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:

GTC-Liquid Biopsy, Hematology

Diagnosis and Monitoring

-**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 |   |

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