

Supplementary Table S1. Information of CMA/MLPA/qPCR for CNV validation.

Sample	Status	Location by NGS (hg19)	Size by NGS (kb)	Validation Method	Kit
S0058	DUP	X:53220502-54071682	851.18	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S0666	DEL	X:2852698-5950791	3098.093	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S0683	DEL	16:29802039-30200397	398.358	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S0703	DUP	X:95939661-111003873	15064.212	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S0928	DEL	X:148558520-148568844	10.324	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S1003	DEL	X:28807436-38546928	9739.492	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S1081	DUP	X:48542167-50659607	2117.44	CMA	Agilent SureTag DNA Labeling kit (4X180K)
S0201	DUP	7:73442118-74191693	749.575	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0434	DEL	7:73442118-74203657	761.539	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0746	DEL	17:17115525-18083116	967.591	MLPA	SALSA MLPA probemix P064-C1 Mental Retardation-1
S0797	DEL	7:73442118-74203657	761.539	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0805	DEL	7:73442118-74203657	761.539	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0818	DEL	7:73442118-74148323	706.205	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0827	DEL	7:73442118-74148323	706.205	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0894	DEL	16:29802039-30200397	398.358	MLPA	SALSA MLPA probemix P343-C3 Autism-1
S0897	DEL	7:73442118-74133260	691.142	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0955	DEL	7:73442118-74173185	731.067	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S0974	DUP	X:153194694-153588955	394.261	MLPA	SALSA MLPA probemix P015-F2 MECP2
S1033	DUP	7:73442118-74203657	761.539	MLPA	SALSA MLPA probemix P029-C1 Williams-Beuren Syndrome
S1055	DUP	X:152801579-153363212	561.633	MLPA	SALSA MLPA probemix P015-F2 MECP2

Sample	Status	Location by NGS (hg19)	Size by NGS (kb)	Validation method	Primer1-F	Primer1-R	Primer2-F	Primer2-R	Primer3-F	Primer3-R
S0027	DEL	19:17986760-17994855	8.095	qPCR	SLC5A5_p1-F: TCTCCACGT GGCTAACTT GC	SLC5A5_p1-R: CTTGCTCCC CGAAACAC TC	SLC5A5_p2-F: AGATGGTG TGGACGGT CTCT	SLC5A5_p2-R: ACCCAGGT CTCCCTGA AACT	SLC5A5_p3-F: AGGAGTC CTTGAGA CCCACA	SLC5A5_p3-R: GCAGGA ACATTCC CAAGATG
S0183	DEL	9:139981378-139982635	1.257	qPCR	MAN1B1-qEx1-P1-F: CCTCTCAGT CGGACTTC CTG	MAN1B1-qEx1-P1-R: CCGAGATG AAGTCCCG ATGA	MAN1B1-qEx2-P1-F: GTGACAGT TGAGACGT TCCG	MAN1B1-qEx2-P1-R: CTGCCAAT CAACCCTC AAC	MAN1B1-qEx3-P-F: AGAGCAG AAGATGA GGCCAG	MAN1B1-qEx3-P-R: GGTAAGT TCTCAGG GTCGGT
S0212	DEL	19:1401295-1401396	0.101	qPCR	GAMT-qEx1-P1-F: GGTGATGG AGCGCTGG GA	GAMT-qEx1-P1-R: TCCCAGGC GAGGAGAC AG	GAMT-qEx2-P-F: AGCGTCAA AGGTGCAG GA	GAMT-qEx2-P-R: ACCTTGTTGT GTCTGCCGT CCTCT	GAMT-qEx3-P-F: ACCTCTCT ACCCTGT CCTCT	GAMT-qEx3-P-R: CCTCACC CCTCACC ATCAA
S0593	DEL	16:74760122-74808729	48.607	qPCR	FA2H-qEx1-P1-F: CTCCCGGG ATGTTAGAG GC	FA2H-qEx1-P1-R: CACGAAGC TGGAGAGG TCG	FA2H-qEx3-P1-F: TGGCTTCAT CTCTGCAG GA	FA2H-qEx3-P1-R: ATTGGCACC GTCAGAAG AGA	FA2H-qEx4-P-F: TACTACCG AACCTTT GCCCA	FA2H-qEx4-P-R: AGTTTGG GGTTCTG TGCTCT
S0607	DEL	X:30709239-30709316	0.077	qPCR	GK-qEx5-p1-F: TGGTCTATA GTGTGGCTT	GK-qEx5-p1-R: TGGCCTCA ATTATCCGG	GK-qEx6-p-F: GTGAAACT TCGTTGGCT CCT	GK-qEx6-p-R: AACAAATAG GCAAGGCA GTCT	GK-qEx4-p-F: TGAGAGC TGTTTTCC	GK-qEx4-p-R: TGCAATG TGAAGA

S0664	DEL	6:129419317-129419560	0.243	qPCR	GATCT	GAA			TGAAGT	AAGGCC
					LAMA2-qEX4-P1-F:	LAMA2-qEX4-P1-R:	LAMA2-qEx3-P-F:	LAMA2-qEx3-P-R:	LAMA2-qEx5-P-F:	LAMA2-qEx5-P-R:
					TGTGAAGG	TGACAGGC	CTTGGTGG	TCCTTTCAC	GCCGATG	TGGGGTC
					CAGCTAAC	CTATTTAC	CAGAGTCC	CCACAAAC	ATCCTTCT	AATTTCT
S0737	DEL	19:12917393-13710781	793.388	qPCR	TCCC	CGT	CA	CAC	CCAGA	CTTGGGT
					RNASEH2A-qEx1-P1-F:	RNASEH2A-qEx1-P1-R:	RNASEH2A-qEx8-P1-F:	RNASEH2A-qEx8-P1-R:	CACNA1A-qEX46-P1-F:	CACNA1A-qEX46-P1-R:
					TAGTTCTTG	TTTCCCACA	CGATATTTT	CATCCCCTA	CTGAGCA	GTTCCCTG
					CAGCTGGT	TCCTCCGTA	CTGGAACG	CGTGTGGTT	TGACCAC	GGCATAG
S0756	DEL	17:1303340-1680868	377.528	qPCR	GGT	C	CGG	CT	CCAATC	CGGT
					MYO1C-qEX6-P1-F:	MYO1C-qEX6-P1-R:	SLC43A2-qEX8-P1-F:	SLC43A2-qEX8-P1-R:	WDR81-qEX8-p1-F:	WDR81-qEX8-p1-R:
					GGCCACAT	AGCCACTC	CCCCAGGG	GTCCTTGG	GGATGCC	TAGTTGT
					CCTCAGTTA	ACCTTCACC	TGAAGATC	CACTCCTCA	CACTTTCA	ACAGCG
S0774	DEL	X:32429869-32430030	0.161	qPCR	CCT	AG	AAGT	T	CTCC	GCCAGAG
					DMD-qEx30-P1-F:	DMD-qEx30-P1-R:	DMD-qEx29-P-F:	DMD-qEx29-P-R:	DMD-qEx31-P-F:	DMD-qEx31-P-R:
					GCTGTAAAG	TTCCCAGAT	ACAGACCC	TCTGAGAG	CTGCCCA	GTCCTCA
					GAGGCAAA	GTACTTGCC	TAACAGAT	GCCTGTATC	AAGAGTC	AATCCAA
S0928	DEL	X:148564277-	4.567	qPCR	AGTTG	TG	GGCG	TGC	CTGTCT	TCTTGCC
					IDS-qEx9-P1-	IDS-qEx9-P1-	IDS-qEx10-	IDS-qEx10-	IDS-qEx8-P-	IDS-qEx8-

		148568844			F:	R:	P1-F:	P1-R:	F:	P-R:
					GGTGGGCT	GAGGCGGA	CATTCTGCT	ACTGAGGG	GCCTCTTG	CCCCAGG
					CTAGGTGA	ATCAAAAG	CTGTCGCTT	ATGTCTGAA	AGTGCTTT	ATCCCAC
					ACAT	GGTC	CC	GGC	GGAC	TTTGTT
					PMM2-	PMM2-	PMM2-	PMM2-	PMM2-	PMM2-
					qEx2p1-F:	qEx2p1-R:	qEx7p1-F:	qEx7p1-R:	qEx8p1-F:	qEx8p1-R:
S1032	DEL	16:8829555-8906963	77.408	qPCR	TGTGTGGCT	CTCAAAGT	CACCTTTTG	TCTCTTGTC	TCTTCACA	GCCACCG
					TATGACTGT	CCGATCCGC	CCTTTGTGT	CCATCCATC	GACCCCA	AATGCCC
					TGT	CTA	GC	AGG	GAACC	TATG
					KIF22-qEx2-	KIF22-qEx2-	KIF22-qEx13-	KIF22-qEx13-	PRRT2-	PRRT2-
S1053	DUP	16:29802039-30081741	279.702	qPCR	P1-F:	P1-R:	P1-F:	P1-R:	qEx2-P1-F:	qEx2-P1-R:
					ATTGGAGCT	GGAGTGGC	ACTCATCTC	ACGATTAGC	TCCCCTCT	GGGTCTC
					ACTCGTCGT	CTGAACCTT	CCTTTGCCT	TGGGCCTTC	CCCATCTC	TGTGGTT
					CC	ACT	CC	TT	AAGA	TCTGGA
					KIF22-qEx2-	KIF22-qEx2-	KIF22-qEx13-	KIF22-qEx13-	PRRT2-	PRRT2-
S1058	DUP	16:29802039-30081741	279.702	qPCR	P1-F:	P1-R:	P1-F:	P1-R:	qEx2-P1-F:	qEx2-P1-R:
					ATTGGAGCT	GGAGTGGC	ACTCATCTC	ACGATTAGC	TCCCCTCT	GGGTCTC
					ACTCGTCGT	CTGAACCTT	CCTTTGCCT	TGGGCCTTC	CCCATCTC	TGTGGTT
					CC	ACT	CC	TT	AAGA	TCTGGA