Panel Design

Gen	Transkript	Genomische Lokalisation	Abdeckung (Exon)	Hotspot Mutationen
ASXL1	NM_015338	chr20:30946146-31027122	1-13	
CALR	NM_004343	chr19:13049413-13055304	8,9	52bp-Deletion (Typ 1) / 5bp-Insertion in Exon 9 (Typ 2)
CSF3R	NM_000760	chr1:36931643-36948915	14,15,16	Kodon 618
	NM_156039	chr1:36931643-36948915	17	
	NM_172313	chr1:36931643-36948915	10,18	
EZH2	NM_004456	chr7:148504463-148581441	2-20	
IDH1	*NM_005896*	chr2:209100952-209118910	3,4	Kodon 132
IDH2	NM_002168	chr15:90627211-90645708	4,6	Kodon 140 und 172
JAK2	NM_004972	chr9:4985244-5128183	Alle Exone	Kodon 617
MPL	NM_015559	chr18:42260862-42648475	Alle Exone	Kodon 515
SETBP1	NM_003016	chr17:74730196-74733493	4	Kodon 868
SF3B1	NM_012433	chr2:198256697-198299771	13-21	Kodon 666 und 700
SRSF2	NM_006758	chr21:44513065-44527688	1	Kodon 95
TET2	NM_017628	chr4:106067031-106163928	3	
	NM_001127208	chr4:106067841-106200960	4-11	
U2AF1	NM_006758	chr21:44513065-44527688	2, 6	Kodon 34 und 157

Beschreibung der untersuchten Gene

Gen	Funktion	TSG/Onkogen*	Ref.
ASXL1 (Additional Sex Combs Like 1)	Epigenetische Modifikation (Histon-Methylierung)	TSG	[1, 2]
CALR	ER Chaperon	Onkogen	[3] [4, 5]
CSF3R	Signaltransduktion (Regulation von Zellproliferation und Differenzierung von Granulozyten)	Onkogen	[6] [7, 8]
EZH2	Epigenetische Modifikation (Methylierung)	TSG / Onkogen	[9, 10]
IDH1 (Isocitrat-Dehydrogenase 1)	Epigenetische Modifikation (Methylierung)	Onkogen	[6] [7, 8]
IDH2 (Isocitrat-Dehydrogenase 1)	Epigenetische Modifikation (Methylierung)	Onkogen	[6] [7, 8]
JAK2 *(Janus Kinase 2)*	Signaltransduktion *(JAK-STAT-Signalweg)*	Onkogen	[11-14]
MPL	Thrombopoetinrezeptor des *JAK-STAT*-Signalwegs	Onkogen	[15, 16]
SETBP1	Epigenetische Modifikation (Teil eines Proteinkomplexes involviert in der Histon-Methylierung)	Onkogen	[17, 18]
SF3B1 *(Splicing Factor 3B Subunit 1)*	Spleiss-Faktor	Onkogen	[19, 20] [21, 22]
SRSF2 *(Serine and Arginine Rich Splice Factor 2)*	Spleiss-Faktor	TSG / Onkogen	[19] [21, 22]

* Gemäss OncoKB Cancer Gene List, TSG=Tumor Suppressor Gen

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