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Myelodysplastic syndromes (MDS)

The "**MDS**" 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 **diagnosis, prognosis and theranostics.**

- The main purpose is **prognosis**, allowing the definition of the molecular factors with a poor (TP53, EZH2, ETV6, RUNX1, ASXL1 mutations) or good prognosis (SF3B1 mutation), which can be used to guide the treatment strategy. It can help with predicting a treatment response (e.g. Effectiveness of hypomethylating agents: positive impact of a TET2 mutation, negative impact of a ASXL1 mutation or unmutated TET2 or DNMT3A, for a response to EPO: positive impact of a SF3B1 mutation, negative impact of a TP53 mutation, for a 5q- syndrome treated with lenalidomide: negative impact of a TP53 mutation).
- This panel can also address the **theranostic** impact (IDH1, IDH2, SF3B1 therapeutic targets).
- Currently, the **diagnostic** assistance of the "MDS" NGS panel is limited to a clinically suspected myelodysplasia without suggestive bone marrow cytology (absence of dysmyelopoiesis) and without the presence of cytogenetic clonal abnormalities suggestive of a MDS (karyotype without clonal chromosomal abnormality). The only two exceptions are the 'myelodysplastic/myeloproliferative neoplasm with ring sideroblasts and thrombocytosis' entity for which one of the WHO 2017 criteria is the presence of a SF3B1 mutation and the 'MDS with ring sideroblasts - SF3B1 mutated' entity in which this diagnosis can be proposed from 5% bone marrow sideroblasts (and not 15%).

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

The "MDS" NGS 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 compared for the diagnosis and/or prognosis of a malignant haemopathy.

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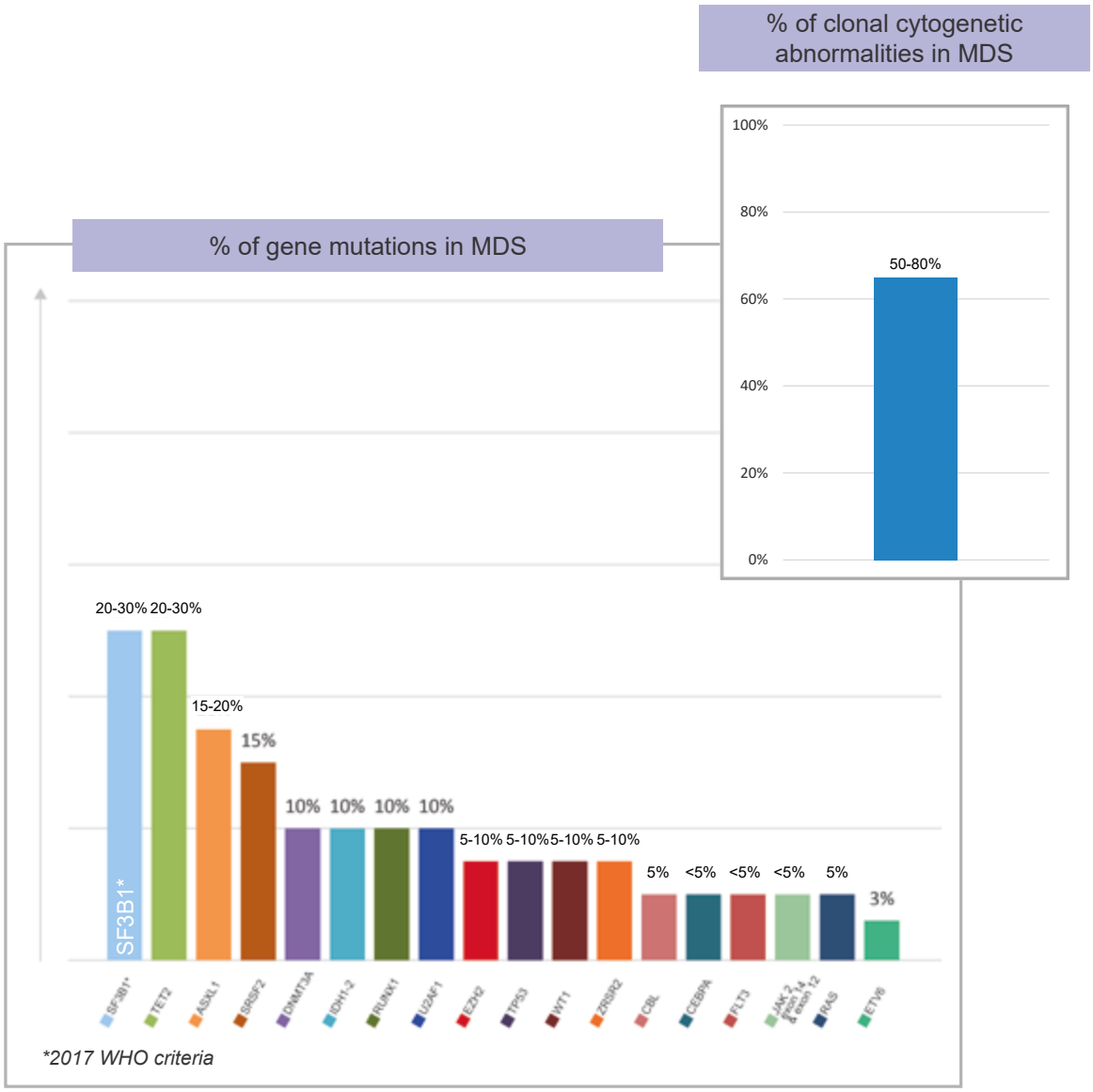
Gene mutations and clonal cytogenetic abnormalities in MDS



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“MDS” 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: MYSMD

Pre-analytical requirements: 2 ml EDTA bone marrow

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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Reference

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

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