

SUPPLEMENTARY TABLES

Supplementary table 1S. Identification of candidate inversions on all chromosomes and chromosome 2

Filters	Number of inversions passing filters	
	All chromosomes	Chromosome 2
Inversions detected by BreakDancer	800	75
Inversions with confidence score >80	352	38
Inversions supported by at least five reads	303	35
Inversions not found in 22 control samples	175	21
Inversions detected by Manta	73	8
Genes near inversion boundaries (<15 kilobases)	39	6
Intonic or exonic breaks near inversion boundaries	31	4

Supplementary table 2S. 31 inversions selected after all the filtration process of whole genome sequence data

CHR#	Left	Annotation	Genes	Left	CHR#	Right	Annotation	Genes	Right	Size
boundary				band	boundary				band	(Mb)
1	17185929	upstream	MIR3675	p36.13	1	145382685	intronic	NBPF10, NBPF20	q21.1	128.2
1	54271697	intronic	NDC1	p32.3	1	55706676	intergenic	MIR4422(dist=15280), PPAP2B(dist=1253743)	p32.3	1.4
1	147731513	intronic	NBPF8	q21.1	1	148559669	ncRNA_intronic	NBPF25P	q21.2	0.8
1	145109494	intronic	NBPF20, NBPF9, SEC22B	q21.1	1	148184080	intronic	NBPF8	q21.2	3.1
2	130944046	intronic	MZT2B	q21.1	2	132246786	ncRNA_intronic	RNU6-81P	q21.2	1.3
2	70184039	intergenic	MXD1(dist=13963), ASPRV1(dist=3185)	p14	2	198761941	intronic	PLCL1	q33.1	128.6
2	126701173	intergenic	CNTNAP5(dist=1028219), GYPC(dist=712338)	q14.3	2	209310827	intronic	PTH2R	q34	82.6
2	126700323	intergenic	CNTNAP5(dist=1027369), GYPC(dist=713188)	q14.3	2	142666659	intronic	LRP1B	q22.2	16.0
3	187860533	intergenic	BCL6(dist=397020), LPP-AS2(dist=8461)	q27.3	3	193081071	exonic	ATP13A5	q28	5.2
3	187860533	intergenic	BCL6(dist=397020), LPP-AS2(dist=8461)	q27.3	3	193081071	exonic	ATP13A5	q28	5.2

3	187860533	intergenic	BCL6(dist=397020), LPP-AS2(dist=8461)	q27.3	3	193080245	exonic	ATP13A5	q28	5.2
3	187860533	intergenic	BCL6(dist=397020), LPP-AS2(dist=8461)	q27.3	3	193080245	exonic	ATP13A5	q28	5.2
4	71878179	intronic	DCK	q13.3	4	71956181	intergenic	DCK(dist=59552), SLC4A4(dist=96822)	q13.3	0.1
7	149737329	intergenic	ATP6V0E2(dist=159528), ACTR3C(dist=206972)	q36.1	7	153760892	intronic	DPP6	q36.2	4.0
8	48844731	intronic	PRKDC	q11.21	8	48847240	intronic	PRKDC	q11.21	0.0
11	24509946	intergenic	MIR8054(dist=1069210), LUZP2(dist=8570)	p14.3	11	34973254	intronic	PDHX	p13	10.5
11	24509946	intergenic	MIR8054(dist=1069210), LUZP2(dist=8570)	p14.3	11	34973254	intronic	PDHX	p13	10.5
12	133141845	intronic	FBRSL1	q24.33	12	133344199	intergenic	ANKLE2(dist=5725), GOLGA3(dist=1296)	q24.33	0.2
12	133141845	intronic	FBRSL1	q24.33	12	133344199	intergenic	ANKLE2(dist=5725), GOLGA3(dist=1296)	q24.33	0.2
14	58259536	intronic	SLC35F4	q23.1	14	58412621	intergenic	SLC35F4(dist=80029), C14orf37(dist=58187)	q23.1	0.2
15	79057976	intronic	ADAMTS7	q25.1	15	82620316	ncRNA_exonic	ADAMTS7P1	q25.2	3.6
16	24121355	intronic	PRKCB	p12.1	16	80277677	ncRNA_intronic	LOC102724084	q23.2	56.2

17	746316	intronic	NXN	p13.3	17	11598770	intronic	DNAH9	p12	10.9
19	53193320	intronic	ZNF83	q13.32	19	53869176	ncRNA_intronic	ZNF525	q13.33	0.7
19	53193320	intronic	ZNF83	q13.32	19	53869176	ncRNA_intronic	ZNF525	q13.33	0.7
19	53193320	intronic	ZNF83	q13.32	19	53870170	ncRNA_intronic	ZNF525	q13.33	0.7
19	53193320	intronic	ZNF83	q13.32	19	53870170	ncRNA_intronic	ZNF525	q13.33	0.7
19	10447827	intronic	ICAM3	p13.2	19	31795439	intronic	TSHZ3	q12	21.3
19	10869740	intronic	DNM2	p13.2	19	54685855	intronic	MBOAT7	q13.33	43.8
X	149570895	intronic	MAMLD1	q28	X	149585449	intronic	MAMLD1	q28	0.0
X	49013681	intergenic	GPKOW(dist=33530), MAGIX(dist=5500)	p11.23	X	49019827	intronic	MAGIX	p11.23	0.0

Supplementary table 3S. List of genes highly affected by homozygous variants

CHR#	Position	Variant ID	Ref	Alt	Effect	Gene
3	194080395	rs113243841	G	A	Nonsense	<i>LRRC15</i>
4	155244401	rs112727159	TTTG	T	Frameshift	<i>DCHS2</i>
4	3590823	rs34083130	GACAC	G	Splice site acceptor	<i>RP3-368B9.1.1</i>
6	96034869	NA	G	GTATA	Frameshift	<i>MANEA</i>
13	100517195	rs3831038	CTG	C	Frameshift	<i>CLYBL</i>
17	71188971	rs57350092	G	GC	Splice site donor	<i>COG1</i>
22	20779973	rs5844418	C	CG	Frameshift	<i>SCARF2</i>

* The 1000 Genomes, and ExAC databases were consulted upon identification of variants.