Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

Supplement to: Wapner RJ, Martin CL, Levy B, et al. Chromosomal microarray versus karyotyping for prenatal diagnosis. N Engl J Med 2012;367:2175-84. DOI: 10.1056/NEJMoa1203382

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Chromosomal Microarray Compared with Karyotyping for Prenatal Diagnosis

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Table S1. All CNVs seen in Karyotypically Normal Pregnancies

A. Known Pathogenic CNVs

Deletions: de novo	**Anomaly**	**ISCN Nomenclature**	**Size in Mb**	**Fetal anomaly detected by US?**	**Consensus Call 2012**
	del Xp21.1 // dup 22q11.23	arr Xp21.1(32,476,856-32,689,852)x0 dn // 22q11.23(22,547,659-23,677,848)x3 pat	0.21 // 1.1	No	pCNC // uCNC
	del Xp22.31	arr Xp22.31(7,094,783-7,443,955)x0 dn	0.35	No	pCNC
	del 3p14.1p13	arr 3p14.1p13(67,917,266-72,754,608)x1 dn	4.8	Yes	pCNC
	del 5q35.1q35.2	arr 5q35.1q35.2(171,483,988-176,409,787)x1 dn	4.9	Yes	pCNC
	del 7q11.23	arr 7q11.23(72,404,248-73,976,981)x1 dn	1.6	Yes	pCNC^{+}
	del 7q21.3	arr 7q21.3(93,870,041-94,962,839)x1 dn	1.1	No	pCNC
	del 8q12.1	arr 8q12.1(61,104,853-62,025,350)x1 dn	0.92	Yes	pCNC
	del 11q24.2q25	arr 11q24.2q25(124,104,318-134,373,618)x1 dn	10.3	Yes	pCNC
	del15q13.2q13.3	arr 15q13.2q13.3(28,606,756-30,686,850)x1 dn	2.1	No	pCNC**$^{+}$
	del 17p13.3 // dup 21q22.3*	arr 17p13.3p13.2(48,338-4,855,684)x1 dn // 21q22.3(43,364,146-46,912,090)x3 dn	4.8 // 3.5	Yes	pCNC // pCNC
	del 17q12	arr 17q12(31,890,368-33,318,471)x1 dn	1.4	Yes	pCNC**$^{+}$
	del 17q12	arr 17q12(31,930,167-33,323,031)x1 dn	1.4	Yes	pCNC**$^{+}$
	del 17q12	arr 17q12(31,501,499-33,678,831)x1 dn	2.2	Yes	pCNC**$^{+}$
	del 17q12	arr 17q12(31,929,967-33,323,171)x1 dn	1.4	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,299,941-19,770,515)x1 dn	2.5	No	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,041,723-19,835,418)x1 dn	2.8	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,041,723-19,835,418)x1 dn	2.8	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,299,741-19,835,557)x1 dn	2.5	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,256,416-19,795,836)x1 dn	2.5	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,085,800-19,891,669)x1 dn	2.8	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,299,741-19,835,557)x1 dn	2.5	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,299,941-19,835,417)x1 dn	2.5	Yes	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,035,860-19,795,836)x1 dn	2.8	No	pCNC**$^{+}$
	del 22q11.21	arr 22q11.21(17,299,941-19,835,417)x1 dn	2.5	Yes	pCNC**$^{+}$

	del 22q11.21	arr 22q11.21(17,256,416-19,795,836)x1 dn	2.5	No	pCNC**+
	del 22q13.32q13.33	arr 22q13.32q13.33(47,562,540-49,513,532)x1 dn	2	No	pCNC**
	del 22q13.33	arr 22q13.33(49,472,416-49,525,855)x1 dn	0.05	Yes	pCNC**
Deletions: inherited					
	del Xp21.1p11.4	arr Xp21.1p11.4(31,972,908-40,736,368)x1 mat	8.8	No	pCNC
	del 1q21.1	arr 1q21.1(144,643,812-146,297,795)x1 mat	1.7	No	pCNC**+
	del 3p26.2	arr 3p26.2(4,395,412-4,598,515)x1 mat	0.2	No	pCNC
	del 17p12	arr 17p12(14,026,349-15,425,584)x1 mat	1.4	No	pCNC+
Duplications: de novo					
	dup 10q26.13q26.2	arr 10q26.13q26.2(125,730,437-129,696,364)x3 dn	4	Yes	pCNC
Duplications: inherited					
	dup Xp21.3 // dup Xp21.1 // dup 4q26	arr Xp21.3(28,356,453-28,859,331)x3 mat // Xp21.1(31,957,886-32,437,407)x3 mat // 4q26(116,357,164-117,646,417)x3 pat	0.5 // 0.5 // 1.3	No	lpCNC // lpCNC // lbCNC
	dup 4q13.2q21.1	arr 4q13.2q21.1(66,871,838-77,744,787)x3 pat	10.9	Yes	pCNC
	dup 17p12	arr 17p12(14,035,412-15,399,448)x3 pat	1.4	No	pCNC+

B. Uncertain CNVs: Referred to Advisory Committee and Reported to Patients

Deletions: de novo	**Anomaly**	**ISCN**	**Size in Mb**	**Fetal anomaly detected by US?**	**Consensus Call 2012**
	del 3q28	arr 3q28(190,362,984-190,910,032)x1 dn	0.55	No	uCNC
	del 6q23.3q24.1	arr 6q23.3q24.1(136,521,187-139,410,871)x1 dn	2.9	Yes	pCNC
	del 7p15.1	arr 7p15.1(28,277,115-30,113,824)x1 dn	1.8	No	pCNC
	del 7q35	arr 7q35(146,082,279-146,189,674)x1 dn	0.11	No	uCNC
	del 8p23.1	arr 8p23.1(11,578,132-11,789,207)x1 dn	0.21	Yes	pCNC
	del 10q11.21q11.23	arr 10q11.21q11.23(45,525,495-51,265,196)x1 dn	5.7	No	uCNC

del 12p11.23p11.22	arr 12p11.23p11.22(27,606,783-29,519,347)x1 dn	1.9	No	pCNC
del 14q32.2q32.31	arr 14q32.2q32.31(99,465,564-100,574,047)x1 dn	1.1	Yes	pCNC
del 15q25.2q25.3	arr 15q25.2q25.3(82,737,136-83,529,848)x1 dn	0.79	No	uCNC
del 16p11.2	arr 16p11.2(29,500,283-30,098,070)x1 dn	0.6	No	pCNC**$^{+}$
del 16p11.2	arr 16p11.2(29,238,063-30,098,209)x1 dn	0.9	Yes	pCNC**$^{+}$
del 16p11.2	arr 16p11.2(29,581,454-30,098,069)x1 dn	0.52	No	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(15,458,602-16,102,219)x1 dn	0.64	No	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(14,796,083-16,535,551)x1 dn	1.7	Yes	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(14,817,705-16,102,080)x1 dn	1.3	No	pCNC**$^{+}$
del 16p13.11p12.3	arr 16p13.11p12.3(15,311,952-18,539,483)x1 dn	3.2	Yes	pCNC**$^{+}$
del 16p13.11p12.3	arr 16p13.11p12.3(15,389,422-18,634,733)x1 dn	3.2	Yes	pCNC**$^{+}$
del 22q11.21	arr 22q11.21(19,084,421-19,835,417)x1 dn	0.75	Yes	uCNC
del 22q11.21q11.22	arr 22q11.21q11.22(19,355,453-20,772,918)x1 dn	1.4	No	uCNC
del 22q11.21	arr 22q11.21(19,046,924-19,941,338)x1 dn	0.89	No	uCNC
del 22q11.21	arr 22q11.21(19,084,421-19,835,417)x1 dn	0.75	No	uCNC
Deletions: Inherited				
del 3p24.1p23	arr 3p24.1p23(30,145,037-31,031,429)x1 mat	0.89	No	uCNC
del 3p26.3	arr 3p26.3(1,467,661-2,660,773)x1 mat	1.2	No	uCNC
del 3q28 // 13q12.12	arr 3q28(190,885,014-190,967,571)x1 mat // 13q12.12(23,214,657-24,123,596)x1 mat	0.08//0.91	No	uCNC // uCNC
del 10q11.21q11.23	arr 10q11.21q11.23(45,525,495-51,265,196)x1 pat	5.7	No	uCNC
del 10q21.1	arr 10q21.1(53,697,770-59,847,798)x1 pat	6.2	Yes	uCNC
del 10q21.1	arr 10q21.1(54,376,822-56,475,582)x1 mat	2.1	Yes	uCNC
del 10q23.33	arr 10q23.33(95,459,308-96,504,070)x1 pat	1	No	uCNC
del 14q22.1	arr 14q22.1(50,309,556-51,739,266)x1 mat	1.4	Yes	uCNC
del 16p12.1	arr 16p12.1(21,744,792-22,315,572)x1 pat	0.57	Yes	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(14,817,705-16,432,850)x1 mat	1.6	No	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(14,817,705-16,679,871)x1 mat	1.9	Yes	pCNC**$^{+}$
del 16p13.11p12.3	arr 16p13.11p12.3(15,311,952-18,577,227)x1 mat	3.3	No	pCNC**$^{+}$
del 16p13.11	arr 16p13.11(15,458,802-15,775,455)x1 mat	0.32	No	pCNC**
del 16p13.12 // 13q12.12	arr 16p13.12p13.11(14,669,793-16,634,816)x1 pat // 13q12.12(22,464,961-23,808,743)x1 mat	2 // 1.3	No	pCNC**$^{+}$ // uCNC
del 22q11.21	arr 22q11.21(19,411,059-19,835,557)x1 pat	0.42	Yes	uCNC

Duplications: De novo				
dup 1q21.1	arr 1q21.1(144,696,283-146,374,546)x3 dn	1.7	No	pCNC**$^{+}$
dup 9p24.1	arr 9p24.1(6,574,138-7,723,826)x3 dn	1.1	No	uCNC
dup 10q11.22q11.23	arr 10q11.22q11.23(49,033,015-51,005,023)x3 dn, 10q11.23(51,407,449-52,135,488)x3 dn	2.7	No	uCNC
dup 15q11.2q13.1	arr 15q11.2q13.1(20,249,885-26,829,558)x3 dn	6.6	No	pCNC^{+}
dup 15q13.1q13.2	arr 15q13.1q13.2(27,040,608-28,441,227)x3 dn	1.4	No	uCNC
dup 16p13.11	arr 16p13.11(14,956,257-16,535,551)x3 dn	1.6	No	uCNC^{+}
dup 19q13.42q13.43	arr 19q13.42q13.43(59,640,794-61,566,588)x3 dn	1.9	Yes	pCNC
dup 22q11.21	arr 22q11.21(17,256,415-19,902,202)x3 dn	2.6	No	pCNC**$^{+}$
Duplications: Inherited				
dup Xp11.1q11.1	arr Xp11.1q11.1(57,307,607-61,734,924)x2 mat	4.4	Yes	uCNC
dup Xp21.2	arr Xp21.2(30,511,110-30,781,353)x2 mat	0.27	No	uCNC
dup Xp22.11	arr Xp22.11(21,911,719-22,875,550)x3 mat	0.96	No	uCNC
dup Xp22.13p22.12	arr Xp22.13p22.12(18,498,513-19,414,777)x2 mat	0.92	Yes	uCNC
dup 1q21.1	arr 1q21.1(144,696,283-146,374,546)x3 mat	1.7	Yes	pCNC**$^{+}$
dup 1q21.1	arr 1q21.1(144,510,899-146,374,545)x3 mat	1.9	No	pCNC**$^{+}$
dup 2q13	arr 2q13(111,085,100-112,782,390)x3 pat	1.7	Yes	uCNC
dup 2q14.2	arr 2q14.2(120,958,376-121,309,793)x3 mat	0.35	No	uCNC
dup 5p13.2	arr 5p13.2(36,984,535-37,151,099)x3 mat	0.17	Yes	uCNC
dup 9q33.3	arr 9q33.3(128,414,271-128,481,044)x3 pat	0.07	No	uCNC
dup 16p13.11	arr 16p13.11(14,805,289-16,203,401)x3 pat	1.4	Yes	uCNC^{+}
dup 16p13.11	arr 16p13.11(14,817,705-16,102,080)x3 pat	1.3	No	uCNC^{+}
dup 16p13.11p12.3	arr 16p13.11p12.3(15,311,952-18,048,552)x3 pat	2.7	Yes	uCNC^{+}
dup 17q12	arr 17q12(32,041,983-33,135,410)x3 pat	1.1	Yes	pCNC**$^{+}$
dup 22q11.2 // 15q13.3	arr 22q11.2(17,041,523-19,891,669)x3 mat // 15q13.3(29,817,814-30,297,358)x3 mat	2.8 // 0.48	Yes	pCNC**$^{+}$ // uCNC
dup 22q11.21	arr 22q11.21(19,038,889-19,770,515)x3 mat	0.73	No	uCNC
dup 22q11.21	arr 22q11.21(19,248,549-19,770,514)x3 mat	0.52	No	uCNC

C. Uncertain CNVs: Referred to Advisory Committee but Not Reported to Patients

Deletions: inherited	Anomaly	ISCN	Size in Mb	Fetal anomaly detected by US?	Consensus Call 2012
	del 5p14.3p14.2	arr 5p14.3p14.2(22,394,512-24,139,699)x1 pat	1.7	No	lbCNC
	del 5p15.2	arr 5p15.2(8,877,417-9,455,871)x1 mat	0.58	No	uCNC
	del 6q26	arr 6q26(162,566,447-163,529,249)x1 mat	0.96	No	uCNC
	del 10q21.1	arr 10q21.1(57,186,532-58,432,936)x1 mat	1.2	No	uCNC
	del 12q24.33	arr 12q24.33(131,718,039-132,287,718)x1 mat	0.57	Yes	uCNC
	del 13q12.12	arr 13q12.12(22,464,961-23,808,744)x1 pat	1.3	No	uCNC
	del 15q11.2	arr 15q11.2(20,316,991-20,697,574)x1 mat	0.38	No	lpCNC[+]
	del 15q11.2	arr 15q11.2(20,316,991-20,610,515)x1 mat	0.29	No	lpCNC[+]
	del 17q12	arr 17q12(31,474,734-33,323,031)x1 pat	1.9	Yes	pCNC**[+]
Duplications: Inherited					
	dup Yq11.223	arr Yq11.223(22,650,981-23,984,067)x2 pat	1.3	Yes	lbCNC
	dup 1q21.1	arr 1q21.1(144,083,907-144,723,881)x3 mat	0.64	Yes	uCNC[+]
	dup 3p26.3	arr 3p26.3(1,054,628-2,745,827)x3 mat	1.7	No	uCNC
	dup 3q22.3	arr 3q22.3(139,923,391-140,181,774)x3 pat	0.26	No	lbCNC
	dup 5q21.1	arr 5q21.1(99,550,860-102,261,779)x3 pat	2.7	No	lbCNC
	dup 6q26	arr 6q26(162,461,141-162,970,533)x3 pat	0.51	No	lbCNC
	dup 8p22p21.3	arr 8p22p21.3(17,905,977-19,482,435)x3 mat	1.6	Yes	lbCNC
	dup 10q11.22q11.23	arr 10q11.22q11.23(46,482,304-51,558,563)x3 mat	5.1	No	uCNC
	dup 13q12.12	arr 13q12.12(22,464,761-23,808,883)x3 pat	1.3	No	uCNC
	dup 15q13.2q13.3	arr 15q13.2q13.3(28,742,017-30,297,218)x3 pat	1.6	No	uCNC[+]
	dup 15q13.3	arr 15q13.3(29,808,902-30,231,488)x3 pat	0.42	No	uCNC
	dup 15q13.3	arr 15q13.3(29,817,814-30,226,375)x3 mat	0.4	Yes	uCNC
	dup 15q13.3	arr 15q13.3(29,759,737-30,297,358)x3 mat	0.54	No	uCNC
	dup 15q13.3	arr 15q13.3(29,810,342-30,299,500)x3 mat	0.49	No	uCNC
	dup 15q13.3	arr 15q13.3(29,798,769-30,231,488)x3 pat	0.43	No	uCNC
	dup 15q13.3	arr 15q13.3(29,768,976-30,231,488)x3 mat	0.46	No	uCNC
	dup 15q13.3	arr 15q13.3(29,885,761-30,186,355)x3 mat	0.3	No	uCNC
	dup 15q13.3	arr 15q13.3(29,817,814-30,297,358)x3 pat	0.48	Yes	uCNC

dup 16p13.11	arr 16p13.11(14,817,705-16,102,080)x3 mat	1.28	No	uCNC+
dup 16p13.11p12.3	arr 16p13.11p12.3(16,445,726-18,192,575)x3 pat	1.8	No	uCNC
dup 16p13.13p13.12	arr 16p13.13p13.12(11,911,449-14,688,144)x3 pat	2.8	No	uCNC
dup 17q12	arr 17q12(29,022,237-29,947,078)x3 pat	0.92	No	lbCNC
dup 17q12	arr 17q12(29,022,237-29,947,078)x3 pat	0.92	No	lbCNC
dup 22q11.22 // 15q13.3	arr 22q11.22q11.23(21,341,812-23,289,967)x3 mat //15q13.3(29,817,814-30,297,358)x3 mat	2.0 // 0.48	No	uCNC // uCNC

D. Other CNVs: Considered Likely Benign in Initial Review and Not Reported to Patients

Deletions: De novo	**Anomaly**	**ISCN**	**Size in Mb**	**Fetal anomaly detected by US?**	**Consensus Call 2012**
	del 19q13.32	arr 19q13.32(52,174,243-52,869,532)x1 dn	0.7	Yes	uCNC
Deletions: Inherited					
	del Yp11.2	arr Yp11.2(6,652,867-8,894,823)x0 pat	2.2	Yes	lbCNC
	del 4q28.3	arr 4q28.3(134,022,764-136,497,342)x1 mat	2.5	No	lbCNC
	del 9p24.3	arr 9p24.3(194,192-244,712)x1 mat	0.05	No	lbCNC
	del 11p14.3p14.2	arr 11p14.3p14.2(22,901,598-26,185,570)x1 pat	3.3	No	lbCNC
	del 16p13.2	arr 16p13.2(7,153,100-7,438,061)x1 pat	0.29	No	lbCNC
	del 17p13.3	arr 17p13.3(513-827,455)x1 mat	0.83	Yes	lbCNC
Duplications: De novo					
	dup 16q23.2	arr 16q23.2(80,764,255-82,220,875)x3 dn	1.5	No	lbCNC
Duplications: Inherited					
	dup Xq21.1	arr Xq21.1(78,832,542-79,279,914)x3 pat	0.45	No	lbCNC
	dup 1p13p12	arr 1p13p12(118,236,263-120,321,801)x3 mat	2.1	No	lbCNC
	dup 1p21.1	arr 1p21.1(103,454,441-105,444,218)x3 pat	2	No	lbCNC
	dup 1p22.1	arr 1p22.1(93,950,420-95,349,885)x3 pat	1.4	No	lbCNC
	dup 1p36.33	arr 1p36.33(850,232-1,644,038)x3 pat	0.79	No	lbCNC
	dup 1q21.1	arr 1q21.1(144,083,908-144,643,814)x3 mat	0.56	Yes	uCNC+
	dup 1q43	arr 1q43(239,139,414-240,072,155)x3 pat	0.94	No	lbCNC
	dup 3p26.3	arr 3p26.3(832,458-1,403,386)x3 mat	0.57	Yes	lbCNC

dup 3p14.1	arr 3p14.1(65,467,941-67,266,258)x3 mat	1.8	No	lbCNC
dup 4p16.3	arr 4p16.3(2,269-494,412)x3 mat	0.49	No	lbCNC
dup 4q28.3	arr 4q28.3(131,752,417-132,819,095)x3 pat	1.1	No	lbCNC
dup 4q35.2	arr 4q35.2(188,115,159-189,660,971)x3 pat	1.5	No	lbCNC
dup 4q35.2	arr 4q35.2(188,142,875-189,145,869)x3 pat	1	No	lbCNC
dup 5q13.2	arr 5q13.2(69,274,232-70,622,914)x3 mat	1.3	No	lbCNC
dup 5q35.3	arr 5q35.3(177,355,106-178,539,600)x3 pat	1.2	No	lbCNC
dup 7p22.1	arr 7p22.1(5,688,165-6,837,608)x3 mat	1.2	No	lbCNC
dup 8p22	arr 8p22(13,400,616-14,702,452)x4 mat	1.3	No	lbCNC
dup 8p22	arr 8p22(13,400,615-14,711,717)x3 mat	1.3	Yes	lbCNC
dup 8p22	arr 8p22(13,446,106-14,659,611)x3 pat	1.2	No	lbCNC
dup 8p23.2	arr 8p23.2(3,674,351-5,938,253)x3 pat	2.3	Yes	lbCNC
dup 8q11.1	arr 8q11.1(46,966,687-47,976,420)x3 pat	1	Yes	lbCNC
dup 9q12-13	arr 9q12-13(68,073,655-70,195,939)x3 pat	2.1	No	lbCNC
dup 12p11.1	arr 12p11.1(33,436,415-34,647,416)x3 mat	1.2	No	lbCNC
dup 17p13.3	arr 17p13.3(513-579,655)x3 pat	0.58	No	lbCNC
dup 18p11.21p11.22	arr 18p11.21p11.22(10,654,393-11,848,195)x3 mat	1.2	No	lbCNC
dup 18q12.3	arr 18q12.3(39,417,443-40,695,766)x3 mat	1.3	No	lbCNC
dup 21q21.1	arr 21q21.1(19,106,129-20,127,326)x3 mat	1	No	lbCNC
dup 21q21.3	arr 21q21.3(27,186,402-28,309,393)x3 mat	1	Yes	lbCNC

The ISCN coordinates listed are based on the National Center for Biotechnology (NCBI) build 36/hg18. US denotes ultrasound.

Multiple findings are separated by "//"

*Unbalanced Translocation

**Significant Variable Expressivity

[+] Recurrent copy number variant associated with autism and neurocognitive alteration

pCNC = Pathogenic Copy Number Change

uCNC = Uncertain Clinical Significance

lbCNC = Likely benign Copy Number Change

TABLE S2: LISTING OF PRE-DETERMINED TARGETED REGIONS

Chromosomal Location	DEL/DUP#	Phenotype	Genomic Coordinates+ (NCBI36/hg18)
Xp22.33/Yp11.32		Leri-Weill dyschondrosteosis	chrX:505,078-540,146
Xp22.32		Autistic Features, X-linked, susceptibility	chrX:5,818,085-6,155,888
Xp22.31		Steroid Sulfatase deficiency	chrX:7,075,293-7,282,680
Xp22.31		Kallmann	chrX:8,456,915-8,660,227
Xp22.2		Microphthalmia with linear skin defects	chrX:11,039,373-11,051,122
Xp21.2		Congenital adrenal hypoplasia / Dosage sensitive sex reversal	chrX:30,232,507-30,237,413
Xp21.2		Glycerol kinase deficiency	chrX:30,581,461-30,658,645
Xp21.2		DMD	chrX:31,048,667-33,277,779
Xq22.1		Bruton agammaglobulinemia tyrosine kinase	chrX:100,491,098-100,532,426
Xq22.2		Pelizaeus-Merzbacher	chrX:102,918,095-102,934,203
Xq23		X-linked lissencephaly	chrX:110,423,663-110,541,962
Xq26.3		X-linked heterotaxy	chrX:136,476,012-136,481,923
Xq27.1		X-linked mental retardation with isolated growth hormone deficiency	chrX:139,411,973-139,415,436
Xq27.3		Fragile X	chrX:146,801,201-146,840,303
Xq28		Rett syndrome	chrX:152,940,458-153,016,323
Yp11.2		SRY - Testis determining factor on Y	chrY:2,714,896-2,715,792
Yq11.223-q11.23		AZF Regions	
1p36		Monosomy 1p36	
1q21.1*	DEL	Thrombocytopenia-absent radius (TAR) syndrome	chr1:144,097,430-144,463,097
1q21.1*	DUP	Variable phenotype	chr1:144,097,430-144,463,097
1q21.1*	DEL	ID, microcephaly, cardiac and cataracts	chr1:145,044,110-145,861,130
1q21.1*	DUP	ID, autism	chr1:145,044,110-145,861,130
2p34.3		Feingold syndrome	chr2:15,998,134-16,004,579
2p21		Holoprosencephaly 2	chr2:45,022,365-45,025,945
2q13		Joubert 4	
2q13		Nephronophthisis 1	chr2:110,180,175-110,500,589
2q22		Mowat-Wilson syndrome	chr2:144,862,056-144,994,386
2q31.1		Split-Hand/Foot Malformation 5 (SHFM 5)	
2q31.1		Synpolydactyly/Syndactyly II	
2q37.1 -q37.3		Holoprosencephaly 6	
2q37.3		Albright hereditary osteodystrophy-like syndrome/Brachydactyly-ID	

3q22.3		Blepharophimosis, ptosis epicanthus inversus (BPE)	chr3:140,144,730-140,149,880
3q24		Dandy-Walker malformation (DWM)	
3q28		Split-Hand/Foot Malformation 4 (SHFM 4)	
3q29	DEL	Intellectual disability, schizophrenia	chr3:197,240,451-198,829,062
3q29	DUP	Intellectual disability	chr3:197,240,451-198,829,062
4p16.3		Wolf-Hirschhorn	
4q25		Rieger 1 (RIEG1)	chr4:111,758,029-111,782,566
5p15.2		Cri-du-Chat	
5p13.2		Cornelia de Lange	chr5:36,912,742-37,101,678
5q22.2		Familial adenomatous polyposis (FAP)/Gardner/ID	chr5:112,071,117-112,209,835
5q35.3		Sotos	chr5:175,661,584-176,946,567
6p21.1		Cleidocranial dysplasia (CCD)	chr6:45,404,032-45,626,796
6q16.3		Prader-Willi -like phenotype	
7p21.1		Saethre-Chotzen	chr7:19,121,616-19,123,550
7p14.1		Greig cephalopolysyndactyly	chr7:41,967,073-42,243,900
7q11.23	DEL	Williams-Beuren	chr7:72,382,390-73,780,449
7q11.23*	DUP	Autism; reciprocal dup. to Williams synd.	chr7:72,382,390-73,780,449
7q21.3		Split-Hand/Foot Malformation 1 (SHFM1)	chr7:95,371,796-96,617,422
7q36.3		Holoprosencephaly 3	chr7:155,288,319-155,297,728
8p22-23.1	DEL	8p22 deletion syndrome	chr8:8,156,705-11,803,128
8p22-23.1	DUP	8p22 duplication syndrome	chr8:8,156,705-11,803,128
8q12.2		CHARGE	chr8:61,753,893-61,942,017
8q13.3		Branchio-Oto-Renal (BOR)/Melnick-Fraser/ Oto-Facio-Cervical (OFC)	chr8:72,272,222-72,437,021
8q24.11		Langer-Giedion / Trichorhinophalangeal	
9q22.32		Basal cell nevus/Gorlin-Goltz/ Holoprosencephaly 7	chr9:97,245,085-97,319,068
9q33.3		Nail Patella syndrome	chr9:128,416,569-128,503,132
10p14		DiGeorge 2	
10p14		Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	chr10:8,136,673-8,157,170
10p12.31		Nebulette	chr10:21,110,094-21,226,537
10q24.32		Split-Hand/Foot Malformation 3 (SHFM3)	
11p15.5		Beckwith-Wiedemann	
11p13		Aniridia / Wilms Tumor (WAGR)	chr11:31,760,085-32,467,564
11p11.2		Potocki-Shaffer	chr11:43,941,853-46,021,136
11q14.2-q14.3		Leukodystrophy	
11q25		Jacobsen/11q terminal deletion disorder	
12p		Pallister-Killian	

12q24.13		Noonan 1	chr12:111,340,919-111,432,099
13q14.2		Retinoblastoma/ID	chr13:47,775,912-47,954,023
13q32.3		Holoprosencephaly 5	chr13:99,432,320-99,437,019
15q11.2	DEL	Angelman	chr15:20,428,073-26,230,781
15q11.2	DEL	Prader-Willi	chr15:20,428,073-26,230,781
15q11.2	DUP	Autism	chr15:20,428,073-26,230,781
15q13.2-q13.3 BP4-BP5*	DEL	ID and epilepsy	chr15:28,924,396-30,232,700
15q13.2-q13.3 BP4-BP5*	DUP	Psychiatric disease	chr15:28,924,396-30,232,700
15q26.2		Congenital diaphragmatic hernia (CDH)	
16p13.3		Tuberous Sclerosis 2	chr16:2,038,600-2,078,713
16p13.3		Polycystic kidney disease 1	chr16:2,078,712-2,125,900
16p13.3		Rubinstein-Taybi	chr16:3,715,057-3,870,122
16p13.11*	DEL	Autism, ID and schizophrenia (1.5 & 3 Mb)	chr16:15,411,955-16,199,769
16p13.11*	DUP	Variable phenotype	chr16:15,411,955-16,199,769
16p12*	DEL	Intellectual disability	Chr16:21,744,792-22,315,572
16p11.2*	DEL	Autism	chr16:29,557,497-30,107,356
16p11.2*	DUP	Autism, schizophrenia	chr16:29,557,497-30,107,356
17p13.3		Miller-Dieker / Isolated Lissencephaly	chr17:1,129,706-2,535,659
17p12	DEL	Hereditary Neuropathy with liability to Pressure Palsies (HNPP)	chr17:14,038,639-15,363,680
17p12	DUP	Charcot Marie Tooth Type 1A	chr17:14,038,639-15,363,680
17p11.2	DEL	Smith-Magenis	chr17:16,697,836-20,160,243
17p11.2*	DUP	Potocki-Lupski syndrome	chr17:16,697,836-20,160,243
17q11.2		Neurofibromatosis 1/ID	chr17:26,186,948-27,242,780
17q12*	DEL	Renal cysts, diabetes, autism and schizophrenia	chr17:31,930,169-33,323,031
17q12*	DUP	Intellectual disability	chr17:31,930,169-33,323,031
17q21.31*	DEL	17q21 deletion syndrome	chr17:41,060,948-41,650,183
17q24.3		Campomelic dysplasia (CMPD)	chr17:67,628,756-67,634,156
18p11.31		Holoprosencephaly 4	chr18:3,402,072-3,448,406
20p12.2		Alagille	chr20:10,566,334-10,602,590
21q22.3		Holoprosencephaly 1	
22q11.1		Cat-eye (Type 1)	chr22:15,772,953-16,971,860
22q11.21	DEL	DiGeorge/Velocardiofacial (VCF)	chr22:17,041,725-19,891,514
22q11.21	DUP	Variable phenotype	chr22:17,041,725-19,891,514
22q12.2		Neurofibromatosis 2	chr22:28,329,565-28,424,583
22q13.3		22q13.3 Microdeletion	chr22:49,392,382-49,525,811

[#] Deletion or duplication noted for recurrent regions to indicate the associated phenotype
[+] Genomic coordinates correspond to either a disease gene or region
*Added Jan 2011

Table S3: Listing of pre-determined common benign copy number changes

Chromosome	Start	Stop	Size*	Location**	Loss/Gain/Both	Genome Build
1	246,695,754	246,932,000	236 kb	telomere	gain	hg18
1	146,814,404	147,989,074	1.17 Mb		both	hg18
2	110,140,524	110,338,194	198 kb	2q13, *NPHP1*	gain	hg18
2	242,579,274	242,717,069	138 kb	telomere	both	hg18
2	88,914,227	89,744,041	829 kb		both	hg18
3	163,990,950	164,108,677	117 kb		both	hg18
4	68,942,656	69,222,608	279 kb		both	hg18
4	70,139,255	70,274,566	135 kb		loss	hg18
5	180,272,529	180,503,659	231 kb	telomere	both	hg18
6	224,313	284,802	60 kb	telomere	both	hg18
8	7,040,595	8,117,330	1.07 Mb	backbone	both	hg18
8	39,344,885	39,516,172	171 kb		loss	hg18
9	67,489,570	69,256,301	1.77 Mb	centromere	gain	hg18
10	46,404,918	47,735,531	1.3 Mb	10q11.22	both	hg18
10	135,053,553	135,254,513	201 kb	telomere	both	hg18
11	133,985,161	134,221,598	236 kb	telomere	gain	hg18
14	18,343,613	19,493,200	148 kb - 1.2 Mb	centromere	both	hg18
14	104,974,236	106,288,923	100-885 kb	telomere	both	hg18
15	18,276,329	20,232,428	1.6 Mb	centromere	both	hg18
15	20,232,428	20,812,097	580 kb	*NIPA2, CYFIP1*	Both***	hg18
16	31,988,362	34,823,346	152 kb-1.2 Mb	centromere	both	hg18
16	87,282,034	87,419,068	137 kb		gain	hg18
17	41,518,904	42,107,467	588 kb		both	hg18
22	20,602,619	21,110,969	508 kb		loss	hg18
X	263,991	837,776	574 kb	Xp22.33, *SHOX*	gain	hg18
X	6,461,675	8,075,124	1.61 Mb	Xp22.31, *STS*	gain	hg18
X	8,561,293	8,665,634	104 kb	Xp22.31, *KAL1*	gain	hg18
Y	25,185,891	26,522,784	1.3 Mb	Yq11.22.q11.23, *DAZ2, DAZ3, DAZ4*	gain	hg18

*May be smaller benign copy number changes within these larger regions
**Not inclusive of all genes in the region
***This was considered benign at the beginning of the study. Recent data suggests that this may not be a benign finding.[1]

[1] Cooper GM, Coe BP, Girirajan S, et al. A copy number variation morbidity map of developmental delay. Nat Genet 2011;43:838-46.

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