

 Universitätsklinikum Leipzig Medizin ist unsere Berufung.	Anlage 01 DNA Panel Solid Tumor Genliste
Institut für Pathologie Molekularpathologie	Liste Genbereiche Stand 01.03.2020

Auflistung der 210 Genbereiche, des Custom Actionable Solid Tumor Panel CDHS-36709Z-1345 (Qiagen) mit insgesamt 19.472 Basen kodierende Sequenz

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
AKT1	chr14	NM_005163.2	AKT1 Exon 3	105.246.541	105.246.562	16 - 20	22	9	COSM33765: c.49G>A (E17K)
							22	13	Basen kodierende Sequenz gesamt
ALK	chr2	NM_004304.4	ALK Exon 5	29.606.669	29.606.689	398 - 404	21		COSM20713: c.1201C>T (R401*)
			ALK Exon 8	29.541.181	29.541.201	539 - 545	21		COSM48230: c.1625C>G (P542R)
			ALK Exon 15	29.455.257	29.455.277	842 - 848	21		COSM148825: c.2535T>C (G845G)
			ALK Exon 18	29.449.809	29.449.829	1009 - 1015	21		COSM148824: c.3036G>A (T1012T)
			ALK Exon 19	29.448.400	29.448.420	1027 - 1033	21		COSM4133807: c.3089A>C (H1030P)
			ALK Exon 20	29.446.286	29.446.306	1088 - 1094	21		COSM28502: c.3271G>A (D1091N)
			ALK Exon 22	29.445.246	29.445.283	1151 - 1160	38	9	COSM98478: c.3452C>T (T1151M)
			ALK Exon 22	29.445.182	29.445.223	1168 - 1172	42	28	COSM28498: c.3512T>A (I1171N)
			ALK Exon 23	29.443.684	29.443.707	1172 - 1178	24	6	COSM28055: c.3522C>A (F1174L)
			ALK Exon 23	29.443.597	29.443.641	1193 - 1207	45		COSM99137: c.3586C>A (L1196M)
			ALK Exon 24	29.436.848	29.436.870	1242 - 1248	23	2	COSM28499: c.3733T>G(F1245V)
			ALK Exon 25	29.432.645	29.432.693	1266 - 1279	49	7	COSM28056: c.3824G>A (R1275Q)
			ALK Exon 29	29.416.605	29.416.625	1443 - 1449	21		COSM5019786: c.4338C>T (T1446T)

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			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			ALK Exon 29	29.416.471	29.416.491	1488 - 1494	21		COSM1130802: c.4472A>G (K1491R)
							389	337	Basen kodierende Sequenz gesamt
BRAF	chr7	NM_004333.4	BRAF Exon 7	140.500.155	140.500.175	323 - 327	21		COSM33964: c.977T>C (I326T)
			BRAF Exon 11*	140.481.376	140.481.503	439 - 477	128	10	COSM460: c.1406G>C (G469A)
			BRAF Exon 12*	140.477.781	140.477.885	478 - 506	105	20	COSM243226: c.1514T>A (L505H)
			BRAF Exon 15*	140.453.065	140.453.203	581 - 620	139	20	COSM476: c.1799T>A (V600E)
			BRAF Exon 16	140.449.140	140.449.180	634 - 646	41		COSM4161857: c.1929A>G (G643G)
							434	384	Basen kodierende Sequenz gesamt
CALR	chr19	NM_004343.3	CALR Exon 9*	13.054.527	13.054.727	352 - 418	201		COSM1738055: c.1099_1150del (L367Tfs*46)
							201	201	Basen kodierende Sequenz gesamt
CTNNB1	chr3	NM_001098209.1	CTNNB1 Exon 3*	41.266.007	41.266.254	5 - 81	248	20	COSM5664: c.121A>G (T41A); COSM5667: c.134C>T (S45F)
							248	228	Basen kodierende Sequenz gesamt
EGFR	chr7	NM_005228.3	EGFR Exon 2	55.210.065	55.210.087	59 - 66	23		COSM35602: c.185T>G (L62R)
			EGFR Exon 3	55.211.070	55.211.107	105 - 117	38		COSM21683: c.323G>A (R108K)
			EGFR Exon 4	55.214.338	55.214.358	155 - 161	21		COSM42978: c.474C>T (N158N)
			EGFR Exon 6	55.220.264	55.220.284	219 - 225	21		COSM35508: c.664C>T (R222C)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			EGFR Exon 7	55.221.700	55.221.720	250 - 255	21	4	COSM35752: c.754C>T (R252C)
			EGFR Exon 7	55.221.733	55.221.753	260 - 266	21		COSM21684: c.787A>C (T263P)
			EGFR Exon 7	55.221.811	55.221.832	286 - 292	22		COSM21687: c.866C>T (A289V)
			EGFR Exon 8	55.223.533	55.223.553	301 - 307	21		COSM2149971: c.910C>T (H304Y)
			EGFR Exon 12	55.227.997	55.228.019	489 - 495	23		COSM236670: c.1476C>A (S492R)
			EGFR Exon 13	55.229.245	55.229.265	518 - 524	21		COSM3721608: c.1562G>A (R521K)
			EGFR Exon 14*	55.231.426	55.231.516	544 - 574	91		ClinVar376211: c.1636C>T (p.Pro546Ser)
			EGFR Exon 15	55.233.027	55.233.053	593 - 601	27		COSM21690: c.1793G>T (G598V)
			EGFR Exon 15	55.233.099	55.233.119	617 - 623	21		COSM35825: c.1859G>A (C620Y)
	NM_201284		EGFR Exon 16	55.238.077	55.238.097	653 - 659	21		COSM54529: c.1957A>T (S653C)
	NM_201284		EGFR Exon 16	55.238.102	55.238.122	662 - 668	21		COSM3942265: c.1993G>T (E665*)
			EGFR Exon 17	55.240.697	55.240.717	648 - 654	21		COSM35840: c.1951G>A (V651M)
			EGFR Exon 17	55.240.776	55.240.816	674 - 687	41		COSM22555: c.2030G>A (R677H)
			EGFR Exon 18*	55.241.617	55.241.637	689 - 695	21		COSM13178: c.2081C>T (P694L)
			EGFR Exon 18*	55.241.646	55.241.736	699 - 728	91		COSM6239: c.2156G>C (G719A)
			Intron 18-19	55.241.745	55.241.765		21	21	
			EGFR Exon 19*	55.242.405	55.242.523	729 - 761	119	20	COSM6223: c.2235_2249del15 (E746_A750del)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			EGFR Exon 20*	55.248.976	55.249.181	762 - 823	206	20	COSM6240: c.2369C>T (T790M)
			EGFR Exon 21*	55.259.402	55.259.577	824 - 875	176	20	COSM6224: c.2573T>G (L858R)
			EGFR Exon 22	55.260.473	55.260.493	881 - 887	21		COSM28575: c.2650G>A (E884K)
			EGFR Exon 23	55.266.459	55.266.481	918 - 924	23		COSM28515: c.2763C>A (S921R)
			EGFR Exon 25	55.268.906	55.268.926	991 - 997	21		COSM3762773: c.2982C>T (D994D)
							1205	1120	Basen kodierende Sequenz gesamt
ERBB2	chr17	NM_004448.2	Intron	37.855.803	37.855.850		48	48	
			ERBB2 Exon 8*	37.868.171	37.868.310	301 - 341	140	20	COSM48358: c.929C>T (S310F)
			ERBB2 Exon 17*	37.879.562	37.879.720	649 - 695	159	20	COSM436498: c.2033G>A (R678Q); COSM4000121: c.1963A>G (I655V)
			ERBB2 Exon 19*	37.880.155	37.880.273	737 - 769	119	20	COSM14060: c.2264T>C (L755S); COSM1251412: c.2305G>T (D769Y)
			ERBB2 Exon 20*	37.880.969	37.881.174	770 - 831	206	20	COSM14062: c.2329G>T (V777L); COSM20959: c.2324_2325ins12
			ERBB2 Exon 21*	37.881.292	37.881.467	832 - 883	176	20	COSM14065: c.2524G>A (V842I)
			ERBB2 Exon 22*	37.881.570	37.881.665	884 - 909	96	20	COSM978678: c.2690G>A (R897Q)
			ERBB2 Exon 23*	37.881.950	37.882.116	909 - 958	167	20	COSM6005645: c.2790G>T (E930D)
							1111	923	Basen kodierende Sequenz gesamt
ESR1	chr6	NM_000125.3	ESR1 Exon 1	152.129.067	152.129.087	7 - 13	21		COSM3761554: c.30T>C (S10S)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			ESR1 Exon 4	152.265.445	152.265.465	300 - 306	21		COSM4745827: c.908A>G (K303R)
			Intron	152.396.026	152.396.046		21	21	
			ESR1 Exon 8	152.419.912	152.419.936	534 - 541	25		COSM94250: c.1613A>G (D538G); COSM1074639: c.1610A>C (Y537S)
			ESR1 Exon 8	152.420.085	152.420.105	591 - 596*	21	4	COSM3761556: c.1782G>A (T594T)
							109	84	Basen kodierende Sequenz gesamt
FGFR1	chr8	NM_023110.2	FGFR1 Exon 11*	38.275.509	38.275.388	477 - 518	122		41 Mutationen in COSMIC, aber keine Hotspots
			FGFR1 Exon 12*	38.274.934	38.274.824	518 - 555	101		26 Mutationen in COSMIC, aber keine Hotspots
			FGFR1 Exon 13*	38.273.578	38.273.388	555 - 618	191		COSM19176: c.1731C>A (N577K)
			FGFR1 Exon 14*	38.272.419	38.272.297	619 - 659	123		34 Mutationen in COSMIC, aber keine Hotspots
			FGFR1 Exon 15*	38.272.147	38.272.077	660 - 683	71		22 Mutationen in COSMIC, aber keine Hotspots
			FGFR1 Exon 16*	38.271.807	38.271.670	683 - 729	138		COSM35673: c.2059A>G (K687E)
							746	746	Basen kodierende Sequenz gesamt
FGFR2	chr10	NM_022970.3	FGFR2 Exon 7*	123.279.683	123.279.493	250 - 313	191		COSM36903: c.755C>G (S252W)
			FGFR2 Exon 8*	123.278.343	123.278.196	314 - 363	148		29 Mutationen in COSMIC, aber keine Hotspots
			FGFR2 Exon 9*	123.274.833	123.274.631	363 - 430	203		COSM36904: c.1127A>G (Y376C) COSM36906: c.1147T>C (C383R)
			FGFR2 Exon 12*	123.258.119	123.258.009	522 -559	111		COSM36912: c.1650T>A (N550K)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)					Wichtige Mutationen (nach COSMIC)	
			Name	Start	Stopp	Aminosäuren	Größe target region		
			FGFR2 Exon 13*	123.256.236	123.256.046	559 - 622	191		53 Mutationen in COSMIC, aber keine Hotspots
			FGFR2 Exon 14*	123.247.627	123.247.505	623 - 663	123		COSM36909: c.1978A>G (K660E) COSM683053: c.1980G>C (K660N)
			FGFR2 Exon 15*	123.246.938	123.246.868	664 - 687	71		21 Mutationen in COSMIC, aber keine Hotspots
			FGFR2 Exon 16*	123.245.046	123.244.909	687 - 733	138		COSM6854598: c.2183A>G (N728S)
			FGFR2 Exon 17*	123.243.317	123.243.212	733 - 768	106		28 Mutationen in COSMIC, aber keine Hotspots
							1282	1282	Basen kodierende Sequenz gesamt
FGFR3	chr8	NM_023110.2	FGFR3 Exon 7*	1803562	1803752	247 - 310	191		
			FGFR3 Intron 8*	1805447	1805546		100	100	
			FGFR3 Exon 9*	1806057	1806247	361 - 424	191		
			FGFR3 Exon 14*	1807778	1807900	615 - 655	123		
			FGFR3 Exon 15*	1807984	1808054	656 - 678	71		
			FGFR3 Exon 16*	1808273	1808410	679 - 725	138		
							814	714	Basen kodierende Sequenz gesamt
FOXL2	chr3	NM_023067.3	FOXL2 Exon 1	138.665.153	138.665.174	131 - 137	22		COSM33661: c.402C>G (C134W)
							22	22	Basen kodierende Sequenz gesamt

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
GNA11	chr19	NM_002067.2	GNA11 Exon 5	3.118.932	3.118.953	206 - 212	22		COSM52969: c.626A>T (Q209L)
							22	22	Basen kodierende Sequenz gesamt
GNAQ	chr9	NM_002072.4	GNAQ Exon 5	80.409.478	80.409.500	205 - 212	23		COSM28757: c.626A>T (Q209L); COSM28758: c.626A>C (Q209P)
							23	23	Basen kodierende Sequenz gesamt
GNAS	chr20	NM_080425.3	GNAS Exon 8*	57.484.405	57.484.478	839 - 863	74		COSM27895: c.2531G>A (R844H) COSM27887: c.2530C>T (R844C)
			GNAS Exon 9*	57.484.576	57.484.634	863 - 883	59		COSM27888: c.2609A>T (Q870L) + weitere Q870
							133	133	Basen kodierende Sequenz gesamt
H3F3A	chr1	NM_002107.4	H3F3A Exon 2*	226.252.053	226.252.180	1 - 43	128		COSM327928: c.83A>T (K28M) + weitere K28 COSM1732355: c.103G>T (G35W) + weitere G35
							128	128	Basen kodierende Sequenz gesamt
HFE	chr6	NM_000410.3	HFE Exon 4*	26.092.903	26.093.198	206 - 298	296		50 Mutationen in COSMIC, aber keine Hotspots
							296	296	Basen kodierende Sequenz gesamt
HRAS	chr11	NM_005343.4	HRAS Exon 2*	534.322	534.212	1 - 37	111		COSM483: c.35G>T (G12V) + weitere G12 COSM486: c.37G>C (G13R) + weitere G13
			HRAS Exon 3*	533.944	533.766	38 - 97	179		COSM499: c.182A>G (Q61R)
			HRAS Exon 4*	533.612	533.453	97 - 150	160		COSM254733: c.351G>C (K117N) + weitere K117 COSM99642: c.355G>A (D119N) + weitere D119
							450	450	Basen kodierende Sequenz gesamt
IDH1	chr2	NM_005896.3	IDH1 Exon 4	209.113.103	209.113.124	128 - 135	22		COSM28746: c.395G>A (R132H); COSM28747: c.394C>T (R132C)

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			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
							22	22	Basen kodierende Sequenz gesamt
IDH2	chr15	NM_002168.2	IDH2 Exon 4	90.631.828	90.631.849	169 - 175	22		COSM33733: c.515G>A (R172K)
			IDH2 Exon 4	90.631.924	90.631.945	137 - 143	22		COSM41590: c.419G>A (R140Q)
							44	44	Basen kodierende Sequenz gesamt
JAK2	chr9	NM_004972.3	JAK2 Exon 12*	5069925	5070052	505 - 547	128		COSM24439: c.1615_1616inv (K539L) + weitere K539
			JAK2 Exon 13*	5072492	5072626	548 - 592	135		83 Mutationen in COSMIC, aber keine Hotspots
			JAK2 Exon 14*	5073698	5073785	593 - 622	88		COSM12600: c.1849G>T (V617F)
			JAK2 Exon 16*	5078306	5078444	665 - 711	139		COSM29300: c.2047A>G (R683G)
			JAK2 Exon 20*	5089674	5089863	858 - 921	190		183 Mutationen in COSMIC, aber keine Hotspots
							680	680	Basen kodierende Sequenz gesamt
KIT	chr4	NM_000222.2	KIT Exon 2	55.561.748	55.561.775	47 - 55	28		COSM1146: c.154G>A (D52N)
			KIT Exon 8	55.589.758	55.589.788	414 - 423	31		COSM29014: c.1255_1257delGAC (D419del)
			KIT Exon 9*	55.592.013	55.592.226	449 - 514	214	20	COSM1326: c.1509_1510insGCCTAT (Y503_F504insAY)
			KIT Exon 10	55.593.454	55.593.491	538 - 549	38	1	COSM28026: c.1621A>C (M541L)
			KIT Exon 11*	55.593.572	55.593.718	550 - 592	147	20	COSM1217: c.1669_1674del6 (W557_K558del)
			KIT Exon 13*	55.594.167	55.594.297	627 - 664	131	20	COSM1304: c.1924A>G (K642E), COSM12706: c.1961T>C (V654A)

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			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			KIT Exon 14*	55.595.491	55.595.661	664 - 714	171	20	COSM12708: c.2009C>T (T670I)
			KIT Exon 15	55.597.485	55.597.507	714 - 718	23	9	COSM1306: c.2143_2145delAGC (S715del)
			Intron	55.599.149	55.599.169		21	21	
			KIT Exon 17*	55.599.226	55.599.368	788 - 828	143	20	COSM1314: c.2447A>T (D816V)
			Intron	55.599.391	55.599.411		21	21	
			KIT Exon 18*	55.602.654	55.602.674	829 - 832	21	10	COSM13172: c.2485G>C (A829P)
			KIT Exon 18*	55.602.684	55.602.704	836 - 842	21		COSM1324: c.2515G>A (E839K)
			KIT Exon 18*	55.602.753	55.602.775	859 - 865	23		COSM1325: c.2586G>C (L862L)
			KIT Exon 20	55.603.362	55.603.382	907 - 913	21		COSM5609257: c.2732C>T (P911L)
			KIT Exon 21	55.604.649	55.604.669	953 - 959	21		COSM71350: c.2867G>A (R956Q)
							1075	913	Basen kodierende Sequenz gesamt
KRAS	chr12	NM_004985.3	KRAS 3'UTR	25.360.214	25.360.234		21	21	
			KRAS Exon 5	25.362.795	25.362.815	161 - 167	21		COSM41307: c.491G>A (p.R164Q)
			KRAS Exon 4*	25.378.538	25.378.717	97 - 150	180	20	COSM19404: c.436G>A (A146T)
			KRAS Exon 3*	25.380.158	25.380.356	38 - 97	199	20	COSM554: c.183A>C (Q61H)
			KRAS Exon 2*	25.398.198	25.398.339	1 - 37	142	20	COSM521: c.35G>A (G12D)

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			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
							563	482	Basen kodierende Sequenz gesamt
MET	chr7	NM_001127500.2	MET Exon 16*	116.417.443	116.417.523	1105 - 1132	81		47 Mutationen in COSMIC, aber keine Hotspots
			MET Exon 17	116.418.875	116.418.884	1147 - 1152	10		18 Mutationen in COSMIC, aber keine Hotspots
			MET Exon 17	116.419.101	116.419.016	1190 - 1192	16		5 Mutationen in COSMIC, aber keine Hotspots
			MET Exon 18*	116.422.042	116.422.151	1193 - 1229	110		34 Mutationen in COSMIC, aber keine Hotspots
			MET Exon 19	116.423.397	116.423.418	1243 - 1249	22		COSM699: c.3743A>G (Y1248C)
			MET Exon 19	116.423.464	116.423.485	1265 - 1271	22		COSM700: c.3757T>G (Y1253D) COSM691: c.3803T>C (M1268T)
							261	261	Basen kodierende Sequenz gesamt
MPL	chr1	NM_5373.2	MPL Exon 10*	43.814.934	43.815.030	490 - 522	97		COSM18918: c.1544G>T (W515L)
							97	97	Basen kodierende Sequenz gesamt
MYD88	chr3	NM_002468.4	MYD88 Exon 5*	38.182.623	38.182.777	259 - 310	155		COSM85940: c.818T>C (L273P)
							155	155	Basen kodierende Sequenz gesamt
NPM1	chr5	NM_002520.6	NPM1 Exon 11*	170.837.531	170.837.569	283 - 295	39		3.608 Mutationen in COSMIC, davon 3.442 frameshift insertions an Position c.859
							39	39	Basen kodierende Sequenz gesamt
NTRK1	chr1	NM_002529.3	NTRK1 Exon 15*	156.845.872	156.846.002	501 - 544	131		55 Mutationen in COSMIC, aber keine Hotspots
			NTRK1 Exon 16*	156.846.192	156.846.364	545 - 602	173		86 Mutationen in COSMIC, aber keine Hotspots

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			NTRK1 Exon 17*	156.848.914	156.849.154	602 - 682	241		109 Mutationen in COSMIC, aber keine Hotspots
							545	545	Basen kodierende Sequenz gesamt
NTRK2	chr9	NM_006180.4	NTRK2 Exon 19*	87.570.198	87.570.432	646 - 724	235		99 Mutationen in COSMIC, aber keine Hotspots
			NTRK2 Exon 20*	87.635.111	87.635.279	725 - 777	169		58 Mutationen in COSMIC, aber keine Hotspots
							404	404	Basen kodierende Sequenz gesamt
NTRK3	chr15	NM_002530.3	NTRK3 Exon 15*	88.483.984	88.483.854	529 - 572	131		76 Mutationen in COSMIC, aber keine Hotspots
			NTRK3 Exon 16*	88.476.415	88.476.243	573 - 630	173		127 Mutationen in COSMIC, aber keine Hotspots
			NTRK3 Exon 17*	88.472.675	88.472.412	630 - 711	264		134 Mutationen in COSMIC, aber keine Hotspots
							568	568	Basen kodierende Sequenz gesamt
NRAS	chr1	NM_002524.4	NRAS Exon 4*	115.252.180	115.252.359	97 - 150	180	20	COSM4170228: c.437C>T (A146V)
			NRAS Exon 3*	115.256.411	115.256.609	38 - 97	199	20	COSM584: c.182A>G (Q61R)
			NRAS Exon 2*	115.258.661	115.258.808	1 - 37	148	20	COSM564: c.35G>A (G12D)
							527	467	Basen kodierende Sequenz gesamt
PDGFRA	chr4	NM_006206.4	PDGFRA Exon 3	55.127.438	55.127.458	76 - 82	21		COSM5019287:c.236G>A (G79D)
			PDGFRA Exon 4	55.130.068	55.130.088	201 - 207	21		COSM4416371:c.612T>C (N204N)
			PDGFRA Exon 5	55.131.108	55.131.128	218 - 224	21		COSM6438148: c.661C>T (L221F)

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Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
			PDGFRA Exon 5	55.131.132	55.131.171	226 - 238	40		COSM42897: c.704G>A (C235Y)
			PDGFRA Exon 7	55.133.716	55.133.736	311 - 316	21	3	COSM1666925: c.939T>G (G313G)
			PDGFRA Exon 10	55.139.761	55.139.781	475 - 481	21		COSM5008347: c.1432T>C (S478P)
			PDGFRA Exon 12*	55.140.998	55.141.150	552 - 596	153	20	COSM12418: c.1698_1712del15; COSM739: c.1682T>A (V561D)
			PDGFRA Exon 13	55.143.567	55.143.587	600 - 606	21		COSM4417622: c.1809G>A (A603A)
			PDGFRA Exon 14*	55.144.053	55.144.183	631 - 668	131	20	COSM22414: c.1977C>G (N659K)
			PDGFRA Exon 15	55.144.537	55.144.557	671 - 677	21		COSM743: c.2021C>T (T674I)
			PDGFRA Exon 18*	55.151.998	55.152.140	814 - 854	143	20	COSM736: c.2525A>T (D842V); COSM22413: c.2472C>T (V824V)
			Intron	55.153.537	55.153.558		22	22	
			PDGFRA Exon 23	55.161.370	55.161.390	1068 - 1074	21		COSM12893: c.3211G>A (D1071N)
							657	572	Basen kodierende Sequenz gesamt
PIK3CA	chr3	NM_006218.2	PIK3CA Exon 2*	178.916.604	178.916.975	1 - 118	372	20	COSM746: c.263G>A (R88Q)
			PIK3CA Exon 3*	178.917.468	178.917.697	118 - 188	230	20	COSM751: c.353G>A (G118D)
			PIK3CA Exon 4*	178.919.068	178.919.338	188 - 271	271	20	COSM86047: c.665C>T (A222V)
			PIK3CA Exon 5*	178.921.322	178.921.587	272 - 353	266	20	COSM754: c.1035T>A (N345K)
			PIK3CA Exon 6*	178.922.281	178.922.386	354 - 382	106	20	COSM86044: c.1093G>A (E365K)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)					Wichtige Mutationen (nach COSMIC)	
			Name	Start	Stopp	Aminosäuren	Größe target region		Größe nicht- kodierende Sequenz
			PIK3CA Exon 7*	178.927.373	178.927.498	382 - 417	126	20	COSM328028: c.1173A>G (I391M)
			PIK3CA Exon 8*	178.927.964	178.928.136	418 - 468	173	20	COSM757: c.1258T>C (C420R)
			PIK3CA Exon 9*	178.928.209	178.928.363	469 - 513	155	20	COSM136387: c.1412C>T (P471L)
			PIK3CA Exon 10*	178.935.988	178.936.132	514 - 555	145	20	COSM763: c.1633G>A (E545K); COSM760: c.1624G>A (E542K)
			PIK3CA Exon 11*	178.936.974	178.937.075	555 - 582	102	20	COSM30790: c.1700A>G (K567R)
			PIK3CA Exon 12*	178.937.349	178.937.533	583 - 637	185	20	COSM1041500: c.1810T>C (C604R)
			PIK3CA Exon 13*	178.937.727	178.937.850	638 - 672	124	20	COSM250052: c.1913T>C (V638A)
			PIK3CA Exon 14*	178.938.764	178.938.955	672 - 729	192	20	COSM87306: c.2176G>A (E726K)
			PIK3CA Exon 15*	178.941.859	178.941.985	730 - 765	127	20	COSM6148: c.2198A>G (K733R)
			PIK3CA Exon 16*	178.942.478	178.942.619	765 - 806	142	20	COSM4598858: c.2352G>C (E784D)
			PIK3CA Exon 17*	178.943.740	178.943.838	806 - 832	99	20	COSM1041508: c.2452C>T (R818C)
			PIK3CA Exon 18*	178.947.050	178.947.240	832 - 889	191	20	COSM4481256: c.2494C>T (R832*)
			PIK3CA Exon 19*	178.947.782	178.947.919	889 - 928	138	20	COSM769: c.2702G>T (C901F)
			PIK3CA Exon 20*	178.948.003	178.948.174	929 - 979	172	20	COSM94980: c. 2908G>A (E970K)
			PIK3CA Exon 21*	178.951.872	178.952.162	979 - 1069*	291	20	COSM775: c.3140A>G (H1047R)
							3607	3207	Basen kodierende Sequenz gesamt

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
POLE	chr12	NM_006231.2	POLE Exon 9*	133.253.239	133.253.132	268 - 303	108		COSM937332: c.857C>G (P286R) + weitere P286
			POLE Exon 11*	133.252.406	133.252.321	341 - 369	86		71 Mutationen in COSMIC, aber keine Hotspots
			POLE Exon 13*	133.250.293	133.250.161	409 - 453	133		COSM204094: c.1231G>T (V411L) + weitere V411
			POLE Exon 14*	133.249.863	133.249.750	454 - 491	114		COSM937318: c.1366G>C (A456P)
								441	441 Basen kodierende Sequenz gesamt
RAF1	chr3	NM_002880.3	RAF1 Exon 17	12.626.113	12.626.133	610 - 616	21		COSM97875: c.1837C>G (L613V)
			RAF1 Exon 7	12.645.689	12.645.709	254 - 260	21		COSM181063: c.770C>T (S257L)
								42	42 Basen kodierende Sequenz gesamt
RET	chr10	NM_020975.5	RET Exon 10	43.609.061	43.609.082	606 - 613	22		COSM95656: c.1827C>A (C609*)
			RET Exon 10	43.609.086	43.609.114	615 - 623	29		COSM29804: c.1858T>C (C620R)
			RET Exon 11	43.609.926	43.609.961	627 - 638	36	2	COSM966: c.1900T>C (C634R)
			RET Exon 13	43.613.830	43.613.851	765 - 772	22		COSM4418405: c.2307G>T (L796L)
			RET Exon 13	43.613.895	43.613.919	787 - 794	25		COSM1159820: c.2372A>T (Y791F)
			RET Exon 15	43.615.558	43.615.579	880 - 886	22		COSM1570338: c. 2651A>T (E884V)
			RET Exon 15	43.615.583	43.615.604	888 - 894	22		COSM918124: c.2672C>T (S891L)
			RET Exon 16	43.617.406	43.617.427	915 - 921	22		COSM965: c.2753T>C (M918T)

Gen	Lage (hg19)	Transkript	Amplifikat (Covered target region)						Wichtige Mutationen (nach COSMIC)
			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
							200	198	Basen kodierende Sequenz gesamt
SRSF2	chr17	NM_003016.4	SRSF2 Exon 1*	74.733.242	74.732.881	1 - 121	640		COSM211504: c.284C>A (P95H) + weitere P95
							640	640	Basen kodierende Sequenz gesamt
STK11	chr19	NM_000455.4	STK11 Exon 1*	1.206.913	1.207.202	1 - 97	289		COSM12925: c.109C>T (Q37*) + weitere Q37
			STK11 Exon 2*	1.218.416	1.218.499	97 - 125	84		50 Mutationen in COSMIC, aber keine Hotspots
			STK11 Exon 3*	1.219.323	1.219.412	125 - 155	90		60 Mutationen in COSMIC, aber keine Hotspots
			STK11 Exon 4*	1.220.372	1.220.504	155 - 199	133		COSM20944: c.580G>T (D194Y) + weitere D194
			STK11 Exon 5*	1.220.580	1.220.716	200 - 245	137		175 Mutationen in COSMIC, aber keine Hotspots
			STK11 Exon 6*	1.221.212	1.221.339	245 - 288	128		163 Mutationen in COSMIC, aber keine Hotspots
			STK11 Exon 7*	1.221.948	1.222.005	288 - 307	58		46 Mutationen in COSMIC, aber keine Hotspots
			STK11 Exon 8*	1.222.984	1.223.171	307 - 370	188		COSM21360: c.1062C>G (F354L) + weitere F354
							1107	1107	Basen kodierende Sequenz gesamt
TERT	chr5	NM_198253.2	TERT Promoter + Exon 1*	1.295.364	1.294.886	c.-255 - c.219	523	304	Chr5:1295228: c.-124C>T (C228T) Chr5:1295250: c.-146C>T (C250T)
							523	219	Basen kodierende Sequenz gesamt
TP53	chr17	NM_000546.5	TP53 Exon 11*	7.572.917	7.573.018	367 - 394*	102	20	COSM13747: c.1146delA (K382fs)
			TP53 Exon 10*	7.573.917	7.574.043	332 - 367	127	20	COSM11073: c.1024C>T (R342*)

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			Name	Start	Stopp	Aminosäuren	Größe target region	Größe nicht- kodierende Sequenz	
		NM_001276699	TP53 Exon 6*	7.576.527	7.576.594	173 - 188*	68	20	
		NM_001126116	TP53 Exon 6*	7.576.615	7.576.667	200 - 210*	53	20	
			TP53 Exon 9*	7.576.843	7.576.936	307 - 331	94	20	COSM10786: c.949C>T (Q317*)
			TP53 Exon 8*	7.577.009	7.577.165	261 - 307	157	20	COSM10660: c.818G>A (R273H)
			TP53 Exon 7*	7.577.489	7.577.618	225 - 261	130	20	COSM10662: c.743G>A (R248Q); COSM10656: c.742C>T (R248W)
			TP53 Exon 6*	7.578.167	7.578.299	187 - 224	133	20	COSM10654: c.637C>T (R213*); COSM10758: c.659A>G (Y220C)
			TP53 Exon 5*	7.578.361	7.578.564	126 - 187	204	20	COSM10648: c.524G>A (R175H); COSM10670: c.469G>T (V157F)
			TP53 Exon 4*	7.579.302	7.579.600	33 - 125	299	20	COSM250061: c.215C>G (P72R); COSM44492: c.273G>A (W91*)
			TP53 Exon 3*	7.579.690	7.579.731	25 - 32	42	20	COSM85573: c.80delC (P27fs)
			TP53 Exon 2*	7.579.829	7.579.922	1 - 25	94	20	COSM11606: c.31G>C (E11Q)
							1503	1263	Basen kodierende Sequenz gesamt

Erklärung: vollständig untersuchte Exone sind mit * markiert, fett hervorgehoben wichtige Hotspotmutationen