

Supplementary Table S7. Information of the pre-published CNV database.

Common recurrent rearrangements associated with genomic disorders							
Cytoband	Status	Syndrome	Causative gene	Phenotype	Location (hg19)	Size (Mb)	Reference (OMIM/PMID)
1q21.1	DEL	Thrombocytopenia-absent radius syndrome	<i>RBM8A</i>	reduction in the number of platelets and absence of the radius; skeletal anomalies	1:144112610-144639480	0.53	274000/17236129
1q21.1	DEL	Chromosome 1q21.1 deletion syndrome	<i>GJA5</i>	mental retardation; microcephaly; cardiac abnormalities; cataracts	1:146512930-147737500	1.22	612474/16906162,18784092
1q21.1	DUP	Chromosome 1q21.1 duplication syndrome	.	mental retardation; autism; congenital anomalies; macrocephaly	1:146512930-147737500	1.22	612475/19029900,18784092
3q29	DEL	Chromosome 3q29 microdeletion syndrome	<i>PAK2,DLG1</i>	mental retardation; dysmorphic facial; autism	3:195672229-197497869	1.83	609425/16906162,18784092
3q29	DUP	Chromosome 3q29 microduplication syndrome	<i>PAK2,DLG1</i>	mental retardation; dysmorphic features	3:195672229-197497869	1.83	611936/24838842
5q35	DEL	Sotos syndrome 1	<i>NSD1</i>	excessively rapid growth; advanced bone age; craniofacial dysmorphic features including macrocephaly, and learning difficulties	5:175130402-177456545	2.33	117550/11896389,19844260

5q35	DUP	5q35 duplication	<i>NSD1</i>	short stature; microcephaly; speech delay; brachydactyly; delayed bone age; intellectual disability supravalvular aortic stenosis (SVAS); mental retardation; distinctive facial features	5:175130402- 177456545	2.33	NA/19844260,2336 9838
7q11.23	DEL	Williams-Beuren syndrome	<i>ELN,GTF21</i>	speech delay and mild craniofacial anomalies; increased incidence of congenital anomalies such as heart defects, diaphragmatic hernia, and cryptorchidism	7:72332743- 74616901	2.28	194050/12952863,1 6236740
7q11.23	DUP	Chromosome 7q11.23 duplication syndrome	<i>ELN</i>	congenital heart malformations congenital heart malformations; characteristic facial phenotype including a prominent forehead and arched eyebrows.	7:72332743- 74616901	2.28	609757/16236740
8p23.1	DEL	8p23.1 deletion	<i>GATA4</i>	behavioral and neurodevelopmental	8:8119295- 11765719	3.65	NA/10090897
8p23.1	DUP	8p23.1 duplication	<i>GATA4</i>		8:8119295- 11765719	3.65	NA/17940555
10q22-q23	DEL	10q22-q23 deletion	<i>NRG3,GRID 1</i>		10:81671774- 88103093	6.32	NA/17436248

15q11-q13	DEL	Angelman syndrome	<i>UBE3A</i>	abnormalities mental retardation; movement or balance disorder; typical abnormal behaviors; severe limitations in speech and language diminished fetal activity; obesity; muscular hypotonia; mental retardation; short stature; hypogonadotropic hypogonadism; small hands and feet autism, mental retardation, ataxia, seizures, developmental delays, and behavioral problems	15:23758390- 28557186	4.79	105830/8988172,14 508708,18500341
15q11-q13	DEL	Prader-Willi syndrome	<i>SNRPN,NDN</i>	mild to moderate mental retardation or learning difficulties, or may have no cognitive deficits; Some individuals have epilepsy	15:23758390- 28557186	4.79	176270/14508708,1 8500341
15q11-q13	DUP	chromosome 15q11-q13 duplication syndrome	.	mental retardation; movement or balance disorder; typical abnormal behaviors; severe limitations in speech and language diminished fetal activity; obesity; muscular hypotonia; mental retardation; short stature; hypogonadotropic hypogonadism; small hands and feet autism, mental retardation, ataxia, seizures, developmental delays, and behavioral problems	15:23758390- 28557186	4.79	608636/9106540,19 166990
15q13.3	DEL	Chromosome 15q13.3 microdeletion syndrome	<i>CHRNA7</i>	mental retardation; movement or balance disorder; typical abnormal behaviors; severe limitations in speech and language diminished fetal activity; obesity; muscular hypotonia; mental retardation; short stature; hypogonadotropic hypogonadism; small hands and feet autism, mental retardation, ataxia, seizures, developmental delays, and behavioral problems	15:30769995- 32701482	1.93	612001/18278044,1 8179902
15q13.3	DUP	Chromosome 15q13.3	<i>CHRNA7</i>	mental retardation; movement or balance disorder; typical abnormal behaviors; severe limitations in speech and language diminished fetal activity; obesity; muscular hypotonia; mental retardation; short stature; hypogonadotropic hypogonadism; small hands and feet autism, mental retardation, ataxia, seizures, developmental delays, and behavioral problems	15:30769995-	1.93	NA/18278044,1817

		duplication		epilepsy; variable facial and digital dysmorphisms	32701482		9902
15q24	DEL	Witteveen-Kolk syndrome	<i>SIN3A</i>	developmental delay, short stature, hypotonia, digital abnormalities, joint laxity, genital abnormalities, and characteristic facial features	15:74377174-761622772	1.79	613406/17360722
15q24	DUP	Chromosome 15q24 duplication	.	developmental delay, short stature, hypotonia, digital abnormalities, joint laxity, genital abnormalities, and characteristic facial features	15:74377174-761622772	1.79	NA/19557438
15q25.2	DEL	Chromosome 15q25.2 microdeletion syndrome	.	congenital diaphragmatic hernia; cognitive deficits; possibly Diamond--Blackfan anaemia	/	3.2	NA/20921022
16p13.11	DEL	Chromosome 16p13.11 deletion	.	developmental disabilities	16:15504454-16284248	0.78	NA/19843651,17704777
16p13.11	DUP	Chromosome 16p13.11 duplication	.	autism and/or mental retardation	16:15504454-16284248	0.78	NA/17480035,18550696
16p11.2	DEL	Chromosome 16p11.2 deletion syndrome	<i>SH2B1</i>	severe early-onset obesity; developmental delay	16:28100000-34600000	0.22	613444/19966786,20130649

16p11.2	DEL	Chromosome 16p11.2 deletion syndrome	.	autism spectrum disorder; developmental delay; severe early-onset obesity	16:29501198-30202572	0.7	611913/19966786,20130649
16p11.2	DUP	Chromosome 16p11.2 duplication syndrome	.	autism spectrum disorder	16:29501198-30202572	0.7	614671/18184952,19914906
16p12.1	DEL	Chromosome 16p12.1 deletion syndrome	.	developmental delay; learning disability; craniofacial dysmorphism; congenital heart defects	/	0.54	136570/229709
16p12.2-p11.2	DEL	Chromosome 16p12.2-p11.2 deletion syndrome	.	dysmorphic facial features; feeding difficulties; recurrent ear infections; developmental delay; cognitive impairment	16:21613956-29042192	7.43	612604/17704777
17p12	DUP	Charcot-Marie-Tooth disease, type 1A	<i>PMP22</i>	abnormalities of peripheral nervous system	17:13968607-15434038	1.47	118220/1677316
17p12	DEL	Chromosome 17p12 deletion	<i>HNPP</i>	abnormalities of peripheral nervous system	17:13968607-15434038	1.47	162500/8422677
17p11.2	DEL	Smith-Magenis syndrome	<i>RAI1</i>	impaired cognitive and adaptive functioning; short stature; ophthalmological, and otolaryngological anomalies; hearing impairment; abnormal	17:16706021-20482061	3.78	182290/14614393

17p11.2	DUP	Potocki-Lupski syndrome	<i>RAI1</i>	EEG; scoliosis developmental disorder characterized by hypotonia, failure to thrive, mental retardation, pervasive developmental disorders, and congenital anomalies cafe-au-lait spots; Lisch nodules in the eye; fibromatous tumors of the skin; dysmorphism;	17:16706021- 20482061	3.78	610883/17357070
17q11.2	DEL	NF1 microdeletion syndrome	<i>NF1</i>	learning disabilities/mental retardation nondiabetic renal disease resulting from abnormal renal development;	17:29162822- 30218667	1.06	162200/10631140
17q12	DEL	Renal cysts and diabetes syndrome	<i>HNF1B</i>	diabetes; mild to moderate mental retardation; epilepsy; focal cortical dysplasia	17:34907366- 36076803	1.17	137920/17924346,1 9844256
17q12	DUP	Chromosome 17q12 duplication syndrome	.	moderate to severe intellectual disability; hypotonia; friendly demeanor; highly	17:34907366- 36076803	1.17	614526/17924346,1 9844256
17q21.31	DEL	Koolen-De Vries syndrome	<i>KANSL1</i>		17:43632466- 44210205	0.58	610443/16906162,1 6906164

17q21.31	DUP	Chromosome 17q21.31 duplication syndrome	<i>KANSL1?</i>	distinctive facial features behavioural problems; poor social interaction	17:43632466-44210205	0.58	613533/19502243
17q23.1-q23.2	DEL	Chromosome 17q23.1-q23.2 deletion syndrome	<i>TBX2, TBX4</i>	mild to moderate developmental delay;	17:57600000-61100000	3.5	613355/20206336
17q23.1-q23.2	DUP	Chromosome 17q23.1-q23.2 duplication syndrome	<i>TBX2, TBX4</i>	mild to moderate developmental delay; hypocalcemia arising from parathyroid hypoplasia, thymic hypoplasia, and outflow tract defects of the heart Microcephaly; Micrognathia; Congenital cardiac malformations;	17:57600000-61100000	3.5	NA/20206336
22q11.2	DEL	DiGeorge syndrome	<i>TBX1</i>	Delayed psychomotor development; Learning disabilities; Mental retardation; facial dysmorphic features; rematurity, prenatal and postnatal growth delay;	22:18546349-22336469	3.79	188400/11925570,14585638
22q11.2	DUP	Chromosome 22q11.2 microduplication syndrome	<i>TBX1</i>	developmental delay; mild skeletal abnormalities	22:18546349-22336469	3.79	608363/14526392
22q11.2	DEL	Chromosome 22q11.2 deletion syndrome, distal	.		22:22115848-23696229	1.58	611867/18179902

22q11.2	DUP	Chromosome 22q11.2 microduplication	.	developmental delay	22:22115848-23696229	1.58	NA/18414210
22q13.3	DEL	Phelan-McDermid syndrome	<i>SHANK3</i>	neonatal hypotonia; global developmental delay; normal to accelerated growth; absent to severely delayed speech; autistic behavior; minor dysmorphic features	22:51045516-511878440	0.14	606230/18505557
Xp11.2-p11.23	DUP	Xp11.2-p11.23 duplication	.	mental retardation; speech delay; EEG anomalies	/	4.5	NA/19716111

Common nonrecurrent rearrangements associated with genomic disorders

Cytoband	CNV	Syndrome	Causative gene	Reference (OMIM/PMID)
1p36	DEL/DUP	chromosome 1p36 microdeletion/microduplication syndrome	<i>GABRD,PRKCZ</i>	613060/20635359
1q32-q41	DEL	van der Woude syndrome	<i>IRF6</i>	119300/12219090
1q41	DEL	1q41-q42 microdeletion syndrome	<i>DISP1</i>	NA/17873649
1q43-q44	DEL	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	<i>AKT3</i>	615937/17668379
1q21.1	DEL/DUP	chromosome 1q21.1 microdeletion/microduplication syndrome	<i>GJA5</i>	121013/15117819
2p24.1	DEL	Feingold syndrome 1	<i>MYCN</i>	164280/15821734
2p16.3	DEL/DUP	Pitt-Hopkins-like syndrome 2	<i>NRXN1</i>	614325/23207424
2q22.3	DEL	Mowat-Wilson syndrome	<i>ZEB2</i>	235730/11595972

2q23.1	DEL	chromosome 2q23.1 deletion syndrome	<i>MBD5</i>	156200/19809484
4p16.3	DEL	Wolf-Hirschhorn syndrome	.	194190/12563561
4q21	DEL	Chromosome 4q21 deletion syndrome	<i>PRKG2,RASGEF1B</i>	613509/20522426
5p15.2	DEL	Cri-du-chat syndrome	<i>TERT</i>	123450/11238681
5p13.1	DEL	Cornelia de Lange syndrome 1	<i>NIPBL</i>	608667/15146185;15821734
5p13.1	DUP	Chromosome 5p13 duplication syndrome	<i>NIPBL</i>	613174/19052029
5q14.3	DEL	Chromosome 5q14.3 deletion syndrome	<i>MEF2C</i>	613443/19592390
6p21	DEL	Cleidocranial dysplasia, forme fruste, with brachydactyly	<i>RUNX2</i>	600211;119600/9182765
7p13	DEL/DUP	Greig cephalopolysyndactyly syndrome	<i>GLI3</i>	175700/1650914
7p21	DEL	Saethre-Chotzen syndrome with or without eyelid anomalies	<i>TWIST1</i>	101400;601622/1650914
7q21.3	DEL	Split hand/foot malformation 1	<i>SHFM1</i>	183600/8023840
7q36.3	DEL	Currarino syndrome	<i>MNX1</i>	176450/10631160
8p23.1-p22	DEL/DUP	Atrial septal defect	<i>GATA4</i>	600576/12845333
8q12.1	DEL	CHARGE syndrome	<i>CHD7</i>	214800/15300250
8q13.3	DEL/DUP	Branchiootorenal syndrome 1, with or without cataracts	<i>EYA1</i>	113650/15146463
8q21.11	DEL	Chromosome 8q21.11 deletion syndrome	<i>ZFHX4</i>	614230/21802062
8q24.12	DEL	Trichorhinophalangeal syndrome	<i>TRPS1,EXT1</i>	190351;150230/11112658
9q22.3	DEL	Basal cell nevus syndrome	<i>PTCH1</i>	109400/8755929
9q22.3	DUP	Chromosome 9q22.3 duplication syndrome	<i>PTCH1</i>	NA/18830227
9q34.1	DEL	Nail-patella syndrome	<i>LMX1B</i>	161200/9590287
9q34.3	DEL	Kleefstra syndrome 1	<i>EHMT1</i>	610253/16826528
10p14	DEL	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	<i>GATA3</i>	146255/10935639

10q23.31	DEL	Cowden syndrome 1	<i>PTEN</i>	158350/9259288
10q24.1	DUP	Split-hand/foot malformation 3, gene duplication syndrome	<i>FBXW4</i>	246560;600095/12913067
11p15	DEL	Beckwith-Wiedemann syndrome	<i>H19, ICRI, KCNQ1OT1, CDKN1C</i>	130650;600856,103280,604115/24154661
11p13	DEL	Wilms tumor, aniridia, genitourinary anomalies and mental retardation syndrome	<i>PAX6, WT1</i>	194072;607108/12386836
11p11.2	DEL	Potocki-Shaffer syndrome	<i>ALX4, EXT2</i>	601224/8882796
11q23	DEL	Jacobsen syndrome	<i>BSX, NRG1</i>	147791/18855024
12p	DUP	Pallister-Killian syndrome	.	601803/2002482
12q14	DEL/DUP	Buschke-Ollendorff syndrome	<i>LEMD3</i>	166700/15489854
12q24.1	DEL	Holt-Oram syndrome	<i>TBX5</i>	142900;601620/8988165
12q24.1	DEL/DUP	Noonan syndrome 1	<i>PTPNL1</i>	163950/18348260
12q24.1	DEL	Ulnar-mammary syndrome	<i>TBX3</i>	181450/16896345
14q12	DEL	Rett syndrome, congenital variant	<i>FOXP1</i>	613454;164874/18571142
14q13	DEL	Chromosome 14q12 deletion syndrome	<i>NKX2-1, PAX9</i>	NA/22636604
15q11.2	DEL/DUP	Chromosome 15q11.2 microdeletion/microduplication syndrom	<i>NIPAI</i>	608145/21187176
15q13.3	DEL/DUP	Chromosome 15q13.3 microdeletion/microduplication syndrom	<i>CHRNA7</i>	612001/24700535
15q23.1 15q26	or DEL	Hernia, congenital diaphragmatic 1	<i>CHD2, NR2F2</i>	142340/15750894
16p13.3	DEL	Rubinstein-Taybi syndrome 1	<i>CREBBP</i>	180849/10699051;8988167
16p13.3	DUP	Chromosome 16p13.3 duplication syndrome	<i>CREBBP</i>	NA/19833603
16p11.2	DEL	Chromosome 16p11.2 deletion syndrome	<i>TBX6</i>	602427/19770079

16q12.1	DEL	Townes-Brocks syndrome 1	<i>SALL1</i>	107480/9425907
16q24	DEL	Alveolar capillary dysplasia with misalignment of pulmonary veins	<i>FOXF1</i>	265380/19500772
17p13.3	DEL	Miller-Dieker lissencephaly syndrome	<i>PAFAH1B1(LIS1), YWAHE</i>	247200/3050093;19584063
17p13.3	DUP	Chromosome 17p13.3 duplication syndrome	<i>PAFAH1B1(LIS1), YWAHE</i>	613215/19136950
17q24.3-q25.1	DEL/DUP	Acampomelic campomelic dysplasia	<i>SOX9</i>	608160/8001137
18q21.1	DEL	Pitt-Hopkins syndrome	<i>TCF4</i>	610954/17436255
20p12	DEL	Alagille syndrome 1	<i>JAG1</i>	118450/11058898
20q13.2	DEL	Duane-radial ray syndrome	<i>SALL4</i>	607323/12393809
22q11.2	DUP	Cat eye syndrome	.	115470/3961499
22q12.2	DEL	Neurofibromatosis, type 2	<i>NF2</i>	101000;607379/19476995
22q13.3	DEL	Phelan-McDermid syndrome	<i>SHANK3 /PROSAP2</i>	606232/17173049
Xpter-p22.32	DEL	Leri-Weill dyschondrosteosis	<i>SHOX</i>	127300/9140395;16826534
Xp22.31	DEL	Kallmann syndrome 1	<i>KALI</i>	308700/3007328
Xp22	DEL	Epileptic encephalopathy, early infantile, 2	<i>CDKL5</i>	300672/19471977
Xp22.2	DEL	Opitz GBBB syndrome, type I	<i>MID1</i>	300000/17221865
Xp21.2	DEL/DUP	Duchenne muscular dystrophy	<i>DMD</i>	300377/2573997
Xp21.3	DEL	Mental retardation, X-linked 21/34	<i>IL1RAPL1</i>	300143/15300857
Xp21.3	DEL	Lissencephaly, X-linked 2	<i>ARX</i>	300215/14722918
Xp21.3-p21.2	DEL	Adrenal hypoplasia, congenital	<i>NR0B1</i>	300200/10361383
Xp21.3-p21.2	DEL	46XY sex reversal 2, dosage-sensitive	<i>NR0B1</i>	300018/7951319

Xp11.4	DEL	Ornithine transcarbamylase deficiency	<i>OTC</i>	311250/8786061
Xp11.4	DEL	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	<i>CASK</i>	300749/17221865
Xp11.23	DEL	Norrie disease	<i>NDP</i>	310600/19165920
Xp11.23	DEL	Goltz syndrome	<i>PORCN</i>	305600/17546030;17546031
Xq22.2	DUP	Pelizaeus-Merzbacher disease	<i>PLP1</i>	312080/15627202
Xq22.3-q23	DEL	Lissencephaly, X-linked	<i>DCX</i>	300067/9489699;9489700
Xq25	DEL	Lymphoproliferative syndrome, X-linked, 1	<i>SH2D1A</i>	308240/11049992
Xq27.1	DEL/DUP	Mental retardation, X-linked, with isolated growth hormone deficiency	<i>SOX3</i>	300123/12428212
Xq28	DEL	Rett syndrome	<i>MECP2</i>	312750/12615169
Xq28	DUP	Mental retardation, X-linked syndromic, Lubs type	<i>MECP2</i>	300260/10398236
