

## Panel Design

Gen	Transkript	Genomische Lokalisation	Abdeckung (Exon)	Hotspot Mutationen
ASXL1	NM_015338	chr20:30946146-31027122	1-13	
	NM_001164603	chr20:30946146-30960352	4	
BCOR	NM_017745	chrX:39910498-39956719	2-15	
CEBPA	NM_004364	chr19:33790839-33793430	1	
DDX41	NM_016222	chr5:176938577-176943967	1-17	
EZH2	NM_004456	chr7:148504463-148581441	2-20	
FLT3	NM_004119	chr13:28577410-28674729	14-20	Kodon 835
IDH1	NM_005896	chr2:209100952-209118910	3,4	Kodon 132
IDH2	NM_002168	chr15:90627211-90645708	4,6	Kodon 140 und 172
NPM1	NM_002520	chr5:170814707-170837888	12	Insertion eines Tetranukleotids in Kodon 288
RUNX1	NM_001122607	chr21:36193573-36260987	5	
	NM_001754	chr21:36160097-36421462	2-9	
SF3B1	NM_012433	chr2:198256697-198299771	13-18	Kodon 666 und 700
SRSF2	NM_003016	chr17:74730196-74733493	1-2	Kodon 95
STAG2	NM_001042749	chrX:123094474-123236505	32	
	NM_006603	chrX:123095555-123236505	2-33	
TP53	NM_001276696	chr17:7571719-7590868	10	
	NM_000546	chr17:7571719-7590868	2-11	Kodon 175, 220, 248, 273*
	NM_000546	chr17:7571719-7590868	1 + UTR	
U2AF1	NM_006758	chr21:44513065-44527688	1-8	Kodon 34 und 157
	NM_001025203	chr21:44513065-44527688	3	
ZRSR2	NM_005089	chrX:15808573-15841382	1-11	

## Beschreibung der untersuchten Gene

Gen	Funktion	TSG/Onkogen*	Ref.
ASXL1 (Additional Sex Combs Like 1)	Epigenetische Modifikation (Histon-Methylierung)	TSG	[1, 2]
BCOR (BCL6 corepressor)	Regulation der Transkription	TSG	[3] [4]
CEBPA (CCAAT/enhancer binding protein)	Transkriptionsfaktor	TSG	[5] [6, 7]
DDX41 (DEAD-Box Helikase 41)	RNA Prozessierung	TSG	[8, 9]
EZH2	Epigenetische Modifikation (Methylierung)	TSG / Onkogen	[10, 11]
FLT3 (Fms-related tyrosine kinase 3)	Signaltransduktion (Tyrosinkinase)	Onkogen	[12, 13] [14]
IDH1 (Isocitrat-Dehydrogenase 1)	Epigenetische Modifikation (Methylierung)	Onkogen	[15] [16, 17]
IDH2 (Isocitrat-Dehydrogenase 1)	Epigenetische Modifikation (Methylierung)	Onkogen	[15] [16, 17]
NPM1 (Nucleophosmin)	Phosphoprotein mit einer Vielzahl von Aufgaben	TSG	[10, 11]
RUNX1 (Runt-related transcription factor 1)	Transkriptionsfaktor	TSG	[18, 19]
SF3B1 (Splicing Factor 3B Subunit 1)	Spleiss-Faktor	Onkogen	[20, 21] [22, 23]
SRSF2 (Serine and Arginine Rich Splice Factor 2)	Spleiss-Faktor	TSG / Onkogen	[20] [22, 23]
STAG2 (Stromal antigen 2)	Chromosomensegregation	TSG	[24]
TP53	DNA-Reparatur	TSG	[25] [22, 23]
U2AF1 (U2 Small Nuclear RNA Auxiliary Factor 1)	Spleiss-Faktor	Onkogen	[20, 26]
ZRSR2	Spleiss-Faktor		

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

## Literatur

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