

Supplementary Data 2. Review of All Reciprocal CNV's

CNV Location	Deletion Phenotype(s)	Duplication Phenotype(s)	†Category	Citation
1q21.1	Microcephaly	Macrocephaly	Additive	Despandhe and Weiss, 2018
	Seizures		Dominant	
		Reduced corpus callosum size	Dominant	
	ID, DD, speech problems, ASD, motor impairment, hypotonia, tremor, ADHD, schizophrenia		U-shaped	
	Autism		U-shaped	Crespi et al., 2010
	Schizophrenia		Dominant	Crespi et al., 2010; Crespi et al., 2018; Golzio and Katsanis, 2013; Lupski, 2015
		Autism	Dominant	Crespi et al., 2018; Lupski, 2015; Golzio and Katsanis, 2013
	Small head size	Large head size	Additive	Golzio and Katsanis, 2013
2q11.2	ASD		U-shaped	Golzio and Katsanis, 2013
3q29	Microcephaly		U-shaped	Golzio and Katsanis, 2013; Tassano et al., 2018
7q11.23	Over friendliness, gregarious personality, relatively normal speech	ASD, expressive and receptive speech and language delay, macrocephaly	Additive	Despandhe and Weiss, 2018
	Hypothyroidism, hypercalcemia		Dominant	

		Schizophrenia Seizures		
	ID, DD, ADHD, hypotonia, anxiety disorders		U-shaped	
	Williams syndrome	Autism	Additive	Mulle et al., 2014
	Distinctive facial appearance Cardiac abnormalities Infantile hypercalcemia Growth retardation/DD	Facial dysmorphism Language and speech delay Autism behavior Epilepsy	U-shaped	Golzio and Katsanis, 2013
8p23.1	Dysmorphic features Developmental delay Heart defects		U-shaped	Golzio and Katsanis, 2013
8q24.3	ASD	ASD Epilepsy	U-shaped	Golzio and Katsanis, 2013
9q34.3	Craniofacial features Microcephaly Speech delay/DD Respiratory failure	Non-syndromic, IQ>70	U-shaped	Golzio and Katsanis, 2013
15q13.3	Aggressive and impulsive behavior		Dominant	Despandhe and Weiss, 2018
	Language delay, ID, DD, schizophrenia, ASD, seizures, ADHD		U-shaped	
	Schizophrenia		Dominant	Crespi et al., 2010
	ID Epilepsy Seizures Variable dysmorphism of the face and digits	ID Autism Recurrent ear infections, low set ears Obesity	U-shaped	Golzio and Katsanis, 2013
16p11.2	Macrocephaly	Microcephaly	Additive	Despandhe and Weiss, 2018; Lupski, 2015
	Obesity	Underweight	Additive	Despandhe and Weiss, 2018
		Schizophrenia	Dominant	
		Speech/language delay, seizures, ID, ADHD, ASD, DD, DCD, anxiety		U-shaped
	Autism	Schizophrenia		Crespi et al., 2010; Crespi et al., 2018;

				Lupski, 2015
	Large head size	Small head size	Additive	Golzio and Katsanis, 2013
	Autism		Dominant	
		Schizophrenia	Dominant	
	Obesity	Underweight	Additive	
16p13.1		Autism, Schizophrenia	Dominant	Crespi et al., 2010
	Microcephaly Severe ID Epilepsy Short stature Cardiac defects Dysmorphism	Microcephaly/macrocephaly Moderate ID Autism spectrum disorders Cardiac defects Dysmorphism	U-shaped	Golzio and Katsanis, 2013
17p11.2	Sleep disturbances Obesity	Sleep apnea Reduced weight	Additive	Golzio and Katsanis, 2013
	Smith-Magenis syndrome (SMS): dysmorphic craniofacial features, DD, severe sleep disturbances, severe behavior disturbances (tantrums, self-injury, aggression)	Potocki-Lupski syndrome (PTLS): ID, behavior disturbances, hypotonia, cognitive impairment, speech delay, autistic features	Additive	Neira-Fresneda and Potocki, 2015
17p13.3	Miller-Dieker syndrome: Lissencephaly ID Facial dysmorphism	Microcephaly Dysgenesis of the corpus callosum Cerebellar atrophy DD/ID ADHD Facial dysmorphism	U-shaped	Golzio and Katsanis, 2013
17q21.31	Mild-to-moderate intellectual disability Distinctive facial features Epilepsy Heart defects Urogenital anomalies	Severe developmental delay Microcephaly Facial dysmorphism Abnormal digits and hirsutism Failure to thrive	U-shaped	Golzio and Katsanis, 2013
22q11.2	Schizophrenia	Protective for schizophrenia	Additive	Despandhe and Weiss, 2018
	ADHD, mood and anxiety disorder, hypothyroidism,		Dominant	

	hyperthyroidism, hypocalcemia, obesity			
	ID, DD, hypotonia, seizures, ASD, language delay		U-shaped	
	Dysmorphic facial features Velocardio-facial syndrome (cleft lip/palate, velopharyngeal insufficiency) Congenital heart disease (conotruncal) Learning disabilities Hearing loss	Dysmorphic facial features Velopharyngeal insufficiency cleft lip/palate Congenital heart disease (conotruncal) intellectual disabilities Speech delay Hearing loss Failure to thrive	U-shaped	Golzio and Katsanis, 2013
22q11.21	Schizophrenia		Dominant	Crespi et al., 2010; Crespi et al., 2018
		Autism	Dominant	
22q13.3	Autism		Dominant	Crespi et al., 2010
		Schizophrenia	Dominant	

†**Additive model:** opposing phenotypes are associated with opposite levels of gene expression; **Dominant model:** gene expression in one direction is associated with a phenotype, while the opposite direction of gene expression has no effect on that trait; **U-shaped model:** identical phenotypes are associated with opposite levels of gene expression (Weiss and Despanthe, 2018)
§**Abbreviations:** ASD- Autism Spectrum Disorder; DD- Developmental Disability; ID- Intellectual Disability; ADHD- Attention Deficit Hyperactivity Disorder; DCD- Developmental Coordination Disorder

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