

# Dissection of major cancer gene variants in subsets of circulating tumor cells in advanced breast cancer

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**Supplementary tables. Sequence variants in major driver genes identified in FFPE primary tumor samples and CTC subsets.**

Abbreviations: M+/E-: CTCs expressing only mesenchymal markers; M-/E-: CTCs which do not express both mesenchymal and epithelial markers; M+/E+: CTCs expressing both mesenchymal and epithelial markers; M-/E+: CTCs expressing only epithelial markers; ND: not detectable.

The sequence variants written in red represent pathogenic mutations, while those in orange are of a uncertain clinical significance. The red boxes indicate the presence of the sequence variant, the green boxes indicate the wild type status. For each sequence variant the allele frequency (%) is also reported.

pt. #123							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E+	CTC M+/E-
APC	c.4479G>A	p.T1493T	COSM3760869	51.13	51.4	44.10	55.7
APC	c.4533C>T	p.Leu1511	rs150089434			8.00	5.80
AKT1	c.480T>C	p.T160T	-				7.45
AKT1	c.543C>A	p.L181L	-				7.86
AKT1	c.474C>A	p.K159K	-				8.05
AKT1	c.486C>G	p.G162G	-				8.29
ATM	c.1023C>T	p.V341V	rs35728619		24.8		11.3
ATM	c.7328G>A	p.R2443Q	COSM20404		25.5		12.3
CDH1	c.1043C>T	p.A348V	-				5.45
CSF1R	c.1841TG>GA	p.?	rs386693509	28.00	27.5	38.93	39.7
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.45	99.6	69.47	99.6
ERBB2	c.2638G>A	p.D880N	COSM7002319				5.32
ERBB4	c.421+58A>G	p.?	rs839541			32.72	
ERBB4	c.729C>T	p.D243D	rs755321591	52.48	44.7	29.09	46.7
FBXW7	c.1737G>A	p.G580G	-				6.00
FBXW7	c.1776T>C	p.N593N	-				6.01
FBXW7	c.1716T>C	p.N572N	-				6.05
FGFR2	c.1654G>A	p.G552R	-				5.85
FGFR3	c.1953G>A	p.T651T	rs7688609	99.58	99.6	99.81	99.7
FLT3	c.1310-3T>C	p.?	rs2491231	48.08	50.3	60.53	55.8
GNA11	c.606-10G>A	p.?	-				5.90
HRA5	c.81T>C	p.H27H	COSM249860	86.22	98.5	60.16	99.7
IDH2	c.483C>T	p.V161V	-				5.75
IDH2	c.477C>T	p.R159R	-				7.05
KDR	c.798+54G>A	p.?	rs7692791	36.89	34.0	60.86	42.7
KDR	c.3849-24C>A	p.?	rs10006115			9.85	
KDR	c.2679T>G	p.G893G	-			5.00	5.33
KIT	c.2538C>T	p.K786K	-			6.50	
MET	c.2962C>T	p.R988C	COSM1666978 rs34589476	46.34	50.1	40.18	43.0
NOTCH1	c.4768T>C	p.F1590L	-				5.04
NOTCH1	c.4702A>C	p.R1568R	-				5.13
NRAS	c.51C>T	p.S17S	-			5.35	
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.8	100	99.9
PDGFRA	c.2472C>T	p.V824V	COSM22413	48.27	50.6	32.82	56.6
PIK3CA	c.352+40A>G	p.?	rs3729674			9.35	
PIK3CA	c.1624G>C	p.E542Q	COSM17442		27.4		
PIK3CA	c.3145G>C	p.G1049R	COSM12597		31.6		
PIK3CA	c.3270G>A	p.?	rs183721599			7.06	
PIK3CA	c.3141T>C	p.H1047H	-			6.35	
PIK3CA	c.3081C>T	p.A1027A	-			6.00	
PIK3CA	c.3054C>T	3'UTR	-			6.30	
PIK3CA	c.3282C>T	3'UTR	-			6.50	
PIK3CA	c.3266C>G	3'UTR	-			6.60	
PIK3CA	c.3245C>T	3'UTR	-			6.85	
RET	c.2307G>T	p.L769L	rs1800861	100	99.0	90.23	99.9
RET	c.2712C>G	p.S904S	rs1800836	48.97	44.7	55.81	46.4
SMAD4	c.1335A>G	p. R445R	-			11.00	17.00
SMAD4	c.1389A>G	p.A463A	-			6.96	
SMAD4	c.1410T>C	p.P470P	-			6.81	
SMAD4	c.1332T>C	p.H444H	-			8.94	
SMAD4	c.1059C>T	p.Y353Y	rs863224400				11.3
SMAD4	c.1062G>A	p.V354V	-				11.35
SMAD4	c.512C>A	p.Ser171*	COSM1151109				6.16
SMAD4	c.1089T>C	p.C363C	-				11.7
SMAD4	c.1035C>T	p.C345C	-				12.4
SMAD4	c.1002G>A	p.Q334Q	rs773598775				12.5
SMAD4	c.990A>G	p.E330E	-				12.6
SMARCB1	c.1119-41G>A	p.?	COSM1090			30.60	
SMO	c.1623G>A	p.T541T	COSM1085436			25.40	34.5
SMO	c.1566G>A	p.L522L	-			25.78	32.8
SMO	c.1539G>C	p.P513P	-			19.06	33.1
SMO	c.1545T>C	p.L515L	-			26.16	33.9
SMO	c.1590_1591delCGG nsTT	p.IA530_531IS	-			25.84	34.2
STK11	c.976C>A	p.P326T	rs771632414	49.77	53.1	39.57	43.9
STK11	c.135C>T	p.L45L	rs1362065365				5.35
STK11	c.174C>G	p.G58G	rs756661578			5.65	5.35
STK11	c.171A>G	p.E57E	rs1060503785			5.70	5.55
STK11	c.177T>G	p.S59S	-			5.00	5.86
STK11	c.72G>A	p.T24T	-			5.30	6.00
STK11	c.141C>A	p.G47G	-			5.71	6.47
STK11	c.84C>G	p.R28R	-			5.00	5.91
STK11	c.114G>C	p.P38P	-			5.41	6.20
TP53	c.215C>G	p.P72R	COSM250061	54.45	49.6	61.36	55.0

pt. #242								
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M-/E+	CTC M+/E+	CTC M+/E-	CTC M-/E-
ABL1	c.750G>C	p.G250G	-				15.24	
ABL1	c.774G>A	p.E258E	-				17.64	
ABL1	c.747C>G	p.G249G	-				17.45	
ABL1	c.714A>G	p.E238E	-				17.58	
APC	c.4479G>A	p.T1493T	COSM3760869	50.30	53.94	49.12	50.82	53.24
ATM	<b>c.1810C&gt;T</b>	<b>p.P604S</b>	<b>COSM22499</b>			6.91		
ATM	c.1874A>G	p.N625S	-				8.85	
CDKN2A	c.234C>T	p.H74H	-				13.78	
CSF1R	c.*1841TG>GA	p.?	rs386693509	99.70	99.57	99.41	99.77	100
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.65	100	100	100	99.45
ERBB4	c.421+58A>G	p.?	rs839541	48.70	54.75	51.35	64.00	49.53
FBXW7	<b>c1702G&gt;A</b>	<b>p.V568M</b>	-				10.60	
FBXW7	c1704G>A	p.V568V	-				10.50	
FBXW7	c.1737G>A	p.G580G	-		8.76		10.60	
FBXW7	c.1776T>C	p.N593N	-		9.00		10.26	
FBXW7	c.1716T>C	p.N572N	-		8.45		10.51	
FGFR3	c.1953G>A	p.T651T	rs7688609	99.86	99.87	99.73	99.70	98.97
FLT3	c.1310-3T>C	p.?	rs2491231	52.55	56.68	57.85	56.90	51.30
HRAS	c.81T>C	p.H27H	COSM249860	50.46	48.79	45.61	50.89	42.46
KDR	c.1416A>T	p.Q47H	COSM149673	14.27			11.70	
KDR	c.2615-36A>CA	p.?	rs3214870	41.91	54.70	39.74	47.30	51.71
KIT	<b>c.1621A&gt;C</b>	<b>p.M541L</b>	<b>COSM28026</b>	51.05	56.51	46.23	38.7	47.71
NRAS	<b>c.181C&gt;A</b>	<b>p.Q61K</b>	<b>COSM580</b>		9.15			
PDGFRA	<b>c.1667A&gt;G</b>	<b>p.E556G</b>	-				8.66	
PDGFRA	c.2017A>C	p.I673L	-				8.82	
PDGFRA	c.1701A>G	p.P567P	rs1873778	99.90	99.90	100	100	99.80
PIK3CA	<b>c.323G&gt;A</b>	<b>p.R108H</b>	<b>COSM27497</b>				5.69	
PIK3CA	c.352+40A>G	p.?	rs3729674	49.85	51.93	54.80	34.34	48.78
PTEN	c.324T>C	p.L108L	-				9.43	
RET	c.1908G>A	p.T382T	rs886038663				9.98	
RET	c.2307G>T	p.L769L	rs1800861	48.97	41.55	51.89	67.86	52.03
SMAD4	c.727G>A	p.G243R	-				10.75	
SMAD4	<b>c.1216G&gt;A</b>	<b>p.A406T</b>	<b>COSM10103</b>				11.76	
SMAD4	c.1335A>G	p.R445R	-		14.00	5.23	10.00	8.00
SMAD4	c.1059C>T	p.Y353Y	rs863224400				13.06	
SMAD4	c.1062G>A	p.V354V	-				12.86	
SMAD4	c.1089T>C	p.C363C	-				13.00	
SMAD4	c.1035C>T	p.C345C	-				13.07	
SMAD4	c.1002G>A	p.Q334Q	rs773598775				13.12	
SMAD4	c.990A>G	p.E330E	-				13.51	
SMO	<b>c.1214T&gt;C</b>	<b>p.L405P</b>	-		6.72			
SMO	c.1623G>A	p.T541T	COSM1085436		6.38		12.48	
SMO	c.1566G>A	p.L522L	-		6.58		12.21	
SMO	c.1539G>C	p.P513P	-		6.62		12.35	
SMO	c.1545T>C	p.L515L	-		6.74		12.32	
SMO	c.1590_1591del CGinsTT	p.IA530_531IS	-		7.00		12.05	
STK11	c.465-51T>C	p.?	rs2075606				18.51	
STK11	<b>c.536C&gt;T</b>	<b>p.P179L</b>	<b>COSM51520</b>				10.87	
STK11	c.174C>G	p.G58G	rs756661578		9.31			
STK11	c.171A>G	p.E57E	rs1060503785		9.35			
STK11	c.177T>G	p.S59S	-		9.37			
STK11	c.72G>A	p.T24T	-		9.35			
STK11	c.141C>A	p.G47G	-		9.46			
STK11	c.84C>G	p.R28R	-		9.40			
STK11	c.114G>C	p.P38P	-		10.21			
TP53	<b>c.388C&gt;A</b>	<b>p.L130I</b>	<b>IARCTP53</b>		5.00		5.96	
TP53	c.215C>G	p.P72R	COSM250061	98.40	97.51	97.10	97.12	96.38
TP53	c.1043T>C	p.L348S	-				5.43	
VHL	c.378T>C	p.D126D	-				6.10	

pt. #253							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M-/E+	CTC M+/E+	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	100	98.09	nc	97.09
APC	c.2601A>G	p.T867T	-			9.57	
ATM	c.9159A>G	p.K3053K	rs1060504315			20.42	
CDH1	c.1027C>A	p.L343M	-			19.64	
CSF1R	c.*1841TG>GA	p.?	rs386693509	99.51	99.78	21.94	99.24
EGFR	c.2361G>A	p.Q787Q	COSM1451600	51.94	51.70	nc	66.73
ERBB4	c.1080C>T	p.T360T	rs183921316			6.15	
FBXW7	c.1451G>T	p.R484M	COSM34016			39.00	
FGFR1	c.518A>G	p.Glu173Gly	-			27.20	
FGFR3	c.1953G>A	p.T651T	rs7688609	100	99.59	99.38	99.60
FGFR3	c.1150T>C	p.F384L	COSM724 rs17881656	45.35	44.89	nc	53.27
FLT3	c.1310-3T>C	p.?	rs2491231	53.02	36.50	99.53	27.56
HNF1A	c.635A>G	p.Q212R	-			52.45	
HRAS	c.81T>C	p.H27H	COSM249860			52.68	
JAK3	c.2156T>C	p.F719S	-			17.86	
KDR	c.798+54G>A	p.?	rs7692791	48.00	40.91	nc	37.64
KDR	c.1416A>T	p.Q47H	COSM149673	99.82	99.75		99.66
KDR	c.2615-36A>CA	p.?	rs3214870	100	99.80		100
KIT	c.1936T>C	p.Y646H	COSM3734987			5.34	
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	100	nc	100
PIK3CA	c.1633G>A	p.E545K	COSM763		48.63		53.55
PIK3CA	c.3140A>T	p.H1047L	COSM776		59.35		68.78
PIK3CA	c.1348C>T	p.H450Y	-			18.75	
PIK3CA	c.3076C>T	p.(=)	-			6.06	
RET	c.2307G>T	p.L769L	rs1800861			99.58	
SMARCB1	c.1119-41G>A	p.?	COSM1090	49.38	32.62		25.79
SMO	c.623A>G	p.E208G	-			5.88	
SMO	c.646C>T	p.Q216Ter	COSM6906070			11.57	
STK11	c.465-51T>C	p.?	rs2075606	49.67	37.45	nc	33.20
TP53	c.215C>G	p.P72R	COSM250061	50.77	52.39	nc	38.33
TP53	c.742C>T	p.R248TW	COSM10656		20.19	nc	38.45

pt. #274							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E+	CTC M+/E-
CSF1R	c.*1841TG>GA	p.?	rs386693509	26.5	41.8	33.93	32.2
EGFR	c.2361G>A	p.Q787Q	COSM1451600	51.8	51.4	52.76	53.4
ERBB4	c.421+58A>G	p.?	rs839541	48.5	55.0	51.43	46.9
FGFR3	c.1953G>A	p.T651T	rs7688609	99.5	99.5	99.70	99.6
HRAS	c.81T>C	p.H27H	COSM249860	52.2	53.6	48.09	48.9
IDH1	c.315C>T	p.G105G	rs11554137	50.4	44.9	43.17	47.0
KDR	c.798+54G>A	p.?	rs7692791	99.9	96.9	100	99.6
KDR	c.4035T>C	p.P1345P	-				16.4
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.9	100	99.9
RET	c.2307G>T	p.L769L	rs1800861	100	99.5	99.95	99.9
RET	c.2712C>G	p.S904S	rs1800836	46.9	46.7	49.65	43.9
SMAD4	c.955+58C>T	p.?	rs948588	51.5	48.4	40.70	48.8
TP53	c.215C>G	p.P72R	COSM250061	51.3	63.4	59.16	50.7
TP53	<b>c.833C&gt;T</b>	<b>p.P278L</b>	<b>COSM10863</b>		13.8		

pt. #279								
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E+	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869				10.07	11.27
ATM	<b>c.1810C&gt;T</b>	<b>p.P604S</b>	<b>COSM22499</b>				7.31	11.1
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	100	99.8	82.94	75.3
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.40	99.2	99.5	98.02	100
EGFR	<b>c.2257C&gt;T</b>	<b>p.P753S</b>	<b>COSM6268</b>					5.14
ERBB4	c.421+51C>T	p.?	-					7.86
ERBB4	c.421+58A>G	p.?	rs839541	99.31	99.7	99.9	81.90	90.1
FGFR3	c.1953G>A	p.T651T	rs7688609	99.68	99.5	99.7	99.75	99.7
FGFR3	c.1959+22G>A	p.?	rs3135898	47.50	46.9	40.3	32.29	40.2
FGFR3	<b>c.2408G&gt;A</b>	<b>p.G803D</b>	<b>COSM7208599</b>			6.95		
FGFR3	<b>c.1150T&gt;C</b>	<b>p.F384L</b>	<b>COSM724</b>				6.91	
FLT3	c.1310-3T>C	p.?	rs2491231	51.05	50.9	44.1	59.03	60.4
HNF1A	<b>c.620G&gt;A</b>	<b>p.G207D</b>	<b>COSM24931</b>			5.05		
HRAS	c.81T>C	p.H27H	COSM249860				6.14	
IDH1	c.315C>T	p.G105G	rs11554137				5.50	
KDR	c.798+54G>A	p.?	rs7692791				8.60	5.00
KIT	<b>c.1621A&gt;C</b>	<b>p.M541L</b>	<b>COSM28026</b>	50.90	48.5	30.9	32.52	39.5
MPL	c.1565+27G>A	p.?	-			6.15		
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.4	100	99.94	100
PDGFRA	c.2472C>T	p.V824V	COSM22413				5.05	9.00
PIK3CA	c.1173A>G	p.I391M	rs2230461	54.45	60.5	49.5	54.09	61.4
PIK3CA	c.352+40A>G	p.?	rs3729674	52.23	48.5	42.6	45.49	41.6
PIK3CA	<b>c.3140A&gt;T</b>	<b>p.H1047L</b>	<b>COSM776</b>		29.5			
RET	c.2307G>T	p.L769L	rs1800861	99.80	99.9	100	93.97	96.2
RET	c.2712C>G	p.S904S	rs1800836	47.62	46.5	56.8	47.29	50.1
SMAD4	c.1059C>T	p.Y353Y	rs863224400					5.10
SMARCB1	c.1119-41G>A	p.?	COSM1090	48.10	72.7	63.3	39.30	46.5
SMO	c.1623G>A	p.T541T	COSM1085436				5.26	
SMO	c.1566G>A	p.L522L	-				5.41	
SMO	c.1539G>C	p.P513P	-				5.40	
SMO	c.1545T>C	p.L515L	-				5.40	
STK11	c.465-51T>C	p.?	rs2075606				14.79	10.7
STK11	c.80A>G	p.H27R	-				5.25	
TP53	c.215C>G	p.P72R	COSM250061	97.85	96.4	98.4	98.65	86.4

pt. #335						
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-
ABL1	c.750G>C	p. G250G	-			7.80
ABL1	c.774G>A	p.E258E	-			10.0
ABL1	c.747C>G	p.G249G	-			9.00
ABL1	c.714A>G	p.E238E	-			9.70
APC	c.4479G>A	p.T1493T	COSM3760869	51.8	51.9	57.3
APC	<b>c.4348C&gt;T</b>	<b>p.R1450*</b>	<b>COSM13127</b>			5.25
AKT1	c.48G>T	p.G16G	-		43.8	
ATM	<b>c.7328G&gt;A</b>	<b>p.R2443Q</b>	<b>COSM20404</b>			12.20
CSF1R	c.*1841TG>GA	p.?	rs386693509	99.8	97.0	64.0
EGFR	c.2361G>A	p.Q787Q	COSM1451600		6.17	
ERBB4	c.421+58A>G	p.?	rs839541			10.0
FGFR3	c.1953G>A	p.T651T	rs7688609	99.8	99.4	96.3
FLT3	c.1310-3T>C	p.?	rs2491231	100	96.7	64.5
HRAS	c.81T>C	p.H27H	COSM249860	98.9	95.8	97.5
KDR	c.798+54G>A	p.?	rs7692791	47	38	49.8
KDR	c.4035T>C	p.P1345P	-			5.36
KDR	c.4008C>T	p.T1336T	COSM35855	52.4	48.0	26.5
KDR	c.4022A>G	p.Q1341R	-			5.01
KDR	<b>c.2946C&gt;T</b>	<b>p.S982S</b>	<b>COSM4992129</b>			5.75
MET	<b>c.2962C&gt;T</b>	<b>p.R988C</b>	<b>COSM1666978</b>			9.0
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.7	99.8
PDGFRA	c.2472C>T	p.V824V	COSM22413			26.4
PTEN	<b>c.959T&gt;A</b>	<b>p.L320*</b>	<b>COSM35671</b>		62.7	6.06
RET	c.2307G>T	p.L769L	rs1800861	46,25	35.1	68.6
RET	c.2712C>G	p.S904S	rs1800836			6.05
SMO	c.1623G>A	p.T541T	COSM1085436			6.25
SMO	c.1539G>C	p.P513P	-			6.05
SMO	c.1545T>C	p.L515L	-			6.30
STK11	c.465-51T>C	p.?	rs2075606		11.7	
STK11	<b>c.976C&gt;A</b>	<b>p.P326T</b>	<b>rs771632414</b>			10.10
STK11	c.598-14C>T	p.?	rs769378511			5.45
TP53	c.215C>G	p.P72R	COSM250061	51,3	44.1	62.1
TP53	<b>c.4G&gt;A</b>	<b>p.Glu2Lys</b>	<b>rs769884991</b>			5.95
VHL	c.321C>A	p.R107R	-			6.04



pt. #337							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E+	CTC M+/E-
ALK	c.3836+1G>T	Splice site	-			5.71	
APC	c.4479G>A	p.T1493T	COSM3760869	99.82	99.8	100	100
APC	c.4114C>A	p.P1372T	-			5.38	
AKT1	c.528C>A	p.Tyr176*	-				6.19
ATM	c.8177C>T	p.Ala2726Val	-				6.80
ATM	c.3878A>G	p.N1293S	COSM5490564				6.95
ATM	c.2578G>T	p.Asp860Tyr	-				8.32
ATM	c.3947C>T	p.Thr1316Ile	-				9.05
BRAF	c.1379G>T	p.Gly460Val	rs121913351				6.29
CDH1	c.1097C>A	p.T366N	-			12.02	
CSF1R	c.*1841TG>GA	p.?	rs386693509	28.43	51.5	29.54	45.2
EGFR	c.2361G>A	p.Q787Q	COSM1451600	53.30	48.1	74.02	49.00
EGFR	c.2469+32G>T	p.?	-				11.8
ERBB2	c.2527C>T	p.His843Tyr	-				8.74
ERBB2	c.2551G>T	p.V851L	-			5.04	
ERBB4	c.421+58A>G	p.?	rs839541			6.98	
ERBB4	c.1084A>G	p.I362V	-				6.32
ERBB4	c.1015A>G	p.T339A	-				7.04
ERBB4	c.915G>A	p.V305V	rs781470325				7.68
FGFR1	c.497_498insTGA	p.D166dup	rs776622310	30.26	42.5	29.50	41.1
FGFR2	c.1130A>G	p.E377G	-				6.97
FGFR2	c.939+41C>A	p.?	-				11.0
FGFR3	c.1953G>A	p.T651T	rs7688609	99.51	99.6	100	99.9
FLT3	c.1310-3T>C	p.?	rs2491231	51.17	51.7	76.10	47.5
FLT3	c.2001G>T	p.Q667H	-			10.95	
FLT3	c.2537G>T	p.G846V	-			8.30	
HNF1A	c.771C>A	p.N257K	-				5.91
HRAS	c.60C>T	p.T20T	ExAC			8.93	
IDH1	c.354C>T	p.P118P	ExAC			9.10	
KDR	c.798+54G>A	p.?	rs7692791	38.37	40.7	27.91	36.6
KDR	c.1416A>T	p.Q47H	COSM149673	51.34	39.1	33.05	72.8
KDR	c.3849-24C>A	p.?	rs10006115	54.85	50.9	51.68	36.7
KDR	c.3454A>G	p.T1152A	-				6.62
KDR	c.2615-36A>CA	p.?	rs3214870	56.6	49.7	33.54	38.5
KDR	c.2654A>G	p.E885G	-			8.64	
KIT	c.1904A>G	p.E635G	COSM144512				5.74
MET	c.3394+3A>G	p.?	COSM5731184				8.22
NOTCH1	c.5026G>A	p.V1676I	COSM327182				10.6
NOTCH1	c.5019-4C>A	p.?	-				6.95
NOTCH1	c.7362C>T	p.H2454H	-			7.33	
NRAS	c.394G>T	p.E132Ter	-			5.17	
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	100	100	100
PDGFRA	c.2472C>T	p.V824V	COSM22413	47.9	55.1	41.78	70.0
PDGFRA	c.1955G>T	p.G652V	COSM3946437			5.47	
PIK3CA	c.352+40A>G	p.?	rs3729674	51.88	46.3	48.60	62.7
PIK3CA	c.352+1G>A	Splice site	-			7.01	
PIK3CA	c.3196G>A	p.A1066T	COSM6941468			18.0	64.0
PIK3CA	c.1238A>G	p.K413R	-				8.04
PTEN	c.63C>A	p.F21L	-				7.24
PTEN	c.209+45A>G	p.?	-			5.17	
PTEN	c.1008C>T	p.Y336Y	-			7.76	
PTPN11	c.1578G>A	p.Q526Q	-			5.62	
PTPN11	c.1478C>A	p.T493N	-			5.90	
RB1	c.1389+37C>T	p.?	rs373682367			5.01	
RET	c.2307G>T	p.L769L	rs1800861	100	100	100	99.9
RET	c.2712C>G	p.S904S	rs1800836	51.67	43.5	46.66	45.5
RET	c.2681G>T	p.G894V	-				5.97
SMAD4	c.949C>A	p.H317AN	-			7.22	
SMAD4	c.955+58C>T	p.?	rs948588	50.95	58.4	24.21	66.8
SMAD4	c.573G>T	p.S191S	COSM6475980				5.60
SMO	c.1545T>C	p.L515L	-			5.66	
SMO	c.1590_1591delCGinsTT	p.IA530_531IS	-				
SMO	c.1592C>T	p.A531V	-			5.35	
SMO	c.677A>G	p.E226G	-				5.50
SMO	c.614G>A	p.S205N	-				6.40
STK11	c.465-51T>C	p.?	rs2075606	47.93	43.9	65.61	60.5
TP53	c.215C>G	p.P72R	COSM250061	97.99	97.2	98.78	99.1

pt. #371							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	99.78	97.7	88.0	87.3
APC	c.4548A>T	p.I1516I	-			6.76	
APC	c.4524T>C	p.A1508A	rs767025305			6.95	
APC	c.4512C>T	p.S1504S	-			7.40	
APC	c.4476C>T	p.A1492A	-			7.84	
ATM	<b>c.1810C&gt;T</b>	<b>p.P604S</b>	<b>COSM22499</b>			6.66	
CSF1R	c.*1841TG>GA	p.?	rs386693509	34.48	23.50	24.4	31.6
EGFR	c.2361G>A	p.Q787Q	COSM1451600	48.80	51.6	59.7	48.1
ERBB2	c.2638G>A	p.D880N	COSM7002319				5.02
ERBB2	c.2642G>A	p.G881E	-				5.19
ERBB2	c.2644G>T	p.G882C	-				5.41
FGFR3	c.1953G>A	p.T651T	rs7688609	100	99.6	99.6	99.5
FLT3	c.1310-3T>C	p.?	rs2491231	100	100	100	100
HRAS	c.81T>C	p.H27H	COSM249860	48.50	58.0	60.6	49.3
KDR	c.798+54G>A	p.?	rs7692791	53.00	43.00	45.4	49.00
KIT	c.103T>C	p.S35P	-				5.20
KIT	<b>c.1638A&gt;G</b>	<b>p.K546K</b>	<b>COSM21983</b>	44.84	50.8	50.7	53.6
MET	c.3798G>A	p.K1266K	-			6.46	
PIK3CA	c.1173A>G	p.I391M	rs2230461			8.50	
PIK3CA	c.352+40A>G	p.?	rs3729674	45.36	50.3	43.1	39.8
PIK3CA	<b>c.1633G&gt;A</b>	<b>p.E545K</b>	<b>COSM763</b>		20.7		
PTEN	<b>c.991G&gt;A</b>	<b>p.D331N</b>	<b>COSM6327339</b>			7.90	
PTEN	<b>c.635-1G&gt;A</b>	<b>p.?</b>	<b>COSM28920</b>			6.75	
PTPN11	c.193C>T	p.L65L	-			5.45	
PTPN11	<b>c.169C&gt;T</b>	<b>p.Q57*</b>	<b>-</b>			6.00	
RET	c.2307G>T	p.L769L	rs1800861	44.58	41.6	53.2	53.3
SMAD4	<b>c.767A&gt;T</b>	<b>p.Q256L</b>	<b>COSM5546625</b>				11.2
SMAD4	c.1335A>G	p. R445R	-			10.0	10.0
SMO	c.1623G>A	p.T541T	COSM1085436			5.15	
SMO	c.1566G>A	p.L522L	-			5.16	
SMO	c.1539G>C	p.P513P	-			5.65	
SMO	c.1545T>C	p.L515L	-			5.70	
STK11	c.465-51T>C	p.?	rs2075606	56.80	46.0	41.2	61.7
TP53	<b>c.388C&gt;A</b>	<b>p.L130I</b>	<b>IARCTP53</b>				9.70
TP53	c.215C>G	p.P72R	COSM250061	97.13	100	98.2	95.7

GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M-/E+	CTC M+/E+	CTC M+/E-
ABL1	c.720G>A	p.T240T	-	-	9.66	-	-
APC	c.4479G>A	p.T1493T	COSM3760869	49.27	71.74	47.17	52.01
APC	c.2613A>G	p.G871G	-	-	-	-	9.79
APC	c.2652A>G	p.A884A	-	-	-	-	9.06
APC	c.3362T>C	p.I1121T	-	-	26.44	-	-
AKT1	c.489G>A	p.k163K	rs576374556	-	9.05	-	-
ATM	c.1810C>T	p.P604S	COSM22499	-	68.97	14.34	-
ATM	c.3927A>G	p.A1309A	rs155093665	-	-	-	6.15
ATM	c.5056A>G	p.L1666V	rs145453814	-	-	-	5.05
ATM	c.5381T>C	p.L1794P	-	-	-	-	9.80
CDHL1	c.2511D>C	p.V95A	rs878854698	-	-	-	10.85
CDHL1	c.1078A>G	p.T360A	-	-	-	-	5.94
CDHL1	c.237A>G	p.T79T	-	-	-	-	7.00
CDKN2A	c.310C>T	p.L104V	-	-	32.16	-	-
CSF1R	c.1841T>C>G>A	p.?	rs386693509	99.69	100	37.36	46.42
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.60	100	99.39	97.08
EGFR	c.2300C>T	p.A787V	COSM26445	-	11.27	-	-
EGFR	c.2469>57C>T	p.?	-	-	-	-	5.35
EGFR	c.2469>27G>A	p.?	rs368440941	-	-	-	5.85
EGFR	c.2459A>G	p.Q820R	COSM85898	-	-	-	5.95
EGFR	c.2436G>A	p.R821I	-	-	16.05	-	-
EGFR	c.875T>C	p.V292A	ExAC	-	6.67	-	-
ERBB2	c.2307>46A>G	p.?	rs4252645	-	5.30	-	-
ERBB2	c.2307>66T>A	p.?	-	-	5.45	-	-
ERBB4	c.421>58A>G	p.?	rs839541	-	28.25	47.47	47.85
FBXW7	c.1319A>G	p.Asp440Gly	-	-	9.89	-	-
FBXW7	c.861>17A>G	p.?	-	-	13.55	-	-
FBXW7	c.1442C>T	p.A481V	COSM4122764	-	-	-	7.10
FGFR3	c.1953G>A	p.T651T	rs7688609	99.60	100	99.53	99.61
FGFR3	c.1959>22G>A	p.?	rs1335898	55.02	-	25.04	32.14
FGFR3	c.1150T>C	p.F384L	COSM7724	-	41.90	6.77	-
FLT3	c.1310>3T>C	p.?	rs2491231	99.75	nc	99.75	94.15
FLT3	c.2541>4A>G	p.?	-	-	24.06	-	-
FLT3	c.2541>11T>C	p.?	-	-	6.58	-	-
GNAQ	c.735>1G>T	Splice site	-	-	-	-	5.75
HNF1A	c.612C>T	p.F204F	-	-	-	-	5.85
HNF1A	c.638T>C	p.I213T	-	-	-	-	6.31
HRA5	c.81T>C	p.H27H	COSM249860	51.49	-	26.70	27.28
JAK3	c.2128G>A	p.T1797ter	-	-	20.90	-	-
KDR	c.798>54C>A	p.?	rs7692791	-	18.09	23.09	9.45
KDR	c.799C>T	p.His267Tyr	?	-	5.95	-	-
KDR	c.1416A>T	p.Q47H	COSM149673	49.32	-	40.06	50.87
KDR	c.3849>24C>A	p.?	rs10006115	-	-	-	-
KDR	c.2615>36A>CA	p.?	rs3214870	45.09	24.00	48.35	43.31
KDR	c.2881C>A	p.R961R	-	-	-	-	5.00
KIT	c.1621A>C	p.M541L	COSM28020	35.74	-	12.74	22.78
KIT	c.2019T>C	p.C679C	-	-	5.50	-	-
KIT	c.2555T>C	p.V852A	ExAC	-	-	-	5.10
KRAS	c.103A>G	p.T35A	COSM1360858	-	4.30	-	-
KRAS	c.113A>G	p.Asp38Gly	-	-	-	-	5.10
KRAS	c.153T>C	p.C51C	COSM5009217	-	-	-	6.16
MET	c.3770A>G	p.N1257S	-	-	-	-	10.24
MET	c.1115A>G	p.D372G	-	-	-	-	16.53
MET	c.1124A>G	p.N375S	COSM5020653	-	-	-	11.24
MET	c.3794T>C	p.V1265A	-	-	-	-	9.82
MLH1	c.1184C>T	p.A395V	rs1064794142	-	65.51	-	-
MLH1	c.1156A>G	p.T386A	-	-	-	-	5.25
NOTCH1	c.5019>83G>T	p.?	-	-	5.25	-	-
NOTCH1	c.7407A>G	p.F2479P	-	-	6.18	-	-
PDGFRA	c.1701A>G	p.P567P	rs1873778	99.00	99.95	100	100
PDGFRA	c.2472C>T	p.V824V	COSM22413	-	26.2	34.39	29.45
PDGFRA	c.1990T>C	p.C664R	-	-	-	-	5.50
PIK3CA	c.1173A>G	p.L391M	rs2230461	34.75	24.40	-	-
PIK3CA	c.266T>C	p.L89P	-	-	7.26	-	-
PIK3CA	c.352>40A>G	p.?	rs3729674	49.60	-	-	-
PIK3CA	c.240A>G	p.E80E	-	-	8.85	-	-
PIK3CA	c.3140A>G	p.H1047R	COSM775	-	-	9.41	7.30
PIK3CA	c.2752A>G	p.N918D	-	-	-	-	5.05
PTEN	c.995A>G	p.L332R	-	-	10.73	-	-
PTEN	c.60A>G	p.C20G	COSM4016447	-	5.44	-	-
PTEN	c.966A>G	p.K322K	rs786201392	-	23.03	-	-
RB1	c.1724A>G	p.Q675R	-	-	6.29	-	-
RB1	c.1975T>C	p.Y659H	COSM327254	-	6.30	-	-
RB1	c.2271C>T	p.N757N	-	-	6.73	-	-
RB1	c.2253T>C	p.S751S	rs373070661	-	6.74	-	-
RB1	c.2259A>C	p.I753I	-	-	6.75	-	-
RB1	c.2232C>T	p.I744I	-	-	6.79	-	-
RB1	c.2234A>G	p.K745R	-	-	7.75	-	-
RB1	c.1050>30A>G	p.?	-	-	-	-	13.91
RB1	c.2293A>G	p.K765E	-	-	-	-	6.31
RET	c.2307C>T	p.L769L	rs1800861	49.59	100	23.37	29.06
SMAD4	c.1332T>C	p.H444H	-	-	-	-	5.72
SMAD4	c.1176A>G	p.K392K	-	-	18.33	-	-
SMAD4	c.525A>G	p.E175E	rs368528856	-	-	-	5.65
SMARCB1	c.172G>A	p.E58K	-	-	15.24	-	-
SMO	c.1870A>T	p.Lys624Ter	-	-	6.52	-	-
SMO	c.1625T>C	p.L542P	-	-	-	-	8.18
SMO	c.1180T>C	p.T394H	COSM3878023	-	-	-	14.17
SMO	c.929A>G	p.E310G	-	-	-	-	9.61
STK11	c.465>51T>C	p.?	rs2075606	-	75.44	50.34	19.92
STK11	c.825G>A	p.P275P	rs202011521	-	11.67	-	-
STK11	c.524A>G	p.K175R	-	-	9.59	-	-
TP53	c.388C>A	p.L130I	IARC1P53	-	9.85	-	-
TP53	c.742C>T	p.R248TW	COSM10856	-	-	11.25	-
TP53	c.311A>G	p.Q104R	-	-	8.59	-	-
TP53	c.376>24T>C	p.?	-	-	5.59	-	-
TP53	c.376>20T>C	p.?	rs1483487754	-	-	-	5.65
TP53	c.467C>T	p.P223S	COSM44816	-	-	-	6.42
VHL	c.303G>C	p.L101L	ExAC	-	10.43	-	-

pt. #382							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M+/E+	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	99.85	100	89.97	99.85
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	99.52	99.-72	99.90
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.70	99.63	92.89	98.98
ERBB2	c.2270A>G	p.E757G	-			5.15	
ERBB2	c.2295A>C	p.K765N	-			5.17	
FBXW7	c.1737G>A	p.G580G	-		6.36		
FBXW7	c.1776T>C	p.N593N	-		6.25		
FBXW7	c.1716T>C	p.N572N	-		6.45		
FGFR3	c.1953G>A	p.T651T	rs7688609	100	99.77	99.21	99.72
FGFR3	<b>c.1150T&gt;C</b>	<b>p.F384L</b>	<b>COSM724</b>			5.56	7.64
FLT3	c.1310-3T>C	p.?	rs2491231		7.20	9.95	
FLT3	c.2510T>A	p.M837K	-			5.46	
HRAS	c.81T>C	p.H27H	COSM249860	47.52	49.78	46.28	38.71
IDH1	c.315C>T	p.G105G	rs11554137	49.85	52.4	41.65	44.50
KDR	c.798+54G>A	p.?	rs7692791	33.53	42.58	39.05	38.12
KDR	c.1416A>T	p.Q47H	COSM149673	49.40	54.73	40.72	50.63
KDR	c.2615-36A>CA	p.?	rs3214870	46.13	55.61	41.79	40.00
KRAS	c.102A>G	p.R102R	-			6.80	
NRAS	c.51C>T	p.S17S	-		7.06		
PDGFRA	c.1701A>G	p.P567P	rs1873778	99.90	100	99.95	100
PIK3CA	c.1173A>G	p.I391M	rs2230461	58.65	50.48	38.82	46.45
PIK3CA	c.352+40A>G	p.?	rs3729674	47.55	53.73	43.62	45.81
PIK3CA	<b>c.3196G&gt;A</b>	<b>p.A1066T</b>	<b>COSM6941468</b>		7.0	8.66	
PIK3CA	<b>c.3140A&gt;G</b>	<b>p.H1047R</b>	<b>rs121913279</b>		5.00	5.00	
RET	c.2307G>T	p.L769L	rs1800861	99.85	99.15	89.29	99.95
RET	c.2712C>G	p.S904S	rs1800836	50.40	49.20	41.37	46.12
SMAD4	c.955+58C>T	p.?	rs948588			12.00	
SMO	c.1623G>A	p.T541T	COSM1085436		21.10		
SMO	c.1566G>A	p.L522L	-		20.91		
SMO	c.1539G>C	p.P513P	-		19.30		
SMO	c.1545T>C	p.L515L	-		20.85		
SMO	c.1590_1591del CGinsTT	p.IA530_531IS	-		21.55		
STK11	c.465-51T>C	p.?	rs2075606	99.95	82.13	86.65	84.52
TP53	c.215C>G	p.P72R	COSM250061	51.28	54.98	53.68	54.66
TP53	<b>c.1009C&gt;T</b>	<b>p.R337C</b>	<b>COSM11071</b>		11.5		

pt. #392							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-	CTC M-/E-
ABL1	c.750G>C	p. G250G	-			21.7	
ABL1	c.774G>A	p.E258E	-			23.0	
ABL1	c.747C>G	p.G249G	-			23.8	
ABL1	c.714A>G	p.E238E	-			24.4	
APC	c.4479G>A	p.T1493T	COSM3760869	49.41	61.9	99.5	50.0
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.71	99.9	nd	100
ERBB4	<b>c.866G&gt;T</b>	<b>p.C289F</b>	-			22.3	
ERBB4	c.421+58A>G	p.?	rs839541	51.06	42.5	nd	51.7
FGFR3	c.1953G>A	p.T651T	rs7688609	100	99.8	99.7	100
FLT3	c.1310-3T>C	p.?	rs2491231	99.76	100	nd	100
HRAS	c.81T>C	p.H27H	COSM249860	51.75	50.5	nd	33.7
KDR	c.798+54G>A	p.?	rs7692791	48.00	23.7	nd	34.00
KIT	c.68-13T>G	p.?	-			19.5	
NOTCH1	c.5019-85T>C	p.?	-			5.04	
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	100	100	100
PIK3CA	c.352+40A>G	p.?	rs3729674			13.89	
PIK3CA	<b>c.1633G&gt;A</b>	<b>p.E545K</b>	<b>COSM763</b>			82.9	
RET	c.2307G>T	p.L769L	rs1800861	100	100	64.0	100
SMAD4	c.1335A>G	p. R445R	-			100	
SMAD4	c.1389A>G	p.A463A	-			100	
SMAD4	c.1410T>C	p.P470P	-			100	
SMAD4	c.594A>G	p.P198P	-			26.1	
SMAD4	c.1332T>C	p.H444H	-			96.7	
SMAD4	c.1353_1359 delGGCTACTin sAGCCACG	p.AAT451_453AAT	-			100	
SMO	c.1623G>A	p.T541T	COSM1085436			100	
SMO	c.1566G>A	p.L522L	-			100	
SMO	c.1539G>C	p.P513P	-			95.5	
SMO	c.1545T>C	p.L515L	-			99.2	
SMO	c.1590_1591del CGinsTT	p.IA530_531IS	-			100	
STK11	c.465-51T>C	p.?	rs2075606	52.24	53.3	nd	55
STK11	c.75C>T	p.F25F	-			5.2	
TP53	c.215C>G	p.P72R	COSM250061	99.40	97.8	98.0	100

pt. #399							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	99.8	99.8	99.8	100
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	98.1	99.6	99.9
EGFR	c.2361G>A	p.Q787Q	COSM1451600	99.6	99.7	99.5	100
FGFR3	c.1953G>A	p.T651T	rs7688609	99.7	99.7	99.8	99.8
FLT3	c.1310-3T>C	p.?	rs2491231	100	99.8	99.9	99.9
KDR	c.798+54G>A	p.?	rs7692791	99.7	99.6	99.9	100
MET	c.3029C>T	p.T1010I	COSM707	49.9	49.3	54.7	52.0
MPL	c.1565+27G>A	p.?	-				5.35
PDGFRA	c.1701A>G	p.P567P	rs1873778	99.9	99.6	99.9	99.9
PDGFRA	c.2472C>T	p.V824V	COSM22413	47.3	42.4	50.9	47.3
RET	c.2307G>T	p.L769L	rs1800861	52.5	44.2	52.3	44.9
STK11	c.465-51T>C	p.?	rs2075606	52.2	45.2	60.5	42.6
TP53	<b>c.711G&gt;A</b>	<b>p.M237I</b>	<b>COSM10834</b>		5.89		

pt. #407								
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E+	CTC M+/E-	CTC M-/E-
ALK	c.3541C>T	p.R181C	rs56315533				6.65	
APC	c.4479G>A	p.T1493T	COSM3760869			20.37		17.56
CSF1R	c.*1841TG>GA	p.?	rs386693509	96.67	96.61	98.25	99.00	100
EGFR	c.2361G>A	p.Q787Q	COSM1451600	100	99.58	100	99.00	100
EGFR	c.1750C>A	p.H584N	-				6.06	
EGFR	c.356T>C	p.L119S	-				6.50	
ERBB4	c.1786G>A	p.G596S	-				8.25	
EZH2	c.1934A>G	p.E645G	-			7.00		
FBXW7	c.1412A>G	p.E471G	COSM305935			7.74		
FGFR3	c.1953G>A	p.T651T	rs7688609	99.48	100	99.66	99.86	99.88
FLT3	c.2053+23A>G	p.?	rs75580865	54.81	64.65	59.23	41.65	74.15
FLT3	c.1310-3T>C	p.?	rs2491231	44.48	55.99	52.20	44.15	58.35
FLT3	c.2531T>C	p.V844A	-				5.26	
HRAS	c.81T>C	p.H27H	COSM249860	44.86	53.70	36.96	48.66	52.23
JAK3	c.420+18G>A	p.?	-			8.01		
JAK3	c.1737G>A	p.V579V	-				6.90	
KDR	c.798+54G>A	p.?	rs7692791	37.68	38.00	29.17	37.31	50.00
KDR	c.3423C>A	p.D1141E	-				9.65	
KDR	c.4004A>G	p.Q1335R	-				17.26	
KIT	<b>c.1638A&gt;G</b>	<b>p.K546K</b>	<b>COSM21983</b>	50.22	49.72	59.93	44.90	45.19
KIT	c.1540+8T>C	p.?	-				5.30	
KIT	c.1994C>A	p.P665H	-				5.35	
KRAS	<b>c.172A&gt;G</b>	<b>p.T58A</b>	-				8.20	
NRAS	c.130G>A	p.V44M	-			5.24		
NRAS	c.192C>T	p.Y64Y	-			5.47		
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.93	100	99.84	100
PIK3CA	<b>c.3140A&gt;T</b>	<b>p.H1047L</b>	<b>COSM776</b>			5.97		
PTEN	<b>c.1001A&gt;G</b>	<b>p.N334S</b>	<b>COSM6948036</b>				6.26	
PTEN	<b>c.511C&gt;T</b>	<b>p.Gln171Ter</b>	<b>COSM5149</b>				5.27	nc
PTPN11	c.1551G>A	p.A517A	COSM6135785				6.30	
RET	c.2307G>T	p.L769L	rs1800861	100	100	95.63	100	100
RET	c.2712C>G	p.S904S	rs1800836	48.11	47.09	nc	46.74	nc
RET	c.2285-8C>T	p.?	-				7.45	
SMAD4	c.1408C>T	p.P470S	-			5.85		
SMAD4	c.955+58C>T	p.?	rs948588	53.74	65.22	51.61	53.83	50.84
SMAD4	c.954T>C	p.P318P	rs773615487	52.79	65.56	49.25	54.15	43.58
SMAD4	c.1197C>T	p.V399V	-				6.86	
SMARCB1	c.573G>A	p.L191L	COSM6506621				7.60	
STK11	c.465-51T>C	p.?	rs2075606	45.31	69.69	41.94	57.26	nc
STK11	c.91T>C	p.S31P	-			6.63		
TP53	<b>c.388C&gt;A</b>	<b>p.L130I</b>	<b>IARCTP53</b>				6.50	
TP53	c.215C>G	p.P72R	COSM250061	45.75	67.35	50	60.90	50.0
TP53	c.376-35T>C	p.?	-				7.43	
TP53	c.615T>C	p.Y205Y	rs786202222			6.4		

pt. #431						
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	100	99.95	80.86
ATM	c.5415G>A	p.W1805*	COSM4551820			5.85
ATM	c.8108A>G	p.D2703G	-			5.95
BRAF	c.1800G>A	p.V600V	COSM249890			5.40
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	100	91.26
EGFR	c.2361G>A	p.Q787Q	COSM1451600	56.22	51.54	57.62
EGFR	c.2283+25G>A	p.?	COSM24212			8.30
EGFR	c.2266A>G	p.N756D	-			8.33
ERBB2	c.2621A>G	p.Glu874Gly	-			5.73
ERBB4	c.421+58A>G	p.?	rs839541		7.06	
FBXW7	c.796A>G	p.Lys266Glu	-			5.80
FGFR3	c.2115G>A	p.K705K	COSM6282664			5.06
FGFR3	c.1953G>A	p.T651T	rs7688609	100	99.86	99.52
FLT3	c.1310-3T>C	p.?	rs2491231	50.00	49.30	59.60
HRAS	c.81T>C	p.H27H	COSM249860	43.22	55.26	58.5
KDR	c.798+54G>A	p.?	rs7692791	37.60	39.03	39.20
KIT	c.1638A>G	p.K546K	COSM21983			14.90
KIT	c.2532T>C	p.C844C	-		5.90	
PDGFRA	c.1701A>G	p.P567P	rs1873778	99.69	100	99.85
PIK3CA	c.352+40A>G	p.?	rs3729674	48.24	35.56	36.91
PIK3CA	c.2765T>A	p.M922L	-		7.06	
PTEN	c.989A>G	p.K330R	-			5.05
RET	c.2307G>T	p.L769L	rs1800861	100	90.53	95.53
RET	c.2712C>G	p.S904S	rs1800836			30.37
RET	c.2691A>G	p.R897R	COSM3723341			5.74
RET	c.2289C>T	p.N763N	COSM918121			6.09
SMAD4	c.955+58C>T	p.?	rs948588	51.33	48.60	44.76
SMO	c.669A>C	p.T229T	ExAC		5.90	
SMO	c.642C>G	p.G214G	-		5.75	
SMO	c.675T>C	p.A225A	-		5.60	
SMO	c.600A>C	p.T200T	-		5.95	
SMO	c.576G>A	p.Q192Q	-		5.85	
SMO	c.573C>T	p.G191G	-		5.95	
STK11	c.465-51T>C	p.?	rs2075606	53.04	38.79	53.48
STK11	c.980A>G	p.D327G	rs864622638			5.15
TP53	c.215C>G	p.P72R	COSM250061	97.81	99.22	99.38



## pt. #454

GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	46.02	51.21	48.82	55.04
CSF1R	c.954G>A	p.G318G	-				5.70
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	99.83	95.56	100
EGFR	c.2361G>A	p.Q787Q	COSM1451600	51.39	40.65	59.59	55.89
FGFR3	c.1953G>A	p.T651T	rs7688609	100	100	99.68	99.03
FLT3	c.1310-3T>C	p.?	rs2491231	100	99.95	99.95	99.90
IDH1	c.315C>T	p.G105G	rs11554137	53.43	50.03	49.95	43.65
KDR	c.798+54G>A	p.?	rs7692791	100	99.83	100	91.28
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.85	100	100
PDGFRA	c.2472C>T	p.V824V	COSM22413	99.46	99.30	98.55	95.37
RET	c.2307G>T	p.L769L	rs1800861	99.49	100	100	99.85
RET	c.2712C>G	p.S904S	rs1800836				7.68
STK11	c.465-51T>C	p.?	rs2075606	54.92	53.05	49.14	52.45
TP53	<b>c.388C&gt;A</b>	<b>p.L130I</b>	<b>IARCTP53</b>			7.03	
TP53	c.215C>G	p.P72R	COSM250061	99.20	97.78	99.09	99.11

pt. #458							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	FFPE	CTC M+/E-	CTC M-/E-
APC	c.4479G>A	p.T1493T	COSM3760869	50.51	67.64	54,06	50.00
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	100	100	84.29
EGFR	c.2361G>A	p.Q787Q	COSM1451600	100	99.62	100	nc
FGFR3	c.1953G>A	p.T651T	rs7688609	50.00	99.82	99.43	100
FLT3	c.1310-3T>C	p.?	rs2491231	50.00	73.59	53.98	44.80
HRAS	c.81T>C	p.H27H	COSM249860	42.29	37.46	43.59	65.43
KIT	c.1638A>G	p.K546K	COSM21983				5.52
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	99.38	99.74	100
PIK3CA	c.352+40A>G	p.?	rs3729674	58.08	35.52	48.41	35.71
RET	c.2307G>T	p.L769L	rs1800861	49.78	48.00	45.41	55.74
TP53	c.215C>G	p.P72R	COSM250061	98.39	97.52	97.86	nc
TP53	c.817C>T	p.R273C	rs121913343		40.86		

pt. #471							
GENE	Nucleotide variant	Amino acid mutation	COSMIC/dbSNP ID number	gDNA	CTC M+/E+	CTC M+/E-	CTC M-/E-
ALK	c.3559C>T	p.S1186S	-		20.59		
APC	c.4479G>A	p.T1493T	COSM3760869	49.21	50.46	44.00	50.75
ATM	c.1065+4C>T	p.?	-		6.72		
ATM	c.5226C>T	p.A1742A	rs750881004		10.57		
CDKN2A	<b>c.241C&gt;T</b>	<b>p.P81S</b>	<b>COSM48299</b>		5.26		
CSF1R	c.941C>T	p.T314I	COSM4513528			6.95	
CSF1R	c.*1841TG>GA	p.?	rs386693509	100	100	100	99.12
CTNNB1	<b>c.136C&gt;T</b>	<b>p.L46L</b>	<b>COSM5988491</b>		13.33		
EGFR	c.2361G>A	p.Q787Q	COSM1451600	45.06	60.21	44.58	47.62
ERBB4	c.421+58A>G	p.?	rs839541	100	99.30	100	91.86
ERBB4	c.2759C>T	p.P920L	-		10.56		
FGFR1	c.844T>C	p.S282P	-			7.40	
FGFR3	c.1953G>A	p.T651T	rs7688609	100	100	100	100
FGFR3	c.1959+22G>A	p.?	rs3135898	62.70	48.35	45.96	51.61
FGFR3	c.1959+21C>T	p.?	-	37.00	36.81	37.27	35.26
FGFR3	c.740-8C>T	p.?	rs886038257		13.25		
FLT3	c.1310-3T>C	p.?	rs2491231	99.79	99.91	99.95	84.82
HNF1A	c.795C>T	p.Y265Y	-		7.99		
HRAS	c.81T>C	p.H27H	COSM249860	50.96	45.59	56.09	41.35
KDR	c.798+54G>A	p.?	rs7692791	47.00	35.00	21.35	32.23
KDR	c.3849-16G>A	p.?	-				5.60
KDR	<b>c.2959G&gt;T</b>	<b>p.E987Ter</b>	-		5.13		
KIT	<b>c.1621A&gt;C</b>	<b>p.M541L</b>	<b>COSM28026</b>	48.22	63.31	37.81	43.87
KIT	c.1899A>G	p.E633E	-		10.44		
MPL	c.1549T>C	p.F517L	-			8.91	
NOTCH1	c.5020T>C	p.S1674P	COSM36230				5.63
NPM1	c.847-5T>C	p.?	rs191111314	50.37	50.00	44.21	48.11
PDGFRA	c.1701A>G	p.P567P	rs1873778	100	100	100	99.72
PIK3CA	c.1173A>G	p.I391M	rs2230461	52.71	47.22	53.05	51.29
PIK3CA	c.352+40A>G	p.?	rs3729674	48.69	62.38	55.56	48.88
PTEN	<b>c.794T&gt;C</b>	<b>p.L265P</b>	<b>rs1335033900</b>				5.54
PTPN11	c.1571C>T	p.T524I	-		6.27		
PTPN11	c.1527A>G	p.A509A	-		6.34		
RB1	c.969A>G	p.E323E	-		7.11		
RET	c.2307G>T	p.L769L	rs1800861	51.66	47.10	46.94	48.50
RET	c.2712C>G	p.S904S	rs1800836	47.80	44.59	59.49	37.57
SMAD4	c.955+58C>T	p.?	rs948588		10.57		6.37
SMARCB1	c.1119-41G>A	p.?	COSM1090	49.11	61.54	47.58	45.13
STK11	c.465-51T>C	p.?	rs2075606	52.31	45.35	42.14	46.30
TP53	<b>c.388C&gt;A</b>	<b>p.L130I</b>	<b>IARCTP53</b>		5.73		5.41
TP53	c.215C>G	p.P72R	COSM250061	99.35	97.78	97.04	96.37
TP53	<b>c.713G&gt;A</b>	<b>p.C238Y</b>	<b>COSM11059</b>		6.65		