

Table S1 Samples with CNVs larger than 5 Mb in size

Sample	Sex	Tissue	Chr	Size (bp)	CNV	Karyotype	Other arrays
119975L	M	L	9p22.1-p21.3	4,998,116	loss	n/a	detected by Affy500K, Marshall <i>et al.</i> 2008
139364L	M	L	21q21.2-q21.1	6,260,886	gain	46,XY,dup(21)(q?q?) or der(21)ins(21;21)(q?q?)	detected by Affy6.0
109332	F	B	18q21.1-q23	31,604,736	gain	46,XX,der(11)t(11;18)(q25;q21.1)	detected by Affy6.0
146451L	F	L	7q21.11 - q36.3	80,056,407	complex (loss/gain)	n/a	Possibly a cell-line artifact, not detected by Affy6.0 (blood DNA was used)
97412	M	B	1q42.3 - q44	13,708,317	gain	n/a	detected by Illumina 1M-single array, Pinto <i>et al.</i> 2010
89853L	M	L	21	33,487,618	gain	47, XY + 21 (Down Syndrome)	detected by Illumina 1M-single array, Pinto <i>et al.</i> 2010; confirmed by karyotyping
50800L	M	L	7q31.1-q31.31	11,033,516	loss	XY, del(7)(q31)	detected by Affy500K, Marshall <i>et al.</i> 2008
85181L	M	L	7q22.2 - q35	38,410,895	gain	46,XY,dup(7)(q22q34)	detected by Affy6.0
60974L	F	L	5p15.33 - p15.2	13,783,361	loss	46,XX,del(5)(p15.1)	detected by Affy500K, Marshall <i>et al.</i> 2008
72871L	M	L	3p14.1	5,375,845	loss	t(6;14)(q13;q21)	detected by Affy500K, Marshall <i>et al.</i> 2008
165457L	M	L	21	33,580,687	gain	n/a	detected by Affy6.0
165445L	F	L	21	33,580,887	gain	n/a	was not run on any other array, but proband has Down syndrome
60433-L	F	L	7q31.1 - q32.1	15,437,215	loss	XX, del(7)(q31.2q31.3)	detected by Affy500K, Marshall <i>et al.</i> 2008
56034	M	B	21	32,875,937	gain	46,XY,+21 (trisomy 21)	detected by Affy500K, Marshall <i>et al.</i> 2008 was not run on any other array,

59172L	M	L	2p13.3-p25.3	70,640,252	gain	n/a	but proband has Down syndrome Possibly a cell-line artifact, not detected by Illumina 1M array, DNA source-cell line for Agilent and blood for Illumina 1M array detected by Affy500K, Marshall <i>et</i> <i>al.</i> 2008
60340	F	B	18q21.32 - q23	20,357,135	loss	46, XX, del (18)(q21)	detected by Affy500K, Marshall <i>et</i> <i>al.</i> 2008
115733L	M	L	15q11.2 - q13.3	11,634,435	gain	46,XY, trp(15)(q11.2q13)	detected by Affy6.0 (same DNA source as Agilent)
82361L	F	L	2p25.3 - p15; Xp22.33 - p22.31	63,366,686; 6,017,794	complex (loss/gain); loss	46,XX,t(11;12)(q23.3;p13.3)	detected by Affy500K, Marshall <i>et</i> <i>al.</i> 2008
57283L	F	L	15q11.1 - q13.3	11,887,780	gain	isodisomy Chr.15 46,XX (17 cells), 46,XX,+ring; Ring chromosome	Marshall <i>et al.</i> 2008
100569L	F	L	1q21.1-1q21.3	8,315,572	gain	1	Marshall <i>et al.</i> 2008

Abbreviations: B-blood; L-cell line