OncoNext™ *23 geni*

Geni investigati e principali tipi di tumore associati

Gene	Tipi di tumore associati
AKT1	Mammella, Polmone, Colon-Retto*
ALK	Polmone, Neuroblastoma, Rhabdomyosarcoma
AR	Prostata
BRAF	Melanoma*, Colon-Retto* Polmone, Ovarico, Gastrico, Glioma, Tiroide, Pancreas, Prostata
CTNNB1	Melanoma
EGFR	Polmone*; Head & Neck, Prostata
ERBB2	Mammella, Polmone
ESR1	Mammella
FOXL2	Ovarico
GNA11	Melanoma
GNAQ	Melanoma
KIT	Gastrico, Melanoma*, Carcinoma Timico
KRAS	Colon-Retto*, Gastrico, Polmone*, Ovarico, Tiroide, Endometrio, Pancreas, Prostata
MEK1 (MAP2K1)	Melanoma, Polmone, Ovarico, Colon-Retto,
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
PTEN	Mammella, Polmone,
RET	Polmone*, Tiroide
ROS1	Polmone
SMAD4	Colon-Retto
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™ *23 geni*

Gene	Mutazione	Esone	Variazione Nucleotidica
AKT1	E17K	3	c.49 G>A
ALK	D1091N	20	c.3271G>A
ALK	I1171N	22	c.3512T>A
ALK	T1151M	22	c.3452C>T
ALK	F1174C	23	c.3521T>G
ALK	F1174I	23	c.3520T>A
ALK	F1174L	23	c.3522C>A
ALK	F1174V	23	c.3520T>G
ALK	D1225N	24	c.3673G>A
ALK	F1245C	24	c.3734T>G
ALK	F1245L	24	c.3735C>G
ALK	F1245V	24	c.3733T>G
ALK	R1275Q	25	c.3824G>A
ALK	Y1278S	25	c.3833A>C
ALK	1151Tins		
ALK	C1156Y		
ALK	G1202R		
ALK	G1269A		
ALK	L1152R		
ALK	L1196M		
ALK	L1198F		
ALK	S1206Y		
AR	L702H	4	c.2105T>A
AR	W742C	5	c.2226G>T
AR	H875Y	8	c.2623C>T
AR	F877L	8	c.2631C>A
AR	T878A	8	c.2632A>G
BRAF	G466V	11	c.1397G>T
BRAF	G469A	11	c.1406G>C
BRAF	G469E	11	c.1406G>A
BRAF	G469L	11	c.1405_1406delGGinsTT
BRAF	G469V	11	c.1406G>T
BRAF	Y472C	11	c.1415A>G
BRAF	D594E	15	c.1782T>A
BRAF	D594E	15	c.1782T>G
BRAF	D594G	15	c.1781A>G
BRAF	D594H	15	c.1780G>C
BRAF	D594N	15	c.1779_1780delTGinsGA
BRAF	D594N	15	c.1780G>A
BRAF	D594V	15	c.1781A>T
BRAF	G596R	15	c.1786G>C
BRAF	K601E	15	c.1801A>G
BRAF	L597Q	15	c.1790T>A
BRAF	L597R	15	c.1790T>G
BRAF	L597S	15	c.1789_1790delCTinsTC
BRAF	L597V	15	c.1789C>G
BRAF	V600D	15	c.1799_1800delTGinsAT
BRAF	V600E	15	c.1799T>A
BRAF	V600E	15	c.1799_1800delTGinsAA
BRAF	V600G	15	c.1799T>G
BRAF	V600K	15	c.1798_1799delGTinsAA
BRAF	V600M	15	c.1798G>A
BRAF	V600R	15	c.1798_1799delGTinsAG

CTNNB1	S37F	3	c.110C>T
CTNNB1	S37Y	3	c.110C>A
CTNNB1	S45P	3	c.133T>C
CTNNB1	S45F	3	c.134C>T
CTNNB1	S45Y	3	c.134C>A
EGFR	G719A	18	c.2156G>C
EGFR	G719C	18	c.2155G>T
EGFR	G719S	18	c.2155G>A
EGFR	Exon 19 Deletions	19	
EGFR	Exon 19 Insertions	19	
EGFR	A763_Y764insFQEA	20	c.2290_2291ins
EGFR	Exon 20 Insertions	20	
EGFR	S768I	20	c.2303G>T
EGFR	T790M	20	c.2369C>T
EGFR	L858R	21	c.2573T>G
EGFR	L861Q	21	c.2582T>A
EGFR	**E746_A750>IP**		c.2235_2248delGGAATTAAGAGAAGinsAATTC
EGFR	**E746_A750del**		c.2235_2249delGGAATTAAGAGAAGC
EGFR	**E746_A750del**		c.2236_2250delGAATTAAGAGAAGCA
EGFR	**E746_P753>VS**		c.2237_2257del21insTCT
EGFR	**E746_S752>A**		c.2237_2254del18
EGFR	**E746_S752>D**		c.2238_2255del18
EGFR	**E746_S752>I**		c.2235_2255delinsAAT
EGFR	**E746_S752>V**		c.2237_2255delinsT
EGFR	**E746_T751>A**		c.2237_2251del15
EGFR	**E746_T751>I**		c.2235_2252delinsAAT
EGFR	**E746_T751>IP**		c.2235_2251delinsAATTC
EGFR	**E746_T751>V**		c.2237_2252delinsT
EGFR	**E746_T751>VA**		c.2237_2253delinsTTGCT
EGFR	**E746_T751del**		c.2236_2253del18
EGFR	**K745_E749del**		c.2233_2247del15)
EGFR	**L747_A750>P**		c.2238_2248delATTAAGAGAAGinsGC
EGFR	**L747_A750>P**		c.2239_2248delTTAAGAGAAGinsC
EGFR	**L747_E749del**		c.2239_2247delTTAAGAGAA
EGFR	**L747_P753>Q**		c.2239_2258delinsCA
EGFR	**L747_S752>Q**		c.2239_2256delinsCAA
EGFR	**L747_S752del**		c.2239_2256del18
EGFR	**L747_T751>Q**		c.2238_2252delinsGCA
EGFR	**L747_T751>S**		c.2240_2251del
EGFR	**L747_T751del**		c.2238_2252del
ERBB2(HER2)	G309A	8	c.926G>C
ERBB2(HER2)	D769H	19	c.2305G>C
ERBB2(HER2)	D769Y	19	c.2305G>T
ERBB2(HER2)	G776S	19	c.2326 G>A
ERBB2(HER2)	L755_T759del	19	c.2264_2278del
ERBB2(HER2)	L755S	19	c.2264T>C
ERBB2(HER2)	Exon 20 Insertions	20	
ERBB2(HER2)	G778_P780dup	20	c.2339_2340ins
ERBB2(HER2)	V777L	20	c.2329G>T
ERBB2(HER2)	V842I	21	c.2524G>A
ERBB2(HER2)	R896C	22	c.2686C>T
ERBB2(HER2)	c.2263_2264delTTinsCC		c.2263_2264delTTinsCC

ERBB2(HER2)	c.2322_2334dupATACGTGATGGC		c.2322_2334dupATACGTGATGGC
ERBB2(HER2)	c.2328_2336dupTGTGGGCTC		c.2328_2336dupTGTGGGCTC
ESR1	S463P		
ESR1	V534E		
ESR1	P535H		
ESR1	L536Q		
ESR1	L536R		
ESR1	Y537C		
ESR1	Y537S		
ESR1	Y537N		
ESR1	D538G		
FOXL2	C134W	1	c.402 C>G
GNA11	R183C	4	c.546_547delCCinsTT
GNA11	R183C	4	c.547C>T
GNA11	Q209L	5	c.626A>T
GNA11	Q209P	5	c.626A>C
GNAQ	R183Q	4	c.548G>A
GNAQ	Q209L	5	c.626A>T
GNAQ	Q209P	5	c.626A>C
GNAQ	Q209R	5	c.626A>G
KIT	A502–Y503insFA	9	c.1507_1508insTTGCCT
KIT	E490K	9	c.1468G>A
KIT	Exon 9 Mutation	9	
KIT	F504L	9	c.1510T>C
KIT	556 ins L	11	
KIT	575 ins PE	11	
KIT	Del 554–558	11	
KIT	Del 554–559	11	
KIT	Del 566–572	11	
KIT	Del 566–574	11	
KIT	Del 579	11	
KIT	Del V559	11	
KIT	E583_E589dupPYDHKWE	11	
KIT	Exon 11 Mutation	11	
KIT	G565V	11	
KIT	K550N	11	
KIT	K558N	11	
KIT	L576P	11	c.1727T>C
KIT	N566D	11	
KIT	P577_D579del	11	c.1730_1738del
KIT	V559A	11	c.1676T>C
KIT	V559D	11	c.1676T>A
KIT	V559G	11	
KIT	V560A	11	
KIT	V560D	11	c.1727T>C (V560D)
KIT	V560del	11	c.1679_1681del
KIT	V560G	11	
KIT	V569G	11	
KIT	W557R	11	c.1669T>A
KIT	W557R	11	c.1669T>C
KIT	Y553N	11	c.1657T>A
KIT	Exon 13 Mutation	13	
KIT	K642E	13	c.1924A>G
KIT	N655	13	
KIT	N655S	13	
KIT	R634W	13	

KIT	V654A	13	
KIT	Exon 14 Mutation	14	
KIT	H697Y	14	c.2089C>T
KIT	D816H	17	c.2446G>C
KIT	D816V	17	
KIT	D820E	17	c.2460T>A
KIT	D820V	17	
KIT	D820Y	17	
KIT	Exon 17 Mutation	17	
KIT	N822I	17	
KIT	N822K	17	
KIT	N822Y	17	
KIT	Y823D	17	
KIT	A829P	18	
KIT	I841V	18	
KIT	S864F	18	
KIT	Other KIT mutations		
KRAS	G12A	2	c.35G>C
KRAS	G12C	2	c.34G>T
KRAS	G12D	2	c.35G>A
KRAS	G12R	2	c.34G>C
KRAS	G12S	2	c.34G>A
KRAS	G12V	2	c.35G>T
KRAS	G13A	2	c.38G>C
KRAS	G13C	2	c.37G>T
KRAS	G13D	2	c.38G>A
KRAS	G13R	2	c.37G>C
KRAS	G13S	2	c.37G>A
KRAS	G13V	2	c.38G>T
KRAS	Q22K	2	c.64C>A
KRAS	Q61H	3	c.183A>C
KRAS	Q61H	3	c.183A>T
KRAS	Q61H	3	c.183A>C
KRAS	Q61K	3	c.181C>A
KRAS	Q61L	3	c.182A>T
KRAS	Q61P	3	c.182A>C
KRAS	Q61R	3	c.182A>G
KRAS	A146P	4	c.436G>C
KRAS	A146T	4	c.436G>A
KRAS	A146V	4	c.437C>T
KRAS	K117N	4	c.351A>C
KRAS	K117N	4	c.351A>T
MEK1 (MAP2K1)	D67N	2	c.199G>A
MEK1 (MAP2K1)	F53L	2	c.157T>C
MEK1 (MAP2K1)	K57N	2	c.171G>T
MEK1 (MAP2K1)	Q56P	2	c.167A>C
MEK1 (MAP2K1)	C121S	3	c.362G>C
MEK1 (MAP2K1)	E203K	3	c.607G>A
MEK1 (MAP2K1)	I111S	3	c.332T>G

MEK1 (MAP2K1)	N382H	3	c.1144A>C
MEK1 (MAP2K1)	P124L	3	c.371C>T
MEK1 (MAP2K1)	P124S	3	c.370C>T
MEK1 (MAP2K1)	P264S	3	c.790C>T
MET	c.2888-6_29del	14	c.2888-6_29del
MET	c. 3028G>C	14	c. 3028G>C
MET	c.2887-18_2887-7del12	14	c.2887-18_2887-7del12
MET	c.2888delA	14	c.2888delA
MET	c.3001_3021delGTAGACTACCGAG CTACTTTT	14	c.3001_3021delGTAGACTACCGAGC TACTTTT
MET	c.3024_3028+7delAGAAGGTATATT	14	c.3024_3028+7delAGAAGGTATATT
MET	c.3028+1G>T	14	c.3028+1G>T
MET	c.3028G>A	14	c.3028G>A
MET	c.3028G>T	14	c.3028G>T
MET	L1213V	18	c.3637 C>G
MET	V1206L	18	c.3616 G>T
NRAS	G12A	2	c.35G>C
NRAS	G12C	2	c.34G>T
NRAS	G12D	2	c.35G>A
NRAS	G12R	2	c.34G>C
NRAS	G12S	2	c.34G>A
NRAS	G12V	2	c.35G>T
NRAS	G13A	2	c.38G>C
NRAS	G13C	2	c.37G>T
NRAS	G13D	2	c.38G>A
NRAS	G13R	2	c.37G>C
NRAS	G13V	2	c.38G>T
NRAS	Q61E	3	c.181C>G
NRAS	Q61H	3	c.183A>C
NRAS	Q61H	3	c.183A>T
NRAS	Q61H	3	c.183A>T
NRAS	Q61K	3	c.181C>A
NRAS	Q61L	3	c.182A>T
NRAS	Q61L	3	c.182_183delAAinsTG
NRAS	Q61P	3	c.182A>C
NRAS	Q61R	3	c.182A>G
NRAS	Q61R	3	c.182_183delAAinsGG
PDGFRA	c.1679_1693delGGGTCATTGAATCA A		
PDGFRA	c.1681_1682insAGAGGG		
PDGFRA	c.1696_1713del18		
PDGFRA	c.2526_2537delCATCATGCATGA		
PDGFRA	c.2533_2544delCATGATTCGAAC		
PDGFRA	D842V	18	c.2525 A>T
PDGFRA	D846Y (c.2536 G>T)	18	
PDGFRA	Exon 12 Mutation	12	
PDGFRA	Exon 14 Mutation	14	
PDGFRA	Exon 18 Mutation	18	
PDGFRA	V561D (c.1682 T>A)		
PDGFRA	Y555C (c.1664 A>G)		
PIK3CA	D549N	9	c.1645G>A
PIK3CA	E542K	9	c.1624G>A

PIK3CA	E545G	9	c.1634A>G
PIK3CA	E545K	9	c.1633G>A
PIK3CA	E545Q	9	c.1633G>C
PIK3CA	E545V	9	c.1634A>T
PIK3CA	Q546E	9	c.1636C>G
PIK3CA	Q546K	9	c.1636C>A
PIK3CA	Q546L	9	c.1637A>T
PIK3CA	Q546P	9	c.1637A>C
PIK3CA	Q546R	9	c.1637A>G
PIK3CA	H1047R	20	c.3140A>G
PIK3CA	H1047L	20	c.3140A>T
PIK3CA	H1047Y	20	c.3139C>T
PIK3CA	M1043I	20	c.3129G>A
PTEN	R130*	5	c.388C>T
PTEN	R130fs*4	5	c.389delG
PTEN	R130G	5	c.388C>G
PTEN	R130Q	5	c.389G>A
PTEN	R159S	6	c.477G>T
PTEN	K267fs*9	7	c.800delA
PTEN	P248fs*5	7	c.741dupA
PTEN	R233*	7	c.697C>T
PTEN	N323fs*2	8	c.968supA
PTEN	N323fs*21	8	c.968delA
RET	C634 Mutations	11	
RET	M918T	16	
ROS1	G2032R		
ROS1	D2033N		
ROS1	L2155S		
SMAD4	E330A		c.989A>C
SMAD4	D351H		c.1051G>C
SMAD4	D351N		c.1051G>A
SMAD4	D355E		c.1065C>A
SMAD4	R361C		c.1081C>T
SMAD4	R361S		c.1081C>A
SMAD4	R361H		c.1082G>A
SMAD4	D537Y		c.1609G>T
TP53	Whole coding region	Exons 2-11	