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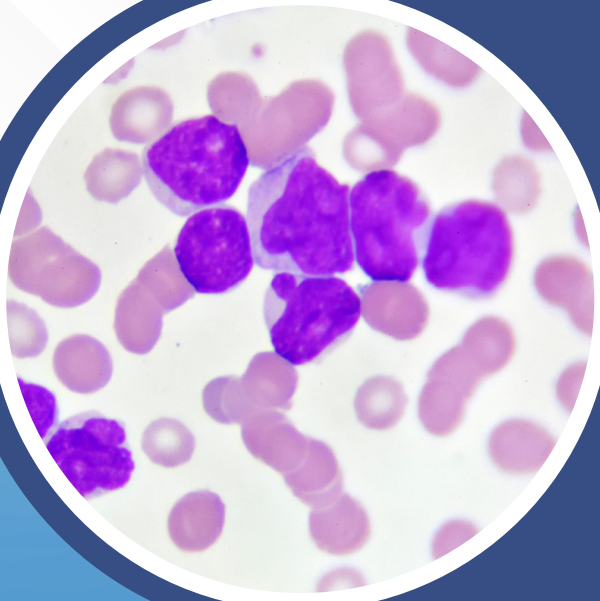
MC-5293

Uncovering Mutations in Your DNA

& providing clinically actionable data for choice of the most optimal treatment



LEUKEMIA

Leukemia is a cancer of the blood cells. Some of the major types of leukemia are Myelo Proliferative Neoplasms, Acute myeloid leukemia (AML), Acute lymphocytic leukemia (ALL), Chronic myeloid leukemia (CML), Chronic lymphocytic leukemia (CLL). The diagnosis of leukemia relies upon a multiparametric approach and Molecular methods are increasingly employed to help refine diagnosis, establish prognosis and determine the most appropriate or treatment. Following are the solutions being offered at Bioserve for molecular/ genetic testing of Leukemia.

Myelo Proliferative Neoplasms (MPN)

1. **JAK -2 Exon 14 Mutation / JAK2 V617F**
   - Detection of JAK2 V617F mutation helps in diagnosis of 70-85% cases Of **Polycythaemia Vera (PV**) and 40-50% cases of **Essential Thrombocythemia (ET**) and **Idiopathic Myelofibrosis (IMF).**
2. **JAK- Exon 12 Mutation**
   - Diagnose Polycythemia Vera (PV) (5-15%) for patients negative for JAK2 V617F to classify further or rule out
3. **Myeloproliferative Leukaemia (MPL) Mutation**
   - Identification of MPL mutations (codon 505, 515) can aid in the diagnosis of a myeloproliferative neoplasm and is highly suggestive of either PMF (8-10%) or ET (3-5%).
4. **Calreticulin Mutation**
   - Detection of mutations in exon 9 of CALR gene.
   - CALR mutated ET and PMF are associated with increased survival

Chronic Myeloid Leukemia (CML)

5. **BCR/ABL or Philadelphia chromosome**
   - Analysis of BCR-ABL fusion gene confirms the diagnosis
   - Effective for monitoring treatment efficacy.
6. **Imatinib Resistance test**
   - Failure of TKI treatment could be due to resistance due to mutations.
   - Identification of the mutations (T315I, Y253H, E255K/V, F359V/C/I, F317L/V/I/C, T315A, and V299L) may suggest need for second generation TKIfor further treatment.

Acute Myeloid Leukemia (AML)

7. **FMS-like tyrosine kinase 3(FLT3)**
   - Identifies internal tandem duplication (ITD) and D835 mutation of the FLT3 gene.
   - Unfavourable prognosis: associated with disease progression or relapse of AML. Target therapy Gilteritinib.
8. **Nucleophosmin 1 (NPMI)**
   - Favourable prognosis in the absence of a FLT3 mutations and normal karyotype
   - NPMI mutation is associated with better response to induction chemotherapy.
9. **CCAAT/Enhancer-binding protein alpha (CEBPA)**
   - CEBPA mutations are associated with favourable prognosis in the absence of associated cytogenetic abnormalities
   - Screening for CEBPA mutations offers a means for risk stratification in AML patients with normal karyotype.
10. **IDH1/IDH2 mutations**
    - Unfavourable prognosis.
    - Enasidenib, Ivosidenib targeted therapy is available for AML with IDH1 or IDH2 mutation

Hereditary Leukemia/ Lymphoma Panel

Leukemia is rarely hereditary. In cases of strong family history or risk prediction needs, NGS panel with 10 genes associated with familial leukemia risk can be profiled.

Genes Covered in this Panel

ATM SBDS PRF1 PTPN11 BRCA2 TP53 PALB2 BRIP1 RUNX1 CBL

Sample Type Required

Peripheral Blood In EDTA tubes or Bone marrow Aspirate

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