

SYNDROMES

Syndrome and Frequency	Physical Characteristics	Behavioral Issues	Medical Problems
<p>Down “Trisomy 21” 1 in 700-1000 live births</p> <p>The most common cause of intellectual disability</p>	<p>Small head with flat face, small ears and mouth, protruding tongue, upward slant to eyes, broad neck, short, hypertonia; flexible joints, single crease across center of palm</p>	<p>Often seems passive, often sociable personality, stubborn/distractible, resistant to change, require stimulation and more attention, excellent memory, early onset of Alzheimer’s (as early as 35), conduct problems</p>	<p>Congenital heart defects-50%, lack of Moro (plantar) reflex, hearing loss 66-89%, seizures-33%, vision problems, hypothyroidism, gastrointestinal problems, obesity, dry skin, dental problems, males-generally sterile, females-low fertility.</p>
<p>Fragile X 1 in 1500 males 1 in 2500 females</p> <p>The most common Hereditary cause of ID</p>	<p>Macrocephaly (large head), large ears, long narrow face, macroorchidism (large testicles), low muscle tone, hyperextensible joints, flat feet, soft skin</p>	<p>Speech pattern of short bursts, repeated phrases, verbal aggression, stalling, delayed reactions, overreactions, attention deficits, gaze aversion beginning at age 2, hyper-arousal, mouthing objects/clothes, hand-flapping</p>	<p>Mitral valve prolapse, seizures, scoliosis, sinus problems, atlantoaxial (neck) dislocation</p>
<p>Fetal Alcohol Syndrome .2-1.5 in 1000 live births</p>	<p>Short stature, long lean body, small head, abnormal facial features-thin upper lip, small eye openings</p>	<p>Poor impulse control, hyperactive, poor sucking and sleep as infant, poor coordination</p>	<p>Heart problems, kidney problems, hearing loss</p>

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<p>Tuberous Sclerosis 1 in 10,000 live births</p>	<p>Small benign tumors grow on organs, eyes, and skin. May experience none or all of the symptoms with varying degrees.</p>	<p>In some cases, neurobehavioral problems.</p>	<p>Multisystem disease that can affect brain, kidneys, heart, eyes, lungs and other organs, skin and eye lesions, seizures.</p>
<p>Rett's disorder 1 in 10,000 female births Normal development for first 6-18 months, then regress</p>	<p>Wide-based gait, scoliosis, normal head size at birth, slowed growth between 5 months and 4 years or age.</p>	<p>Loss of acquired hand and speech skills by age 1-2, hand wringing, mouthing, hand washing, often unable to use hands, SIB-40-50%.</p>	<p>Seizures, hyperventilation, central breathing dysfunction, sleep disorders, constipation, scoliosis, poor circulation, often cold, high metabolic rate.</p>
<p>PKU Phenylketonuria 1 in 15,000 live births in US</p>	<p>Nothing remarkable</p>	<p>OCD, ADHD, Autistic-like features, fine motor problems</p>	<p>Eczema, seizures, ataxia, vulnerable to depression</p>
<p>Lesch-Nyhan 1 in 38,000 live births Symptoms appear between 3-6 months</p>	<p>Often have deformed lips and fingers due to biting themselves.</p>	<p>Compulsive drive to self-injure: SIBs include lip and finger biting, normal experience of pain, often highly social; good sense of humor, empathy for others, head banging, irritability, uncontrolled aggression, involuntary writhing movements, repetitive movements, facial grimacing.</p>	<p>Kidney stones, pain and swelling of joints, difficulty swallowing and eating, hypotonia.</p>

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