

## 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	<a href="#">E17K</a>	3	c.49 G>A
ALK	<a href="#">D1091N</a>	20	<a href="#">c.3271G&gt;A</a>
ALK	<a href="#">I1171N</a>	22	<a href="#">c.3512T&gt;A</a>
ALK	<a href="#">T1151M</a>	22	<a href="#">c.3452C&gt;T</a>
ALK	<a href="#">F1174C</a>	23	<a href="#">c.3521T&gt;G</a>
ALK	<a href="#">F1174I</a>	23	<a href="#">c.3520T&gt;A</a>
ALK	<a href="#">F1174L</a>	23	<a href="#">c.3522C&gt;A</a>
ALK	<a href="#">F1174V</a>	23	<a href="#">c.3520T&gt;G</a>
ALK	<a href="#">D1225N</a>	24	c.3673G>A
ALK	<a href="#">F1245C</a>	24	<a href="#">c.3734T&gt;G</a>
ALK	<a href="#">F1245L</a>	24	<a href="#">c.3735C&gt;G</a>
ALK	<a href="#">F1245V</a>	24	<a href="#">c.3733T&gt;G</a>
ALK	<a href="#">R1275Q</a>	25	<a href="#">c.3824G&gt;A</a>
ALK	<a href="#">Y1278S</a>	25	<a href="#">c.3833A&gt;C</a>
ALK	1151Tins		
ALK	C1156Y		
ALK	G1202R		
ALK	G1269A		
ALK	L1152R		
ALK	L1196M		
ALK	L1198F		
ALK	S1206Y		
AR	<a href="#">L702H</a>	4	c.2105T>A
AR	<a href="#">W742C</a>	5	c.2226G>T
AR	<a href="#">H875Y</a>	8	c.2623C>T
AR	<a href="#">F877L</a>	8	c.2631C>A
AR	<a href="#">T878A</a>	8	c.2632A>G
BRAF	<a href="#">G466V</a>	11	c.1397G>T
BRAF	<a href="#">G469A</a>	11	c.1406G>C
BRAF	<a href="#">G469E</a>	11	c.1406G>A
BRAF	<a href="#">G469L</a>	11	c.1405_1406delGGinsTT
BRAF	<a href="#">G469V</a>	11	c.1406G>T
BRAF	<a href="#">Y472C</a>	11	c.1415A>G
BRAF	<a href="#">D594E</a>	15	c.1782T>A
BRAF	<a href="#">D594E</a>	15	c.1782T>G
BRAF	<a href="#">D594G</a>	15	c.1781A>G
BRAF	<a href="#">D594H</a>	15	c.1780G>C
BRAF	<a href="#">D594N</a>	15	c.1779_1780delTGinsGA
BRAF	<a href="#">D594N</a>	15	c.1780G>A
BRAF	<a href="#">D594V</a>	15	c.1781A>T
BRAF	<a href="#">G596R</a>	15	c.1786G>C
BRAF	<a href="#">K601E</a>	15	c.1801A>G
BRAF	<a href="#">L597Q</a>	15	c.1790T>A
BRAF	<a href="#">L597R</a>	15	c.1790T>G
BRAF	<a href="#">L597S</a>	15	c.1789_1790delICTinsTC
BRAF	<a href="#">L597V</a>	15	c.1789C>G
BRAF	<a href="#">V600D</a>	15	c.1799_1800delTGinsAT
BRAF	<a href="#">V600E</a>	15	c.1799T>A
BRAF	<a href="#">V600E</a>	15	c.1799_1800delTGinsAA
BRAF	<a href="#">V600G</a>	15	c.1799T>G
BRAF	<a href="#">V600K</a>	15	c.1798_1799delGTinsAA
BRAF	<a href="#">V600M</a>	15	c.1798G>A
BRAF	<a href="#">V600R</a>	15	c.1798_1799delGTinsAG

CTNNB1	<a href="#">S37F</a>	3	c.110C>T
CTNNB1	<a href="#">S37Y</a>	3	c.110C>A
CTNNB1	<a href="#">S45P</a>	3	c.133T>C
CTNNB1	<a href="#">S45F</a>	3	c.134C>T
CTNNB1	<a href="#">S45Y</a>	3	c.134C>A
EGFR	<a href="#">G719A</a>	18	c.2156G>C
EGFR	<a href="#">G719C</a>	18	c.2155G>T
EGFR	<a href="#">G719S</a>	18	c.2155G>A
EGFR	<a href="#">Exon 19 Deletions</a>	19	
EGFR	<a href="#">Exon 19 Insertions</a>	19	
EGFR	<a href="#">A763_Y764insFQEA</a>	20	c.2290_2291ins
EGFR	<a href="#">Exon 20 Insertions</a>	20	
EGFR	<a href="#">S768I</a>	20	c.2303G>T
EGFR	<a href="#">T790M</a>	20	c.2369C>T
EGFR	<a href="#">L858R</a>	21	c.2573T>G
EGFR	<a href="#">L861Q</a>	21	c.2582T>A
EGFR	<b>E746_A750&gt;IP</b>		c.2235_2248delGGAATTAAGAGAAGInsAATTC
EGFR	<b>E746_A750del</b>		c.2235_2249delGGAATTAAGAGAAGC
EGFR	<b>E746_A750del</b>		c.2236_2250delGAATTAAGAGAAGCA
EGFR	<b>E746_P753&gt;VS</b>		c.2237_2257del21insTCT
EGFR	<b>E746_S752&gt;A</b>		c.2237_2254del18
EGFR	<b>E746_S752&gt;D</b>		c.2238_2255del18
EGFR	<b>E746_S752&gt;I</b>		c.2235_2255delinsAAT
EGFR	<b>E746_S752&gt;V</b>		c.2237_2255delinsT
EGFR	<b>E746_T751&gt;A</b>		c.2237_2251del15
EGFR	<b>E746_T751&gt;I</b>		c.2235_2252delinsAAT
EGFR	<b>E746_T751&gt;IP</b>		c.2235_2251delinsAATTC
EGFR	<b>E746_T751&gt;V</b>		c.2237_2252delinsT
EGFR	<b>E746_T751&gt;VA</b>		c.2237_2253delinsTTGCT
EGFR	<b>E746_T751del</b>		c.2236_2253del18
EGFR	<b>K745_E749del</b>		c.2233_2247del15)
EGFR	<b>L747_A750&gt;P</b>		c.2238_2248delATTAAGAGAAGInsGC
EGFR	<b>L747_A750&gt;P</b>		c.2239_2248delTTAAGAGAAGInsC
EGFR	<b>L747_E749del</b>		c.2239_2247delTTAAGAGAA
EGFR	<b>L747_P753&gt;Q</b>		c.2239_2258delinsCA
EGFR	<b>L747_S752&gt;Q</b>		c.2239_2256delinsCAA
EGFR	<b>L747_S752del</b>		c.2239_2256del18
EGFR	<b>L747_T751&gt;Q</b>		c.2238_2252delinsGCA
EGFR	<b>L747_T751&gt;S</b>		c.2240_2251del
EGFR	<b>L747_T751del</b>		c.2238_2252del
ERBB2(HER2)	<a href="#">G309A</a>	8	c.926G>C
ERBB2(HER2)	<a href="#">D769H</a>	19	c.2305G>C
ERBB2(HER2)	<a href="#">D769Y</a>	19	c.2305G>T
ERBB2(HER2)	<a href="#">G776S</a>	19	c.2326 G>A
ERBB2(HER2)	<a href="#">L755_T759del</a>	19	c.2264_2278del
ERBB2(HER2)	<a href="#">L755S</a>	19	c.2264T>C
ERBB2(HER2)	<a href="#">Exon 20 Insertions</a>	20	
ERBB2(HER2)	<a href="#">G778_P780dup</a>	20	c.2339_2340ins
ERBB2(HER2)	<a href="#">V777L</a>	20	c.2329G>T
ERBB2(HER2)	<a href="#">V842I</a>	21	c.2524G>A
ERBB2(HER2)	<a href="#">R896C</a>	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	<a href="#">R183C</a>	4	c.546_547delCCinsTT
GNA11	<a href="#">R183C</a>	4	c.547C>T
GNA11	<a href="#">Q209L</a>	5	c.626A>T
GNA11	<a href="#">Q209P</a>	5	c.626A>C
GNAQ	<a href="#">R183Q</a>	4	c.548G>A
GNAQ	<a href="#">Q209L</a>	5	c.626A>T
GNAQ	<a href="#">Q209P</a>	5	c.626A>C
GNAQ	<a href="#">Q209R</a>	5	c.626A>G
KIT	A502-Y503insFA	9	c.1507_1508insTTGCCT
KIT	<a href="#">E490K</a>	9	c.1468G>A
KIT	<a href="#">Exon 9 Mutation</a>	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	<a href="#">Exon 11 Mutation</a>	11	
KIT	G565V	11	
KIT	K550N	11	
KIT	K558N	11	
KIT	<a href="#">L576P</a>	11	c.1727T>C
KIT	N566D	11	
KIT	<a href="#">P577_D579del</a>	11	c.1730_1738del
KIT	<a href="#">V559A</a>	11	c.1676T>C
KIT	<a href="#">V559D</a>	11	c.1676T>A
KIT	V559G	11	
KIT	V560A	11	
KIT	V560D	11	c.1727T>C (V560D)
KIT	<a href="#">V560del</a>	11	c.1679_1681del
KIT	V560G	11	
KIT	V569G	11	
KIT	<a href="#">W557R</a>	11	c.1669T>A
KIT	<a href="#">W557R</a>	11	c.1669T>C
KIT	<a href="#">Y553N</a>	11	c.1657T>A
KIT	<a href="#">Exon 13 Mutation</a>	13	
KIT	<a href="#">K642E</a>	13	c.1924A>G
KIT	N655	13	
KIT	N655S	13	
KIT	R634W	13	

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

MEK1 (MAP2K1)	<a href="#">N382H</a>	3	c.1144A>C
MEK1 (MAP2K1)	<a href="#">P124L</a>	3	c.371C>T
MEK1 (MAP2K1)	<a href="#">P124S</a>	3	c.370C>T
MEK1 (MAP2K1)	<a href="#">P264S</a>	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_3021delGTAGACTACCGAGCTACTTTT	14	c.3001_3021delGTAGACTACCGAGCTACTTTT
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	<a href="#">G12A</a>	2	c.35G>C
NRAS	<a href="#">G12C</a>	2	c.34G>T
NRAS	<a href="#">G12D</a>	2	c.35G>A
NRAS	<a href="#">G12R</a>	2	c.34G>C
NRAS	<a href="#">G12S</a>	2	c.34G>A
NRAS	<a href="#">G12V</a>	2	c.35G>T
NRAS	<a href="#">G13A</a>	2	c.38G>C
NRAS	<a href="#">G13C</a>	2	c.37G>T
NRAS	<a href="#">G13D</a>	2	c.38G>A
NRAS	<a href="#">G13R</a>	2	c.37G>C
NRAS	<a href="#">G13V</a>	2	c.38G>T
NRAS	<a href="#">Q61E</a>	3	c.181C>G
NRAS	<a href="#">Q61H</a>	3	c.183A>C
NRAS	<a href="#">Q61H</a>	3	c.183A>T
NRAS	<a href="#">Q61H</a>	3	c.183A>T
NRAS	<a href="#">Q61K</a>	3	c.181C>A
NRAS	<a href="#">Q61L</a>	3	c.182A>T
NRAS	<a href="#">Q61L</a>	3	c.182_183delAAinsTG
NRAS	<a href="#">Q61P</a>	3	c.182A>C
NRAS	<a href="#">Q61R</a>	3	c.182A>G
NRAS	<a href="#">Q61R</a>	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	<a href="#">D842V</a>	18	c.2525 A>T
PDGFRA	D846Y (c.2536 G>T)	18	
PDGFRA	<a href="#">Exon 12 Mutation</a>	12	
PDGFRA	<a href="#">Exon 14 Mutation</a>	14	
PDGFRA	<a href="#">Exon 18 Mutation</a>	18	
PDGFRA	V561D (c.1682 T>A)		
PDGFRA	Y555C (c.1664 A>G)		
PIK3CA	<a href="#">D549N</a>	9	c.1645G>A
PIK3CA	<a href="#">E542K</a>	9	c.1624G>A

PIK3CA	<a href="#">E545G</a>	9	c.1634A>G
PIK3CA	<a href="#">E545K</a>	9	c.1633G>A
PIK3CA	<a href="#">E545Q</a>	9	c.1633G>C
PIK3CA	<a href="#">E545V</a>	9	c.1634A>T
PIK3CA	<a href="#">Q546E</a>	9	c.1636C>G
PIK3CA	<a href="#">Q546K</a>	9	c.1636C>A
PIK3CA	<a href="#">Q546L</a>	9	c.1637A>T
PIK3CA	<a href="#">Q546P</a>	9	c.1637A>C
PIK3CA	<a href="#">Q546R</a>	9	c.1637A>G
PIK3CA	<a href="#">H1047R</a>	20	c.3140A>G
PIK3CA	<a href="#">H1047L</a>	20	c.3140A>T
PIK3CA	H1047Y	20	c.3139C>T
PIK3CA	M1043I	20	c.3129G>A
PTEN	<a href="#">R130*</a>	5	c.388C>T
PTEN	<a href="#">R130fs*4</a>	5	c.389delG
PTEN	<a href="#">R130G</a>	5	c.388C>G
PTEN	<a href="#">R130Q</a>	5	c.389G>A
PTEN	<a href="#">R159S</a>	6	c.477G>T
PTEN	<a href="#">K267fs*9</a>	7	c.800delA
PTEN	<a href="#">P248fs*5</a>	7	c.741dupA
PTEN	<a href="#">R233*</a>	7	c.697C>T
PTEN	<a href="#">N323fs*2</a>	8	c.968supA
PTEN	<a href="#">N323fs*21</a>	8	c.968delA
RET	<a href="#">C634 Mutations</a>	11	
RET	<a href="#">M918I</a>	16	
ROS1	G2032R		
ROS1	D2033N		
ROS1	L2155S		
SMAD4	<a href="#">E330A</a>		c.989A>C
SMAD4	<a href="#">D351H</a>		c.1051G>C
SMAD4	<a href="#">D351N</a>		c.1051G>A
SMAD4	<a href="#">D355E</a>		c.1065C>A
SMAD4	<a href="#">R361C</a>		c.1081C>T
SMAD4	<a href="#">R361S</a>		c.1081C>A
SMAD4	<a href="#">R361H</a>		c.1082G>A
SMAD4	<a href="#">D537Y</a>		c.1609G>T
TP53	Whole coding region	Exons 2-11	