Table 1: Interesting rearrangements identified by the CHERISH Consortium.

a) Lituania

Patient ID	Chromosome band	Genomic position	Origin	Comment
LIT-001-138	del2q24.2 - q24.3	162118995-164461141	De novo	Previously reported deletion associated with epilepsy, ID
LIT-001-227	del2q37.3	239525614-242717216	De novo	2q37 monosomy syndrome
LIT-001-025	del4q28.3	137417138-138947393	Maternal	Private inherited CNV, PCDH18 possible candidate gene for ID
LIT-001-117	del4q21.22	83373844-84097897	De novo	Previously reported deletion associated with ID, epilepsy, hypotony, obesity, small hands and feet
LIT-001-030	del5p14.3 - p14.1	23025478 - 26938536	De novo	Cri du chat syndrome in complex chromosomal rearrangement
LIT-001-216	del5q14.3 del6q21 del16q23.1	86456211-89104733 104995949-105493496 81507514-81784949	De novo De novo De novo	Complex chromosomal rearrangement
LIT-001-123	del7q35 - q36.1	146392196-149441454	De novo	Duplication in the region of Sotos syndrome
LIT-001-156	del16p11.2	29563985-30106254	De novo	Autism susceptibility locus
LIT-001-029	del16p11.2	29215334-30106254	Maternal	Autism susceptibility locus
LIT-001-054	del5q35.3	176728738-176992730	De novo	Duplication in the region of Sotos syndrome
LIT-001-150	dup7p22.1	5337072-6316915	De novo	Novel syndrome
LIT-001-019	dup16p13.3	2589524-3911387	De novo	Duplication in the region of Rubinstein-Taybi syndrome
LIT-001-010	dup14q32.12	90907243-105866436	Unknown	Unbalanced translocation
LIT-001-012	del4q34.3 dup8p23.1	179503254-191162351 0- 10772254	De novo	Unbalanced translocation
LIT-001-013	del17q22.31	45682246-47544816	De novo	Novel syndrome
LIT-001-292	dupXq27.1	138271793-138525255	Maternal	Prieto syndrome

b) Russia

Array-CGH findings in Russian ID patients using 44K and 60K arrays.

Patient ID	Chromosome band	Genomic position (hg18)	Origin	Comment
RUS 008	del11p13	35,137-36,292 Mb	undefined	close to WAGR locus
RUS 018	del11p15.4	9,718-9,967 Mb	de novo	Charcot-Marie-Tooth Type 4B region
RUS 036	del22q11.21	17,277-19,713 Mb	undefined	DiGeorge locus
RUS 045	del15q24.1q25.2	71,791-79,856 Mb	undefined	15q24 recurrent microdeletion syndrome
RUS 053	dup15q11.1-q11.2	18,741,716 -20,335,946 kb	undefined	Close to PWS/AS deletion breakpoints
	dup22q11.22	20,613,292 -20,899,881 kb	undefined	22q11 duplication syndrome
RUS 077	del15q11.1-q11.2	18,741,716 -20,010,618 kb	undefined	Close to PWS/AS deletion breakpoints
	del15q24.1-q24.2	70,766,177 -73,754,412 kb	undefined	15q24 recurrent microdeletion syndrome
	del15q24.1-q24.2	73,035,132 -73,419,685 kb	undefined	15q24 recurrent microdeletion syndrome
RUS 081	del15q11.1-q11.2	18,741,716 -20,010,618 kb	undefined	Close to PWS/AS deletion breakpoints
RUS 091	del10q24.32	104,147-104,668 Mb	undefined	Desmoplastic medulloblastoma region
RUS 098	dup3p26.4	701,645 -1,467,721 kb	undefined	*CNTN6* gene
	del15q11.1-q11.3	18,835,660 -20,010,618 Mb	undefined	Close to PWS/AS deletion breakpoints
RUS 120	del16p11.2	29,581-30,105 Mb	undefined	16p11.2 autism susceptibility locus
RUS 141	dup14q11.2	20,767,632 -22,722,130 kb	undefined	Lysinuric protein intolerance
RUS 153	dup(1)(q25.1q25.2)	172,269-178,409 Mb	undefined	
RUS 154	del3p26.3	1,164-1,533 Mb	undefined	*CNTN6* gene
RUS 165	del16p12.2	21,507-21,647 Mb	undefined	16p11.2-p12.2 microdeletion syndrome
RUS 167	del16p11.2	29,233-30,106 Mb	undefined	16p11.2 autism susceptibility locus

RUS 193	del5p15.33-p15.2	0,148-12,692 Mb	undefined	Cri du Chat Syndrome
	del5q13.3	74,021-74,103 Mb	paternal	Sandhoff disease
Unclear variants				
RUS 015	del11q22.3	107,970-108,140 Mb	undefined	
RUS 031	dup18p11.32	2,322-2,673 Mb	undefined	
RUS 104	dup6p22.2	25,677-26,393 Mb	undefined	
RUS 105	dup5q33.1	148,620-148,735 Mb	undefined	
RUS 195	dup7q21.3	94,769-94,901 Mb	undefined	Region associated with coronary artery disease
RUS 199	del2q12.3	107,971-108,435 Mb	undefined	

c) Czech Republic

Patient ID	**Chr band**	**Genomic position**	**Origin**	**Comment**
cze004	del16p12.1	21858916-22338671	pat	known microdeletion syndrome
cze016	del1q21	144927798-147656069	mat	known microdeletion syndrome
cze020	del1q44	242951908-243763883	dn	known microdeletion syndrome
cze022	del10pter	125708-338041	dn	known microdeletion syndrome
cze033	del2p15-16.1	60543481-60981483	dn	known microdeletion syndrome
cze054A,B	del17q21	41063083-41571673	dn	known microdeletion syndrome
cze057	dupXq28	152303338-153557763	mat	known microduplication syndrome
cze075	dup22q11	17257787-19792353	pat	known microduplication syndrome
cze075	del15q13.1	27234439-28443879	dn	debated in literature
cze099	del16p11.2	29535521-30107306	mat	known microdeletion syndrome
cze140A,B	del16p11.2	29575754-30107306	pat	known microdeletion syndrome
cze001	del8pter	166818-6601195	mat	MCPH1 known MR gene
cze025	del15	29806023-30383373	mat	CHRNA7 known MR gene
cze050	del2p	50867151-51111920	mat	NRXN1 known MR gene
cze087	del2p	50603932-50774974	mat	NRXN1 known MR gene
cze159	del2p	50450619-50894976	dn	NRXN1 known MR gene

cze021	dup20p	1-7100000	mat	very large aberration (7 Mb)
cze047	invdupdel3p	41894-14852539	dn	very large aberration (11+4 Mb)
cze089	del9p	36587-13647571	dn	very large aberration (14 Mb)
cze139	del2p	9977295-17926516	dn	very large aberration (8 Mb)
Unclear variants				
cze013	del12q13	51801299-52737892	dn	including HOXC, two case reports in the literature, several specific phenotypic features
cze036	del1q24.3	170194319-170797310	dn	DNM3 codes a postsynaptic protein associated with MR
cze036	del6q24.1	142211174-144665837	dn	PLAGL1 is a maternally imprinted gene associated with growth and developmental delay
cze049	delXq27.1	138351842-139709011	mat	SOX3 codes a transcription factor involved in neural development, has been involved in XLMR
cze053	del9q21.33	85784890-87547315	dn	NTRK2 signalling is involved in several neuropsychiatric diseases, de novo point mutation was reported in a patient with ID and obesity
cze061A,B	del15q25	82743638-83515133	mat	suspected novel microdeletion syndrome, inherited from a normal mother
cze072	del15q25	82743638-83515133	mat	suspected novel microdeletion syndrome, inherited from a very mildly affected mother
cze073	del2p14-p15	62013232-65731466	dn	two case reports in the literature, limited amount of specific phenotypic features
cze110	del 17q24.3	62712112-65528608	dn	MAP2K6, other members of the MAPK pathway were associated with ID
cze002	dup2q12.3	106765625-107275024	mat	possible accumulation of multiple hits?
cze002	dup9q34.3	138794348-139629602	dn or pat	possible accumulation of multiple hits?
cze002	del15q26.2	93288056-94374146	mat	possible accumulation of multiple hits?
cze017	dup5p13.2	33977113-34785173	pat	possible accumulation of

				multiple hits?
cze017	del13q12.12	22437563-23802315	pat	possible accumulation of multiple hits?
cze017	del14q32.2	96283870-99185361	mat	possible accumulation of multiple hits?
cze017	dup19p13.3	3737824-4591983	pat	possible accumulation of multiple hits?
cze077A	del9q31.2	107449221-107518947	mat	possible accumulation of multiple hits?
cze077A	dup10q11.23	51738304-51830868	pat	possible accumulation of multiple hits?
cze077A	del15q11.2	20305686-20667673	pat	possible accumulation of multiple hits?

("dn" indicates *de novo* origin, "mat" and "pat" indicate maternal and paternal inheritance, respectively)

d) Estonia

Patient ID	Gender	Rearrangement	Chromosome band	Genomic position	Size	Confirmation
EST-050-004	female	Del	22q11.21	17 118 296 - 19 963 350	2.85 Mb	qPCR
EST-053-001	female	Del	22q11.21	17 118 296 -19 963 350	2.85 Mb	qPCR
EST-042-001	male	Del	11q24.2-qter	123 280 701 - 134 439 273	11.16 Mb	FISH
EST-042-001	male	Dup	20q13.32-qter	56 361 884 - 62 419 232	6.06 Mb	FISH
EST-117-001	female	Dup	8p23.1	7 882 322 - 10 186 977	2.3 Mb	qPCR
EST-006-001	male	Del	16p11.2	29 767 385 - 30 123 121	0.36 Mb	qPCR
EST-054-001	female	Del	16p13.11	14 837 226 - 16 199 483	1.36 Mb	qPCR
EST-004-001	female	Dup	Xq13.1	69 158 806 - 69 621 096	0.46 Mb	qPCR
EST-016-001	female	Del	Xq25	126 846 693 - 127 333 008	0.49 Mb	qPCR
EST-028-001	male	Del	10q21.1	55 279 774 - 55 355 078	0.075 Mb	qPCR
EST-035-001	male	Dup	5p14.3	21 228 514 - 23 215 032	1.99 Mb	qPCR
EST-040-001	male	Del	17p12	14 041 753 - 15 411 904	1.37 Mb	qPCR
EST-048-001	male	Del	14q21.3	44 513 286 - 44 856 588	0.34 Mb	qPCR
EST-049-001	male	Del	4q35.2	188 944 085 - 189 987 027	1.04 Mb	qPCR
EST-127-001	female	Del	2q31.2-q32.1	178 159 154 - 185 704 503	7.55 Mb	qPCR
EST-120-001	male	Del	Xp22.11	23 298 146-23 516 211	0.22 Mb	qPCR
EST-156-001	male	Del	1p32.3-p32.2	52 763 932-58 347 905	5.58 Mb	qPCR
EST-161-001	male	Del	20p12.3	8 047 252-8 523 333	0.48 Mb	qPCR
EST-136-001	male	ROH	10q26.11-q26.3	118 816 431-134 429 016	15.61 Mb	-

e) Ukraine

Patient ID	Chromosome band	Genomic position	Origin	Comment
UKR 003	Dup 12q24.33	130868308-131117235	maternal	The region of rearrangement involved only one gene - *RIMBP2* (RIMS-binding protein 2) that is associated with speech delay. Mother has no MR, but speech delay.
UKR 022	Dup 7p21.1	16455637-16819776	maternal	Deletion in *MECP2* gene. The patient shows some features of Rett syndrome, although her phenotype is not typical.
UKR 031	Del 5q15-q22.1	92,766-110,875	novel	several SNPs within the region were associated with psychiatric disorders (bipolar disorder, Asperger syndrome, attention deficit disorder with hyperactivity, panic disorder).
UKR 050	Dup 8p23.1-p23.2	5,878-6,572	paternal	

UKR 112	Del Xq28	152,975-153,062	not maternal	*MECP2* gene deletion. Father is not available for the analysis
UKR 119	Del 2q32.3-q33.1	196,509-201,522	novel	The deletion overlaps with a rearrangement described in DECIPHER, but phenotypic features are different
UKR 124	Del 1p36.32-pter	0,749-3,552	paternal	Region described for 1p36 Deletion Syndrome. Paternal origin of the rearrangement was confirmed by qPCR (father is healthy)
UKR 143	Del 19q12	34,283-34,717	maternal	
UKR 173	Del 16p11.2	29,581,455-30,106,101	maternal	
UKR 057	Del 10q26.3-qter	132,997-135,254	paternal	Proband has an inbalanced translocation, inherited from father. Confirmed by FISH
UKR096	Dup 2q35-qter	218,312-242,690		
UKR 202	Del 5p15.2/ Dup 10q25.3-26.3	10 Mb 18Mb	paternal	Proband has an inbalanced translocation, inherited from father. Confirmed by FISH
UKR 044	Del 8pter-p23.1	0,181-6,901	not maternal	Suspected inv dup del 8p syndrome as a result of unbalanced translocation
	Dup 8p23.1-p21.2	12,267-26,684	maternal	
UKR 091	Dup Xp22.12-p22.11	786,847-21,925,361	maternal (mother has no phenotype)	region contains considerable amount of genes previously not reported to cause ID : *SMS, MBTPS2.*
	Dup 12q21.31	84,791,739-84,846,143	maternal (mother has no phenotype)	region contains considerable amount of genes previously not reported to cause ID : neurotensin gene (*NTS*)
UKR 136	Del 2q37.1-qter	233,055-242,690	paternal	Both regions contain considerable amount of genes previously not reported to cause ID
	Dup 3q27.3-qter	187,764-199,288	novel	
UKR 160	Del 1p36.13	16,814-16,921	maternal	Region described for 1p36 Deletion Syndrome.
	Dup Xp21.1	32,948-33,139	maternal	

f) Cyprus

Patient ID	Chromosome band	Size	Origin	Comment
CYP 001-003	dup17p13.3	2.1Mb	paternal	Partial overlap with Miller-Dieker region Father is borderline/mild ID
CYP 004-005	dup19p13.2	102kb	undefined	
CYP 005-005	del12q24.3	536kb	maternal	Acquiring clinical details of mother
CYP 009-003	dup12q21.1-12q21.3	7Mb	paternal	Region includes Bardet-Biedl syndrome
	delXq28	198kb	paternal	Polymorphic within study cohort
CYP 011-003	del9q33.1	164kb	paternal	Non-segregating
CYP 012-003	dup1q21.3	99kb	paternal	Acquiring clinical details of father
CYP 013-003	dup17q22	145kb	maternal	Non-segregating

NOTE: analysis included parents to facilitate first-time analysis and evaluation of 400K platform

g) Poland

Patient ID	Chromosome band	Genomic position	Origin	Comment
326-POL-001-060	dup16q22.3 q24.3	71,438,065-88,685,456 Mb	de novo	large aberration
237-POL-001-019	dup22q11.21	17,187,197-19,654,308 Mb	de novo	known syndrome (22q11 duplication syndrome)
355-POL-001-079	del16p11.2	29,563,594-30,731,722 Mb	de novo	autism susceptibility locus
255-POL-001-029	del2q37.3	238,251,381-242,604,099 Mb	de novo	large aberration
	dup10p15.3 p15.1	125,708-6,683,822 Mb	de novo	large aberration
2128-POL-001-	del9p24.3p22.2	36,587-18,206,477	de novo	large aberration
	dup9p22.2p	18,214,660-	de	large aberration

238	12	41,513,872 Mb	novo	
328-POL-001-062	del22q13.33	48,483,337-49,515,911 Mb	de novo	known syndrome (Phelan Mcdermid)
259-POL-001-033	dicentric isochromosome X, dupXp21.1q28		de novo	large aberration
655-POL-001-124	del11q13.1	65,952,841-66,768,341	de novo	Genes that are involved or may be involved in ID located in this region: *NPAS4*, *BBS1*,
644-POL-001-122	del15q11.2	18,818,086-22,219,100 Mb	de novo	known syndrome (Angelman syndrome)
1351-POL-001-188	dup15q26.3	99,841,659-100,092,020	undefined	known syndrome (15q26 overgrowth syndrome)

h) Italy

Patient ID	Chromosome band	Genomic position	Origin	Comment
ITA.003.01	del16p11.2	29,581-30,105 Mb	de novo	Autism susceptibility locus
ITA.004.01	del7q31.32	121,788-122,030 Mb	likely maternal	Familial CNV, not reported in Decipher
ITA.029.01	del1q44	242,748-243,979 Mb	de novo	
ITA.089.01	del(14)(q22.3q23.1)	54,493-58,868 Mb	de novo	
ITA.098.01	dup22q11.21	17,276-19,712 Mb	paternal	microdup 22q11 syndrome
ITA.101.01	dup7q11.23	72,338-73,777 Mb	de novo	
ITA.116.01	dup22q11.21	19,084-19,712	paternal	microdup 22q11 syndrome (smaller)
ITA.121.01	dup15q11.2q13.1	21,250-26,199	maternal	Debated – susceptibility to ID
ITA.128.01	del6q26qter	161,079-170,734	de novo	2 similalr deletions reported in DECIPHER
ITA.129.01	del9q31.1q31.3	107,009-113,380	de novo	
ITA.131.01	dupXp11.23p11.22	47,937-52,858	familial	
ITA.152.01	dup16q22.3q24.1	71,334-84,917	de novo	
ITA.154.01	dup1q21.1	145,031-146,201	maternal	Debated – susceptibility to ID
ITA.158.01	del3p25.3	9,391-9,474	de novo	
ITA.159.01	del17pterp13.3	0,05-2,361	de novo	Miller-Dieker Syndrome region
ITA.161.01	del8p23.1	7,290-12,285	de novo	
	del21q22.3qter	46,380-46,880	de novo	
ITA.163.01	dup6q26qter	162,314-170,734	de novo	
	del8pterp23.1	0,181-6,901	de novo	invdupdel
	dup8p23.1p21.3	12,627-20,512	de novo	
ITA.165.01	dup15q11.2q13.1	21,208-26,199	de novo	
ITA.170.01	del16p13.11p12.3	15,399-18,020	paternal	epilepsy associated region
ITA.176.01	del16p11.2	29,581-30,106	paternal	Autism susceptibility locus
Unclear variants				
ITA.086.01	del6q14.3	85,740-87,097 Mb	undefined	Previously unreported CNV

ITA.098.01	dup16q23.3	82,387-82,593 Mb	maternal	Could be a modifier of the other variant
ITA.115.01	dupXp22.31	6,561-7,992 Mb	undefined	Debated pathogenicity
ITA.130.01	dup3p24.1	27,301-27,517	maternal	Previously unreported CNV
	dup9p24.2	3,817-4,577	paternal	One single patient in DECIPHER
	del16q22.1	67,335-67,433	maternal	Previously unreported CNV
ITA.164.01	del2q37.3	241,961-242,053		Previously unreported CNV
ITA.169.01	dup3q23	143,649-143,760	maternal	Previously unreported CNV
	del7q11.21	64,329-64,503	paternal	Known CNV
	dup12q12	38,705-38,770	maternal	Previously unreported CNV
	dup15q13.3	29,809-30,298	maternal	Debated pathogenicity
ITA.176.01	dup(8)(q24.3)	145,976-146,037	maternal	Debated pathogenicity

***Coverage proportions*. Number of target bases covered 0X-100X for the 21 CHERISH samples.**