E**	*BRAF*/MEK inhibitors (e.g. dabrafenib-trametinib)
***BRAF* non V600E**	Mutations in codons near V600 have shown response to MEK inhibitors and *BRAF* and MEK inhibitor combinations
***BRAF* fusions**	Fusions in *BRAF* have also shown responses to MEK inhibitors and nonspecific RAF inhibitors (e.g. sorafenib)
***BRAF* amplification**	Resistance to *BRAF* inhibitors
KIT	- Exon 11 and 13 mutations are associated with sensitivity to *KIT* inhibitors - D816H mutation is associated with resistance to *KIT* inhibitors - KIT amplifications appear to have minimal or no sensitivity to *KIT* inhibitors
NRAS	May be associated with response to MEK inhibitors in some patients
KRAS	May be associated with response to MEK inhibitors in some patients
PTEN	Associated with resistance to targeted and immunotherapy
NF1	Potential therapeutic target - novel *NF1* binding partner: Calpain1 (CAPN1)
ALK	Fusion-directed therapy (e.g. crizotinib)
ROS1	Fusion-directed therapy (e.g. crizotinib or entrectinib)
NTRK 1/2/3	Fusion-directed therapy (e.g. larotrectinib or entrectinib)