آغا خان یونیورسٹی ہسپتال
The Aga Khan University Hospital

Myeloid Neoplasms by Next Generation Sequencing

Department of Pathology and Laboratory Medicine

UPDATE NO:10,VOL NO:XXXI,2025

November 2025

The information contained in this flyer is intended for healthcare professionals.

INTENT OF USE:

Myeloid NGS panel is intended for the molecular evaluation of myeloid neoplasms, including myeloproliferative neoplasms (MPN), myelodysplastic syndromes (MDS), myelodysplastic/myeloproliferative neoplasms (MDS/MPN), acute myeloid leukemias (AML), systemic mastocytosis, and myeloid neoplasms associated with eosinophilia and gene rearrangements. This assay, that interrogates 58 genes of interest, is most relevant to myeloid cancers.

INTRODUCTION:

Next-generation sequencing (NGS) is a comprehensive molecular diagnostic approach that allows simultaneous analysis of multiple genomic regions in tumor DNA within a single assay. While many hematologic neoplasms share morphologic or phenotypic features, they may harbor distinct somatic mutations that enable more precise classification. Moreover, several myeloid neoplasms present with a normal karyotype at diagnosis but can still be identified, confirmed, and classified based on their mutational profiles. Patients with unexplained cytopenia may exhibit acquired genetic alterations in hematopoietic cells—referred to as clonal cytopenia of uncertain significance (CCUS)—which carry a risk of progression to overt myeloid malignancies. The detection and interpretation of gene mutations in suspected or confirmed myeloid neoplasms provide essential diagnostic, prognostic, and therapeutic insights that guide clinical management.

IMPORTANT NOTE:

This test is for the evaluation of known or suspected hematologic neoplasms of myeloid origin (e.g., AML, MDS, MPN, MDS/MPN, and unexplained cytopenia) at the time of initial diagnosis or at disease relapse.
Its clinical utility includes:

- Assisting in accurate diagnostic classification
- Providing prognostic and therapeutic information to guide patient management
- Detecting newly acquired, clinically significant gene mutations at relapse

PRINCIPLE:

Next Generation Sequencing (NGS)

SPECIMEN TYPE:

Whole Blood in EDTA tubes

CHARGES:

PKR 150,000/ *Revisions may apply

SCHEDULE:

Test is performed 1st Monday of the month. Report will be issued after 20 days.

ADDITIONAL INFORMATION :

Myeloid Panel (58 genes)

ABL1	BRAF	CEBPA	ETV6	HRAS	KDM6A	NPM1	PTEN	SMC1A	TP53
ANKRD26	CALR	CSF3R	EZH2	IDH1	KIT	NRAS	PTPN11	SMC3	U2AF1
ASXL1	CBL	CUX1	FLT3	IDH2	KMT2A	PDGFRA	RAD21	SRSF2	WT1
ATRX	CBLB	DDX41	GATA1	IKZF1	KRAS	PHF6	RUNX1	STAG1	ZRSR2
BCOR	CBLC	DNMT3A	GATA2	JAK2	MPL	PIGA	SETBP1	STAG2	
BCORL1	CDKN2A	ETNK1	GNAS	JAK3	NF1	PPM1D	SF3B1	TET2	

Gene marked in green fully covers the coding sequence

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