

Panel Design

Gen	Transkript	Genomische Lokalisation	Abdeckung (Exon)	Hotspot Mutationen
ASXL1	NM_015338	chr20:30946146-31027122	1-13	
	NM_001164603	chr20:30946146-30960352	4	
DNMT3A	NM_153759	chr2:25455829-25475184	1,2	Kodon 882
	NM_022552	chr2:25455829-25564784	2-23	Kodon 882
GNAS	NM_000516	chr20:57466425-57486250	8-11	Kodon 201
IDH2	NM_002168	chr15:90627211-90645708	4,6	Kodon 140 und 172
SF3B1	NM_012433	chr2:198256697-198299771	13-21	Kodon 666 und 700
SH2B3	NM_005475.2	chr12:111843751-111889427	2-8	
SRSF2	NM_003016	chr17:74730196-74733493	1,2	Kodon 95
TET2	NM_001127208	chr4:106067031-106163928	4-11	
	NM_017628	chr4:106067841-106200960	3	
TP53	NM_000546	chr17:7571719-7590868	1	
	NM_000546	chr17:7571719-7590868	2-11	
	NM_001276696	chr17:7571719-7590868	10	
U2AF1	NM_006758	chr21:44513065-44527688	2,6,7	Kodon 34 und 157
UBA1	NM_003334	chrX:47053200-47074527	3	Kodon 41

Beschreibung der untersuchten Gene

Gen	Funktion	TSG/Onkogen*	Ref.
ASXL1 (Additional Sex Combs Like 1)	Epigenetische Modifikation (Histon-Methylierung)	TSG	[1, 2]
DNMT3A (DNA Methyltransferase 3A)	Epigenetische Modifikation (DNA-Methylierung)	TSG	[3] [4-6]
GNAS (G-Protein Alpha Subunit)	Signaltransduktion	Onkogen	
IDH2 (Isocitrat-Dehydrogenase 1)	Epigenetische Modifikation (Methylierung)	Onkogen	[7] [8, 9]
SF3B1 (Splicing Factor 3B Subunit 1)	Spleiss-Faktor	Onkogen	[10, 11] [12, 13]
SH2B3	Signaltransduktion (negative Regulation von JAK-STAT-Signalweg)	TSG	[14]
SRSF2 (Serine and Arginine Rich Splice Factor 2)	Spleiss-Faktor	TSG / Onkogen	[10] [12, 13]
TET2 (TET Methylcytosin Dioxygenase 2)	Epigenetische Modifikation (De-Methylierung)	TSG	[15, 16]
TP53	DNA-Reparatur	TSG	[17] [12, 13]
U2AF1 (U2 Small Nuclear RNA Auxiliary Factor 1))	Spleiss-Faktor	Onkogen	[10, 18]
UBA1 (Ubiquitin-activating enzyme))	DNA-Reparatur		[19]

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

Literatur

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