

**Γονίδια που αναλύονται και η συσχέτισή τους με διαφορετικούς τύπους καρκίνου.**

Γονίδιο	Τύποι καρκίνου
AKT1	Μαστού, Πνεύμονα, Ορθοκολικός*
ALK	Πνεύμονα, Νευροβλάστωμα, Ραβδομυοσάρκωμα
AR	Προστάτη
BRAF	Μελάνωμα*, Ορθοκολικός* Πνεύμονα, Ωοθηκών, Στομάχου, Γλίωμα, Θυροειδούς, Πάγκρεας,
CTNNB1	Μελάνωμα
EGFR	Πνεύμονα*; Κεφαλής και τραχήλου, Προστάτη
ERBB2	Μαστού, Πνεύμονα
ESR1	Μαστού
FOXL2	Ωοθηκών
GNA11	Μελάνωμα
GNAQ	Μελάνωμα
KIT	Στομάχου, Μελάνωμα*, Θύμου αδένα
KRAS	Ορθοκολικός*, Στομάχου, Πνεύμονα*, Ωοθηκών, Θυροειδούς, Ενδομητρίου, Πάγκρεας, Προστάτη
MEK1 (MAP2K1)	Μελάνωμα, Πνεύμονα, Ωοθηκών, Ορθοκολικός,
MET	Πνεύμονα*, Ορθοκολικός, Στομάχου
NRAS	Ορθοκολικός*, Πνεύμονα, Μελάνωμα, Θυροειδούς
PDGFRA	Στομάχου, Μελάνωμα,
PIK3CA	Πνεύμονα, Μαστού, Προστάτη, Ορθοκολικός, Ωοθηκών, Κεφαλής και τραχήλου, Πάγκρεας,
PTEN	Μαστού, Πνεύμονα,
RET	Πνεύμονα*, Θυροειδούς
ROS1	Πνεύμονα
SMAD4	Ορθοκολικός
TP53	Πνεύμονα, Μελάνωμα, Ωοθηκών, Ορθοκολικός, Μαστού; Ενδομητρίου, Κεφαλής και τραχήλου, Νεφρού, Πάγκρεας, Προστάτη, Θυροειδούς

\*Σύμφωνα με τις κατευθυντήριες οδηγίες της NCNN.

Μεταλλάξεις που αναλύονται με το OncoNext™ Liquid 23 γονίδια

GENE	MUTATION Protein	MUTATION cDNA
<b>AKT1</b>	E17K	c.49 G>A
<b>ALK</b>	1151Tins	
<b>ALK</b>	L1152R	c.3455T>G
<b>ALK</b>	D1091N	c.3271G>A
<b>ALK</b>	T1151M	c.3452C>T
<b>ALK</b>	C1156Y	c.3467G>A
<b>ALK</b>	I1171N	c.3512T>A
<b>ALK</b>	F1174I	c.3520T>A
<b>ALK</b>	F1174V	c.3520T>G
<b>ALK</b>	F1174C	c.3521T>G
<b>ALK</b>	F1174L	c.3522C>A
<b>ALK</b>	L1196M	c.3586C>A
<b>ALK</b>	L1198F	c.3592C>T
<b>ALK</b>	S1206Y	c.36+A4:C2317C>A
<b>ALK</b>	G1202R	c.3604G>A
<b>ALK</b>	D1225N	c.3673G>A
<b>ALK</b>	F1245V	c.3733T>G
<b>ALK</b>	F1245C	c.3734T>G
<b>ALK</b>	F1245L	c.3735C>G
<b>ALK</b>	G1269A	c.3806G>C
<b>ALK</b>	R1275Q	c.3824G>A
<b>ALK</b>	Y1278S	c.3833A>C
<b>AR</b>	L702H	c.2105T>A
<b>AR</b>	W742C	c.2226G>T
<b>AR</b>	H875Y	c.2623C>T
<b>AR</b>	F877L	c.2631C>A
<b>AR</b>	T878A	c.2632A>G
<b>BRAF</b>	G466V	c.1397G>T
<b>BRAF</b>	G469A	c.1406G>C
<b>BRAF</b>	G469E	c.1406G>A
<b>BRAF</b>	G469L	c.1405_1406delGGinsTT
<b>BRAF</b>	G469V	c.1406G>T
<b>BRAF</b>	Y472C	c.1415A>G
<b>BRAF</b>	D594E	c.1782T>A
<b>BRAF</b>	D594E	c.1782T>G
<b>BRAF</b>	D594G	c.1781A>G
<b>BRAF</b>	D594H	c.1780G>C
<b>BRAF</b>	D594N	c.1779_1780delTGinsGA
<b>BRAF</b>	D594N	c.1780G>A
<b>BRAF</b>	D594V	c.1781A>T
<b>BRAF</b>	G596R	c.1786G>C
<b>BRAF</b>	K601E	c.1801A>G
<b>BRAF</b>	L597Q	c.1790T>A

<b>BRAF</b>	L597R	c.1790T>G
<b>BRAF</b>	L597S	c.1789_1790delCTinsTC
<b>BRAF</b>	L597V	c.1789C>G
<b>BRAF</b>	V600D	c.1799_1800delTGinsAT
<b>BRAF</b>	V600E	c.1799T>A
<b>BRAF</b>	V600E	c.1799_1800delTGinsAA
<b>BRAF</b>	V600G	c.1799T>G
<b>BRAF</b>	V600K	c.1798_1799delGTinsAA
<b>BRAF</b>	V600M	c.1798G>A
<b>BRAF</b>	V600R	c.1798_1799delGTinsAG
<b>CTNNB1</b>	S37F	c.110C>T
<b>CTNNB1</b>	S37Y	c.110C>A
<b>CTNNB1</b>	S45P	c.133T>C
<b>CTNNB1</b>	S45F	c.134C>T
<b>CTNNB1</b>	S45Y	c.134C>A
<b>EGFR</b>	Exon 19 Deletions	
<b>EGFR</b>	Exon 19 Insertions	
<b>EGFR</b>	Exon 20 Insertions	
<b>EGFR</b>	G719S	c.2155G>A
<b>EGFR</b>	G719C	c.2155G>T
<b>EGFR</b>	G719A	c.2156G>C
<b>EGFR</b>	K745_E749del	c.2233_2247del15)
<b>EGFR</b>	E746_A750>IP	c.2235_2248delGGAATTAAGAGAAGinsAATTC
<b>EGFR</b>	E746_A750del	c.2235_2249delGGAATTAAGAGAAGC
<b>EGFR</b>	E746_T751>IP	c.2235_2251delinsAATTC
<b>EGFR</b>	E746_T751>I	c.2235_2252delinsAAT
<b>EGFR</b>	E746_S752>I	c.2235_2255delinsAAT
<b>EGFR</b>	E746_A750del	c.2236_2250delGAATTAAGAGAAGCA
<b>EGFR</b>	E746_T751del	c.2236_2253del18
<b>EGFR</b>	E746_T751>A	c.2237_2251del15
<b>EGFR</b>	E746_T751>V	c.2237_2252delinsT
<b>EGFR</b>	E746_T751>VA	c.2237_2253delinsTTGCT
<b>EGFR</b>	E746_S752>A	c.2237_2254del18
<b>EGFR</b>	E746_S752>V	c.2237_2255delinsT
<b>EGFR</b>	E746_P753>VS	c.2237_2257del21insTCT
<b>EGFR</b>	L747_A750>P	c.2238_2248delATTAAGAGAAGinsGC
<b>EGFR</b>	L747_T751del	c.2238_2252del
<b>EGFR</b>	L747_T751>Q	c.2238_2252delinsGCA
<b>EGFR</b>	E746_S752>D	c.2238_2255del18
<b>EGFR</b>	L747_E749del	c.2239_2247delTTAAGAGAA
<b>EGFR</b>	L747_A750>P	c.2239_2248delTTAAGAGAAGinsC
<b>EGFR</b>	L747_S752del	c.2239_2256del18
<b>EGFR</b>	L747_S752>Q	c.2239_2256delinsCAA

EGFR	L747_P753>Q	c.2239_2258delinsCA
EGFR	L747_T751>S	c.2240_2251del
EGFR	A763_Y764insFQEA	c.2290_2291ins
EGFR	S768I	c.2303G>T
EGFR	T790M	c.2369C>T
EGFR	L858R	c.2573T>G
EGFR	L861Q	c.2582T>A
ERBB2(HER2)	Exon 20 Insertions	
ERBB2(HER2)		c.2263_2264delTTinsCC
ERBB2(HER2)	L755_T759del	c.2264_2278del
ERBB2(HER2)	p.L755S	c.2264T>C
ERBB2(HER2)	D769H	c.2305G>C
ERBB2(HER2)	D769Y	c.2305G>T
ERBB2(HER2)		c.2322_2334dupATACGTGATGGC
ERBB2(HER2)	G776S	c.2326 G>A
ERBB2(HER2)		c.2328_2336dupTGTGGGCTC
ERBB2(HER2)	V777L	c.2329G>T
ERBB2(HER2)	G778_P780dup	c.2339_2340ins
ERBB2(HER2)	V842I	c.2524G>A
ERBB2(HER2)	R896C	c.2686C>T
ERBB2(HER2)	G309A	c.926G>C
ESR1	S463P	c.1387T>C
ESR1	V534E	c.1601T>A
ESR1	P535H	c.?
ESR1	L536Q	c.1607_1608TC>AG
ESR1	L536R	c.1607T>G
ESR1	Y537N	c.1609T>A
ESR1	Y537S	c.1610A>C
ESR1	Y537C	c.1610A>G
ESR1	D538G	c.1613A>G
FOXL2	C134W	c.402 C>G
GNA11	R183C	c.546_547delCCinsTT
GNA11	R183C	c.547C>T
GNA11	Q209L	c.626A>T
GNA11	Q209P	c.626A>C
GNAQ	R183Q	c.548G>A
GNAQ	Q209L	c.626A>T
GNAQ	Q209P	c.626A>C
GNAQ	Q209R	c.626A>G
KIT	Del 554–558	
KIT	Del 554–559	
KIT	Del 566–572	
KIT	Del 566–574	
KIT	Del 579	
KIT	Del V559	
KIT	Exon 9 Mutations	

KIT	Exon 11 Mutations	
KIT	Exon 13 Mutations	
KIT	Exon 14 Mutations	
KIT	Exon 17 Mutations	
KIT	Other KIT mutations	
KIT	E490K	c.1468G>A
KIT	A502–Y503insFA	c.1507_1508insTTGCCT
KIT	F504L	c.1510T>C
KIT	K550N	c.1650A>C
KIT	Y553N	c.1657T>A
KIT	556 ins L	
KIT	W557R	c.1669T>A
KIT	W557R	c.1669T>C
KIT	K558N	c.1674G>C
KIT	V559D	c.1676T>A
KIT	V559A	c.1676T>C
KIT	V559G	c.1676T>G
KIT	V560del	c.1679_1681del
KIT	V560A	c.1679T>C
KIT	V560G	c.1679T>G
KIT	G565V	c.1694G>T
KIT	N566D	c.1696A>G
KIT	V569G	c.1706T>G
KIT	575 ins PE	
KIT	L576P	c.1727T>C
KIT	V560D	c.1727T>C (V560D)
KIT	P577_D579del	c.1730_1738del
KIT	E583_E589dupPYDHKWE	
KIT	R634W	c.1900C>T
KIT	K642E	c.1924A>G
KIT	V654A	c.1961T>C
KIT	N655K	c.1965T>A,c.1965T>G
KIT	N655S	
KIT	H697Y	c.2089C>T
KIT	D816H	c.2446G>C
KIT	D816V	c.2447A>T
KIT	D820Y	c.2458G>T
KIT	D820V	c.2459A>T
KIT	D820E	c.2460T>A
KIT	N822Y	c.2464A>T
KIT	N822I	c.2465A>T
KIT	N822K	c.2466T>G, c.2466T>A
KIT	Y823D	c.2467T>G
KIT	A829P	c.2485G>C
KIT	I841V	
KIT	S864F	c.2591C>T
<b>KRAS</b>	G12S	c.34G>A

KRAS	G12R	c.34G>C
KRAS	G12C	c.34G>T
KRAS	K117N	c.351A>C
KRAS	K117N	c.351A>T
KRAS	G12D	c.35G>A
KRAS	G12A	c.35G>C
KRAS	G12V	c.35G>T
KRAS	G13S	c.37G>A
KRAS	G13R	c.37G>C
KRAS	G13C	c.37G>T
KRAS	G13D	c.38G>A
KRAS	G13A	c.38G>C
KRAS	G13V	c.38G>T
KRAS	Q22K	c.64C>A
KRAS	Q61K	c.181C>A
KRAS	Q61P	c.182A>C
KRAS	Q61R	c.182A>G
KRAS	Q61L	c.182A>T
KRAS	Q61H	c.183A>C
KRAS	Q61H	c.183A>C
KRAS	Q61H	c.183A>T
KRAS	A146T	c.436G>A
KRAS	A146P	c.436G>C
KRAS	A146V	c.437C>T
<hr/>		
MEK1 (MAP2K1)	F53L	c.157T>C
MEK1 (MAP2K1)	Q56P	c.167A>C
MEK1 (MAP2K1)	K57N	c.171G>T
MEK1 (MAP2K1)	D67N	c.199G>A
MEK1 (MAP2K1)	I111S	c.332T>G
MEK1 (MAP2K1)	C121S	c.362G>C
MEK1 (MAP2K1)	P124S	c.370C>T
MEK1 (MAP2K1)	P124L	c.371C>T
MEK1 (MAP2K1)	E203K	c.607G>A
MEK1 (MAP2K1)	P264S	c.790C>T
MEK1 (MAP2K1)	N382H	c.1144A>C
<hr/>		
MET		c.2887-18_2887-7del12
MET		c.2888-6_29del
MET		c.2888delA
MET		c.3001_3021delGTAGACTACCGAGC TACTTTT
MET		c.3024_3028+7delAGAAGGTATATT
MET		c.3028+1G>T
MET		c.3028G>A
MET		c.3028G>C
MET		c.3028G>T
MET	V1206L	c.3616 G>T

<b>MET</b>	L1213V	c.3637 C>G
<b>NRAS</b>	G12S	c.34G>A
<b>NRAS</b>	G12R	c.34G>C
<b>NRAS</b>	G12C	c.34G>T
<b>NRAS</b>	G12D	c.35G>A
<b>NRAS</b>	G12A	c.35G>C
<b>NRAS</b>	G12V	c.35G>T
<b>NRAS</b>	G13R	c.37G>C
<b>NRAS</b>	G13C	c.37G>T
<b>NRAS</b>	G13D	c.38G>A
<b>NRAS</b>	G13A	c.38G>C
<b>NRAS</b>	G13V	c.38G>T
<b>NRAS</b>	Q61K	c.181C>A
<b>NRAS</b>	Q61E	c.181C>G
<b>NRAS</b>	Q61R	c.182_183delAAinsGG
<b>NRAS</b>	Q61L	c.182_183delAAinsTG
<b>NRAS</b>	Q61P	c.182A>C
<b>NRAS</b>	Q61R	c.182A>G
<b>NRAS</b>	Q61L	c.182A>T
<b>NRAS</b>	Q61H	c.183A>C
<b>NRAS</b>	Q61H	c.183A>T
<b>NRAS</b>	Q61H	c.183A>T
<b>PDGFRA</b>	Exon 12 Mutations	
<b>PDGFRA</b>	Exon 14 Mutations	
<b>PDGFRA</b>	Exon 18 Mutations	
<b>PDGFRA</b>	Y555C	c.1664 A>G
<b>PDGFRA</b>		c.1679_1693delGGGTCATTGAATCAA
<b>PDGFRA</b>		c.1681_1682insAGAGGG
<b>PDGFRA</b>	V561D	c.1682 T>A
<b>PDGFRA</b>		c.1696_1713del18
<b>PDGFRA</b>	D842V	c.2525 A>T
<b>PDGFRA</b>		c.2526_2537delCATCATGCATGA
<b>PDGFRA</b>		c.2533_2544delCATGATTCGAAC
<b>PDGFRA</b>	D846Y	c.2536 G>T
<b>PIK3CA</b>	E542K	c.1624G>A
<b>PIK3CA</b>	E545K	c.1633G>A
<b>PIK3CA</b>	E545Q	c.1633G>C
<b>PIK3CA</b>	E545G	c.1634A>G
<b>PIK3CA</b>	E545V	c.1634A>T
<b>PIK3CA</b>	Q546K	c.1636C>A
<b>PIK3CA</b>	Q546E	c.1636C>G
<b>PIK3CA</b>	Q546P	c.1637A>C
<b>PIK3CA</b>	Q546R	c.1637A>G
<b>PIK3CA</b>	Q546L	c.1637A>T
<b>PIK3CA</b>	D549N	c.1645G>A
<b>PIK3CA</b>	M1043I	c.3129G>A

<b>PIK3CA</b>	H1047Y	c.3139C>T
<b>PIK3CA</b>	H1047R	c.3140A>G
<b>PIK3CA</b>	H1047L	c.3140A>T
<b>PTEN</b>	R130G	c.388C>G
<b>PTEN</b>	R130*	c.388C>T
<b>PTEN</b>	R130fs*4	c.389delG
<b>PTEN</b>	R130Q	c.389G>A
<b>PTEN</b>	R159S	c.477G>T
<b>PTEN</b>	R233*	c.697C>T
<b>PTEN</b>	P248fs*5	c.741dupA
<b>PTEN</b>	K267fs*9	c.800delA
<b>PTEN</b>	N323fs*21	c.968delA
<b>PTEN</b>	N323fs*2	c.968supA
<b>RET</b>	C634 Mutations	c.1900T>C c.1901G>A c.1902C>G c.1900T>A c.1901G>T
<b>RET</b>	M918T	c.2753T>C
<b>ROS1</b>	G2032R	
<b>ROS1</b>	D2033N	
<b>ROS1</b>	L2155S	
<b>SMAD4</b>	E330A	c.989A>C
<b>SMAD4</b>	D351N	c.1051G>A
<b>SMAD4</b>	D351H	c.1051G>C
<b>SMAD4</b>	D355E	c.1065C>A
<b>SMAD4</b>	R361S	c.1081C>A
<b>SMAD4</b>	R361C	c.1081C>T
<b>SMAD4</b>	R361H	c.1082G>A
<b>SMAD4</b>	D537Y	c.1609G>T
<b>TP53</b>	Whole coding region	



