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Acute myeloid leukaemia (AML)

The "**AML**" NGS panel includes the analysis of 29 genes: ASXL1/BRAF/CALR/CBL/CEBPA/CSF3R/DNMT3A/ETV6/EZH2/FLT3/HRAS/IDH1/IDH2/JAK2/KIT/KRAS/MPL/NPM1/NRAS/PTPN11/RUNX1/SETBP1/SF3B1/SRSF2/TET2/TP53/U2AF1/WT1/ZRSR2.

It can be used in the three areas of **prognosis**, **theranostics** and **diagnosis** (as per the WHO 2017 classification).

- According to the WHO 2017, the mutation status of NPM1, CEBPA and RUNX1 are included in the **diagnostic criteria** for the WHO classification of acute myeloid leukaemias associated with a somatic mutation ('AML with gene mutation').
- The "AML" NGS panel can also assist with **prognosis** (2017 ELN Recommendations) by allowing the definition of molecular factors with a poor (NPM1 unmutated and FLT3-ITD high, RUNX1 mutation, ASXL1 mutation, TP53 mutation) or good prognosis (NPM1 mutation without FLT3- ITD or with FLT3-ITD low, biallelic CEBPA mutation). This molecular prognosis stratification supplements the cytogenetic prognosis stratification. The WHO 2017 also places the prognostic value of molecular abnormalities within cytogenetic groups: poor prognostic value of a CKIT mutation in AML with t(8;21)(q22;q22.1) or inv(16)(p13.1q22)/t(16;16)(p13.1;q22), of a WT1, TET2, ASXL1, DNMT3A or IDH1/2 mutation in AML with normal karyotypes, of aTP53 mutation in AML with complex karyotypes.
- In **theranostics**, testing for the FLT3 mutation is already dependant on a targeted treatment. KIT, IDH1, IDH2 or NPM1 can also be therapeutic targets.

Note: The BCOR and STAG2 genes are not included in the panel offered.

The "AML" panel thus allows a comprehensive analysis of somatic 'driver' mutations (it is not adapted to testing for germ mutations).

Please note that the morphology (peripheral blood, bone marrow aspiration, bone marrow trephine biopsy), the immunophenotypic analysis and genetic data must be must be compared for a diagnosis and/or prognosis of a malignant haemopathy.

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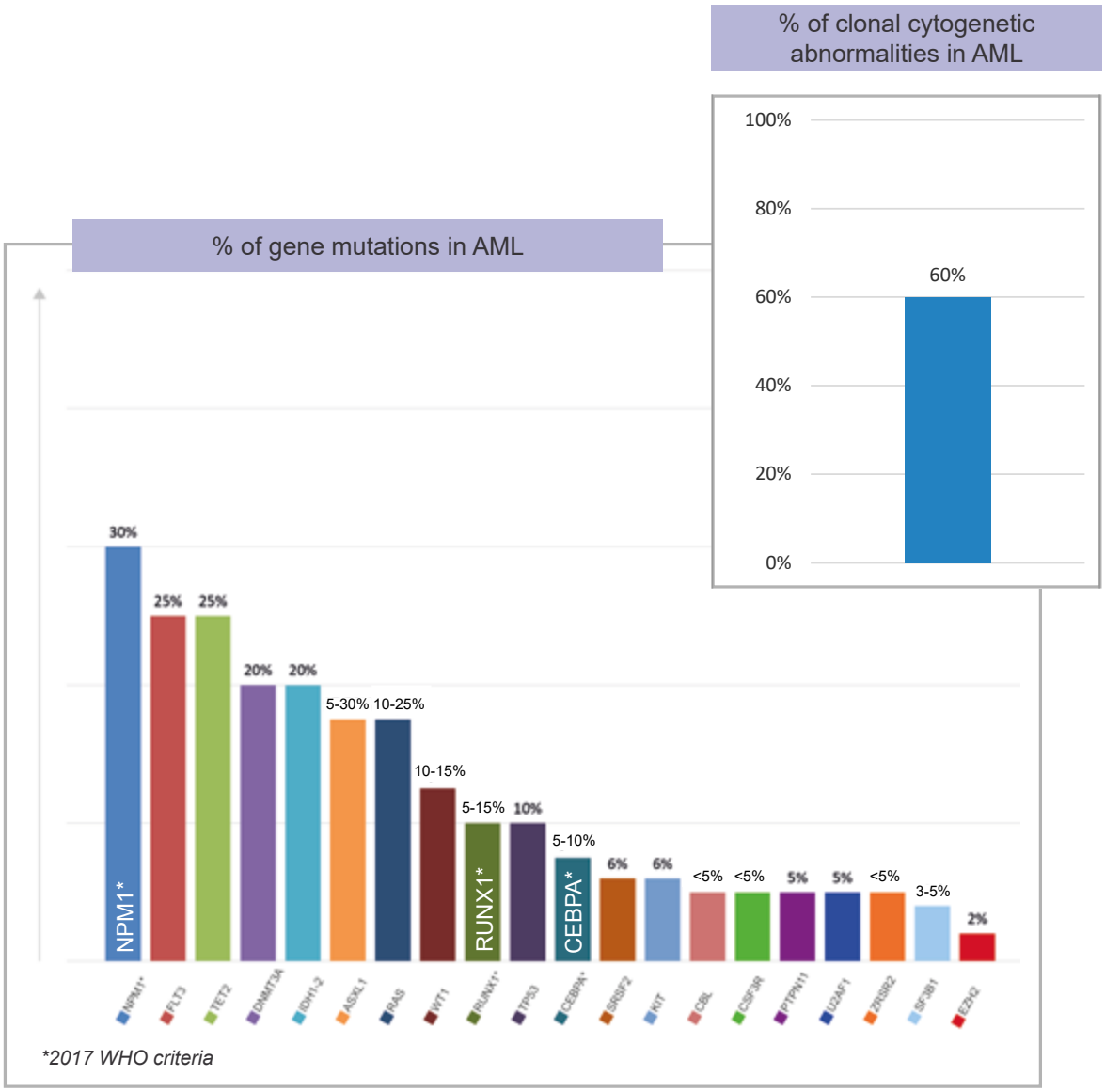
Gene mutations and clonal cytogenetic abnormalities in AML



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“AML” NGS Panel –Targeted genes

Gene	Transcript	Exon rank
CEBPA	NM_004364	Full coding region
CSF3R	NM_000760	Full coding region
DNMT3A	NM_022552	Full coding region
ETV6	NM_001987	Full coding region
EZH2	NM_001203247	Full coding region
JAK2	NM_004972	Full coding region
RUNX1	NM_001754	Full coding region
TET2	NM_001127208	Full coding region
TP53	LRG_TP53 (LRG-specific mixed numbering)	Full coding region
ZRSR2	NM_005089	Full coding region
ASXL1	NM_015338	9, 11, 12, 14
BRAF	NM_004333	15
CALR	NM_004343	9
CBL	NM_005188	8, 9
FLT3	NM_004119	13, 14, 15, 20

Gene	Transcript	Exon rank
HRAS	NM_176795	2, 3
IDH1	NM_005896	4
IDH2	NM_002168	4
KIT	NM_000222	2, 8, 9, 10, 11, 13, 17, 18
KRAS	NM_033360	2, 3
MPL	NM_005373	10
NPM1	NM_002520	10, 11
NRAS	NM_002524	2, 3
PTPN11	NM_002834	3, 7, 8, 9, 10, 11, 12, 13
SETBP1	NM_015559	4
SF3B1	NM_012433	10, 11, 12, 13, 14, 15, 16
SRSF2	NM_003016	1
U2AF1	NM_006758	2, 6
WT1	NM_024426	6, 7, 8, 9, 10

Test code: MYSLA

Pre-analytical: 2 ml EDTA bone marrow or 2 x 5 ml EDTA whole blood if blast infiltration into the periphery is significant.

Turnaround time: 10 days (Results may require an extended turnaround time of one week, depending on the confirmation tests required by Sanger sequencing)

Contact

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References

1. Swerdlow SH, Campo E, Harris NL, Jaffe ES, Pileri SA, Stein H, Thiele J. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues (Revised 4th edition) IARC Lyon 2017
2. Döhner H. et al, Diagnosis and management of AML in adults: 2017 ELN recommendations from an international expert panel. Blood. 2017 Jan 26;129(4):424-447

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