

Supplementary Table 2: Chromosomal imbalances and molecular alterations detected in 29 GIST submitted to CGH analysis.

ID	Gene	Exon	Mutation*	CGH findings**
2	<i>KIT</i>	9	p.Ala502_Tyr503dup	rev ish dim(X)(p11p22),dim(1)(p13p36),enh(1)(q21),enh(1)(q32),dim(2)(q11q37),enh(3)(p21),enh(3)(q21),enh(3)(q28q29),enh(6)(p21),enh(7)(q22),enh(7)(q36),enh(8)(p11p23),enh(9)(p12p13),enh(9)(q22q34),enh(10)(q22q26),dim(11)(p14p15),enh(11)(q13q14),enh(12)(p13),enh(12)(q13),enh(12)(q22q24),dim(13)(q),dim(14)(q),dim(15)(q15q26),enh(16),enh(17),enh(19),enh(20)(q),dim(21)(q)
18a	<i>KIT</i>	9	p.A502_Y503dup	rev ish dim(1)(p13p36),dim(2)(p25q13),dim(11),enh(12)(p12p13),dim(13)(q),dim(15)(q),enh(21)(q)
55	<i>KIT</i>	9	p.Ala502_Tyr503dup	rev ish dim(1)(p13p36),dim(6)(q12q27),dim(9)(p21p24),dim(14)(q),dim(18)(p11q21),dim(19)(p13),enh(20)(p12p13),enh(20)(q),dim(22)(q)
3	<i>KIT</i>	11	p.Trp557_Lys558del	rev ish enh(Y),dim(1)(p21p36),enh(1)(q21q23),enh(1)(q42q44),dim(2)(q11q37),enh(6)(p12p25),dim(6)(q24q27),enh(8)(p21p22),enh(12)(q21q24),dim(13)(q),dim(14)(q),dim(15)(q21q26),enh(17)(q11q24),dim(18)(q21q23),dim(22)(q)
4	<i>KIT</i>	11	p.Lys550_Gln556del	rev ish enh(6)(q26q27),dim(10),dim(14)(q),dim(15)(q),dim(22)(q)
44	<i>KIT</i>	11	p.Val559_Glu561del	rev ish dim(14)(q),dim(15)(q),dim(22)(q)
60	<i>KIT</i>	11	p.Asp579del	rev ish dim(14)(q)
62	<i>KIT</i>	11	p.Lys550_Lys558del	rev ish dim(11)(p),dim(14)(q),dim(22)(q)
13	<i>KIT</i>	11	p.Tyr553_Leu576delinsAsnCysLeuHis LeuTyrSerSerGln	rev ish dim(14)(q)
34	<i>KIT</i>	11	p.Trp557_Val559delinsPhe	rev ish dim(1)(p13p36),enh(1)(q21q44),dim(2)(p),enh(2)(q14q36),enh(3),enh(4),enh(5)(p12p15),enh(5)(q12q35),enh(6)(p12p25),dim(6)(q16q27),enh(7)(p13p22),enh(7)(q21q35),enh(8)(p22p23),enh(8)(q13q24),enh(10)(p13q22),enh(10)(q24),enh(11)(p14p15),dim(11)(q22q23),enh(12)(q14q22),dim(13)(q),dim(14)(q),dim(15)(q),dim(18)(q),dim(21)(q),dim(22)(q)
45	<i>KIT</i>	11	p.Lys558_Thr574delinsAsnArgSer	rev ish dim(1)(p12p36),dim(2)(p22p23),dim(3)(q12q22),dim(4)(p12p16),dim(7)(p),dim(9)(q22q32)
10	<i>KIT</i>	11	p.Asp572_His580dup	rev ish dim(X)(q21q28),dim(14)(q)
72	<i>KIT</i>	11	p.Pro585_Arg586insThrThr GlnLeuProTyrAspHisLysTrpGluPhePro	rev ish dim(14)(q),enh(20)(q)
5a	<i>KIT</i>	11	p.Trp557Arg	rev ish dim(1)(p13p36),dim(3)(p21p25),dim(14)(q22q32),dim(15)(q12q22),enh(17)(q21q25),dim(22)(q)
54	<i>KIT</i>	11	p.Val559Asp	rev ish dim(14)(q),dim(22)(q)
15b	<i>KIT</i>	11	p. Trp557Gly	rev ish dim(1)(p36q21),enh(10)(q22q23),dim(10)(q24q26),dim(14)(q),dim(15)(q21q26),dim(22)(q)
14	<i>KIT</i>	17	p.Asp820Tyr	rev ish dim(1)(p21p34),dim(3)(q12q26),dim(8)(p21p23),dim(11),enh(12),dim(22)(q)
40	<i>KIT</i>	17	p.Asp820Tyr	rev ish dim(14)(q),dim(15)(q)
21	<i>PDGFRA</i>	12	p.Ser566_Glu571delinsArg	rev ish dim(14)(q)

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22	<i>PDGFRA</i>	14	p.Asn569Tyr	No copy number changes
7	<i>PDGFRA</i>	18	p.Met844_Ser847del	rev ish dim(12)(p),enh(12)(q12q24),dim(14)(q)
6	<i>PDGFRA</i>	18	p.Asp842Val	rev ish dim(14)(q),dim(22)(q)
9	<i>PDGFRA</i>	18	p.Asp842Val	rev ish dim(14)(q22q32)
50	<i>PDGFRA</i>	18	p.Asp842Val	rev ish enh(10)(q26),dim(14)(q)
53	<i>PDGFRA</i>	18	p.Asp842Val	rev ish dim(1)(p),enh(5),dim(10)(q21q26),dim(14)(q12q32)
63	-	-	-	rev ish dim(X),dim(1)(p12p36),enh(1)(q21q44),dim(3)(q13q29),dim(9)(q22q34),enh(11)(p15), enh(11)(q13q25),dim(13)(q),dim(14)(q)
1	-	-	-	No copy number changes
8	-	-	-	No copy number changes
32	-	-	-	No copy number changes

*Mutant sequences at the protein level are deduced from the mutations identified at the DNA level. Mutation nomenclature follows the recommendations of the Human Genome Variation Society (<http://www.hgvs.org>). **CGH descriptions follow the guidelines proposed by the International System for Human Chromosome Nomenclature (ISCN) 2005. Abbreviations: delins, deletion insertion mutation; rev, reverse; ish, in situ hybridization; dim, diminished; enh, enhanced.