

Panel Design

Gen	Transkript	Genomische Lokalisation	Abdeckung (Exon)	Hotspot Mutationen
ASXL1	NM_015338	chr20:30946146-31027122	1-13	
	NM_001164603	chr20:30946146-30960352	4	
CBL	NM_005188	chr11:119076985-119178859	2-5, 7-9, 16	
CHEK2	NM_001005735	chr22:29083730-29137822	3	
	NM_007194	chr22:29083730-29137822	2-15	
DNMT3A	NM_022552	chr2:25455829-25564784	2-23	Kodon 882
	NM_175630	chr2:25504320-25565459	4	
	NM_153759	chr2:25455829-25475184	1, 2	
GNAS	NM_000516	chr20:57466425-57486250	8-11	Kodon 201
GNB1	NM_002074	chr1:1716724-1822526	3-11	
JAK2	NM_004972	chr9:4985244-5128183	12-16, 19-25	Kodon 617
PPM1D	NM_003620	chr17:58677543-58743640	6	
RAD21	NM_006265	chr8:117858172-117887105	2-14	
SF3B1	NM_012433	chr2:198256697-198299771	13-21	Kodon 666 und 700
SRSF2	NM_003016	chr17:74730196-74733493	1, 2	Kodon 95
TET2	NM_017628	chr4:106067031-106163928	3	
	NM_001127208	chr4:106067841-106200960	4-11	
TP53	NM_001276696	chr17:7571719-7590868	10 + UTR	
	NM_000546	chr17:7571719-7590868	1-11 + UTR	Kodon 175, 248, 273

Beschreibung der untersuchten Gene

Gen	Funktion	TSG/Onkogen*	Ref.
ASXL1 (Additional Sex Combs Like 1)	Epigenetische Modifikation (Histon-Methylierung)	TSG	[1, 2]
CBL (Casitas B-cell lymphoma)	Signaltransduktion (E3 Ubiquitin-Protein Ligase)	TSG	[3, 4]
CHEK2 (Checkpoint-Kinase 2)	DNA-Reparatur & Zellzykluskontrolle	TSG	
DNMT3A (DNA Methyltransferase 3A)	Epigenetische Modifikation (DNA-Methylierung)	TSG	[5] [6-8]
GNAS (G-Protein Alpha Subunit)	Signaltransduktion	Onkogen	
GNB1 (Guanin Nukleotid Bindeprotein)	Signaltransduktion	Onkogen	
JAK2 (Janus Kinase 2)	Signaltransduktion (JAK-STAT-Signalweg)	Onkogen	[9-12]
PPM1D (Protein Phosphatase 1D)	DNA-Reparatur	Onkogen	[13]
RAD21 (RAD21 cohesin complex component)	Zellteilung / DNA Reparatur (Kohesin-Komplexes)	TSG	[14]
SF3B1 (Splicing Factor 3B Subunit 1)	Spleiss-Faktor	Onkogen	[15, 16] [17, 18]
SRSF2 (Serine and Arginine Rich Splice Factor 2)	Spleiss-Faktor	TSG / Onkogen	[15] [17, 18]
TET2 (TET Methylcytosin Dioxygenase 2)	Epigenetische Modifikation (De-Methylierung)	TSG	[19, 20]
TP53	DNA-Reparatur	TSG	[21] [17, 18]

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

Literatur

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