

Common Genetic Conditions with Psychiatric Manifestations					
Condition	Genetics	Physical Features	Medical Comorbidities	Psych/Neuro Features	Resources
22q11.2 Deletion Syndrome (DiGeorge Syndrome)	Deletion on 22q11.2	Facial dysmorphism (eg, hooded eyelids, small ears), palate abnormalities	Heart defects (tetralogy of Fallot), immune deficiency due to thymic hypoplasia; hypocalcemia from parathyroid hypoplasia	Psychosis, anxiety, ADHD, autism	International 22q Foundation www.22q.org
Trisomy 21 (Down Syndrome)	Extra copy of chromosome 21	Characteristic facial features eg, eyes that slant upward, a flat bridge of the nose, and a round face with a small chin	Heart defects, early-onset Alzheimer's	ID, anxiety, depression	National Down Syndrome Society (NDSS) www.ndss.org
Fragile X Syndrome	Inactivation of FMR1 gene due to CGG trinucleotide repeat expansion	Long face, large ears, macroorchidism (enlarged testes). Physical features are more prominent in males.	Mitral valve prolapse, joint laxity	ID, autism, ADHD, anxiety, hand-flapping (mainly in males); female carriers may exhibit mild symptoms, like anxiety or learning difficulties	National Fragile X Foundation fragilex.org
Prader-Willi Syndrome	Paternal 15q11.2-q13 loss or dysfunction	Hypotonia at birth, hyperphagia	Excessive daytime sleepiness, obesity	ID, compulsivity (eg, skin-picking, hoarding), emotional dysregulation, anxiety, depression, psychosis	Prader-Willi Syndrome Association USA pwsausa.org
Rett Syndrome	MECP2 mutation (X-linked) It only affects females, as mutations are typically lethal in males.	Gait abnormalities, stereotypic handwringing	Seizures, spasticity, autonomic dysfunction (eg, breathing irregularities, heart rate issues)	Normal development in the first year of life, followed by regression in motor skills, language development, and social functioning. Severe ID.	International Rett Syndrome Foundation rettsyndrome.org
Turner Syndrome (Monosomy X, 45,X, or mosaic Turner syndrome)	Missing/abnormal X chromosome	Short stature, webbed neck	Heart defects (eg, coarctation of the aorta), ovarian insufficiency	ADHD, anxiety, social difficulties	Turner Syndrome Society of the United States turnersyndrome.org
Williams Syndrome	Deletion on 7q11.23, which includes the elastin gene	"Elfin" facial features, short stature	Cardiac, renal, thyroid abnormalities	Mild ID. Individuals are often described as socially gregarious but anxious, with a strong affinity for music. ADHD, specific phobias, and depressive symptoms are common.	Williams Syndrome Association williams-syndrome.org

From the article:
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REFERENCE TABLE**

Treatment Approaches for Select Genetic Conditions	
Genetic Condition	Example Treatment Approaches
Fragile X Syndrome	SSRIs for anxiety; stimulants or guanfacine for ADHD; risperidone/aripiprazole for irritability; behavioral therapy and sensory supports
22q11.2 Deletion (DiGeorge)	Antipsychotics for psychosis; SSRIs for anxiety; cautious use of stimulants or non-stimulants for ADHD; coordinate with neurology/cardiology
Prader-Willi Syndrome	SSRIs for compulsivity and anxiety; behavioral interventions for hyperphagia and emotional dysregulation; structured environment with food access management
Down Syndrome	SSRIs for depression or anxiety; monitor thyroid, sleep apnea; structured behavioral interventions
Turner Syndrome	CBT for social anxiety and self-esteem; SSRIs as needed; academic and social skill supports
Rett Syndrome	Supportive behavioral interventions; seizure management; PT/OT; limited role for psychiatric meds

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