Id	Karyotype (ISCN 2016)	nb breakpoints (karyotype)	Mean sequencing depth	WGS detection	Revised karyotype (ISCN 2016)	nb breakpoints (WGS)	Chromosomal position	Sequencing depth at breakpoint	Breakpoints (GRCh37)	Loss (bp)	Gain (bp)	Gene disruption	Repeated elements (repeat masker)	TAD disruption	ABCR interpretation	Causative gene
IS/0101	46,XX,t(9;17)(p13;q21)	2	10.79X	1st analysis	t(9;17)(p13.3;q21.31)	4	9p13.3	10.00X	chr9:35773305		5208	0 -	SINE (AluJb)	+	Pathogenic	*KANSL1* Koolen-de Vries syndrome (MIM 610443)
							9p13.3	8.00X	chr9:35779222_35779227		5	0 -	LTR (LTR78B ERV1)	+		
							17q21.31	5.10X	chr17:44134030_44134039		4	3 *KANSL1* (intron 7/15)	SINE (MIRb)	+		
							17q21.31	8.00X	chr17:44139543		3479	0 *KANSL1* (intron 7/15)	SINE (AluSx1)	+		
EB/0501	46,X,t(X;1)(p22.1;q12)	2	9.70X	Failure	NS	NS	NS	NS	NS		NS	NS NS	NS	NS	NS	NS
IL/1901	46,XX,der(3)inv(3)(p13p22)inv(3)(p12q26.3)	4	11.36X	1st analysis	der(3)inv(3)(p13p22.1)inv(3)(p12.3q26.1)	13	3p22.1	8.31X	chr3:39909637_39909662		24	0 *MYRIP* (intron 1/16)	-	+	Pathogenic	*FOXP1* Mental retardation with language impairment (MIM 613670)
							3p14.1	6.03X	chr3:65925648_65929005		3356	0 *MAGI1* (intron 1/24)	SINE (AluSx3)	+		
							3p14.1	6.33X	chr3:68270000_68270008		7	0 *FAM19A1* (intron 2/4)	-	+		
							3p13	5.23X	chr3:70044598_70044658		59	0 -	SINE (AluSp)	-		
							3p13	5.70X	chr3:70051680-70051712		31	0 *BC015590* (intron 1/3)	-	-		
							3p13	1.51X	chr3:70054609_70054684		74	0 *BC015590* (intron 1/3)	LTR (MLT1H2 ERVL-MaLR)	-		
							3p13	5.63X	chr3:71156417_71156446		28	0 *FOXP1* (intron 7/20)	DNA (Charlie4z hATcharlie)	+		
							3p13	8.20X	chr3:71884474_71884465		0	11 -	-	+		
							3p12.3	8.80X	chr3:74225415_74225419		3	0 -	DNA (Tigger1 TcMar-Tigger)	+		
							3p12.3	5.51X	chr3:75494494_75498225		3730	0 -	Segmental duplication	+		
							3p12.3	3.29X	chr3:75498415_75498424		8	0 -	SINE (AluYa8)	+		
							3p26.1	8.00X	chr3:163559725_163559729		3	-	LTR (THE1D-int ERVL-MaLR)	+		
							3p26.1	7.00X	chr3:163583574_163583575		0	0 -	-	+		
LV/1701	46,XX,t(3;9)(q26.33;p22)	2	10.78X	1st analysis	t(3;9)(q26.33;p21.3)	2	3p26.33	6.13X	chr3:179816182_179818874		2691	0 -	-	+	VUS	-
							9p21.3	8.00X	chr9:22010919_22010942		21	0 *CDKN2B-AS1* (intron 1/18)	-	+		
JB/0102	46,XY,t(6;8;9;13)(q26;p23;p21;q21)	4	10.01X	1st analysis	t(6;8;9;13)(q27;p23.2;p21.3;q31.1)	8	6q27	3.41X	chr6:166365424_166365823		399	0 *AK090688* (intron 1/1)	-	+	VUS	-
							6q27	0.00X	chr6:168444979_168444979		0	1 *KIF25* (intron 8/8)	-	-		
							8p23.2	6.00X	chr8:2388818_2388819		0	0 *LOC101927815* (intron 5/5)	-	+		
							9p21.3	10.5X	chr9:23875494_23875495		0	0 -	LINE (L1MB2)	+		
							9p21.3	8.50X	chr9:25276660_25276661		0	0 -	LINE (L1M1)	-		
							11q22.3	14.33X	chr11:103440006_103440008		1	0 -	-	+		
							13q31.1	7.29X	chr13:80742954_80742970		15	0 -	-	+		
							13q31.1	11.00X	chr13:82220728_82220727		0	2 -	-	+		
CM/0103	46,XY,t(3;22)(q13-21;p11)	2	9.82X	2nd analysis	t(3;22)(q13.33;p11.2)	2	3q13.33	5.42X	chr3:120947492_120947490 (GRCh38)		17	0 *STXBP5L* (intron 2/27)	LINE (L1PB2)	+	VUS	-
							22p11.2	84.75X	chr22:12041367_12041370 (GRCh38)		2	0 -	Segmental duplication	NS		
MG/1001	46,XX,t(14;20)(p13;p11)	2	10.96X	Failure	NS	NS	NS	NS	NS		NS	NS NS	NS	NS	NS	NS
PS/0502	46,XY,t(20;21)(q11.2;q21)	2	9.37X	Failure	NS	NS	NS	NS	NS		NS	NS NS	NS	NS	NS	NS
VJ/0601	46,XY,t(4;14)(p15;q32.1)	2	7.85X	1st analysis	t(4;14)(p15.2;q32.2)	2	4p15.2	6.00X	chr4:24332309_24332315	5	0	*PPARGC1A* (intron 1/14)	SINE (MIR)	+	Likely pathogenic	*BCL11B* Intellectual developmentam disorder and T-cell abnormalities disorder (MIM 618092)
							14q32.2	7.00X	chr14: 98758657_98758653	0	5	-	-	+		
CS/1002	46,XX,ins (15;12) (q15 or q21;q24.1q24.2)	3	10.95X	1st analysis	t(12;15)(q23.2q24.21;q14q14)	6	12q23.2	4.92X	chr 12: 101739873_101739885		11	0 *UTP20* (intron 37/61)	SINE (AluSp)	-	Pathogenic	*SPRED1* Legius syndrome (MIM 611431)
							12q24.21	4.29X	chr 12: 115799079_115799085		7	0 -	-	+		
							15q14	11.00X	chr 15: 35043064_35043063		0	2 -	-	+		
							15q14	12.50X	chr15 : 36618916_36618919		2	0 -	-	+		
							15q14	8.80X	chr15 : 38605305_38605309		3	0 *SPRED1* (intron 2)	LINE (L1MC4)	+		
							15q14	6.67X	chr15:39186047_39186049		1	0 -	-	+		
CL/2001	46,XX,t(11;15)(p14;q14)	2	10.58X	1st analysis	t(11;15)(p15.5;q14)	2	11p15.1	15.00X	chr11: 20219439_20219438		0	2 -	-	+	Pathogenic	*MEIS2* Cleft palate, cardiac defect and
							15q14	12.00X	chr15: 37377784_37377785		0	0 *MEIS2* (intron 6/12)	-	+		
MD/0104	46,X,t(X;7)(p11.1;q36.1)	2	5.53X	1st analysis	t(X;7)(p11.1;q36.1)	2	7q36.1	6.00X	chr7:149451597_149451594		0	4 -	SINE (AluJr4)	+	VUS	-
							Xp11.1	0.00X	chrX:58568911_58568915		3	0 -	Satellite (ALR/alpha)	+		
LF/2002	46,XX,t(2;8)(p24;q12)	2	10.91X	1st analysis	t(2;8)(p24.1;q11.23)	2	2p24.1	2.00X	chr2:21497656_21497667		10	0 -	-	-	VUS	-
							8q11.23	5.57X	chr8:55047229_55052711		4981	0 *MRPL15* (exons 1-3/5)	-	+		
SL/0105	46,XX,inv(2)(p12q22)	2	12.84X	1st analysis	inv(2)(p13.1q23.1)	2	2p13.1	5.80X	chr2:74789593_74789726		132	0 *M1AP* (intron 7/10)	-	+	Pathogenic	*MBD5* Mental retardation, autosomal dominant 1 (MIM 156200)
							2q23.1	6.79X	chr2:148888435_148888477		41	0 *MBD5* (intron 2/14)	DNA (Tigger15a TcMar-Tigger), Other (Helitron3Na_Mam)	+		
EB/0401	46,XY,ins(5)(q15q23.3q34)	3	10.76X	1st analysis	ins(5)(q14.2q23.2q34)	23	5q14.2	6.86X	chr 5: 82639865_82639871		5	0 *XRCC4 (intron 7/7)*	-	+	Likely pathogenic	*MEF2C* Mental retardation, autosomal dominant 20 (MIM 613443)
							5q14.2	7.63X	chr5:82727718_82727779		60	0 -	-	+		
							5q14.3	6.33X	chr5: 88428853_88428855		1	0 -	LINE (L2b)	+		
							5q14.3	8.00X	chr 5:90444779_90444779		0	1 *GPR98* (intron 87/89)	LTR (THE1D-int)	+		
							5q23.1	3.63X	chr5:119887503_119887510		6	0 *PRR16* (intron 2/2)	-	-		
							5q23.1	8.00X	chr 5:121056498_121056498		0	1 -	LINE (L1PA8)	+		
							5q23.1	12.67X	chr5:121094016_121094014		0	3 -	LTR (MLT1J ERVL-MaLR)	+		
							5q23.1	11.00X	chr5:121175464_121175466		1	0 -	LTR (LTR87)	+		
							5q23.2	12.00X	chr 5: 122429965_122429970		4	0 *PRDM6* (intron 2/7)	-	+		
							5q23.2	14.50X	chr 5:122565263_122565264		0	0 -	LTR (MLT1N2)	+		
							5q23.2	6.09X	chr5:123494994_123495004		9	0 -	-	+		
							5q23.2	7.80X	chr 5:123913719_123913723		3	0 -	SINE (AluSz)	+		
							5q23.3	8.38X	chr5:128098589_128098596		6	0 -	-	+		
							5q32	9.53X	chr 5:144980454_144980468		13	0 -	SINE (AluJr4)	-		
							5q32	11.25X	chr 5:145115091_145115094		2	0 -	-	+		
							5q32	10.75X	chr5:145663114_145663117		2	0 *RBM27* (intron 19/20)	-	+		
							5q34	11.50X	chr5:164462059_164462058		0	2 -	-	+		
							5q34	9.33X	chr5:165945117_165945122		4	0 -	LTR (LTR1D ERVL1)	+		
							5q34	11.50X	chr5:166779229_166779230		0	0 *TENM2* (intron 1/28)	DNA (MER102c hAT-Charlie)	+		
							5q34	19.83X	chr5:167429301_167429296		0	6 *TENM2* (intron 5/28)	-	+		
							5q34	13.00X	chr5:167436338_167436338		0	1 *TENM2* (intron 5/28)	-	+		
							5q35.2	8.00X	chr5:176428596_176428600		3	0 *UIMC1* (intron 1/14)	LINE (L1MD)	-		
							5q35.2	4.00X	chr5: 176446837_176446831		0	7 -	SINE (AluSx1)	-		
TS/1101	46,XY,t(3;4;10;14;18)(p23;q24;p13;q32;q23)	5	10.25X	1st analysis	t(3;4;10;14;18)(p23;q22.1;p14;q31.3;22.3)	18	3p26.3	5.25X	chr3:2693744_2693747		2	0 *CNTN4* (intron 4/24)	-	+	VUS	-
							3p26.1	12.00X	chr3:4235296_4235295		0	2 *SUMF1* (intron 8/12)	-	+		
							3p23	10.00X	chr3: 31560763_31560762		0	2 -	-	+		
							3q24	7.45X	chr3:146852555_146852592		36	0 -	LTR (THE1B-int, fa ERVL-MaLR)	-		
							4q22.1	7.00X	chr4:91933971_91933972		0	0 *CCER1* (intron9/10)	LINE (L1M2a)	+		
							10p14	10.00X	chr10:9559538_9559539		0	0 -	LTR (LTR41B, fam ERVL)	+		
							10p14	9.68X	chr10:10315687_10315964		6	0 -	LINE (L1MC4)	+		
							10p14	11.00X	chr10:10334273_10334272		0	2 -	-	+		
							10p14	8.17X	chr10:10355464_10355498		33	0 -	-	+		
							10p14	5.00X	chr10:10607349_10607348		0	2 *CELF2* (intron 1/17) RefSeq	-	+		
							10p14	8.50X	chr10:11359832_11359833		0	0 *CELF2* (intron 10/14) UCSC	-	+		
							10p13	10.00X	chr10:12437410_12437412		1	0 *CAMK1D* (intron 1/10)	LTR (MER21B ERVL)	+		
							10p13	9.33X	chr10:12474190_12474192		1	0 *CAMK1D* (intron 1/10)	-	+		
							10p13	4.08X	chr10:12981895_12981920		24	0 *CCDC3* (intron 2/2)	-	+		
							10p13	3.20X	chr10:12993998-12994051		52	0 *CCDC3* (intron 2/2)	-	+		
							14q31.3	7.67X	chr14:86864001_86863996		0	6 -	SINE (MIR)	+		
							18q22.3	11.80X	chr18:71840451_71840447		0	5 -	-	+		
							18q23	6.42X	chr18:75078221_75078232		10	0 -	-	+		

CG/0106	46,X,t(X;13;15)(p21;q22;q22),t(6;11)(q21;q24)	5	13.63X	1st analysis	t(X;13;15)(p21.2;q21.33;q22.2),t(6;11)(q21;q23.2)	5	6q21 11q23.2 Xp21.2 13q21.33 15q22.2	9.38X 4.83X 11.71X 9.00X 3.50X	chr6:109890177_109890184 chr11:113314551_113314556 chrX: 31363619_31363625 chr13:72481968_72481969 chr15:60191405_60191406	6 4 5 0 0	0 *AK9* (intron 21/40) 0 *DRD2* (intron1/7) 0 *DMD* (intron 61/78) 0 - 0 -	LINE (L1PA5) DNA (Tigger13a TcMar-Tigger) - SINE (AluY) -	+ + + + +	Pathogenic	*DMD* Muscular dystrophy, Duchenne type (MIM 310200)
AG/1003	46,XY,t(5;14)(q34;q23)	2	11.99X	1st analysis	t(5;14)(q35.1;q24.1)	2	5q35.1 14q24.1	8.00X 4.64X	chr5:170866372_170866372 chr14:69584205_69584218	0 12	1 *FGF18* (intron 3/4) 0 *DCAF5* (intron 4/8)	- -	+ +	VUS	-
VL/1102	46,XX,t(7;10)(p22;q26)	2	10.52X	Failure	NS	NS	NS	NS	NS	NS	NS NS	NS	NS	NS	NS
MH/1103	46,XX,inv(2)(q22;q32.3)	2	9.19X	1st analysis	inv(2)(q23.1q32.2)	2	2q23.1 2q32.2	8.00X 6.70X	chr2:148869298_148869296 chr2:190089580_190089589	0 8	3 *MBD5* (intron 2/14) 0 -	LINE (L1MC4) LINE (L1M5)	+ +	Pathogenic	*MBD5* Mental retardation, autosomal dominant 1 (MIM 156200)
JP/0107	46,XX,t(4;17)(q27;q23)	2	8.71X	1st analysis	t(4;17)(q27;q23.2)	2	4q27 17q23.2	4.93X 5.00X	chr4:122332907_122332920 chr17:60581315_60581319	12 3	0 - 0 *TLK2* (intron 2/21)	LINE L1 (L1PB1) SINE (AluSp), segmental duplication	- +	Pathogenic	*TLK2* Mental retardation (MIM 618050)
CS/1902	46,XY,t(4;14)(q31.1;q32.2)	2	10.47X	1st analysis	t(4;14)(q31.22;q32.2)	2	4q31.22 14q32.2	8.67X 10.60X	chr4:148252852_148252854 chr14:99097976_99097980	1 3	0 - 0 -	- DNA (HSMAR1)	+ +	Likely pathogenic	*BCL11B* Intellectual developmentam disorder and T-cell abnormalities disorder (MIM
AD/1601	46,XY,t(9;17)(q33;q22)	2	11.37X	1st analysis	t(9;17)(q33.3;q21.23)	2	9q33.3 17q21.32	9.67X 8.17X	chr9:129869403_129869408 chr17:45807432_45807437	4 4	0 *RALGPS1* (intron 8/18)/ *ANGPTL2* (intron 2/4) 0 -	- SINE (AluSx1)	+ +	VUS	-
LD/0108	46,X,t(X;1)(p11.2;p36.1)	2	12.91X	Failure	NS	NS	NS	NS	NS	NS	NS NS	NS	NS	NS	NS
HB/1104	46,XX,t(11;12)(q24;p12.2)	2	13.08X	1st analysis	t(11;12)(q24.1;p12.3)	2	11q24.1 12p12.3	7.20X 6.22X	chr11:123156790_123156794 chr12:19733004_19733012	3 7	0 - 0 -	- SINE (AluSz6)	+ +	VUS	-
OL/2202	46,XX,t(1;14)(q32;q22)	2	12.55X	1st analysis	t(1;14)(q32.1;q21.3)	5	1q32.1 5q14.3 5q14.3 5q14.3 14q21.3	8.00X 13.00X 6.00X 10.64X 5.54X	chr1:199578194_199578195 chr5:87638776_87638779 chr5:87791275_87791277 chr5:88527549_88527562 chr14:48231747_48231803	0 2 1 12 55	0 - 0 *TMEM161B-AS1* (intron 2/3) 0 - 0 - 0 -	LINE (L2a) LINE (L1MD1) SINE (AluJb) LINE (L1MCb) LINE (L1MEd)	- + + + +	Likely pathogenic	*MEF2C* Mental retardation, autosomal dominant 20 (MIM 613443)
AS/0301	46,XY,t(5;6)(q31.3;p22.1)	2	16.41X	1st analysis	t(5;6)(q33.1;p22.3)	2	5q33.1 6p22.3	10.2X 10.65X	chr5:152196581_152196590 chr6:17401275_17401294	8 18	0 *AK123816* (intron 3/4) 0 *CAP2* (intron 1/12)	DNA (MER121) -	+ +	VUS	-
JP/2301	46,XY,t(9;18)(p12;q12)	2	13.65X	1st analysis	t(9;18)(p23;q12.2)	2	9p23 18q12.2	5.07X 7.00X	chr9:13697578_13697592 chr18:35615585_35615587	13 1	0 - 0 -	LINE (L1PA13) -	+ -	VUS	-
NM/0201	46,XY,t(1;2)(p13.2;q31.2)	2	15.33X	1st analysis	t(1;2)(p11.2;q31.2)	19	1p11.2 2q11.2 2q12.1 2q22.3 2q22.3 2q22.3 2q23.3 2q23.3 2q24.1 2q24.1 2q24.1 2q24.1 2q24.1 2q24.1 2q24.1 2q24.1 2q31.2 3p21.31	16.00X 5.61X 10.00X 8.08X 17.00X 14.00X 14.00X 11.50X 14.00X 11.00X 8.90X 17.50X 7.15X 7.09X 17.00X 10.00X 13.00X 8.00X 0.00X	chr1:121247972_121247972 chr2:101663576_101663717 chr2:103067084_103067091 chr2:146853147_146853158 chr2:147559888_147559889 chr2:148318076_148318077 chr2:148346753_148346754 chr2:150753371_150753372 chr2:150803421_ chr2:156072831_156072831 chr2:156265463_156265493 chr2:156427981_156427982 chr2:156459486_156459545 chr2:157269856_157269866 chr2:157274489_157274589 chr2:157275115_157275114 chr2:157372068-157372067 chr2:179849948_ chr3:49454928-49454929	0 140 6 10 0 0 0 0 NS 0 29 0 58 9 0 0 0 NS 0	1 - 0 *TBC1D8* (intron 5/19) 0 *IL18RAP* (intron 10/11) 0 - 0 - 0 - 0 - 0 - NS NS 1 - 0 - 0 - 0 - 1 - 2 - 2 *GPD2* (intron 6/16) NS *CCDC141* (intron 2/23) 0 *AMT* (exon 9/9)	LINE (L1PA12) - - - LTR (LTR26 ERV1) - - LTR (LTR16A ERVL) LINE (L1PA2) - LTR (MER51-int, ERV1) LINE (L1PA5) SINE (AluSx) SINE (AluSx) SINE (AluSx3) LTR (LTR29 ERV1) - LINE (L1PA3) -	+ + + + + + + + + + + + + + + + + + +	VUS	-
VD/2401	46,XX,inv(7)(q22q34)	2	13.69X	2nd analysis	inv(7)(q21.3q35)	2	7q21.3 7q35	13.00X 23.00X	chr7:96420534_96420531 chr7:143282648_143282647	0 0	4 - 2 -	- LINE (L1P3), segmental duplication	+ -	Likely pathogenic	*DLX5* Split hand/foot malformation 1 (MIM 183600)
AL/2402	46,XY,inv(3)(p24q25.2)	2	16.34X	1st analysis	inv(3)(p24.3q26.1)	3	3p24.3 3p24.3 3q26.1	9.99X 8.30X 5.94X	chr3:16657768_16657884 chr3:16652378_16652421 chr3:162351577_162351610	116 43 33	0 - 0 - 0 -	LTR (THE1D-int ERVL-MaLR) - SINE (AluY)	+ + +	VUS	-
MD/2203	46,XX,t(1;5;3)(p21;q?34;p24)	3	14.20X	1st analysis	t(1;3;5)(p22.2;p24.3;q33.2)	18	1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 1p22.2 3p24.3 3p24.3 5q14.3 5q14.3 5q14.3 5q33.2	12.00X 20.50X 8.11X 24.00X 10.57X 7.26X 8.25X 12.21X 17.00X 11.20X 7.65X 10.00X 12.00X 7.50X 14.20X 25.00X 16.67X 9.50X	chr1:88987532_88987533 chr1:89228244_89228243 chr1:89292127_89292144 chr1:89318168_89318161 chr1:89407129_89407142 chr1:89475928_89478645 chr1:89479919_89497087 chr1:90835669_90835682 chr1:90912340_90912339 chr1:90917424_90917428 chr1:90920858_90924676 chr1:90927238_90927239 chr3:18842071_18842070 chr3:18845976_18845977 chr5:88188531_88188522 chr5:88189081_88189081 chr5:89299080_89299075 chr5:155211974_155211975	0 0 16 0 12 2716 17167 12 0 3 3817 0 0 0 0 0 0 0	0 - 2 *PKN2* (intron 3/21) 0 *PKN2* (intron 18/21) 8 - 0 *CCBL2* (intron 13/13) 0 *GBP3* (exons 7-8) 0 *GBP3* (exons 1-4) 0 - 2 - 0 - 0 - 2 - 2 - 0 - 10 *MEF2C* (intron 1/11) 1 *MEF2C* (intron 1/11) 6 - 0 *SGCD* (intron 1/8)	SINE (MIR3) - LINE (L1PB4) - LINE (L1MEf) SINE (AluSc/AluSz6) LINE (L1PA6) - - LINE (L1ME2) SINE (MIRb) LINE (L1PA15) - - - Simple (TA)n LINE (L1ME3C)	+ + + + + + + + + + + + - - + + + +	Pathogenic	*MEF2C* Mental retardation, autosomal dominant 20 (MIM 613443)
JB/1602	46,XX,t(2;16)(p25.3;q12.2)	2	14.91X	1st analysis	t(2;16)(p25.3;q12.2)	2	2p25.3 16q12.2	15.00X 15.50X	chr2:245819_245820 chr16:56288704_56288703	0 0	0 *SH3YL1* (intron 4/9) 2 *GNAO1* (intron 2/8)	- -	+ +	VUS	-
EC/2302	46,XX,t(4;14)(q24;q32.2)	2	14.89X	1st analysis	t(4;14)(q24;q32.12)	2	4q24 14q32.12	12.00X 13.00X	chr4:105548686_105548687 chr14:94170010_94170011	0 0	0 *AK094561* (intron 1/7) 0 *UNC79* (intron 48/49)	LTR (MLT2B1 ERVL) LTR (MLT1C ERVL-MaLR)	+ +	VUS	-
TP/0801	46,XY,t(1;8)(p21;q21.1)	2	15.78X	1st analysis	t(1;8)(p21.3;q21.2)	2	1p21.3 8q21.2	11.46X 13.00X	chr1:97975912_97975924 chr8:85384078_85384078	11 0	0 *DPYD* (intron 13/22) 1 *RALYL* (intron 2/9)	LTR MLT1H-int (fam ERVL MaLR) SINE (MIR)	+ -	VUS	-
DM/0109	46,XY,t(1;19)(p36;q13.1)	2	15.25X	1st analysis	t(1;19)(p36.32;q13.11)	2	1p36.32 19q13.11	6.00X 5.00X	chr1:3199348_3199352 chr19:32805952_32805963	3 10	0 *PRDM16* (intron 3/16) 0 -	- -	+ +	Pathogenic	*PRDM16*
BA/2303	46,XY,t(2;8)(p16.3;p11.23)	2	14.68X	1st analysis	t(2;8)(p16.3;q11.23)	4	2p16.3 8p11.23 8q11.23 8q12.1	17.50X 7.35X 6.38X 10.75X	chr2:50899875_50899876 chr8:38240669_38240691 chr8:53594094_53594109 chr8:57742496_57742499	0 21 14 2	0 *NRXN1* (intron 5/22) 0 - 0 *RB1CC1* (intron 5/23) 0 -	- - - LINE (L1MC1)	- + + +	Pathogenic	*NRXN1* Autism (MIM 209850)
JE/1401	46,X,t(X;10)(q?27;q?23)	2	12.83X	1st analysis	t(X;10)(q27.3;q22.3)	2	Xq27.3 10q22.3	14.75X 8.00X	chrX:143274164_143274161 chr10:80552343_80552344	0 0	4 - 0 -	Segmental duplication LINE (L2b)	- +	Likely pathogenic	*ZMIZ1** Syndromic neurodevelopmental disorder
MG/1801	46,XX,t(8;9)(p21.2;q34)	2	17.91X	1st analysis	t(8;9)(p12;q33.1)	2	8p12 9q33.1	9.25X 20.67X	chr8:32608533_32608536 chr9:118797860_118797858	2 0	0 *NRG1* (intron 7/11) 3 -	- -	+ +	VUS	-
EM/2403	46,XX,t(4;19)(q33;p13.2)	2	14.30X	1st analysis	t(4;19)(q34.1;p13.2)	2	4q34.1 19p13.2	11.00X 19.29X	chr4:173612939_173612937 chr19:13191093_13191099	0 5	3 *GALNT16* (intron 5/12) 0 *NFIX* (intron 7/10)	- SINE (AluJr4)	+ +	Pathogenic	*NFIX* Sotos syndrome 2 (MIM 614753)

BM/1603	46,X,t(X;13)(p22.1;q22)	2	13.59X	1st analysis	t(X;13)(p22.31;q31.3)	2	Xp22.31	8.00X	chrX:6560456_6560460	3	0 -	-	+	VUS	-
							13q31.3	9.50X	chr13:79950804_79950807	2	0 *RBM26* (intron 3/20)	-	+		
CF/2304	46,XY,t(2;15;8;10)(q32.3;q26;q21.3;q23)	4	17.83X	1st analysis	t(2;15;8;10)(q33.1;q26.3;q22.1;q23.31)	5	2q33.1	15.50X	chr2:199780459_199780460	0	0 -	LTR (MLT1G1 ERVL MaLR)	+	Likely pathogenic	*SATB2* Glass syndrome (MIM 612313)
							8q22.1	21.00X	chr8:98883797_98883795	0	3 *MATN2* (1/18)	-	+		
							10q23.31	12.25X	chr10:90093734_90093731	0	4 *RNLS* (intron 5/6)	LTR (MLT1A0 ERVL MaLR)	+		
							15q26.3	7.76X	chr15:99757847_99763974	6126	0 *TTC23* (intron 4-7/12)	LTR (MLT1G1 ERVL MaLR)	+		
							15q26.3	9.69X	chr15:99781580_99783832	2251	0 *TTC23* (intron 2/12)	LINE (L1MA3)	+		
ML/1402	46,XX,t(14;20)(p13;q13.2)	2	14.07X	Failure	NS	NS	NS	NS	NS	NS	NS NS	NS	NS	NS	NS
KT/1403	46,XY,der(8)t(8;22)(q12;q12),der(13)t(8;13)(q31;q23),der(14)t(14;15)(q11.2;q25),der(15)t(14;15)(q21;q24),der(22)t(13;22)(q31.1;p11.2)	6	12.40X	1st analysis	t(8;13)(q23.1;q31.1),t(14;15)(q12;q26.2)	14	8q12.2	10.00X	chr8:61885817_61885817	0	1 -	-	+	VUS	-
							8q21.13	11.67X	chr8:80663818_80663823	4	0 -	LINE (L1MA9)	+		
							8q21.13	7.19X	chr8:80741443_80754254	12810	0 *AK055332* (intron 2/5)	LINE (L1PA10)	+		
							8q22.1	13.50X	chr8:98566061_98566062	0	0 -	-	+		
							8q23.1	13.50X	chr8:109560519_109560518	0	2 -	-	+		
							13q31.1	20.00X	chr13:83214192_83214192	0	1 -	DNA (Tigger TcMar)	-		
							13q31.3	8.40X	chr13:93493574_93493578	3	0 *GPC5* (intron 7/7)	-	+		
							14q12	15.67X	chr14:25904165_25904167	1	0 -	LINE (L1P3)	-		
							14q12	11.00X	chr14:31225226_31225226	0	1 -	-	+		
							14q21.1	4.50X	chr14:42074504_42074505	0	0 -	-	+		
							14q21.3	10.00X	chr14:49059674_49059674	0	1 -	LINE (L1PA5)	+		
							14q12	8.57X	chr14:32709589_32709602	12	0 -	LINE (L1PA4)	+		
							14q13.1	18.00X	chr14:33757311_33757312	0	0 *NPAS3* (intron 3/11)	-	+		
							15q26.2	11.78X	chr15:94885854_94885846	0	9 *MCTP2* (intron 8/23)	Simple (DNA)	+		
AL/1802	46,XX,t(6;14)(p12;q21)	2	9.17X	1st analysis	t(6;14)(p12.1;q21.1)	2	6p12.1	7.40X	chr6:53155903_53155917	13	0 *ELOVL5*	-	+	VUS	-
							14q21.1	7.44X	chr14:42469830_42469838	7	0 -	LINE (L1MB8)	+		
MC/1803	46,XY,t(7;10)(q21.1;q26.1)	2	13.35X	1st analysis	t(7;10)(q11.22;q24.33)	2	7q11.22	11.00X	chr7:69507717_69507715	0	3 *AUTS2* (intron 2/18)	-	+	Pathogenic	*AUTS2* Mental retardation, autosomal dominant 26 (MIM 615834)
							10q24.33	6.33X	chr10:105031287_105031289	1	0 *BC040734* (intron 1/1)	-	+		
GS/2305	46,XY,t(5;12)(q34;q23)	2	15.10X	1st analysis	t(5;12)(q33.2;q22)	2	5q33.2	11.00X	chr5:153874165_153874166	0	0 -	-	+	VUS	-
							12q22	13.10X	chr12:92834979_92834988	8	0 *CLLU1* (intron 1/1)	-	+		
OP/0701	46,XX,t(3;14)(q26.3;q32.3)	2	13.83X	1st analysis	t(3;14)(q26.2;q32.12)	2	3q26.2	8.57X	chr3:169239241_169239247	5	0 *MECOM* (intron 1/16)	-	+	VUS	-
							14q32.12	13.50X	chr14:94528185_94528188	2	0 *DDX24* (intron 3/8)	DNA (MER58A hAT Charlie)	+		
JB/1404	46,XX,t(10;12)(q22.2;q24.3)	2	14.64X	1st analysis	t(10;12)(q22.3;q24.32)	2	10q22.3	4.29X	chr10:81010865_81010871	5	0 *ZMIZ1* (intron 7/24)	-	+	Pathogenic	*ZMIZ1** Syndromic neurodevelopmental disorder
							12q24.32	9.70X	chr12:128533508_128533517	8	-	-	-		
MD/0110	mos 46,X,t(X;18)(q28;q11.2)/46,XX	2	24.90X	1st analysis	t(X;18)(q28;q11.2)	2	Xq28	18.87X	chrX:151730337_151756693	26355	0 -	-	+	VUS	-
							18q11.2	31.40X	chr18:20256560_20185222	0	71339 -	DNA (MER58A hAT Charlie)	+		
JJ/0402	46,XY,t(3;6)(q21.3;p21.33)	2	12.83X	1st analysis	t(3;6)(q21.1;p21.32)	2	3q21.1	10.33X	chr3:122821837_122821839	1	0 *PDIA5* (intron 5/16)	-	+	Pathogenic	*SYNGAP1* Mental retardation, autosomal dominant 5 (MIM 612621)
							6p21.32	3.33X	chr6:33409201_33409206	4	0 *SYNGAP1* (intron 12/18)	-	+		
MCP/1105	46,XX,t(2;15)(q32.2;q26.1)	2	13.60X	1st analysis	t(2;15)(q32.2;q26.2)	2	2q32.2	5.67X	chr2:191104110_191104112	1	0 *HIBCH* (intron 11/13)	LINE (L1PA8)	+	VUS	-
							15q26.2	11.33X	chr15:96475803_96475805	1	0 -	-	+		
BM/1201	46,X,idic(Y)(q11.22),der(5)t(5;8)(q23;q24)t(5;11)(p12;p11),der(8)t(5;8),der(11)t(5;11)	4	13.97X	1st analysis	der(5)t(5;8)(q23.1;q24.21)t(5;11)(p12;p11.2),der(8)t(5;8),der(11)t(5;11)	5	5p13.3	7.00X	chr5:29792806_29792807	0	0 -	LINE (L1MA8)	-	Pathogenic	*GHR* Partial Growth Hormone Insensitivity (MIM 604271)
							5p12	3.70X	chr5:42642909_42643063	153	0 *GHR* (intron 4/10)	LTR (MLT1C MaLR)	+		
							5q23.1	7.80X	chr5:115867110_115867114	3	0 *SEMA6A* (intron 1/18)	-	+		
							8q24.21	7.00X	chr8:128815229_128815228	0	2 *PVT1* (intron 1/2)	-	+		
							11p11.2	7.54X	chr11:47868788_47868800	11	0 *NUP160* (intron 2/35)	LINE (L2c)	+		
AB/1604	46,XY,t(1;2)(p13;p13)	2	15.48X	1st analysis	t(1;2)(p13.1;p16.1)	2	1p13.1	7.67X	chr1:108658558_108658563	4	0 -	LTR (MER21C ERVL)	+	VUS	-
							2p16.1	12.00X	chr2:60225092_60225095	2	0 -	-	+		
AB/2404	46,XX,t(7;18)(q21;q21)	2	14.48X	1st analysis	t(7;18)(q21.3;q21.2)	2	7q21.3	7.94X	chr7:93672128_93676082	3953	0 -	LINE (L1MeC)	+	VUS	-
							18q21.2	7.95X	chr18:52640089_52643123	3033	0 -	LINE (L2a)	+		

Supplementary Table S2: genome sequencing results and breakpoints characteristics

nb: number; GS: genome sequencing; NS: not specified;
TAD: topologically associated Domains (GM12878; Rao et al., 2014)
* Carapito R, et al. ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. Am J Hum Genet 2018