

Specifications Sanquin Molecular diagnostics for MDS/MPN (X068)

Design AML-MDS NGS Panel

The AML-MDS Panel (IAD245850_243), request code X068, exists of 348 amplicons and is in silico covering 100% of submitted areas (all coding regions (exons)) and is able to analyze variants in 31 genes related to AML. Indicated exons (Table 1) include flanking intronic regions based on 10 base exon padding. For some genes only a hotspot location is covered. See Table 1 and 2 for detailed coverage information about these aberrant regions.

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Table 1. Design AML-MDS panel



Table 2. Aberrant covered regions AML-MDS panel

Gene	Exon	Coding DNA region
SETBP1	4	c.2477-c.2760

Coverage of the NGS AML-MDS Panel

Coverage is the number of times a base is sequenced. The deeper the coverage of each base the greater the reliability and sensitivity of the sequencing assay. The minimum depth of coverage required for detection of somatic variants with the AML-MDS AmpliSeq Panel is 500X. The percentage of Target Base coverage (%Base500x) is the percentage of target bases in a panel that is covered at least 500 times.

The percentage of target bases that is covered at least 500 times (%Base500x) is at least 99% at 2.200.000 Mapped Reads. With this acceptance criteria two regions failed to yield >500 times coverage, specific locations are listed in Table 3. One amplicon covering exon 1 in ASXL1 failed to reach >500 times coverage. A region in the middle of exon 1 of CEBPA also did not reach >500 times coverage. This does **not** concern the bZIP domain of CEBPA, which is used for the AML risk classification. The bZIP domain is covered >500 times. In the regions of ASXL1 and CEBPA that are covered <500 times, germline variants will be called, but for somatic variants the coverage might be too low.

	GRCh37/hg19 coordinates				
Chr	Start	End	Gene	Number of bases	Exon
Chr20	30946569	30946645	ASXL1	77	1
Chr19	33792690	33793122	СЕВРА	433	1

Table 3. Information amplicons AML-MDS panel that fail to reach >500 times coverage



Reporting: addition hematological malignancies variants

This test does not distinguish between somatic and germ line alterations in analyzed gene regions, particularly when variant allele frequencies (VAF) are near 50% or 100%. If nucleotide alterations in genes associated with germline mutation syndromes are present and there is also a strong clinical suspicion or family history of malignant disease predisposition, appropriate genetic counselling may be indicated.

Variants detected between 5% and 10% may indicate subclonal tumor populations. However, the clinical significance of these findings may not always be distinct. It is demonstrated that in blood DNA samples from individuals with advancing age and who do not have a hematologic neoplasm, a low incidence of gene variants that are associated with myeloid neoplasms can be detected. This phenomenon of clonal hematopoiesis of indeterminate potential (CHIP) may not be clearly distinguishable from tumor-associated mutations, especially if detected as a sole abnormality (DOI: 10.1182/blood-2015-03-631747).

Correlation with clinical, histopathologic and additional laboratory findings is required for final interpretation of the results. The final interpretation of results for clinical management of the patient is the responsibility of the managing physician.

NGS data are interpreted with the current knowledge concerning variants in relation to disease or as explanation of a phenotype. For reporting variants, the following guidelines will be followed: 'Best Practice Guidelines for Reporting Molecular Genetics results' written by R.J.L. Treacy and D.O. Robinson. The authorization of the results is done by a recognized Clinical molecular geneticist. All variants are annotated and reported as designated by the Human Genome Variation Society (HGVS) nomenclature, as described at <u>their</u> website.