Patient	Sex	Age (year)	Rearrangement	Karyotype (ISCN 2016)	CMA	Inheritance	Phenotype
IS/0101	F	3	Translocation	46,XX,t(9;17)(p13;q21)	Agilent 180K	dn	IUGR, post-natal growth retardation, developmental delay, hypotonia, facial dysmorphism, eczema, dystrophic hair
EB/0501	F	4	Translocation	46,X,t(X;1)(p22.1;q12)	Agilent 180K	mat	Mild growth delay (-2SD), mild ID, feeding difficulties
IL/1901	F	41	CCR	46,XX,der(3)inv(3)(p13p22)inv(3)(p12q26.3)	Agilent 105K	familial	Mild ID, speech delay, cleft palate, single palmar crease, strabismus. Translocation transmitted to her daughter, with the same phenotype.
LV/1701	F	6	Translocation	46,XX,t(3;9)(q26.33;p22)	Agilent 105K	dn	Mild ID, facial dysmorphism, strabismus, hypermetropia
JB/0102	M	3	CCR	46,XY,t(6;8;9;13)(q26;p23;p21;q21)	Agilent 180K	dn	Mild ID, thin motor skills troubles, autistic spectrum disorder (lonely, irritable), repeated otitis media
CM/0103	M	23	Translocation	46,XY,t(3;22)(q13-21;p11)	Agilent 180K	dn	Moderate ID with neurocognitive regression, pharmacoresistant epilepsy
MG/1001	F	1	Translocation	46,XX,t(14;20)(p13;p11)	Agilent 60K	dn	Pierre-Robin syndrome
PS/0502	M	23	Translocation	46,XY,t(20;21)(q11.2;q21)	Agilent 60K	dn	ID, autistic spectrum disorder, hyperactivity, food behavioral troubles, epilepsy, strabismus, pain insensitivity
VJ/0601	M	11	Translocation	46,XY,t(4;14)(p15;q32.1)	Agilent 60K	dn	Moderate ID, speech delay, autistic spectrum disorder (anxiety), facial dysmorphism, multiple allergies
CS/1002	F	12	Insertion	46,XX,ins (15;12) (q15 or q21;q24.1q24.2)	Agilent 44K	dn	Relative macrocephaly (OFC +2SD), moderate ID, facial dysmorphism, café-au-lait spots
CL/2001	F	19	Translocation	46,XX,t(11;15)(p14;q14)	Affymetrix CytoScan HD Array	dn	Severe ID, absent speech, epilepsy, microcephaly, facial dysmorphism, atrial and ventricular septal defect, growth retardation, delayed myelinsation of corpus callosum, larynx malposition, hiatal hernia
MD/0104	F	15	Translocation	46,X,t(X;7)(p11.1;q36.1)	Illumina SNP-array Human CNV370-quad V3	dn	Moderate ID, hyperactivity attention deficit disorder, pharmacoresistant epilepsy, ataxia, tremor, auto-immune hepatitis
LF/2002	F	23	Translocation	46,XX,t(2;8)(p24;q12)	Agilent 180K	mat	Learning difficulties (dyslexia, dysorthographia); delay in walking acquisition
SL/0105	F	13	Inversion	46,XX,inv(2)(p12q22)	Agilent 180K	dn	Microcephaly, severe ID, behavioral troubles (anxiety, aggressivity, sleep disturbance), pharmacoresistant epilepsy, ataxia, hearing loss, strabismus, constipation, feeding difficulties
EB/0401	M	9	Insertion	46,XY,ins(5)(q15q23.3q34)	Integragen Intrachip V6 (1Mb)	dn	IUGR, post-natal growth retardation, microcephaly, ID, sleeping and feedings difficulties, epilepsy (hyperthermic seizures), choreic and dystonic abnormal movements, stabismus
TS/1101	M	5	CCR	46,XY,t(3;4;10;14;18)(p23;q24;p13;q32;q23)	Illumina Human OmniExpress24 - 715K	dn	Mild ID, behavioral troubles, facial dysmoprhism
CG/0106	F	16	CCR	46,X,t(X;13;15)(p21;q22;q22),t(6;11)(q21;q24)	Agilent 180K	dn	Severe ID, absent speech, attention deficit, Duchenne muscular dystrophy
AG/1003	M	16	Translocation	46,XY,t(5;14)(q34;q23)	Agilent 60K	dn	Neonatal stridor, feeding difficulties, facial dysmorphism, hypertrichosis, club feet, left thumb ankylosis, Chrohn disease
VL/1102	F	11	Translocation	46,XX,t(7;10)(p22;q26)	Illumina Human OmniExpress24 - 715K	dn	ID, speech delay, facial dysmorphism (retrognathia), high-arched palate
MH/1103	F	3	Inversion	46,XX,inv(2)(q22;q32.3)	Illumina Human OmniExpress24 - 715K	dn	ID, speech delay, troubles of thin motor skills, pharmacoresistant epilepsy
JP/0107	F	14	Translocation	46,XX,t(4;17)(q27;q23)	Agilent 244K	dn	Overweight, learning difficulties, attention disorder, facial dysmorphism, coronal craniosynostosis (surgicallay treated)
CS/1902	M	29	Translocation	46,XY,t(4;14)(q31.1;q32.2)	Agilent 180K	dn	Learning difficulties, mild speech delay, relational troubles, psychiaric disorders, facial dysmorphism
AD/1601	M	18	Translocation	46,XY,t(9;17)(q33;q22)	Illumina Human Cyto SNP -12v2 - 300K	mat	Spondylo-epiphysal dysplasia, acro-osteolysis
LD/0108	F	4	Translocation	46,X,t(X;1)(p11.2;p36.1)	Agilent 180K	dn	ID, facial dysmorphism, axial hypotonia, dyschromia Blaschko line, strabismus
HB/1104	F	18	Translocation	46,XX,t(11;12)(q24;p12.2)	Illumina HumanCytoSNP - 300K	dn	moderate ID, speech delay, obesity, facial dysmorphism (blepharophimosis)
OL/2202	F	5	Translocation	46,XX,t(1;14)(q32;q22)	Agilent 60K	dn	Severe ID, absent speech, sterotypy, epileptic encephlopathy, constipation, myopia
AS/0301	M	7	Translocation	46,XY,t(5;6)(q31.3;p22.1)	Agilent 60K	dn	Learning difficulties (delay in thin motor skills, dyspraxia), behavioral troubles, sleeping difficulties, strabismus, hyperlaxity
JP/2301	M	14	Translocation	46,XY,t(9;18)(p21;q12)	Agilent 244K	dn	Goldenhar syndrome
NM/0201	M	7	Translocation	46,XY,t(1;2)(p13.2;q31.2)	Agilent 180K avec focus chrX	dn	ID, speech delay, hypotonia, anxiety
VD/2401	F	44	Inversion	46,XX,inv(7)(q22q34)	Agilent 180K	familial	Mandibulo-facial dysostosis, toes syndactyly, hearing loss. Inversion transmitted to her son presenting a more severe phenotype
AL/2402	M	11	Inversion	46,XY,inv(3)(p24q25.2)	Agilent 60K	dn	Macrocrania, overweight, ID, sleeping apnea, encopresia, asthme, corpus callosum agenesis, brachydactyly, abnormal palmar creases, asthma

MD/2203	F	11	CCR	46,XX,t(1;5;3)(p21;q?34;p24)	Agilent 44K	dn	Severe ID (walking at 9 years old), absent speech, autistic spectrum disorder, epilepsy, facial dysmorphism, constipation
JB/1602	F	31	Translocation	46,XX,t(2;16)(p25.3;q12.2)	Illumina HumanCytoSNP-12v2 300K	dn	Mild ID, repeated miscarriages
EC/2302	F	15	Translocation	46,XX,t(4;14)(q24;q32.2)	Agilent 105K	dn	mild ID, psychiatric troubles, facial dysmorphism
TP/0801	M	2	Translocation	46,XY,t(1;8)(p21;q21.1)	Agilent 60K	dn	ID, abnormal brain MRI : pachygyria mainly in anterior regions and vermian atrophy, axial hypotonia, peripheral spasticity
DM/0109	M	18	Translocation	46,XY,t(1;19)(p36;q13.1)	Agilent 180K	dn	Mild ID, learning difficulties, dyslexia, behavioral troubles (anxiety)
BA/2303	M	21	Translocation	46,XY,t(2;8)(p16.3;p11.23)	Agilent 105K	dn	Mild ID, behavioral troubles, hyperactivity, cryptorchidism
JE/1401	F	15	Translocation	46,X,t(X;10)(q?27;q?23)	Agilent 105K	dn	ID, feeding behavior troubles, facial dysmorphism, hands and feet anomalies, advenced menarche, obesity
MG/1801	F	7	Translocation	46,XX,t(8;9)(p21.2;q34)	Agilent 180K	dn	Macrocephaly, facial dysmorphism, motor delay, learning difficulties, hypoglycemia
EM/2403	F	8	Translocation	46,XX,t(4;19)(q33;p13.2)	Agilent 60K	dn	Fetal macrosomia, post-natal overgrowth, macrocephaly, moderate ID, speech delay, facial dysmorphism (hypertelorism), Chiari type 1 malformation, nystagmus, strabismus, fetal pads
BM/1603	F	3	Translocation	46,X,t(X;13)(p22.1;q22)	Illumina HumanCytoSNP12v2 - 220K	dn	Moderate ID, facial dysmorphism (narrow palpebral fissures, posteriorly rotated ears), philtrum angioma, atrial septal defect, patent ductus arteriosus, cerebral ventricular dilatation, bilateral single palmar crease, fetal pads
CF/2304	M	30	CCR	46,XY,t(2;15;8;10)(q32.3;q26;q21.3;q23)	Agilent 105K	dn	Mild ID, speech delay, major atopic dermatitis
ML/1402	F	3	Translocation	46,XX,t(14;20)(p13;q13.2)	Illumina HumanCytoSNP-12v2.1 - 300K	dn	ID, speech delay, synophris, café-au-lait spot
KT/1403	M	8	CCR	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)	Affymetrix CYTOSCAN HD - SNP	dn	Macrocephaly, ID, psychomotor delay, speech delay, left club-foot, café-au-lait spot
AL/1802	F	13	Translocation	46,XX,t(6;14)(p12;q21)	Agilent 180K	dn	Growth retardation, dyspraxia, facial dysmorphism, sandal gap, camptodactyly
MC/1803	M	4	Translocation	46,XY,t(7;10)(q21.1;q26.1)	Agilent 60K	dn	Microcephaly, severe ID, absent speech,facial dysmorphism (short philtrum, everted lower lip, flat face) strabismus
GS/2305	M	9	Translocation	46,XY,t(5;12)(q34;q23)	Agilent 105K	familial	Immune deficiency (alymphopénia B) hearing loss, cochlear malformation. Translocation transmitted to his son, with the same phenotype.
OP/0701	F	19	Translocation	46,XX,t(3;14)(q26.3;q32.3)	Agilent 180K	dn	Moderate ID, speech dealy, behavioral troubles (anxiety), strabismus
JB/1404	F	2	Translocation	46,XX,t(10;12)(q22.2;q24.3)	Agilent 60K	dn	Post-natal micocephaly, ID, hypertonia, stereotypy, hyperlactacidemia
MD/0110	F	14	Translocation	mos 46,X,t(X;18)(q28;q11.2)/46,XX	Agilent 180K	dn	ID, learning difficulties, speech delay, behavioral troubles (anxiety), facial dysmorphism, advanced menarche
JJ/0402	M	27	Translocation	46,XY,t(3;6)(q21.3;p21.33)	Agilent 44K	dn	ID, hypotonia, facial dysmorphism, behavioral troubles (anxiety, tamper tantrum)
MCP/1105	F	16	Translocation	46,XX,t(2;15)(q32.2;q26.1)	Illumina Human OmniExpress24 - 715K	dn	Pyelic dilatation, ureteral duplication, bipartite uterus, evolutive scoliosis, myopia, strabismus
BM/1201	M	7	CCR	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)	Agilent Whole Genome ISCA 60K	dn	Growth retardation, ID, speech delay
AB/1604	M	3	Translocation	46,XY,t(1;2)(p13;p13)	Illumina SNP-array 700K	dn	ID, speech delay,plexus choroid cysts, pericerebral area enlargement
AB/2404	F	8	Translocation	46,XX,t(7;18)(q21;q21)	Agilent 60K	dn	Growth retardation, post-natal microcephaly, moderate ID, behavioral troubles (hyperactivity, aggressivity, sleeping difficulties), enuresia, facial dysmorphism (asymmetric); angiomas

Supplementary Table S1: Patients phenotype and karyotype
M: male; F: female; CCR: complex chromosomal rearrangement; dn: de novo; mat: maternally inherited; IUGR: intra-uterine growth retardation; ID: intellectual disability; OFC: occipito-frontal circumference