

Supplemental Data

FISH Mapping of de novo Apparently Balanced Chromosome Rearrangements Identifies Characteristics Associated with Phenotypic Abnormality

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Table S1. Cytogenetic and FISH Localization of Breakpoints in Cases and Controls

Group	Number	AB	Code	ABCR (Cyto BP)	ABCR (FISH BP)
Cases	1	XAT	GILLE	t(X;11)(p22.3;p12)	t(X;11)(p22.2; p11.2)
	2	Recip	MARTA	t(8;9)(q21;q21)	t(8;9)(q22.1;q21.13)
	3	Recip	SG-3301	t(2;17)(q32;q24)pat	t(2;17)(q32.1;q24.3)pat
	4	Recip	ROOJA	t(5;7)(q21;q33)	t(5;7)(q14.3;q22.1)
	5	Recip	MAGAN	t(3;4)(q23;q31)	t(3;4)(q23;q31.22)
	6	Inv	CRENA	inv(4)(q21q35)	inv(4)(q21.22q35)
	7	Recip	F92-2253	t(5;11)(q15;p15.5)	t(5;11)(q15;p15.5)
	8	Recip	T86-0593	t(5;12)(q11.2;q12)	t(5;12)(q11.1;q11)
	9	Inv	GILL	inv(12)(p11.2q24.3)	inv(12)(p11.21q24.31)
	10	Recip	B97-1182	t(2;10)(q11.2;q22.3)	t(2;10)(q11.2;q22.3)
	11	Recip	B01-2958	t(4;14)(q31.22;q11.2)	t(4;14)(q31.22;q11.2)
	12	Recip	B96-0581	t(2;3)(q21.3;q21.3)	t(2;3)(q11.2;q13.31)
	13	XAT	F03-0432	t(X;8)(q26.2;p23.2)	t(X;8)(q26.2;p23.2)
	14	Recip	B01-2957	t(14;18)(q24.1;p11.32)	t(14;18)(q24.2;p11.32)
	15	Recip	B99-1983	t(13;18)(q31.1/q22.3;q22.1)	t(13;18)(q31.1;q22.2)
	16	Recip	B04-0611	t(1;9)(p32.3;q33.2)	t(1;9)(p31.1;q31.3)
	17	Inv	B90748	inv(1)(p36.1q25.3)	inv(1)(p31.3q25.1)

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18	Recip	CMS5859	t(3;7)(q28;p21.3)	t(3;7)(q28;p21.3)
19	Recip	B04-0880	t(2;3)(q32.2/q33;q26.3/q27)	t(2;3)(q33.1;q26.33)
20	Recip	T86-0404	t(2;6)(p23;q14)	t(2;6)(p22.3;q12)
21	Recip	CV1012	t(5;17)(q15;q23)pat	t(5;17)(q15;q24.3)pat
22	Recip	NCL-B04-2316	t(3;6)(q23;p21.1)	t(3;6)(q21.3;p21.1)
23	Recip	COLJA	t(11;13)(p15.3;q22)	t(11;13)(p15.4;q22.1)
24	Recip	B00213	t(2;7)(q33;p21)	t(2;7)(q33.1;p21.1)
25	Recip	BB45	t(2;11)(q32;p14)	t(2;11)(q33.1;p13)
26	XAT	BL02-1299	t(X;10)(q22.3;q24.3)	t(X;10)(q23;q25.1)
27	XAT	BL02-0828	t(X;11)(q21.2;q13.5)	t(X;11)(q21.31;q14.1)
28	Recip	F0124/00	t(2;16)(p15;q22)	t(2;16)(p14;q21/q22.1)
29	Recip	BL02-3104	t(9;22)(q34.2;q11.21)	t(9;22)(q34.2;q11.21)
30	Recip	AC114	t(10;11)(q24;p13)	t(10;11)(q24.2;p12)
31	Recip	BL03-1791	t(7;8)(q32.2;q22)	t(7;8)(q22.1;q22.1)
32	Recip	CV345	t(6;9)(p23;q22.3)	t(6;9)(p24.3;q22.33)
33	Inv	BL02-2567	inv(1)(q32.1q44)	inv(1)(q41q42.2)inv(1)(q42.3q44)
34	Recip	BL03-0967	t(9;20)(p13.1;p13)	t(9;20)(p13.3;p13)
35	Recip	BL03-1789	t(5;9)(p13.1;q22.1)	t(5;9)(p12;q21.11)
36	Recip	BL03-2425	t(11;13)(p15;q32)	t(11;13)(p15.4;q32.1)
37	Recip	CV1222	t(1;7)(q21.3;q34)	t(1;7)(q23.2;q34)
38	Recip	BL04-0107	t(5;6)(q13;q23)	t(5;6)(q14.3;q25.1)
39	Recip	BL03-3077	t(1;20)(q32.1;q13.3)	t(1;20)(q31.3;q13.32)
40	Recip	CV1456	t(1;2)(q24.2;q31.3)	t(1;2)(q24.1;q31.1)
41	Recip	BL04-2026	t(5;6)(q15;q25.1)	t(5;6)(q14.3;q25.1)
42	Recip	BL04-2240	t(9;15)(q21.2;q26)	t(9;15)(q21.33;q26.2)
43	XAT	BL04-3772	t(X;4)(p11.4;p16)	t(X;4)(p21.1;p16.2)
44	Recip	BL04-1899	t(6;16)(q15;q13)	t(6;16)(q16.1;q21)

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	45	Recip	BL05-0692	t(1;13)(q44;q32)	t(1;13)(q44;q32.1)
	46	Recip	BL03-2183	t(5;14)(q15;q24)	t(5;14)(q21.3/q22.1;q31.1)
Controls	1	Recip	B98-0026	t(1;15)(q24.3;q22.3)	t(1;15)(q25.2;q24.2)
	2	Recip	F00-1558	t(15;16)(q22.3;q22.1)	t(15;16)(q24.2;q22.1)
	3	Recip	B97-0349	t(3;11)(p13;q12.2)	t(3;11)(p14.1;q11)
	4	Recip	B04-0088	t(3;9)(p26;p22)	t(3;9)(p26.3;p21.3)
	5	Recip	F96-1781	t(5;6)(q31.3;p21.3)	t(5;6)(q32;p22.2)
	6	Inv	SISCO	inv(1)(p31q43)	inv(1)(p13.3q32.3)
	7	Recip	PETEN	t(7;12)(q32;p13)	t(7;12)(q32.2;p13.32)
	8	Recip	EDINN	t(1;14)(q42;q11)	t(1;14)(q41;q11.2)
	9	Recip	JOGAR	t(X;14)(p11.21;q32.2/32.3)	t(X;14)(p11.3;q32.33)
	10	Recip	B01-2804	t(4;12)(p16.1;q24.31)	t(4;12)(p16.1;q24.23)
	11	Recip	BL03-0187	t(2;8)(p15;q13)	t(2;8)(p16.3;q21.11)
	12	Recip	BL03-0186	t(1;7)(p32;q34)	t(1;7)(p32.1;q35)
	13	Recip	BL03-0185	t(2;4)(q37.1;p15.32)	t(2;4)(q37.2;p15.33)
	14	Recip	BL03-0362	t(2;5)(q31;q33.3)	t(2;5)(q32.1;q34)
	15	Recip	BL03-0356	t(3;12)(q27;p13)	t(3;12)(q27.1;p13.32)
	16	Recip	BL03-0426	t(4;11)(q31;p13)	t(4;11)(q28.3;p12)
	17	Recip	BL03-1970	t(6;15)(q23.1;q21.2)	t(6;15)(q23.2;q21.3)
	18	Recip	BL03-1979	t(6;14)(q32;q32.1)	t(6;14)(q22.33;q31.3)
	19	Recip	F0440	t(8;15)(q22;q22.1)	t(8;15)(q21.11;q21.1)
	20	Recip	BL03-0876	t(1;11)(q42.1;p15.5)	t(1;11)(q44;p15.5)
	21	Recip	BL03-3032	t(5;18)(p15.1;p11.32)	t(5;18)(p15.2;p11.31)
	22	Recip	BL04-0339	t(5;6)(q11.2;q23.1)	t(5;6)(q12.3;q24.1)
	23	Inv	BL03-2725	inv(14)(q22.1q32.1)	inv(14)(q21.3q31.3)
	24	Recip	BL04-2024	t(5;7)(p15.1;q31.2)	t(5;7)(p14.1;q31.2)
	25	Recip	BL04-2025	t(3;6)(q25;q15)	t(3;6)(q26.1;q15)

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26	Recip	CV1510	t(6;14)(p23;q13)	t(6;14)(p22.2;q12)
27	Recip	BL04-3994	t(3;9)(p25;q13)	t(3;9)(p26.3/p26.2;q21.13)
28	Recip	BL04-4270	t(6;8)(q25;q12)	t(6;8)(q24.2;q12.1)
29	Recip	BL05-0379	t(5;9)(p14;p13.1)	t(5;9)(p14.1;p13.2)
30	Recip	BL03-2435	t(6;12)(q21;q24.1)	t(6;12)(q16.3;q23.1)

“Recip” denotes “reciprocal translocation.”

“Inv” denotes “inversion.”

“XAT” denotes “X-autosome translocation.”

Table S2. Breakpoint-Localizing FISH Probes for Cases and Controls

Group	Number**	Gene Interrupted	FISH BP	Proximal Probe*	BP Spanning 1	BP Spanning 2	BP Spanning 3	Distal Probe*
Cases	1	Yes	Xp22.2		RP11-311A17	RP11-50B1	RP11-81G16	
	1	Yes	11p11.2		RP11-618K13			
	2	No	8q22.1		RP11-328L11			
	2	Yes	9q21.13		RP11-71A24			
	3	No	2q32.1		CTD-2053I13			
	3	No	17q24.3		RP11-1003J3			
	4	Yes	5q14.3		RP11-433H20	111M24	159L1	
	4	Yes	7q22.1		RP11-984K9	RP11-401L13		
	5	Yes	3q23		RP11-316C10	RP11-831N10		
	5	Yes	4q31.22		RP11-552I10			
	6	No	4q21.22		RP11-263F19			
	6	Unknown	4q35		cT379			
	7	Yes	5q15		RP11-126G10	RP11-641C22		
	7	No	11p15.5		RP4-539G11	RP11-889I17		
	8	No	5q11.1	Centromeric Repeats				RP11-185I4
	8	No	12q11	Centromeric Repeats				Centromeric Repeats
	9	No	12p11.21		RP13-359K18			
	9	Yes	12q24.31		RP11-665J20			
	10	Unknown	2q11.2	RP11-549H5				RP11-292K15
	10	Unknown	10q22.3		RP11-428P16			
	11	No	4q31.22		RP11-184E7			
	11	Unknown	14q11.2	RP11-1080M7	RP11-244E17			RP11-929J10

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12	Unknown	2q11.2					
12	Unknown	3q13.31		RP11-553L6	RP11-107J18		
13	Yes	Xq26.2		RP3-448E20			
13	No	8p23.2		RP11-728L1			
14	Unknown	14q24.2		RP11-41B1			
14	No	18p11.32		RP11-267C19	RP11-78F17		
15	No	13q31.1	RP11-521J24				RP11-421K11
15	Del/Dup	Del	RP11-365J7				RP11-632L2
15	No	18q22.2		RP11-384E22	RP11-430H7		
16	No	1p31.1		RP11-386O9			
16	Yes	9q31.3		RP11-272D1			
17	No	1p31.3		RP4-535B20			
17	Yes	1q25.1		RP5-1141O19	RP1-262D12		
18	No	3q28		RP11-332P22			
18	Del/Dup	Del	RP11-496M9				RP1-296J4
18	Unknown	7p21.3		RP11-135E21			
19	No	2q33.1		RP11-258L23			
19	No	3q26.33		RP11-43F17			
20	No	2p22.3	RP11-257N21				RP11-153O16
20	No	6q12		RP11-365P20			
21	No	5q15		RP11-496M2			
21	No	17q24.3		RP11-420O5			
22	Yes	3q21.3	RP11-177O2				RP3-519C2
22	Yes	6p21.1		RP1-244F24	427I13		
23	No	11p15.4		RP11-92N11	RP11-120E20		
23	No	13q22.1		RP11-309H15			

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24	Yes	2q33.1		RP11-530J6			
24	No	7p21.1		RP11-455A9	RP11-455K15	RP11-30M5	
25	No	2q33.1		RP11-505H14			
25	No	11p13		RP1-305G21			
26	No	10q25.1		RP11-469M2			
26	No	Xq23		RP11-537J24			
27	No	11q14.1	RP11-730C14				RP11-816L8
27	No	Xq21.31	RP13-212L9				RP13-24G8
27	Del/Dup	Del		RP11-179A16			
28	No	2p14		RP11-30P5			
28	No	16q21	RP11-256I9				RP11-298C15
29	Yes	9q34.13		G248P88004A10			
29	Unknown	22q11.21	RP11-466H9				RP11-481H20
30	Yes	10q24.2		RP11-34A14			
30	No	11p12		RP11-64I17			
31	No	8q22.1		RP11-347C18			
31	Yes	7q22.1	RP11-333G13				RP11-163E9
32	No	6p24.3	RP11-12I14				RP1-90O12
32	Yes	9q22.33	G248P87972G12				G248P80977B9
33	Del/Dup	Del q42.2		RP11528D17			
33	Del/Dup	Del q43-44	RP11-553N16				RP11-411G13
33	Del/Dup	Inv q32.3;q42.2	RP11-66M7				RP11-99J16
33	Del/Dup	Inv q42.3;q44	RP11-509I1				CTB-160H23
34	No	9p13.3		RP11-251H13			
34	Yes	20p13	RP5-1187M17				RP11-794I6
35	No	5p12	RP11-143E24				RP11-34D23

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	35	Unknown	9q21.11	RP11-265B8				RP11-141J10
	36	Yes	13q32.1	RP11-318K19				RP11-83D23
	36	Unknown	11p15.4		RP11-89E5			
	37	No	1q23.2	RP11-550P17				RP11-180D21
	37	Yes	7q34	RP11-759K14				RP4-592P3
	38	Yes	6q25.1	RP1-144O21				RP1-281H8
	38	No	5q14.3		RP11-348K15			
	39	Del/Dup	20q13.32	G248P83716B4	RP5-1059L7			RP11-402F1
	39	Unknown	1q31.3		RP11-332L8			
	40	Yes	Del 2q22.2	RP11-285H23				RP11-107E5
	40	Unknown	2q31.1	RP11-158L8				RP11-205B19
	40	Unknown	1q24.1	RP11-306I1				RP11-702J19
	41	No	5q14.3	RP11-24O11				RP11-302L17
	41	Yes	6q25.1	RP11-99E18				RP1-297M16
	42	No	15q26.2	RP11-522B15				RP11-78F1
	42	No	9q21.31		RP11-394O9			
	43	Unknown	4p16.2	G248P8903E10				G248P81939D6
	43	No	Xp21.1	RP11-281B1				RP11-330k13
	44	No	16q21	RP11-133B5				RP11-22H12
	44	No	6q16.1		G248P8547E3			
	45	No	13q32.1	G248P81419A12				G248P86855F5
	45	No	1q44	G248P89661B9				
	46	Yes	Dup 5q21.3 - q22.1	RP11-319P13				RP11-252I13
	46	Yes	Del 5q21.3 - q22.1	RP11-174L14				RP11-231G15
	46	Del/Dup	Ins 6q	RP11-276O18				RP11-1146A18

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	46	Unknown	14q31.1		RP11-299L17			
Controls	1	Yes	1q25.2		RP11-52I18			
	1	Unknown	15q24.2		RP11-593F23	RP11-685G9		
	2	Unknown	15q24.2		RP11-24M17			
	2	Unknown	16q22.1		RP11-1145H8	RP11-510M2		
	3	Yes	3p14.1		RP11-253K11	RP11-40E22		
	3	Unknown	11q11		RP11-367C21			
	4	Unknown	3p26.3		RP11-86C13	RP11-952H14		
	4	No	9p21.3		RP11-594J14			
	5	Unknown	5q32	RP11-25F13	RP11-153J20			CTC-370H24
	5	Unknown	6p22.2		RP11-95P3			
	6	Yes	1p13.3		RP11-352P4	RP11-911H2	RP11-297O4	
	6	No	1q32.3		RP11-372M18			
	7	No	7q32.2		RP11-2E11			
	7	Yes	12p13.32		RP11-916H17	RP5-1180D12		
	8	No	1q41		RP11-421L10			
	8	Unknown	14q11.2		RP11-98N22			
	9	Yes	Xp11.3		RP5-1158H2	RP11-573N21		
	9	Yes	14q32.33		RP11-982M15	RP11-716K17		
	10	No	4p16.1	RP11-770E1				RP11-136I13
	10	Yes	12q24.23	RP11-110J12				RP11-465L8
	11	No	2p16.3	RP11-460M2				RP11-152D8
	11	No	8q21.11		RP11-758M4			
	12	Yes	1p32.1		RP11-242B9			
	12	Yes	7q35	RP4-545C24				RP4-721B17
	13	Yes	2q37.2	RP11-84G18				RP11-473L20

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13	Yes	4p15.33	RP11-484O2				RP11-1087G16
14	Yes	2q32.1	RP11-363K21				RP11-1148B13
14	No	5q34	RP11-118E6				RP11-511M9
15	Yes	12p13.32	G248P84904G3				G248P8258A7
15	No	3q27.1		RP11-307B12			
16	No	11p12	RP11-330I10				RP11-313M3
16	No	4q28.3	RP11-356A12				RP11-782E2
17	No	6q23.2	RP1-258L1				RP3-352A20
17	No	15q22.2	G248P87450F6				G248P86114E3
18	Yes	6q23.2	RP11-197K23				RP11-103C16
18	No	14q31.3	RP11-126I9				RP11-504I1
19	Unknown	8q21.11	G248P89985B9				G248P8997H8
19	No	15q21.1	RP11-81G13				RP11-90J19
20	Yes	1q44		RP11-522M21			
20	Unknown	11p15.4		G248P81220F7	G248P82039E3		
21	No	5p15.2	G248P84750F11				G248P80586E8
21	Unknown	18p11.31	G248P8540A4				RP11-106J7
22	No	6q24.1	RP11-515I12				RP11-432L12
22	Yes	5q12.3	RP11-19B7				RP11-5N2
23	Unknown	14q21.3	G248P80298H4				G248P87607C11
23	Yes	14q31.3		G248P83151G6			
24	Yes	7q31.2	RP11-76C12				CTB-1013N12
24	No	5p14.1	RP11-500H9				RP11-192H6
25	No	3q26.1	RP11-535I24				RP11-90M7
25	No	6q15		RP11-565A2			
26	Unknown	6p22.2	RP11-111F20				G248P82409E12

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	26	Yes	14q12	RP11-452K13				RP11-159B13
	27	Yes	3p26.2	RP11-6301				RP11-785A7
	27	No	9q21.3	RP11-217F16	RP11-429L21			RP11-28P17
	28	Unknown	6q24.2	RP11-485E9				RP11-3B11
	28	No	8q12.1	RP11-342K10				RP11-513O17
	29	No	5p14.1	RP11-560A7				RP11-100E22
	29	Yes	9p13.2		RP11-392E22			
	29	Yes	Dup	G248P82869H12	G248P81638E3	G248P89472F3	G248P86044F7	RP13-198D9
	30	No	6q16.3	RP11-451D3				RP11-167K4
	30	Unknown	12q23.1		RP11-175P13			

*Where no breakpoint-spanning clone was identified, the proximal and distal flanking clones are given.

**Number is the same as that used in Table S1.

“Del/Dup” indicates that a deletion or duplication was detected at or in *cis* with the breakpoint.

“BP” denotes “breakpoint.”