

HUGO Gen Symbol	Hotspot of volledig gen	RefSeq (NCBI) hg38	ensembl transcript ID e95	Exon	Coördinaten (chromosomale posities) van de nucleotiden van interesse (GRCh38/hg38)	hotspots
AKT1	hotspot	NM_005163.2	ENST0000055528.5	3	chr14:104780088-104780216	E17K, L52
				4	chr14:104776659-104776770	Q79
				11	chr14:104772878-104773092	D323
				19	chr2:29225461-29225565	T1026, H1030
ALK	hotspot	NM_004304.5	ENST00000389048.7	20	chr2:29223342-29223528	D1091
				21	chr2:29222517-29222607	
				22	chr2:29222344-29222408	T1151, L1152, C1156, I1171
				23	chr2:29220706-29220835	F1174, L1196, L1198, G1202, D1203, S1206
				24	chr2:29213984-29214081	F1245
				25	chr2:29209786-29209878	G1269, R1275, Y1278
				26	chr2:29207171-29207272	
				27	chr2:29197542-29197676	K1491
				28	chr2:29196770-29196860	
				29	chr2:29193223-29193922	
				APC	hotspot	NM_000038.6
7	chr5:112792446-112792529	R232				
9	chr5:112815495-112815593	R283, R302				
12	chr5:112827108-112827247	R499				
14	chr5:112828856-112828972	R554, R564				
16, deels	chr5:112837999-112840099	5805, 5836, R876, Y935, K993, R1114, Q1378, R1450, R1556, E1577				
AR	hotspot	NM_000044.4	ENST00000374690.8	4	chrX:67711402-67711689	L702
				5	chrX:67717478-67717622	W742
				8	chrX:67723686-67723841	H875, T878
				1	chr1:26696402-26697540	
ARID1A	volledig	NM_006015.6	ENST00000324856.13	2	chr1:26729651-26729863	
				3	chr1:26731152-26731604	
				4	chr1:26732676-26732792	
				5	chr1:26760856-26761096	
				6	chr1:26761384-26761473	
				7	chr1:26762152-26762319	
				8	chr1:26762973-26763285	
				9	chr1:26766221-26766366	
				10	chr1:26766457-26766566	
				11	chr1:26767790-26767999	
				12	chr1:26771119-26771326	
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				14	chr1:26772812-26772987	
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				17	chr1:26773802-26773898	
				18	chr1:26774329-26775220	
				19	chr1:26775577-26775707	
				20	chr1:26779023-26780758	
				ATM	volledig	NM_000051.4
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4	chr11:108229177-108229323					
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8	chr11:108246963-108247127					
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11	chr11:108251836-108252031					
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27	chr11:108287599-108287715					
28	chr11:108288976-108289103					
29	chr11:108289601-108289801					
30	chr11:108292618-108292793					
31	chr11:108293312-108293477					
32	chr11:108294926-108295059					
33	chr11:108297286-108297382					
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35	chr11:108301647-108301789					
36	chr11:108302852-108303029					
37	chr11:108304674-108304852					
38	chr11:108307896-108307984					
39	chr11:108310159-108310315					
40	chr11:108312410-108312498					
41	chr11:108315822-108315911					
42	chr11:108316010-108316113					
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45	chr11:108321300-108321420					
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62	chr11:108365081-108365218					
63	chr11:108365324-108365508					
BRAF	hotspot	NM_004333.5	ENST00000646891.1	6	chr7:140801412-140801560	D287
				7	chr7:140800362-140800481	D287
				11	chr7:140781576-140781693	G466, G469, Y472
				12	chr7:140777991-140778075	
				14	chr7:140754187-140754233	N581
				15	chr7:140753275-140753393	D594, G596, L597, V600, K601
				1	chr3:52409842-52409880	
				2	chr3:52409714-52409743	
				3	chr3:52409554-52409608	
				4	chr3:52408474-52408606	
5	chr3:52407958-52408077					

<b>BAP1</b>	volledig, vraag Anne Hoorens	NM_004656.4	ENST00000460680.5	6	chr3:52407399-52407460	
				7	chr3:52407174-52407316	
				8	chr3:52406829-52406907	
				9	chr3:52406253-52406376	
				10	chr3:52405765-52405912	
				11	chr3:52405110-52405294	
				12	chr3:52404453-52404586	
				13	chr3:52403416-52403894	
				14	chr3:52403138-52403298	
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11	chr17:43090924-43091052					
12	chr17:43082384-43082595					
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17	chr17:43063854-43063971					
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19	chr17:43057032-43057155					
20	chr17:43051043-43051137					
21	chr17:43049101-43049214					
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5	chr13:32326081-32326170					
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9	chr13:32330899-32331050					
10	chr13:32332252-32333407					
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12	chr13:32344538-32344673					
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17	chr13:32362503-32362713					
18	chr13:32363159-32363553					
19	chr13:32370382-32370577					
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22	chr13:32379297-32379535					
23	chr13:32379730-32379933					
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27	chr13:32398142-32398792					
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11	chr17:39519956-39520087					
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14	chr17:39530604-39531318					
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2	chr9:21970902-21971208					
3	chr9:21968229-21968242					
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2	chr9:22005987-22006247					
3	chr3:41224067-41224081					
3	chr3:41224536-41224753	D32-545				
4	chr3:41224954-41225207	delV22-A97, M12-D144, M8-K132				
7	chr3:41227208-41227352	K335				
8	chr3:41233341-41233444	W383, N387				
26	chr14:95093888-95094156	E1705, D1709				
27	chr14:95091203-95091365	G1809, E1813				
14	chr1:97450008-97450108	*2A, c.1905+1G>A, rs3918290 (nonfunctional) c.1905+1G>A				
int10-11	chr1:97579843-97579943	HapB3 (rs75017182) (nonfunctional) c.1129-5923C>G				
11	chr1:97573813-97573913	c.1236G>A (rs56038477) (linkage disequilibrium HapB3				
13	chr1:97515737-97515837	*13 (rs55886062 A>C) (nonfunctional) c.1679T>G p.Ile560Ser				
22	chr1:97082341-97082441	rs67376798T>A (nonfunctional) c.2846A>T p.Asp949Val				
3	chr7:55143305-55143488	R108				
7	chr7:55140111-55141512	M277, A289				
12	chr7:55160139-55160338	G465, S492				
15	chr7:55165280-55165437	G598				
18	chr7:55173921-55174043	E709, G719				
19	chr7:55174722-55174820					
20	chr7:55181293-55181478	A763-A764,S768, T790				



KIT	hotspot	NM_000222.2	ENST00000288135.5	int10-11	chr4:54727325-54727415	
				11	chr4:54727416-54727542	
				13	chr4:54728011-54728121	
				14	chr4:54729335-54729485	
				17	chr4:54733069-54733192	
				18	chr4:54736498-54736609	
KRAS	hotspot	NM_04985.4	ENST00000311936.7	2	chr12:25245274-25245384	G12, G13
				3	chr12:25227234-25227412	A59, Q61
				4	chr12:25225614-25225773	K117, A146
				1	chr15:66387348-66387427	
MAP2K1	volledig	NM_002755.3	ENST00000307102.9	2	chr15:66435027-66435237	L42, F53, L54, Q56, K57, K59, D67, E73, G91, A95, R96
				3	chr15:66436746-66436892	E102, P105, R108, T111, L116, L124, P124, T226, G126, P129, T130, T134, T135
				4	chr15:66443280-66443357	P162
				5	chr15:66444656-66444707	G176
				6	chr15:66481755-66481879	R189
				7	chr15:66484990-66485191	P193, E203, F223
				8	chr15:66487228-66487292	W247, M256, P264, R291, P293, G294
				9	chr15:66489215-66489276	
				10	chr15:66489718-66489763	S331
				11	chr15:66490502-66490615	E367, N382, P387
MET	hotspot	NM_001127500.3	ENST00000318493.10	2	chr7:116699085-116700284	E168, N375
				13	chr7:116771498-116771654	I959
				int13-14	chr7:116771655-116771848	
				14 + extra int14-15	chr7:116771849-116772139	T1010, Y1021
				15	chr7:116774881-116775111	
				16	chr7:116777389-116777469	H1112
				17	chr7:116778776-116778957	R1188
				18	chr7:116781988-116782097	
				19	chr7:116783304-116783469	D1246, Y1248, Y1253, M1268
				20	chr7:116795655-116795791	D1304
MYODI	volledig	NM_002478.5	ENST00000250003.4	21	chr7:116795887-116796124	A1357, P1382
				1	chr11:17719782-17720412	
				2	chr11:17720902-17720980	
NRAS	hotspot	NM_002524.5	ENST00000369535.4	3	chr11:17721255-17721510	
				2	chr1:114716050-114716160	G12, G13
				3	chr1:114713800-114713978	A59, Q61
				4	chr1:114709569-114709728	K117, A146
NTRK1	hotspot	NM_002529.3	ENST00000524377.5	14	chr1:156876400-156876572	V573M, F589L, G595R
				15	chr1:156879122-156879362	G667C, G667S
NTRK2	hotspot	NM_001018064.3	ENST00000323115.9	14	chr9:84948462-84948634	Q596E, V601G, G623S, R630K
				15	chr9:84955283-84955517	R630K, G693S
NTRK3	hotspot	NM_001012338.2	ENST00000360948.6	16	chr15:87933012-87933184	G623R
				17	chr15:87929191-87929434	G696A
PDGFRA	hotspot	NM_006206.6	ENST00000257290.9	12	chr4:54274841-54274973	V561
				14	chr4:54277896-54278006	N659
				18	chr4:54285841-54285963	D842
				12	chr5:150125445-150125577	R561, P584
PDGFRB	hotspot TK domein	NM_002609.4	ENST00000261799.9	13	chr5:150124727-150124831	
				14	chr5:150124250-150124360	V665, N666
				15	chr5:150123042-150123201	
				16	chr5:150121880-150122040	
				17	chr5:150121204-150121322	
				18	chr5:150120888-150121010	C843
				19	chr5:150120012-150120123	
				20	chr5:150119467-150119566	
PIK3CA	hotspot	NM_006218.4	ENST00000263967.3	21	chr5:150118747-150118852	
				2	chr3:179198827-179199177	R388, R88, K111
				3	chr3:179199690-179199899	G118
				5	chr3:179203544-179203789	N345
				8	chr3:179210186-179210338	C420, E453
				10	chr3:179218210-179218334	E542, E545, Q546
				14	chr3:179220986-179221157	E726
PIK3R1	volledig	NM_181523.3	ENST00000521381.5	21	chr3:179234094-179234364	M1043, H1047, G1049
				2	chr5:68226676-68227009	
				3	chr5:68273390-68273482	
				4	chr5:68273939-68274013	
				5	chr5:68279602-68279733	
				6	chr5:68280528-68280729	
				7	chr5:68280927-68281006	
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				10	chr5:68293303-68293483	
				11	chr5:68293709-68293834	
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				13	chr5:68295148-68295324	
				14	chr5:68295420-68295488	
POLE	hotspot	NM_006231.3	ENST00000320574.9	15	chr5:68296171-68296341	
				16	chr5:68297412-68297603	
				9	chr12:132676546-132676653	P286, S297
				10	chr12:132676094-132676204	
				11	chr12:132675735-132675820	
PTEN	volledig	NM_000314.7	ENST00000371953.7	12	chr12:132675398-132675517	
				13	chr12:132673757-132673707	V411, L424
				14	chr12:132673164-132673277	A456, S459, A465
				1	chr10:87864470-87864548	
				2	chr10:87894025-87894109	
				3	chr10:87925513-87925557	
RB1	volledig, longtumor	NM_000321.2	ENST00000267163.5	4	chr10:87931046-87931089	
				5	chr10:87933013-87933251	R130, R159
				6	chr10:87952118-87952259	
				7	chr10:87957853-87958019	
				8	chr10:87960894-87961118	R233, K267
				9	chr10:87965287-87965472	
				1	chr13:48303913-48304049	
				2	chr13:48307280-48307406	
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17	chr13:48381247-48381443					
18	chr13:48452993-48453111					
19	chr13:48456204-48456349					
20	chr13:48459688-48459833					
21	chr13:48463731-48463835					
22	chr13:48464998-48465111					
23	chr13:48465205-48465368					
24	chr13:48473360-48473390					
25	chr13:48476701-48476843					
26	chr13:48477355-48477404					

Gene	Completeness	Accession	ENST ID	Coordinates	Associated Variants	
<b>RET</b>	volledig	NM_020975.6	ENST00000355710.7	27	chr13:48479998-48480071	
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				2	chr10:43100459-43100722	
				3	chr10:43102342-43102629	
				4	chr10:43104952-43105193	
				5	chr10:43106376-43106571	
				6	chr10:43109031-43109230	
				7	chr10:43111207-43111465	
				8	chr10:43112099-43112224	
				9	chr10:43112583-43112963	
				10	chr10:43113556-43113675	C611, C618, C620
				11	chr10:43114480-43114736	L629, C630, D631, E632, L633, C634
				12	chr10:43116584-43116731	
				13	chr10:43118373-43118480	
				14	chr10:43119531-43119745	
				15	chr10:43120081-43120203	A883
				16	chr10:43121946-43122016	M918
				17	chr10:43123671-43123808	
				18	chr10:43124883-43124982	
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20	chr10:43128112-43128272					
<b>RNF43</b>	volledig	NM_017763.5	ENST00000407977.6	2	chr17:58415326-58415577	
				3	chr17:58370911-58371033	R113
				4	chr17:58363526-58363600	R145
				5	chr17:58363275-58363406	Q152, Y177
				6	chr17:58362544-58362648	S216
				7	chr17:58360783-58360944	
				8	chr17:58360149-58360251	
				9	chr17:58357468-58358823	R371, R519, S607
				10	chr17:58354943-58354986	
				36	chr6:117319868-117320030	
<b>ROS1</b>	hotspot	NM_002944.2	ENST00000368508.7	37	chr6:117318188-117318252	S1986
				38	chr6:117317143-117317272	G2032, D2033
				41	chr6:117308794-117308928	L2155
				2	chr18:51047047-51047295	
				3	chr18:51048686-51048860	A118, R135
<b>SMAD4</b>	volledig	NM_005359.5	ENST00000342988.7	4	chr18:51049295-51049324	
				5	chr18:51054781-51054993	
				6	chr18:51058125-51058244	
				7	chr18:51058340-51058456	
				8	chr18:51059866-51059916	Q311
				9	chr18:51065423-51065606	E330, K340, D351, D355, R361
				10	chr18:51067019-51067187	G386, G419
				11	chr18:51076638-51076776	R445
				12	chr18:51078256-51078467	R496, G510, D537, E538
				<b>SMO</b>	volledig, basal cell carcinoma, medulloblastoma (mycancergenome), precision	NM_005631.5
2	chr7:129203384-129203589					
3	chr7:129205203-129205412					
4	chr7:129205610-129205782					
5	chr7:129206150-129206369					
6	chr7:129206464-129206587	L412F				
7	chr7:129208759-129208851					
8	chr7:129209289-129209397					
9	chr7:129210363-129210548	W535L				
10	chr7:129210965-129211113					
11	chr7:129211636-129211770					
12	chr7:129212024-129212453					
<b>SPOP</b>	hotspot	NM_003563.3	ENST00000393328.6	5	chr17:49619234-49619385	Y83, Y87, F102
				6	chr17:49618981-49619108	W131, F133
				3	chr17:42346569-42346713	L78
<b>STAT3</b>	hotspot	NM_139276.2	ENST00000264657.9	6	chr17:42338731-42338812	E166
				17	chr17:42324711-42324846	N502
				20	chr17:42323004-42323143	S614, G618
				21	chr17:42322282-42322494	Y640, Y657, K658, M660, D661
				1	chr19:1206914-1207203	Q37
<b>STK11</b>	volledig	NM_000455.4	ENST00000326873.11	2	chr19:1218417-1218500	
				3	chr19:1219324-1219413	
				4	chr19:1220373-1220505	Q170, G171, D194, E199
				5	chr19:1220581-1220717	S216, Q220
				6	chr19:1221213-1221340	P281
				7	chr19:1221949-1222006	
				8	chr19:1222985-1223172	F354
				9	chr19:1226454-1226647	
				promoter	chr5:1295016-1295205	C228T, C250T, C242T
<b>TP53</b>	volledig	NM_000546.5	ENST00000269305.8	2	chr17:7676521-7676594	
				3	chr17:7676382-7676403	
				4	chr17:7675994-7676272	
				5	chr17:7675053-7675236	
				6	chr17:7674859-7674971	
				7	chr17:7674181-7674290	
				8	chr17:7673701-7673837	
				9	chr17:7673535-7673608	
				10	chr17:7670609-7670715	
				11	chr17:7669609-7669690	
				beta/gamma exoner	chr17:7673219-7673339	
<b>VHL</b>	volledig	NM_000551.3	ENST00000256474.2	1	chr3:10141848-10142187	
				2	chr3:10146514-10146636	
				3	chr3:10149787-10149965	