

## Overzicht van onderzochte genen/exonen van het Myeloid NGS haloplex HS panel.

### Referentiegenoom GRCh37(hg19)

Gen	RefSeq	LRG	Exon(en)	Type	Indicatie (WHO klasse)
ASXL1	NM_015338.5	LRG_630 t1	13 (laatste exon)	Tumor Suppressor	AML, MDS, MDS/MPN, MPN, mastocytose
BCOR	NM_017745.5		Alle	Tumor Suppressor	MDS, AML
CALR	NM_004343.3	LRG_828 t1	9	Oncogen	MPN, MDS/MPN, mastocytose
CBL	NM_005188.3	LRG_608 t1	8-9	Oncogen/ Tumor Suppressor	MDS, MDS/MPN, mastocytose
CEBPA	NM_004364.3	LRG_456 t1	1	Tumor Suppressor	AML
CSF3R	NM_156039.3	LRG_144 t1	14,17	Oncogen	MPN, MDS/MPN
DNMT3A	NM_175629.2	LRG_459 t4	4,8-23	Tumor Suppressor	AML, MDS, MDS/MPN, MPN
ETNK1	NM_018638.4		3	Oncogen	MDS/MPN
EZH2	NM_004456.4	LRG_531 t1	Alle	Tumor Suppressor/ Oncogen	AML, MDS, MPN, MDS/MPN
FLT3	NM_004119.2	LRG_457 t1	14-15, 20	Oncogen	AML
IDH1	NM_005896.3	LRG_610 t1	4	Oncogen/ Tumor Suppressor	AML, MPN
IDH2	NM_002168.3	LRG_611 t2	4	Oncogen	AML, MPN, mastocytose
JAK2	NM_004972.3	LRG_612 t1	12,14	Oncogen	MPN, MDS/MPN, mastocytose
KIT	NM_000222.2	LRG_307 t1	2,8-11,13,17	Oncogen	AML, mastocytose
KMT2A	NM_005933.3		3-9	Tumor Suppressor	AML
KRAS	NM_004985.4		Alle	Oncogen	AML, MDS/MPN, mastocytose
MPL	NM_005373.2	LRG_510 t1	10	Oncogen	MPN, MDS/MPN, mastocytose
NPM1	NM_002520.6	LRG_458 t1	11	Oncogen	AML
NRAS	NM_002524.3	LRG_92 t1	2-3	Oncogen	AML, MDS/MPN, mastocytose
RUNX1	NM_001754.4	LRG_482 t1	Alle	Tumor Suppressor	AML, MDS, MDS/MPN, mastocytose
SETBP1	NM_015559.3		4	Oncogen	MDS/MPN
SF3B1	NM_012433.3	LRG_624 t2	13-17	Oncogen	AML, MPN, MDS, mastocytose
SRSF2	NM_003016.4	LRG_640 t1	1	Oncogen	AML, MPN, MDS, MDS/MPN, mastocytose
STAG2	NM_001042749.2	LRG_782 t1	Alle	Tumor Suppressor	AML
TET2	NM_001127208.2	LRG_626 t1	3, 9-11	Tumor Suppressor	AML, MDS, MPN, MDS/MPN, mastocytose
TP53	NM_000546.5	LRG_321 t1	3-9	Tumor Suppressor	AML, MDS, MPN
U2AF1	NM_006758.2	LRG_615 t2	2,6-7	Oncogen	AML, MDS
WT1	NM_024426.4		7-9	Tumor Suppressor/ Oncogen	AML
ZRSR2	NM_005089.3	LRG_618 t1	Alle	Tumor Suppressor	MDS

AML= Acute myeloïde leukemie, MDS = Myelodysplastisch syndroom, MPN = myeloproliferatieve neoplasië,  
LRG= Locus Reference Genomic

Arber et al Blood 2016, Papaemmanuil et NEJM 2016, WHO 2017

De volgende regio's zijn systematische drop out regio's: voor deze regio's wordt er systematisch onvoldoende diep gesequeneerd en kan er niet worden geanalyseerd:

Gene	exon	cDNA	proteïn
CEBPA	1	c.340_380	p.114-p.127
CEBPA	1	c.527_530	p.176-p.177
CEBPA	1	c.605_641	p.202-p.214
CEBPA	1	c.735_743	p.245-p.248
EZH2	7	c.654_728	p.218-p.243
KRAS	5	c.558_567	p.186-p.189
RUNX1	9	c.1314_1363	p.439-p.455
RUNX1	9	c.1424_1443	p.475-p.481
STAG2	17	c.1535_1560	p.513-p.521
TP53	4	c.227_235	p.76-p.79