

Table SI. PCR primers used for amplification and sequencing of *KIT* (exons 9, 11, 13 and 17) and *PDGFRA* (exons 12, 14 and 18).

Gene	Exon	Forward primer sequence	Reverse primer sequence	Amplicon size, bp
<i>KIT</i>	9	5'-TTCTTCCCTTTAGATGCT-3'	5'-GATATGGTAGACAGAGCCTAA-3'	262
<i>KIT</i>	11	5'-CTCCAGAGTGCTCTAATG-3'	5'-CACTGTTATGTGTACCCA-3'	251
<i>KIT</i>	13	5'-ACATCAGTTTGCCAGTTG-3'	5'-GCAGTTTATAATCTAGCATTG-3'	256
<i>KIT</i>	17	5'-TCTCCTCCAACCTAATAG-3'	5'-ACTGTCAAGCAGAGAATG-3'	168
<i>PDGFRA</i>	12	5'-CATTTATAGAAACCGAGGTATG-3'	5'-AAATTGTAAAGTTGTGTGC-3'	203
<i>PDGFRA</i>	14	5'-CAGGAAGTTGGTAGCTCAGCT-3'	5'-TGTGTCCAGTGAAAATCCTCA-3'	203
<i>PDGFRA</i>	18	5'-CTGAGTCATTTCTTCCTTT-3'	5'-TGAGGGAAGTGAGGAC-3'	171

Table SII. Summarized list of hot spot mutations included in Colorectal Cancer Mutation Detection Panel v1.3.

Gene	Exon	Nucleotide Change (c.notation)	Amino Acid Change (p.notation)	Cosmic ID
<i>KRAS</i>	2	c.35G>C	p.(Gly12Ala)	522
<i>KRAS</i>	2	c.35G>A	p.(Gly12Asp)	521
<i>KRAS</i>	2	c.34G>C	p.(Gly12Arg)	518
<i>KRAS</i>	2	c.34G>T	p.(Gly12Cys)	516
<i>KRAS</i>	2	c.34G>A	p.(Gly12Ser)	517
<i>KRAS</i>	2	c.35G>T	p.(Gly12Val)	520
<i>KRAS</i>	2	c.38G>A	p.(Gly13Asp)	532
<i>KRAS</i>	3	c.175G>A	p.(Ala59Thr)	546
<i>KRAS</i>	3	c.176C>A	p.(Ala59Glu)	547
<i>KRAS</i>	3	c.176C>G	p.(Ala59Gly)	28518
<i>KRAS</i>	3	c.183A>C	p.(Gln61His)	554
<i>KRAS</i>	3	c.183A>T	p.(Gln61His)	555
<i>KRAS</i>	3	c.182A>T	p.(Gln61Leu)	553
<i>KRAS</i>	3	c.182A>G	p.(Gln61Arg)	552
<i>KRAS</i>	4	c.351A>C	p.(Lys117Asn)	19940
<i>KRAS</i>	4	c.351A>T	p.(Lys117Asn)	28519
<i>KRAS</i>	4	c.350A>G	p.(Lys117Arg)	4696722

<i>KRAS</i>	4	c.349A>G	p.(Lys117Glu)	N/A
<i>KRAS</i>	4	c.436G>A	p.(Ala146Thr)	19404
<i>KRAS</i>	4	c.436G>C	p.(Ala146Pro)	19905
<i>KRAS</i>	4	c.437C>T	p.(Ala146Val)	19900
<i>NRAS</i>	2	c.35G>A	p.(Gly12Asp)	564
<i>NRAS</i>	2	c.34G>A	p.(Gly12Ser)	563
<i>NRAS</i>	2	c.34G>T	p.(Gly12Cys)	562
<i>NRAS</i>	2	c.37G>C	p.(Gly13Arg)	569
<i>NRAS</i>	2	c.38G>T	p.(Gly13Val)	574
<i>NRAS</i>	3	c.175G>A	p.(Ala59Thr)	578
<i>NRAS</i>	3	c.176C>A	p.(Ala59Asp)	253327
<i>NRAS</i>	3	c.181C>A	p.(Gln61Lys)	580
<i>NRAS</i>	3	c.182A>T	p.(Gln61Leu)	583
<i>NRAS</i>	3	c.182A>G	p.(Gln61Arg)	584
<i>NRAS</i>	3	c.183A>C	p.(Gln61His)	586
<i>NRAS</i>	3	c.183A>T	p.(Gln61His)	585
<i>NRAS</i>	4	c.350A>G	p.(Lys117Arg)	N/A
<i>NRAS</i>	4	c.436G>A	p.(Ala146Thr)	27174
<i>BRAF</i>	15	c.1799T>A	p.(Val600Glu)	476
<i>BRAF</i>	15	c.1799_1800delinsAA	p.(Val600Glu)	N/A
<i>BRAF</i>	15	c.1799_1800delinsAT	p.(Val600Asp)	477

<i>BRAF</i>	15	c.1798_1799delinsAA	p.(Val600Lys)	473
<i>PIK3CA</i>	9	c.1624G>A	p.(Glu542Lys)	760
<i>PIK3CA</i>	9	c.1633G>A	p.(Glu545Lys)	763
<i>PIK3CA</i>	9	c.1633G>C	p.(Glu545Gln),	27133
<i>PIK3CA</i>	20	c.3140A>G	p.(His1047Arg)	775
<i>PIK3CA</i>	20	c.3140A>T	p.(His1047Leu)	776
<i>AKT1</i>	4	c.49G>A	p.(Glu17Lys)	33765

N/A, not available.

Table SIII. Summary of detected variants in 62 patients with gastrointestinal tumour.

GIST number	Gene	Location of the alteration			Type of mutation	Pathogenicity	Database	
		Exon	Nucleotide Change (c. notation)	Amino Acid Change (p. notation)				
1	<i>KIT</i>	11	c.1668_1673del	p.(Trp557_Lys558del)	P	N/A	COSMIC: COSM1211 ClinVar: / CIViC Score: /	PMID: 21286759
2	<i>KIT</i>	13	c.1961T>C	p.(Val654Ala)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM12706 ClinVar: 375921 CIViC Score: 18	PMID: 17363509

3	<i>KIT</i>	11	c.1673_1674insTCC	p.(Lys558delinsAsnPro)	P	N/A	COSMIC: COSM21976 ClinVar: / CIViC Score: /	PMID: 18715619
4	<i>KIT</i>	11	c.1653_1660delinsAA	p.(Met552_Glu554delinsLys)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
5	<i>KIT</i>	11	c.1674_1676del	p.(Lys558_Val559delinsAsn)	P	N/A	COSMIC: COSM1244 ClinVar: / CIViC Score: /	PMID: 21725412
6	<i>KIT</i>	11	c.1648_1668del	p.(Lys550_Gln556del)	P	Pathogenic	COSMIC: COSM327596 ClinVar: 13857 CIViC Score: /	PMID: 28553740
7	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355 PMID: 24061512
8	<i>KIT</i>	11	c.1676_1720del	p.(Val559_Thr574delinsAla)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A

9	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355 PMID: 24061512
10	<i>KIT</i>	11	c.1648_1674del	p.(Lys550_Lys558del)	P	Pathogenic	COSMIC: COSM1163 ClinVar: 13857 CIViC Score: /	PMID: 25970686
11	<i>KIT</i>	11	c.1663_1674del	p.(Gln556_Val559)	P	N/A	COSMIC: COSM1199 ClinVar: / CIViC Score: /	PMID: 21286759
12	<i>PDGFRA</i>	14	c.1936A>G	p.(Lys627Glu)	D	N/A	COSMIC: COSM33840 ClinVar: / CIViC Score: /	PMID: 25239601
	<i>PDGFRA</i>	14	c.1975A>C	p.(Asn659His)	D	Pathogenic	COSMIC: COSM22416 ClinVar: / CIViC Score: /	PMID: 20453818
13	<i>KIT</i>	13	c.1924A>G	p.(Lys642Glu)	P	Pathogenic	COSMIC:	PMID: 17824795

							ClinVar: 13866 CIViC Score: 5	
	<i>KIT</i>	17	c.2464A>T	p.(Asn822Tyr)	S	Pathogenic	COSMIC: COSM19109 ClinVar: / CIViC: /	DOI: 10.1200/jco.2015
14	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355 PMID: 24061512
15	<i>KIT</i>	11	c.1727T>C	p.(Leu576Pro)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1290 ClinVar: 375919 CIViC Score: 9	PMID: 26942271
16	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355
17	<i>KIT</i>	11	c.1670_1675del	p.(Trp557_Val559delinsPhe)	P	N/A	COSMIC: COSM1226 ClinVar: / CIViC Score: /	PMID: 12949711

18	<i>KIT</i>	11	c.1668_1724del	p.(Trp557_Gln575del)	P	N/A	COSMIC: COSM96891 ClinVar: / CIViC Score: /	PMID: 20861712
19	<i>KIT</i>	11	c.1670_1675del	p.(Trp557_Val559delinsPhe)	P	N/A	COSMIC: COSM1226 ClinVar: / CIViC Score: /	PMID: 12949711
20	<i>KIT</i>	11	c.1648-5_1672del	p.?	P	N/A	COSMIC: COSM5352267 ClinVar: / CIViC Score: /	PMID: 15507676
21	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355 PMID: 24061512
22	<i>KIT</i>	11	c.1672_1680del	p.(Lys558_Val560del)	P	N/A	COSMIC: COSM1234 ClinVar: / CIViC Score: /	PMID: 29299364
	<i>KIT</i>	17	c.2467T>G	p.(Tyr823Asp)	S	Likely Pathogenic	COSMIC: COSM18681	DOI: 10.1111/j.13 7006.2008.00727

							ClinVar: 376101 CIViC Score: 7.5	
23	<i>KIT</i>	11	c.1722_1766dup	p.(Gln575_Leu589dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
24	<i>KIT</i>	11	c.1669_1683del	p.(Trp557_Glu561del)	P	N/A	COSMIC: COSM1332 ClinVar: / CIViC Score: /	PMID: 20861712
25	<i>KIT</i>	11	c.1652_1660del	p.(Pro551_Glu554delinsGln)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 25970686
26	<i>KIT</i>	11	c.1727T>C	p.(Leu576Pro)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1290 ClinVar: 375919 CIViC Score: 9	PMID: 26942271 DOI: 10.1158/153
	<i>KIT</i>	13	c.1961T>C	p.(Val654Ala)	S	Pathogenic	COSMIC: COSM12706	PMID: 17363509

							ClinVar: 375921 CIViC Score: 18	
27	<i>KIT</i>	11	c.1669_1674del	p.(Trp557_Lys558del)	P	Likely Pathogenic	COSMIC: COSM1217 ClinVar: 222957 CIViC Score: /	PMID: 9438854
28	<i>KIT</i>	11	c.1673_1687del	p.(Lys558_Glu562del)	P	N/A	COSMIC: COSM18896 ClinVar: / CIViC Score: /	PMID: 25970686
29	<i>KIT</i>	11	c.1701_1726delinsGGAAG	p.(Asn567_Leu576delinsLysGluVal)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 25970686
30	<i>KIT</i>	11	c.1721_1765dup	p.(Arg588_Leu589ins15)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
	<i>KIT</i>	17	c.2466T>A	p.(Asn822Lys)	S	Pathogenic	COSMIC: COSM1321	PMID: 26316776

							ClinVar: 375931 CIViC Score: 7.5	
31	<i>KIT</i>	11	c.1676T>A	p.(Val559Asp)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1252 ClinVar: 13856 CIViC Score: 9.5	PMID: 29340041
32	<i>KIT</i>	11	c.1676_1714del	p.(Val559_Ile571del)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 28203166
33	<i>KIT</i>	11	c.1679T>A	p.(Val560Asp)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1257 ClinVar: 375914 CIViC Score: /	PMID: 25157968
34	<i>PDGFRA</i>	18	c.2525A>T	p.(Asp842Val)	P	Pathogenic	COSMIC: COSM736 ClinVar: 13543 CIViC Score:	PMID: 25970686

							84	
35	<i>KIT</i>	11	c.1679T>A	p.(Val560Asp)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1257 ClinVar: 375914 CIViC Score: /	PMID: 10086344
36	<i>KIT</i>	9	c.1504_1509dup	p.(Ala502_Tyr503dup)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 18312355 PMID: 24061512
37	<i>KIT</i>	11	c.1669_1674del	p.(Trp557_Lys558del)	P	Likely Pathogenic	COSMIC: COSM1217 ClinVar: 222957 CIViC Score: /	PMID: 9438854
38	<i>KIT</i>	11	c.1684G>A	p.(Glu562Lys)	P	Pathogenic	COSMIC: COSM17946 ClinVar: / CIViC Score: /	PMID: 20861712
39	<i>KIT</i>	11	c.1673_1714del	p.(Lys558_Asp572delinsAsn)	P	N/A	COSMIC: COSM25070	PMID: 29568401

							ClinVar: / CIViC Score: /	
40	<i>KIT</i>	11	c.1676T>A	p.(Val559Asp)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1252 ClinVar: 13856 CIViC Score: 9.5	PMID: 29340041
41	<i>KIT</i>	11	c.1669T>C	p.(Trp557Arg)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1219 ClinVar: 375909 CIViC Score: /	PMID: 10665649
42	<i>KIT</i>	11	c.1672_1680del	p.(Lys558_Val560del)	P	N/A	COSMIC: COSM1234 ClinVar: / CIViC Score: /	PMID: 10485475 PMID: 25109408
43	<i>KIT</i>	11	c.1661_1675del	p.(Glu554_Lys558del)	P	N/A	COSMIC: COSM1329 ClinVar: / CIViC Score: /	PMID: 9916918 BRAGGIO E et al RESEARCH 30: :

44	<i>KIT</i>	11	c.1668_1686del	p.(Trp557*)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
45	<i>KIT</i>	11	c.1652_1672del	p.(Pro551_Lys558delinsGln)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
46	<i>KIT</i>	11	c.1648_1674del	p.(Lys550_Lys558del)	P	Pathogenic	COSMIC: COSM1163 ClinVar: 13857 CIViC Score: /	PMID: 9438854 PMID: 22307903
47	<i>KIT</i>	11	c.1679T>A	p.(Val560Asp)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1257 ClinVar: 375914 CIViC Score: /	PMID: 25157968
48	<i>KIT</i>	11	c.1679_1681del	p.(Val560del)	P	Likely Pathogenic/ Pathogenic	COSMIC: / ClinVar: 375913 CIViC Score: 12.5	PMID: 19861435

49	<i>KIT</i>	11	c.1652_1672del	p.(Pro551_Lys558delinsGln)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
	<i>KIT</i>	13	c.1961T>C	p.(Val654Ala)	S	Pathogenic	COSMIC: COSM12706 ClinVar: 375921 CIViC Score: 18	PMID: 17363509
50	<i>KIT</i>	11	c.1665_1673del	p.(Gln556_Lys558del)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	PMID: 25970686
	<i>KIT</i>	13	c.1961T>C	p.(Val654Ala)	S	Pathogenic	COSMIC: COSM12706 ClinVar: 375921 CIViC Score: 18	PMID: 17363509
51	<i>KIT</i>	11	c.1679_1681del	p.(Val560del)	P	Likely Pathogenic/ Pathogenic	COSMIC: / ClinVar: 375913	PMID: 19861435

							CIViC Score: 12.5	
	<i>KIT</i>	11	c.1667_1672del	p.(Trp557_Lys558del)	P	N/A	COSMIC: COSM1210 ClinVar: / CIViC Score: /	PMID: 11023095
53	<i>KIT</i>	11	c.1715_1756dup	p.(Pro585_Arg586ins14)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A
54	<i>KIT</i>	11	c.1667_1674delinsCCACT	p.(Gln556_Lys558delinsProThr)	P	N/A	COSMIC: COSM51534 ClinVar: / CIViC Score: /	PMID: 20470368
55	<i>KIT</i>	11	c.1669T>A	p.(Trp557Arg)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1216 ClinVar: 375908 CIViC Score: /	PMID: 10665649
56	<i>KIT</i>	11	c.1671_1679del	p.(Trp557_Val560delinsCys)	P	N/A	COSMIC: COSM1232 ClinVar: /	PMID: 25886408

							CIViC Score: /	
57	<i>KIT</i>	11	c.1713_1739dup	p.(Asp579_His580ins9)	P	N/A	COSMIC: COSM1169434 ClinVar: / CIViC Score: /	PMID: 23291969
58	<i>KIT</i>	11	c.1697_1720del	p.(Asn566_Pro573del)	P	N/A	COSMIC: COSM6948216 ClinVar: / CIViC Score: /	BRAGGIO E et al RESEARCH 30: :
59	<i>KIT</i>	11	c.1669T>A	p.(Trp557Arg)	P	Likely Pathogenic/ Pathogenic	COSMIC: COSM1216 ClinVar: 375908 CIViC Score: /	PMID: 10665649
60	<i>KIT</i>	11	c.1735_1737del	p.(Asp579del)	P	Likely Pathogenic	COSMIC: COSM1294 ClinVar: 409725 CIViC Score: 0	PMID: 20861712
61	<i>KIT</i>	11	c.1669_1683del	p.(Trp557_Glu561del)	P	N/A	COSMIC: COSM1332	PMID: 20861712

							ClinVar: / CIViC Score: /	
62	<i>KIT</i>	11	c.1665_1672delinsCC	p.(Trp557_Lys558del)	P	N/A	COSMIC: / ClinVar: / CIViC Score: /	N/A

N/A, not available; P, primary mutation; S, secondary mutation; D, double mutation.