SYNDROMES

Syndromes: Intellectual disability is associated with more than 500 genetic problems. The most common have been classified as syndromes, some of which are described in the following chart.

Many of the syndromes have predictable behavioral issues and medical problems. These are called medical and behavioral phenotypes. They are as much a part of a person's personality and physical make-up as eye color.

Some of the syndrome related issues can be treated, even resolved. For example: People with Down syndrome are often born with heart defects. These days, most individuals' defects are corrected with surgery in the first years of life. Many behavioral issues can be changed through careful teaching. People with Williams syndrome, for example, are often overly social and therefore vulnerable to people who have ill intent. They can be taught appropriate boundaries and more cautious social interactions.

Some syndromes include behaviors and conditions that cannot be treated or cured. They can only be managed. People with Prader-Willi syndrome, for example, are genetically burdened with a never ending appetite. They are constantly hungry and therefore ever on the prowl for food. Unchecked, these people can become morbidly, dangerously obese. To help them, access to food must be carefully controlled. People with Lesch-Nehan syndrome are unable to stop themselves from self harming behaviors. As much as they want to, they cannot prevent themselves from hitting themselves and biting themselves. It's crucial that staff and family members have a compassionate understanding of their inability to control their self-harming impulses. They cannot learn to control their self injurious behavior. Helpers, therefore, must provide external structures to keep them safe.

The following chart will introduce you to some of the most common syndromes and what you should expect will be issues to either treat or manage (or both). Chapter 3 of the DM-ID includes a more comprehensive explanation of the syndromes. No, you don't have to memorize these charts to be an effective helper (although I do strongly suggest you memorize the first three - Down, Fetal Alcohol and Fragile X – as they are the most common). But when you are responsible for the care of someone, it's important to read the record and determine if the person has a syndrome. If so, it's only good practice to be aware of the potential problems as well as what the person can and can't control. You will save both yourself and the individual lots of frustration.

SYNDROMES

SYNDROME and FREQUENCY	PHYSICAL CHARACTERISTICS	BEHAVIORAL ISSUES	MEDICAL PROBLEMS
Down "Trisomy 21" 1 in 700-1000 live births The most common cause of intellectual disability	Small head with flat face Small ears and mouth Protruding tongue Upward slant to eyes Broad neck Short Hypotonia; flexible joints Single crease across center of palm	Often seem passive Often sociable personality Stubborn/distractible Resistant to change Require stimulation and more attention Excellent memory Early onset of Alzheimers (as early as age 35) Conduct problems	Congenital heart defects -50% Lack of Moro reflex Hearing loss 66-89% Seizures – 33% Vision problems Hypothyroidism Gastrointestinal problems Obesity Dry skin Dental problems Males - generally sterile Females - low fertility
Fragile X 1 in 1500 males 1 in 2500 females The most common hereditary cause of ID	Macrocephaly (large head) Large ears Long, narrow face Macroorchidism (large testicles) Low muscle tone Hyperextensible joints Flat feet Soft skin	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	Mitral valve prolapse Seizures Scoliosis Sinus problems Atlantoaxial dislocation
Fetal Alcohol Syndrome .2-1.5 in 1000 live births	Short stature Long lean body Small head Abnormal facial features - thin upper lip, small eye openings	Poor impulse control Hyperactive Poor sucking and sleep as infant Poor coordination	Heart problems Kidney problems Hearing loss
Tuberous Sclerosis 1 in 10,000 live births	Small benign tumors grow on organs, eyes, and skin. Individuals with TS may experience none or all of the symptoms with varying degrees of severity.	In some cases, neurobehavioral problems.	Multi-system disease that can affect brain, kidneys, heart, eyes, lungs and other organs Skin and eye lesions Seizures
Rett's Disorder 1 in 10,000 female births Normal development for first 6 – 18 months, then regress	Wide-based gait (similar to Angelman) Scoliosis Normal head size at birth Slowed growth between 5 mo and 4 yrs	Loss of acquired hand and speech skills by age 1-2 Hand wringing, mouthing, hand washing Often unable to use hands SIB – 40-50%	Seizures Hyperventilation Central breathing dysfunction Sleep disorders Constipation Scoliosis Poor circulation; often cold

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PKU Phenylketonuria 1 in 15,000 live births in US	Nothing remarkable	OCD ADHD Autistic -like features Fine motor problems	Eczema Seizures Ataxia Vulnerable to depression
Lesch-Nyhan 1 in 38,000 live births Symptoms appear between 3-6 months	Often have deformed lips and fingers due to biting themselves	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	Kidney stones Pain and swelling of joints Difficulty swallowing & eating Hypotonia
Hurler Syndrome **Mucopolysaccha-rido-sis Type I (MP1)** 1 in 100,000 births	Short stature Enlarged tongue Stiff joints Clawed hands Enlarged rib cage	In later stages, aggression	Heart disease Breathing problems Damage to internal organs and brain Vulnerable to pneumonia Joint problems Ear infections Clouding of cornea Difficulty walking
Hunter Syndrome Type A **Mucopolysaccha rido-sis Type II (MP2)** 1 in 65,000 to 1-135,000 births; slightly higher incidence in Jewish population living in Israel. X linked disorder Most often seen in males	Coarse facial features Short stature Ivory colored skin -lesions on the upper back and arms and thighs Joint stiffness Short neck Broad chest Large head	In later stages, aggression	Progressive deafness Atypical retinitis pigmentosa Vision problems Enlarged liver and spleen Respiratory problems Stiffness in joints

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Angelman Syndrome 1 in 15,000 – 20,000 live births	Wide mouth, wide-spaced teeth Protruding tongue Light hair and eye color Scoliosis Sometimes called the "happy puppet syndrome" due to happy presentation and jerky movements Unsteady gait Balance disorder	Frequent laughter, smiling Excitable Hand flapping Hyperactive (ADHD) Excessive chewing/ mouthing Drooling Attraction to or fascination with water Grabbing, pinching and biting	Sleep disturbance Seizures
Tay-Sachs Disease Primarily among Jews of Eastern European descent; French Canadians from St. Lawrence region; certain Cajuns in Louisiana, and some Amish **LOTS (late onset T-S)** - symptoms appear in late childhood or early adulthood. Life expectancy is not affected. Often misdiagnosed as MS or MD	Unsteady gait Red spot on retina In Late Onset: most become wheelchair users due to paralysis	In Late Onset: Schizophrenic-like psychosis About 40% have bi-polar illness Dementia	Blindness Paralysis Epileptic seizures Difficulty breathing Difficulty swallowing
Williams Syndrome 1 in 20,000 live births	Cute, attractive facial appearance: full lips, small jaw, puffy cheeks, elongated face Starburst pattern in iris of eyes Raspy, hoarse voice Short stature Joint contractures Sloping shoulders Spinal curve Far-sighted 2/3 have inward deviation of eye Soft stretchy skin	Sensitivity to sound (causing fear or discomfort) leads to crying, screaming, covering ears Friendly and outgoing Talks too much Empathic and emotionally sensitive Emotionally labile Over-demonstrative Anxiety disorders and phobias Excessive worrying Some are musical savants	Cardiovascular abnormalities Abnormally elevated levels of calcium in the blood High blood pressure Chronic constipation Gastrointestinal problems Kidney problems Hypothyroidism Incontinence

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Cri du Chat 1 in 20,000 to 1 in 50,000 live births	Microcephaly (small head) Wide set eyes Downward slant to the eyes Small jaw Skin tags just in front of ears Single line in palm Partial webbing of fingers or toes Low set ears (may be malformed) Low muscle tone Low birth weight and slow growth Abnormal angle to base of skull	High pitched cry sounds like a cat Tactile defensive Sensitive to sound Slow or incomplete development of motor skills Half learn enough verbal skills to communicate Unable to do self-care Obsessive, repetitive and sometimes self-harming behaviors such as head-banging and hitting, biting or scratching self Hyperactivity (25%)	Heart defects Cleft palate (rare) Scoliosis Hernias (inguinal and abdominal) Bowel problems Epilepsy Swallowing and sucking problems that persist beyond childhood
Prader-Willi Syndrome 1 in 12,000-15,000 births	Low muscle tone If food is uncontrolled, morbidly obese	In infants, poor eating due to sucking problems – At around age 3, develop intense interest in food Compulsive drive to eat OCD Repetitive thoughts and compulsions Hoarding Skin picking Strong need for routine Low frustration tolerance	Morbid obesity Diabetes Flaw in the hypothalamus part of their brain which normally registers feelings of hunger and satiety.
Smith-Lemli-Opitz 1 in 20,000	Distinctive facial features, small head size (microcephaly) Malformations of the heart, lungs, kidneys, gastrointestinal tract, and genitalia Weak muscle tone Most have fused second and third toes (syndactyly), and some have extra fingers or toes (polydactyly).	Aggression Property destruction SIBs Self-stimulation Irritability ADHD	Death from multi-system failure in first weeks of life Congenital heart disease Vision problems Hearing problems Gastro-intestinal problems, constipation, vomiting