

## 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	<a href="#">E17K</a>	c.49 G>A	340.000	
			<a href="#">D594E</a>	c.1782T>A	308.000	
			<a href="#">D594E</a>	c.1782T>G	308.000	
			<a href="#">D594G</a>	c.1781A>G	308.000	
			<a href="#">D594H</a>	c.1780G>C	308.000	
			<a href="#">D594N</a>	c.1779_1780delTGinsGA	308.000	
			<a href="#">D594N</a>	c.1780G>A	308.000	
			<a href="#">D594V</a>	c.1781A>T	308.000	
			<a href="#">G596R</a>	c.1786G>C	308.000	
			<a href="#">K601E</a>	c.1801A>G	308.000	
			<a href="#">L597Q</a>	c.1790T>A	308.000	
			<a href="#">L597R</a>	c.1790T>G	308.000	
			<a href="#">L597S</a>	c.1789_1790delCTinsTC	308.000	
			<a href="#">L597V</a>	c.1789C>G	308.000	
NM_004333	BRAF	15	<a href="#">V600D</a>	c.1799_1800delTGinsAT	308.000	
			<a href="#">V600E</a>	c.1799T>A	308.000	
			<a href="#">V600E</a>	c.1799_1800delTGinsAA	308.000	
			<a href="#">V600G</a>	c.1799T>G	308.000	
			<a href="#">V600K</a>	c.1798_1799delGTinsAA	308.000	
			<a href="#">V600M</a>	c.1798G>A	308.000	
			<a href="#">V600R</a>	c.1798_1799delGTinsAG	308.000	
			<a href="#">G719A</a>	c.2156G>C	84.200	
			<a href="#">G719C</a>	c.2155G>T	84.200	
			<a href="#">G719S</a>	c.2155G>A	84.200	
			<a href="#">Exon 19 Deletions</a>		50.000	
			<a href="#">Exon 19 Insertions</a>		50.000	
			<a href="#">A763_Y764insFQEA</a>	c.2290_2291ins	50.000	
			<a href="#">Exon 20 Insertions</a>		50.000	
			<a href="#">S768I</a>	c.2303G>T	50.000	
			<a href="#">T790M</a>	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_2248delATTAAGAGAAGinsG 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	
	<a href="#">D769H</a>	c.2305G>C	150.000			
	<a href="#">D769Y</a>	c.2305G>T	150.000			
	<a href="#">G776S</a>	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	<a href="#">L755_T759del</a>	c.2264_2278del	150.000
		19	<a href="#">L755S</a>	c.2264T>C	150.000
		20	<a href="#">Exon 20 Insertions</a>		150.000
		20	<a href="#">G778_P780dup</a>	c.2339_2340ins	150.000
		20	<a href="#">V777L</a>	c.2329G>T	150.000
NM_023067	FOXL2	1	C134W	c.402 C>G	500.000
NM_002067	GNA11	5	<a href="#">Q209L</a>	c.626A>T	90.000
		5	<a href="#">Q209P</a>	c.626A>C	90.000
NM_002072	GNAQ	5	<a href="#">Q209L</a>	c.626A>T	60.000
		5	<a href="#">Q209P</a>	c.626A>C	60.000
		5	<a href="#">Q209R</a>	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	<a href="#">Exon 11 Mutation</a>		25.000
		11	G565V		25.000
		11	K550N		25.000
		11	K558N		25.000
		11	<a href="#">L576P</a>	c.1727T>C	25.000
NM_000222	KIT	11	N566D		25.000
		11	<a href="#">P577_D579del</a>	c.1730_1738del	25.000
		11	<a href="#">V559A</a>	c.1676T>C	25.000
		11	<a href="#">V559D</a>	c.1676T>A	25.000
		11	V559G		25.000
		11	V560A		25.000
		11	V560D	c.1727T>C (V560D)	25.000
		11	<a href="#">V560del</a>	c.1679_1681del	25.000
		11	V560G		25.000
		11	V569G		25.000
		11	<a href="#">W557R</a>	c.1669T>A	25.000
		11	<a href="#">W557R</a>	c.1669T>C	25.000
		11	<a href="#">Y553N</a>	c.1657T>A	25.000
		14	<a href="#">Exon 14 Mutation</a>		25.000
		14	<a href="#">H697Y</a>	c.2089C>T	25.000
		2	<a href="#">G12A</a>	c.35G>C	25.000
		2	<a href="#">G12C</a>	c.34G>T	25.000
		2	<a href="#">G12D</a>	c.35G>A	25.000
		2	<a href="#">G12R</a>	c.34G>C	25.000
		2	<a href="#">G12S</a>	c.34G>A	25.000
		2	<a href="#">G12V</a>	c.35G>T	25.000
		2	<a href="#">G13A</a>	c.38G>C	25.000
		2	<a href="#">G13C</a>	c.37G>T	25.000
		2	<a href="#">G13D</a>	c.38G>A	25.000
		2	<a href="#">G13R</a>	c.37G>C	25.000
		2	<a href="#">G13S</a>	c.37G>A	25.000
		2	<a href="#">G13V</a>	c.38G>T	25.000
NM_004985	KRAS	2	<a href="#">Q22K</a>	c.64C>A	25.000
		3	<a href="#">Q61H</a>	c.183A>C	25.000
		3	<a href="#">Q61H</a>	c.183A>T	25.000
		3	<a href="#">Q61H</a>	c.183A>C	25.000
		3	<a href="#">Q61K</a>	c.181C>A	25.000
		3	<a href="#">Q61L</a>	c.182A>T	25.000
		3	<a href="#">Q61P</a>	c.182A>C	25.000
		3	<a href="#">Q61R</a>	c.182A>G	25.000
		4	<a href="#">A146P</a>	c.436G>C	25.000
		4	<a href="#">A146T</a>	c.436G>A	25.000
		4	<a href="#">A146V</a>	c.437C>T	25.000
		4	<a href="#">K117N</a>	c.351A>C	25.000
		4	<a href="#">K117N</a>	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	<a href="#">G12A</a>	c.35G>C	60.000
		2	<a href="#">G12C</a>	c.34G>T	60.000
		2	<a href="#">G12D</a>	c.35G>A	60.000
		2	<a href="#">G12R</a>	c.34G>C	60.000
		2	<a href="#">G12S</a>	c.34G>A	60.000
		2	<a href="#">G12V</a>	c.35G>T	60.000
		2	<a href="#">G13A</a>	c.38G>C	60.000
		2	<a href="#">G13C</a>	c.37G>T	60.000
		2	<a href="#">G13D</a>	c.38G>A	60.000
		2	<a href="#">G13R</a>	c.37G>C	60.000
NM_002524	NRAS	2	<a href="#">G13V</a>	c.38G>T	60.000
		3	<a href="#">Q61E</a>	c.181C>G	60.000
		3	<a href="#">Q61H</a>	c.183A>C	60.000
		3	<a href="#">Q61H</a>	c.183A>T	60.000
		3	<a href="#">Q61H</a>	c.183A>T	60.000
		3	<a href="#">Q61K</a>	c.181C>A	60.000
		3	<a href="#">Q61L</a>	c.182A>T	60.000
		3	<a href="#">Q61L</a>	c.182_183delAAinsTG	60.000
		3	<a href="#">Q61P</a>	c.182A>C	60.000
		3	<a href="#">Q61R</a>	c.182A>G	60.000
		3	<a href="#">Q61R</a>	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	<a href="#">D842V</a>	c.2525 A>T	70.000
		18	<a href="#">D846Y</a>	c.2536 G>T	70.000
		12	<a href="#">Exon 12 Mutation</a>		100.000
		14	<a href="#">Exon 14 Mutation</a>		100.000
		18	<a href="#">Exon 18 Mutation</a>		70.000
		9	<a href="#">D549N</a>	c.1645G>A	110.000
		9	<a href="#">E542K</a>	c.1624G>A	110.000
		9	<a href="#">E545G</a>	c.1634A>G	110.000
		9	<a href="#">E545K</a>	c.1633G>A	110.000
		9	<a href="#">E545Q</a>	c.1633G>C	110.000
		9	<a href="#">E545V</a>	c.1634A>T	110.000
		9	<a href="#">Q546E</a>	c.1636C>G	110.000
NM_006218	PIK3CA	9	<a href="#">Q546K</a>	c.1636C>A	110.000
		9	<a href="#">Q546L</a>	c.1637A>T	110.000
		9	<a href="#">Q546P</a>	c.1637A>C	110.000
		9	<a href="#">Q546R</a>	c.1637A>G	110.000
		20	<a href="#">H1047R</a>	c.3140A>G	110.000
		20	<a href="#">H1047L</a>	c.3140A>T	110.000
		20	H1047Y	c.3139C>T	110.000
		20	M1043I	c.3129G>A	110.000
NM_020975	RET	16	<a href="#">M918T</a>	c.2753 T>C	350.000
NM_000546	TP53		Intera regione codificante		30.000 – 380.000