iBio DIAGNOSTICS

iBio - MYELOID TUMOR PANEL

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MYELOID TUMOR PANEL

- Mutations of 59 genes involved in myeloid neoplasia are investigated.
- Based on national collaboration.
- Genes required for diagnosis, relevant for prognosis and indentifying myeloid neoplasm with germline predisposition are included.
- Mutations relevant in myeloproliferative neoplasms are included.

Mininum diagnostic requirement

ASXL1, CEBPA, FLT3, NPM1, RUNX1, TP53

Important prognostic markers or potential targets of therapy

BRAF, DNMT3A, EZH2, IDH1, IDH2, KIT, MT2A, NRAS, STAG2, PTPN11, RAD21, SF3B1, SRSF2, U2AF1, WT1, ZRSR2

Predisposition identifiers

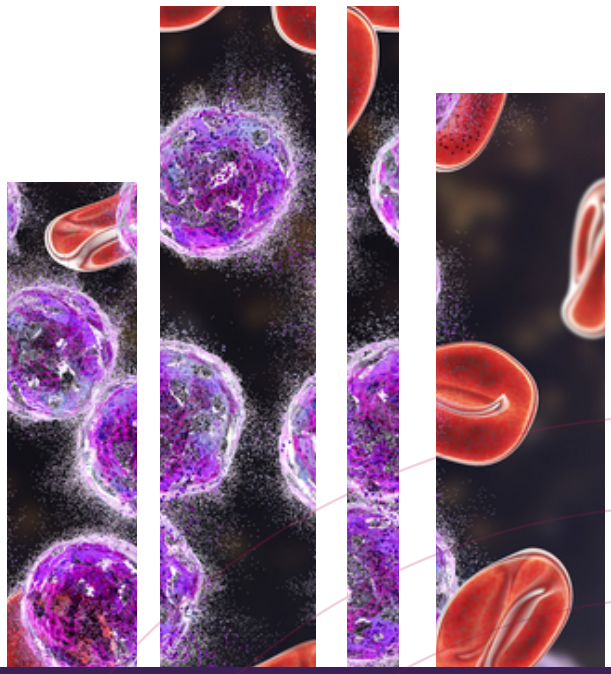
ANKRD26, ATG28, DDX41, ETV6, GATA2, SRP72, TERT

Myeloproliferative neoplasia genes

CALR, CSF3R, JAK2, MPL, SH2B3

ACUTE MYELOID LEUKEMIA

- Acute myeloid leukemia (AML) is a multitude of diseases with distinct genetic background and prognosis.
- Only 25% of patients are expected to survive 5 or more years after diagnosis.
- >90% of cases show translocations or mutations.
- Besides translocations, ELN recommends routine testing for mutations of 6 genes, over 20 are recognized as relevant for prognosis or identifying myeloid neoplasm with germline predisposition category.



MÓDSZER

- Isolated genomic DNA or bone marrow aspirate/peripheral blood sample in EDTA.
- Recommended tumor cell ratio: ≥20%
- Bioinformatic identification of single nucleotide variants **(SNV)**, short insertions and deletions as well as copy number variations **(CNV)**.
- Variant classification and annotation (e.g., ClinVar, COSMIC, HGMD, etc.).

LIST OF INVESTIGATED GENES

FULL CODING REGION:
ASXL1, BCOR, CEBPA, DNMT3A, ETV6, EZH2, GATA1, GATA2, IKZF1, PHF6, RAD21, RUNX1, SETD2, SRSF2, STAG2, TET2, TP53, ZRSR2, DDX41, TERC
HOTSPOTS:
ABL1, ACD, ANKRD26, BRAF, CALR, CBL, CSF3R, CUX1, ERCC6L2, ETNK1, FLT3, GNAS, GNB1, IDH1, IDH2, JAK2, KDM6A, KIT, KRAS, MPL, MYC, NF1, NPM1, NRAS, PDGFRA, PRPF8, PTPN11, SAMD9, SAMD9L, SETBP1, SF3B1, SH2B3, SMC1A, SMC3, SRP72, TERT, TINF2, U2AF1, WT1

COVERAGE: >95% (>500×)

AVERAGE SEQUENCING DEPTH: >1000×

TURNAROUND TIME: 2-3 WEEKS

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iBio SCIENCE

UNIVERSITY OF PÉCS
SZENTÁGOTHAI RESEARCH CENTRE

WORKFLOW

Our mission is advancing scientific research in the fields of **BIOTECHNOLOGY** and **MEDICINE** as well as applying the latest innovative technologies in diagnostics. **IBIOSCIENCE LTD.** in collaboration with **UNIVERSITY OF PÉCS SZENTÁGOTHAI RESEARCH CENTER** provides state-of-the-art next generation sequencing services and expertise for the Hungarian scientific community.

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SAMPLE DISPATCH
DNA, bone marrow aspirate or peripheral blood

03

SAMPLE PROCESSING
microscopical control of tumor cell ratio, DNA isolation

04

GENETIC ANALYSIS
bioinformatic analysis, variant identification and annotation

05

REPORT
categorization of variants based on guidelines (pathogenic, likely pathogenic, VUS etc.)

REFERENCES

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