GTC-Liquid Biopsy, Hematology

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Diagnosis and Monitoring

This test uses cell-free DNA (cfDNA) for detecting abnormalities in hematologic diseases. The test is particularly designed and offered to reduce the need for bone marrow biopsy. It is highly useful for patients presenting with cytopenia and to rule out MDS/CMML, MPN or other hematologic neoplasms. It can also be used for monitoring patients with hematologic neoplasms. Based on multiple studies, cfDNA can be more accurate in detecting abnormalities in bone marrow than bone marrow actual biopsy. Bone marrow biopsy might be limited to site of the biopsy, while the cfDNA reflects abnormalities in the entire body. Furthermore, based on our investigation, plasma is enriched by cancer-specific DNA/RNA due to the high turnover of tumor cells as compared with normal cells. This test is recommended for the diagnosis and follow up of:

-**Myelodysplastic syndrome (MDS)/Chronic myelomonocytic leukemia (CMML):** To determine if the patient has reactive cytopenia and to distinguish between CHIP (Clonal Hematopoiesis of Indeterminate Potential) or CCUS (Clonal Cytopenia of Unknown Significance) and MDS.

-Acute Myeloid Leukemia (AML): To confirm diagnosis of AML and helps in determining eligibility for treatment with FLT3 and IDH1/2 inhibitors and evaluate minimal/measurable residual disease (MRD). It is particularly useful for pediatric and elderly patients.

-Myeloproliferative Neoplasms (MPN): To confirm diagnosis and monitor MPN and evaluate levels of JAK2, CALR and MPL mutations.

-Lymphoma: Liquid biopsy and cfDNA analysis is recommended for patients with lymphoma and specific mutations. The levels of the detected mutations can be used to monitor these diseases and evaluate therapy. Analysis of the original diagnostic sample is required for proper and sensitive monitoring of lymphoma.

Specimen Requirements:

Peripheral blood: 5-10 mL. EDTA tube is preferred.

Shipping:

Ship using cold pack. The cold pack should not directly contact Blood. Ship As soon as sample collected with overnight delivery.

Turn Around Time:

5-7 days

Tested Genes

Hematology Genes Tested for Abnormalities in coding sequence												
ABL1	BCL2	CBL	CDKN2C	DICER1	FAS	IDH2	KMT2A	MPL	PAX5	PTCH1	SMAD2	TGFBR2
AKT1	BCL2L1	CBLB	CEBPA	DNMT3A	FBXW7	IGF1R	KMT2B	MRE11A	PBRM1	PTEN	SMAD4	TP53
AKT2	BCL6	CBLC	CHEK1	EP300	FLT3	IKZF1	KMT2C	MTOR	PDGFRA	PTPN11	SMARCA4	TSC1
AKT3	BCOR	CCND1	CHEK2	ERG	GATA1	IKZF3	KMT2D	MUTYH	PDGFRB	RAD21	SMARCB1	TSC2
ALK	BCORL1	CCND3	CIC	ETV6	GATA2	IRF4	KRAS	MYC	PHF6	RAD50	SMC1A	TSHR
AMER1	BCR	CD274	CREBBP	EZH2	GATA3	JAK1	MAP2K1	MYD88	PIK3CA	RAD51	SMO	WT1
APC	BIRC3	CD79A	CRLF2	FAM175A	GEN1	JAK2	MAP2K2	NFKBIA	PIK3R1	RB1	SOCS1	ZNF217
ARID1A	BLM	CD79B	CSF1R	FAM46C	GNAQ	JAK3	MAP2K4	NOTCH1	PIK3R2	RHOA	SRC	ZRSR2
ARID1B	BRAF	CDH1	CSF3R	FANCA	GNAS	KAT6A	MAP3K1	NOTCH2	PIM1	RNF43	SRSF2	MEF2B
ARID2	BRCA1	CDK12	CTNNA1	FANCC	H3F3A	KDM5C	MAP3K14	NOTCH3	PLCG1	RUNX1	STAG2	
ASXL1	BRCA2	CDK4	CTNNB1	FANCD2	HNF1A	KDM6A	MAPK1	NPM1	POLD1	SDHB	STAT3	
ATM	BTK	CDK6	CUX1	FANCE	HOXB13	KDR	MCL1	NRAS	POLE	SETBP1	STK11	
ATRX	CALR	CDKN2A	CXCR4	FANCF	HSP90AA1	KEAP1	MDM2	NSD1	PPM1D	SETD2	TERT	
B2M	CARD11	CDKN2B	DDR2	FANCG	IDH1	KIT	MDM4	PALB2	PPP2R1A	SF3B1	TET2	

GENOMIC TESTING COOPERATIVE

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