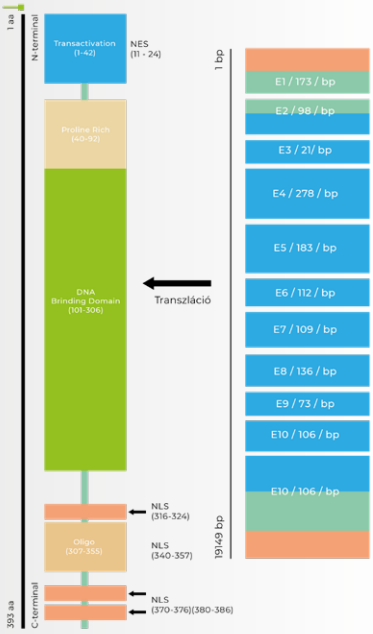
iBio DIAGNOSTICS

iBio - TP53 PANEL

EN

TP53 gene

- *TP53* is the most renowned tumor suppressor gene, its product, the p53 protein is called the guardian of the genome.
- In response to various cell stress, the p53 protein influences many cellular functions most notably cell cycle, apoptosis, cell senescence, DNA repair and metabolism.
- *TP53* gene alteration is present in approximately 50% of malignancies.
- Pathogenic mutations may occur in any of the 11 exons of the 32.8 kb long gene.
- **iwCLL (International Workshop on CLL) recommends TP53 mutation testing before initiating therapy in case of every chronic lymphocytic leukemia patient.**

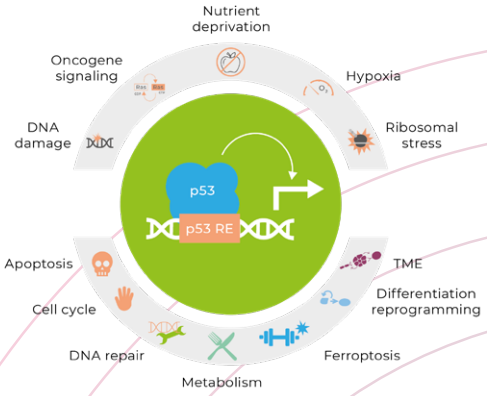


CHRONIC LYMPHOCYTIC LEUKEMIA

- Chronic lymphocytic leukemia (CLL) is the most common adult leukemia, it is genetically heterogeneous.
- *TP53* mutation is detected in 4-7% of patients at diagnosis, the prevalence may be as high as 40-50% at treatment failure or progression.
- *TP53* mutation is the most important prognostic and predictive genetic alteration; it is associated with unfavorable disease course and failure of chemoimmunotherapy.

ACUTE LEUKEMIA

- Acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL) are highly malignant, genetically heterogeneous diseases in adults.
- *TP53* gene alterations occur in 5-15% of acute leukemias and are associated with adverse prognosis.
- *TP53* mutation testing should be considered in every case of AML and ALL.



METHOD

- Isolated genomic DNA or bone marrow aspirate/peripheral blood sample in EDTA, or formalin fixed, paraffin embedded (FFPE) tumor tissue block.
- Tumor cell ratio of ≥20% is needed.
- Sequencing of total coding regions as well as 3'/5' UTRs of *TP53* gene.
- Bioinformatic identification of single nucleotide variants (SNV), short insertions and deletions.
- Variant classification and annotation (e.g. ClinVar, COSMIC, HGMD, IARC TP53).

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.

01

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02

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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