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
NM_004333	BRAF	15	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
			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
NM_005228	EGFR	18	G719A	c.2156G>C	84.200
		18	G719C	c.2155G>T	84.200
		18	G719S	c.2155G>A	84.200
		19	Exon 19 Deletions		50.000
		19	Exon 19 Insertions		50.000
		20	A763_Y764insFQEA	c.2290_2291ins	50.000
		20	Exon 20 Insertions		50.000
		20	S768I	c.2303G>T	50.000
		20	T790M	c.2369C>T	29.400
		19	E746_A750>IP	c.2235_2248delGGAATTAAGAGAAG insAATTC	50.000
		19	E746_A750del	c.2235_2249delGGAATTAAGAGAAG C	50.000
		19	E746_A750del	c.2236_2250delGAATTAAGAGAAGC A	50.000
		19	E746_P753>VS	c.2237_2257del21insTCT	50.000
		19	E746_S752>A	c.2237_2254del18	50.000
		19	E746_S752>D	c.2238_2255del18	50.000
		19	E746_S752>I	c.2235_2255delinsAAT	50.000
		19	E746_S752>V	c.2237_2255delinsT	50.000
		19	E746_T751>A	c.2237_2251del15	50.000
		19	E746_T751>I	c.2235_2252delinsAAT	50.000
		19	E746_T751>IP	c.2235_2251delinsAATTC	50.000
		19	E746_T751>V	c.2237_2252delinsT	50.000
		19	E746_T751>VA	c.2237_2253delinsTTGCT	50.000
		19	E746_T751del	c.2236_2253del18	50.000
		19	K745_E749del	c.2233_2247del15)	50.000
		19	L747_A750>P	c.2238_2248delATTAAGAGAAGinsG C	50.000
		19	L747_A750>P	c.2239_2248delTTAAGAGAAGinsC	50.000
		19	L747_E749del	c.2239_2247delTTAAGAGAA	50.000
		19	L747_P753>Q	c.2239_2258delinsCA	50.000
		19	L747_S752>Q	c.2239_2256delinsCAA	50.000
		19	L747_S752del	c.2239_2256del18	50.000
		19	L747_T751>Q	c.2238_2252delinsGCA	50.000
		19	L747_T751>S	c.2240_2251del	50.000
		19	L747_T751del	c.2238_2252del	50.000
NM_004448	ERBB2	19	D769H	c.2305G>C	150.000
		19	D769Y	c.2305G>T	150.000
		19	G776S	c.2326 G>A	150.000
		19	c.2263_2264delTTinsCC	c.2263_2264delTTinsCC	150.000
		19	c.2322_2334dupATACGT GATGGC	c.2322_2334dupATACGTGATGGC	150.000
		19	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
NM_000222	KIT	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_E589dupPYDHKWE		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
		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
NM_004985	KRAS	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
		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_1693delGGGTCATTGAATCAA	c.1679_1693delGGGTCATTGAATCAA	100.000
		12	c.1681_1682insAGAGGG	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_2537delCATCATGCATGA	c.2526_2537delCATCATGCATGA	70.000
		14	c.2533_2544delCATGATTCGAAC	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