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	*RBM8A*	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	*GJA5*	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	*PAK2,DLG1*	mental retardation; dysmorphic facial; autism	3:195672229-197497869	1.83	609425/16906162,18784092
3q29	DUP	Chromosome 3q29 microduplication syndrome	*PAK2,DLG1*	mental retardation; dysmorphic features	3:195672229-197497869	1.83	611936/24838842
5q35	DEL	Sotos syndrome 1	*NSD1*	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	*NSD1*	short stature; microcephaly; speech delay; brachydactyly; delayed bone age; ntellectual disability	5:175130402-177456545	2.33	NA/19844260,23369838
7q11.23	DEL	Williams-Beuren syndrome	*ELN,GTF21*	supravalvular aortic stenosis (SVAS); mental retardation; distinctive facial features	7:72332743-74616901	2.28	194050/12952863,16236740
7q11.23	DUP	Chromosome 7q11.23 duplication syndrome	*ELN*	speech delay and mild craniofacial anomalies; increased incidence of congenital anomalies such as heart defects, diaphragmatic hernia, and cryptorchidism	7:72332743-74616901	2.28	609757/16236740
8p23.1	DEL	8p23.1 deletion	*GATA4*	congenital heart malformations	8:8119295-11765719	3.65	NA/10090897
8p23.1	DUP	8p23.1 duplication	*GATA4*	congenital heart malformations; characteristic facial phenotype including a prominent forehead and arched eyebrows.	8:8119295-11765719	3.65	NA/17940555
10q22-q23	DEL	10q22-q23 deletion	*NRG3,GRID1*	behavioral and neurodevelopmental	10:81671774-88103093	6.32	NA/17436248

15q11-q13	DEL	Angelman syndrome	*UBE3A*	abnormalities mental retardation; movement or balance disorder; typical abnormal behaviors; severe limitations in speech and language	15:23758390-28557186	4.79	105830/8988172,14508708,18500341
15q11-q13	DEL	Prader-Willi syndrome	*SNRPN,NDN*	diminished fetal activity; obesity; muscular hypotonia; mental retardation; short stature; hypogonadotropic hypogonadism; small hands and feet	15:23758390-28557186	4.79	176270/14508708,18500341
15q11-q13	DUP	chromsome 15q11-q13 duplication syndrome	.	autism, mental retardation, ataxia, seizures, developmental delays, and behavioral problems	15:23758390-28557186	4.79	608636/9106540,19166990
15q13.3	DEL	Chromosome 15q13.3 microdeletion syndrome	*CHRNA7*	mild to moderate mental retardation or learning difficulties, or may have no cognitive deficits; Some individuals have epilepsy	15:30769995-32701482	1.93	612001/18278044,18179902
15q13.3	DUP	Chromosome 15q13.3	*CHRNA7*	mental retardation;	15:30769995-	1.93	NA/18278044,1817

		duplication		epilepsy; variable facial and digital dysmorphisms	32701482		9902
15q24	DEL	Witteveen-Kolk syndrome	*SIN3A*	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	*SH2B1*	severe early-onset obesit; 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 dysmorphology; 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	*PMP22*	abnormalities of peripheral nervous system	17:13968607-15434038	1.47	118220/1677316
17p12	DEL	Chromosome 17p12 deletion	*HNPP*	abnormalities of peripheral nervous system	17:13968607-15434038	1.47	162500/8422677
17p11.2	DEL	Smith-Magenis syndrome	*RAI1*	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	*RAI1*	EEG; scoliosis developmental disorder characterized by hypotonia, failure to thrive, mental retardation, pervasive developmental disorders, and congenital anomalies	17:16706021-20482061	3.78	610883/17357070
17q11.2	DEL	NF1 microdeletion syndrome	*NF1*	cafe-au-lait spots; Lisch nodules in the eye; fibromatous tumors of the skin; dysmorphism; learning disabilities/mental retardation	17:29162822-30218667	1.06	162200/10631140
17q12	DEL	Renal cysts and diabetes syndrome	*HNF1B*	nondiabetic renal disease resulting from abnormal renal development; diabetes;	17:34907366-36076803	1.17	137920/17924346,19844256
17q12	DUP	Chromosome 17q12 duplication syndrome	.	mild to moderate mental retardation; epilepsy; focal cortical dysplasia	17:34907366-36076803	1.17	614526/17924346,19844256
17q21.31	DEL	Koolen-De Vries syndrome	*KANSL1*	moderate to severe intellectual disability; hypotonia; friendly demeanor; highly	17:43632466-44210205	0.58	610443/16906162,16906164

17q21.31	DUP	Chromosome 17q21.31 duplication syndrome	*KANSL1?*	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	*TBX2,TBX4*	mild to moderate developmental delay;	17:57600000-61100000	3.5	613355/20206336
17q23.1-q23.2	DUP	Chromosome 17q23.1-q23.2 duplication syndrome	*TBX2,TBX4*	mild to moderate developmental delay;	17:57600000-61100000	3.5	NA/20206336
22q11.2	DEL	DiGeorge syndrome	*TBX1*	hypocalcemia arising from parathyroid hypoplasia, thymic hypoplasia, and outflow tract defects of the heart	22:18546349-22336469	3.79	188400/11925570,14585638
22q11.2	DUP	Chromosome 22q11.2 microduplication syndrome	*TBX1*	Microcephaly; Micrognathia; Congenital cardiac malformations; Delayed psychomotor development; Learning disabilities; Mental retardation;	22:18546349-22336469	3.79	608363/14526392
22q11.2	DEL	Chromosome 22q11.2 deletion syndrome, distal	.	facial dysmorphic features; rematurity, prenatal and postnatal growth delay; developmental delay; mild skeletal abnormalities	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	*SHANK3*	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	*GABRD,PRKCZ*	613060/20635359
1q32-q41	DEL	van der Woude syndrome	*IRF6*	119300/12219090
1q41	DEL	1q41-q42 microdeletion syndrome	*DISP1*	NA/17873649
1q43-q44	DEL	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	*AKT3*	615937/17668379
1q21.1	DEL/DUP	chromosome 1q21.1 microdeletion/microduplication syndrome	*GJA5*	121013/15117819
2p24.1	DEL	Feingold syndrome 1	*MYCN*	164280/15821734
2p16.3	DEL/DUP	Pitt-Hopkins-like syndrome 2	*NRXN1*	614325/23207424
2q22.3	DEL	Mowat-Wilson syndrome	*ZEB2*	235730/11595972

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

10q23.31		DEL	Cowden syndrome 1	*PTEN*	158350/9259288
10q24.1		DUP	Split-hand/foot malformation 3, gene duplication syndrome	*FBXW4*	246560;600095/12913067
11p15		DEL	Beckwith-Wiedemann syndrome	*H19, ICR1, KCNQ1OT1, CDKN1C*	130650;600856,103280,604115/24154661
11p13		DEL	Wilms tumor, aniridia, genitourinary anomalies and mental retardation syndrome	*PAX6,WT1*	194072;607108/12386836
11p11.2		DEL	Potocki-Shaffer syndrome	*ALX4,EXT2*	601224/8882796
11q23		DEL	Jacobsen syndrome	*BSX,NRGN*	147791/18855024
12p		DUP	Pallister-Killian syndrome	.	601803/2002482
12q14		DEL/DUP	Buschke-Ollendorff syndrome	*LEMD3*	166700/15489854
12q24.1		DEL	Holt-Oram syndrome	*TBX5*	142900;601620/8988165
12q24.1		DEL/DUP	Noonan syndrome 1	*PTPNL1*	163950/18348260
12q24.1		DEL	Ulnar-mammary syndrome	*TBX3*	181450/16896345
14q12		DEL	Rett syndrome, congenital variant	*FOXG1*	613454;164874/18571142
14q13		DEL	Chromosome 14q12 deletion syndrome	*NKX2-1,PAX9*	NA/22636604
15q11.2		DEL/DUP	Chromosome 15q11.2 microdeletion/microduplication syndrom	*NIPA1*	608145/21187176
15q13.3		DEL/DUP	Chromosome 15q13.3 microdeletion/microduplication syndrom	*CHRAN7*	612001/24700535
15q23.1	or 15q26	DEL	Hernia, congenital diaphragmatic 1	*CHD2*, *NR2F2*	142340/15750894
16p13.3		DEL	Rubinstein-Taybi syndrome 1	*CREBBP*	180849/10699051;8988167
16p13.3		DUP	Chromosome 16p13.3 duplication syndrome	*CREBBP*	NA/19833603
16p11.2		DEL	Chromosome 16p11.2 deletion syndrome	*TBX6*	602427/19770079

16q12.1	DEL	Townes-Brocks syndrome 1	*SALL1*	107480/9425907
16q24	DEL	Alveolar capillary dysplasia with misalignment of pulmonary veins	*FOXF1*	265380/19500772
17p13.3	DEL	Miller-Dieker lissencephaly syndrome	*PAFAH1B1(LIS1)*, *YWAHE*	247200/3050093;19584063
17p13.3	DUP	Chromosome 17p13.3 duplication syndrome	*PAFAH1B1(LIS1)*, *YWAHE*	613215/19136950
17q24.3-q25.1	DEL/DUP	Acampomelic campomelic dysplasia	*SOX9*	608160/8001137
18q21.1	DEL	Pitt-Hopkins syndrome	*TCF4*	610954/17436255
20p12	DEL	Alagille syndrome 1	*JAG1*	118450/11058898
20q13.2	DEL	Duane-radial ray syndrome	*SALL4*	607323/12393809
22q11.2	DUP	Cat eye syndrome	.	115470/3961499
22q12.2	DEL	Neurofibromatosis, type 2	*NF2*	101000;607379/19476995
22q13.3	DEL	Phelan-McDermid syndrome	*SHANK3 /PROSAP2*	606232/17173049
Xpter-p22.32	DEL	Leri-Weill dyschondrosteosis	*SHOX*	127300/9140395;16826534
Xp22.31	DEL	Kallmann syndrome 1	*KAL1*	308700/3007328
Xp22	DEL	Epileptic encephalopathy, early infantile, 2	*CDKL5*	300672/19471977
Xp22.2	DEL	Opitz GBBB syndrome, type I	*MID1*	300000/17221865
Xp21.2	DEL/DUP	Duchenne muscular dystrophy	*DMD*	300377/2573997
Xp21.3	DEL	Mental retardation, X-linked 21/34	*IL1RAPL1*	300143/15300857
Xp21.3	DEL	Lissencephaly, X-linked 2	*ARX*	300215/14722918
Xp21.3-p21.2	DEL	Adrenal hypoplasia, congenital	*NR0B1*	300200/10361383
Xp21.3-p21.2	DEL	46XY sex reversal 2, dosage-sensitive	*NR0B1*	300018/7951319

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