Table S1. Limited overview of most common recurrent microdeletion and -duplication syndromes

Chromosomal location	Rearrangement	Main phenotypic features	MIM#/reference
1q21.1	Deletion	Microcephaly, ID, ocular anomalies, cardiac problems	612474
	Deletion (additional RBM8A SNV) (thrombocytopenia-absent radius, TAR)	Thrombocytopenia, aplasia of radii (long forearm bones), other skeletal defects, cardiac problems	274000
	Duplication	Macrocephaly, ID, autism, schizophrenia	612475
5q35.3	Deletion (Sotos)	Childhood overgrowth, mental retardation, facial dysmorphism, hyperinsulinemic hypoglycemia	117550
	Duplication	Short stature, microcephaly, facial dysmorphism, ID	[15]
7q11.23	Deletion (Williams-Beuren)	Facial dysmorphism, ID, cardiac problems, 'sociable phenotype'	194050
	Duplication	Facial dysmorphism, speech delay, cardiac problems, cryptorchidism	609757
8p23.1	Deletion	Cardiac problems, diaphragmatic hernia, ID	[16]
	Duplication	Cardiac problems, ID, learning difficulties, facial dysmorphism	[17]
9q34	Deletion (Kleefstra 1)	Epileptic seizures, ID, cardiac problems, facial dysmorphism	610253

	Duplication	Hyperactivity, psychomotor retardation, musculoskeletal abnormalities, facial dysmorphism	[18]
15q11.2	Paternal deletion (Prader-Willi)	Hypotonia, ID, obesity, small hands and feet, hypogonadotropic hypogonadism	176270
	Maternal deletion (Angelman)	ID, speech and language limitations, ‘happy personality’	105830
	Duplication	Autism, ID, seizures, ataxia	608636
16p11.2	Deletion	Autism, developmental delay, obesity	611913
	Duplication	Autism, attention-deficit hyperactivity disorder, facial dysmorphism	614671
17p11.2	Deletion (Smith-Magenis)	ID, facial dysmorphism, behavioral problems (anxiety, aggression, self-destructive behavior)	182290
	Duplication (Potocki-Lupski)	Hypotonia, congenital anomalies, ID	610883
17p12	Deletion (Hereditary neuropathy with liability to pressure palsies)	Neuropathy with liability to pressure palsies	600361
	Duplication (Charcot-Marie-Tooth disease type 1A)	Muscle weakness and atrophy, reduced sensation	601098
17q11.2	Deletion (NF1 microdeletion)	ID, facial dysmorphism, early-onset neurofibromas	613675
	Duplication	ID, facial dysmorphism, seizures	618874

	(NF1 microduplication)		
17q21.31	Deletion (Koolen-De Vries)	Hypotonia, ID, ‘friendly personality’, facial dysmorphism, cardiac problems, seizures	610443
	Duplication	Psychomotor delay, poor social interaction, ID, facial dysmorphism, congenital malformations	613533
22q11.2	Deletion	Cardiac problems, immunodeficiency, palatal anomalies, neuropsychiatric disease, ID	192430
	Duplication	Behavioral problems, facial dysmorphism, velopharyngeal insufficiency, ID	608363
Yq11.2	Deletion (AZFa microdeletion)	Male infertility	400042

This limited overview of recurrent microdeletion and -duplication syndromes in the human genome is based on Harel and Lupski [2] and the syndrome-specific pages of the OMIM website (Online Mendelian Inheritance in Man catalog, http://www.omim.org).