

OncoNext™ 15 Geni

Geni investigati e principali tipi di tumore associati

Gene	Tipi di tumore associati
AKT1	Mammella, Polmone, Colon-Retto*
BRAF	Melanoma*, Colon-Retto* Polmone, Ovarico, Gastrico, Glioma, Tiroide, Pancreas, Prostata
EGFR	Polmone*; Head & Neck, Prostata
ERBB2	Mammella, Polmone
FOXL2	Ovarico
GNA11	Melanoma
GNAQ	Melanoma
KIT	Gastrico, Melanoma*, Carcinoma Timico
KRAS	Colon-Retto*, Gastrico, Polmone*, Ovarico, Tiroide, Endometrio, Pancreas, Prostata
MET	Polmone*, Colon-Retto, Gastrico
NRAS	Colon-Retto*, Polmone, Melanoma, Tiroide
PDGFRA	Gastrico, Melanoma,
PIK3CA	Polmone, Mammella, Prostata, Colon-Retto, Ovarico, Head & Neck, Pancreas, Tiroide
RET	Polmone*, Tiroide
TP53	Polmone, Melanoma, Ovarico, Colon-Retto, Mammella; Endometrio, Head & Neck, Rene, Pancreas, Prostata, Tiroide

* Linee guida NCCN per tipo di tumore.

Mutazioni hotspot ricercate nel test OncoNext™ 15 Geni

REFSEQ	GENE	Esone	Mutazione	Variazione Nucleotidica	Profondità di sequenziamento	
NM_005163	AKT1	3	E17K	c.49 G>A	340.000	
			D594E	c.1782T>A	308.000	
			D594E	c.1782T>G	308.000	
			D594G	c.1781A>G	308.000	
			D594H	c.1780G>C	308.000	
			D594N	c.1779_1780delTGinsGA	308.000	
			D594N	c.1780G>A	308.000	
			D594V	c.1781A>T	308.000	
			G596R	c.1786G>C	308.000	
			K601E	c.1801A>G	308.000	
			L597Q	c.1790T>A	308.000	
			L597R	c.1790T>G	308.000	
			L597S	c.1789_1790delCTinsTC	308.000	
			L597V	c.1789C>G	308.000	
NM_004333	BRAF	15	V600D	c.1799_1800delTGinsAT	308.000	
			V600E	c.1799T>A	308.000	
			V600E	c.1799_1800delTGinsAA	308.000	
			V600G	c.1799T>G	308.000	
			V600K	c.1798_1799delGTinsAA	308.000	
			V600M	c.1798G>A	308.000	
			V600R	c.1798_1799delGTinsAG	308.000	
			G719A	c.2156G>C	84.200	
			G719C	c.2155G>T	84.200	
			G719S	c.2155G>A	84.200	
			Exon 19 Deletions		50.000	
			Exon 19 Insertions		50.000	
			A763_Y764insFQEA	c.2290_2291ins	50.000	
			Exon 20 Insertions		50.000	
			S768I	c.2303G>T	50.000	
			T790M	c.2369C>T	29.400	
				E746_A750>IP	c.2235_2248delGGAATTAAGAGAAG insAATTC	50.000
				E746_A750del	c.2235_2249delGGAATTAAGAGAAG C	50.000
	E746_A750del	c.2236_2250delGAATTAAGAGAAGC A	50.000			
	E746_P753>VS	c.2237_2257del21insTCT	50.000			
	E746_S752>A	c.2237_2254del18	50.000			
	E746_S752>D	c.2238_2255del18	50.000			
NM_005228	EGFR		E746_S752>I	c.2235_2255delinsAAT	50.000	
			E746_S752>V	c.2237_2255delinsT	50.000	
			E746_T751>A	c.2237_2251del15	50.000	
			E746_T751>I	c.2235_2252delinsAAT	50.000	
			E746_T751>IP	c.2235_2251delinsAATTC	50.000	
			E746_T751>V	c.2237_2252delinsT	50.000	
			E746_T751>VA	c.2237_2253delinsTTGCT	50.000	
			E746_T751del	c.2236_2253del18	50.000	
			K745_E749del	c.2233_2247del15)	50.000	
			L747_A750>P	c.2238_2248delATAAGAGAAGinsG C	50.000	
			L747_A750>P	c.2239_2248delITTAAGAGAAGinsC	50.000	
			L747_E749del	c.2239_2247delITTAAGAGAA	50.000	
			L747_P753>Q	c.2239_2258delinsCA	50.000	
			L747_S752>Q	c.2239_2256delinsCAA	50.000	
			L747_S752del	c.2239_2256del18	50.000	
			L747_T751>Q	c.2238_2252delinsGCA	50.000	
			L747_T751>S	c.2240_2251del	50.000	
			L747_T751del	c.2238_2252del	50.000	
	D769H	c.2305G>C	150.000			
	D769Y	c.2305G>T	150.000			
	G776S	c.2326 G>A	150.000			
NM_004448	ERBB2		c.2263_2264delTTinsCC	c.2263_2264delTTinsCC	150.000	
			c.2322_2334dupATACGT GATGGC	c.2322_2334dupATACGTGATGGC	150.000	
			c.2328_2336dupTGTGG	c.2328_2336dupTGTGGGCTC	150.000	

			GCTC		
		19	L755_T759del	c.2264_2278del	150.000
		19	L755S	c.2264T>C	150.000
		20	Exon 20 Insertions		150.000
		20	G778_P780dup	c.2339_2340ins	150.000
		20	V777L	c.2329G>T	150.000
NM_023067	FOXL2	1	C134W	c.402 C>G	500.000
NM_002067	GNA11	5	Q209L	c.626A>T	90.000
		5	Q209P	c.626A>C	90.000
NM_002072	GNAQ	5	Q209L	c.626A>T	60.000
		5	Q209P	c.626A>C	60.000
		5	Q209R	c.626A>G	60.000
		11	556 ins L		25.000
		11	575 ins PE		25.000
		11	Del 554-558		25.000
		11	Del 554-559		25.000
		11	Del 566-572		25.000
		11	Del 566-574		25.000
		11	Del 579		25.000
		11	Del V559		25.000
		11	E583_E589dupPYDHWKE		25.000
		11	Exon 11 Mutation		25.000
		11	G565V		25.000
		11	K550N		25.000
		11	K558N		25.000
		11	L576P	c.1727T>C	25.000
NM_000222	KIT	11	N566D		25.000
		11	P577_D579del	c.1730_1738del	25.000
		11	V559A	c.1676T>C	25.000
		11	V559D	c.1676T>A	25.000
		11	V559G		25.000
		11	V560A		25.000
		11	V560D	c.1727T>C (V560D)	25.000
		11	V560del	c.1679_1681del	25.000
		11	V560G		25.000
		11	V569G		25.000
		11	W557R	c.1669T>A	25.000
		11	W557R	c.1669T>C	25.000
		11	Y553N	c.1657T>A	25.000
		14	Exon 14 Mutation		25.000
		14	H697Y	c.2089C>T	25.000
		2	G12A	c.35G>C	25.000
		2	G12C	c.34G>T	25.000
		2	G12D	c.35G>A	25.000
		2	G12R	c.34G>C	25.000
		2	G12S	c.34G>A	25.000
		2	G12V	c.35G>T	25.000
		2	G13A	c.38G>C	25.000
		2	G13C	c.37G>T	25.000
		2	G13D	c.38G>A	25.000
		2	G13R	c.37G>C	25.000
		2	G13S	c.37G>A	25.000
		2	G13V	c.38G>T	25.000
NM_004985	KRAS	2	Q22K	c.64C>A	25.000
		3	Q61H	c.183A>C	25.000
		3	Q61H	c.183A>T	25.000
		3	Q61H	c.183A>C	25.000
		3	Q61K	c.181C>A	25.000
		3	Q61L	c.182A>T	25.000
		3	Q61P	c.182A>C	25.000
		3	Q61R	c.182A>G	25.000
		4	A146P	c.436G>C	25.000
		4	A146T	c.436G>A	25.000
		4	A146V	c.437C>T	25.000
		4	K117N	c.351A>C	25.000
		4	K117N	c.351A>T	25.000
NM_001127	MET	18	L1213V	c.3637 C>G	70.000

500		18	V1206L	c.3616 G>T	70.000
		2	G12A	c.35G>C	60.000
		2	G12C	c.34G>T	60.000
		2	G12D	c.35G>A	60.000
		2	G12R	c.34G>C	60.000
		2	G12S	c.34G>A	60.000
		2	G12V	c.35G>T	60.000
		2	G13A	c.38G>C	60.000
		2	G13C	c.37G>T	60.000
		2	G13D	c.38G>A	60.000
		2	G13R	c.37G>C	60.000
NM_002524	NRAS	2	G13V	c.38G>T	60.000
		3	Q61E	c.181C>G	60.000
		3	Q61H	c.183A>C	60.000
		3	Q61H	c.183A>T	60.000
		3	Q61H	c.183A>T	60.000
		3	Q61K	c.181C>A	60.000
		3	Q61L	c.182A>T	60.000
		3	Q61L	c.182_183delAAinsTG	60.000
		3	Q61P	c.182A>C	60.000
		3	Q61R	c.182A>G	60.000
		3	Q61R	c.182_183delAAinsGG	60.000
		12	Y555C	c.1664 A>G	100.000
		12	c.1679_1693delGGGTC ATTGAATCAA	c.1679_1693delGGGTCATTGAATCA A	100.000
		12	c.1681_1682insAGAGG G	c.1681_1682insAGAGGG	100.000
		12	V561D	c.1682 T>A	100.000
		12	c.1696_1713del18	c.1696_1713del18	100.000
NM_006206	PDGFRA	14	c.2526_2537delCATCAT GCATGA	c.2526_2537delCATCATGCATGA	70.000
		14	c.2533_2544delCATGATT CGAAC	c.2533_2544delCATGATTCGAAC	70.000
		18	D842V	c.2525 A>T	70.000
		18	D846Y	c.2536 G>T	70.000
		12	Exon 12 Mutation		100.000
		14	Exon 14 Mutation		100.000
		18	Exon 18 Mutation		70.000
		9	D549N	c.1645G>A	110.000
		9	E542K	c.1624G>A	110.000
		9	E545G	c.1634A>G	110.000
		9	E545K	c.1633G>A	110.000
		9	E545Q	c.1633G>C	110.000
		9	E545V	c.1634A>T	110.000
		9	Q546E	c.1636C>G	110.000
NM_006218	PIK3CA	9	Q546K	c.1636C>A	110.000
		9	Q546L	c.1637A>T	110.000
		9	Q546P	c.1637A>C	110.000
		9	Q546R	c.1637A>G	110.000
		20	H1047R	c.3140A>G	110.000
		20	H1047L	c.3140A>T	110.000
		20	H1047Y	c.3139C>T	110.000
		20	M1043I	c.3129G>A	110.000
NM_020975	RET	16	M918T	c.2753 T>C	350.000
NM_000546	TP53		Intera regione codificante		30.000 – 380.000