TABLE 1*

*** These are some of the more common genetic syndromes associated with Intellectual and Developmental Disabilities but not all people with these genetic syndromes will have an intellectual disability.**

Genetic Syndrome	Physical Appearance or "Dysmorphic" Features	Risk for Medical Problems	Risk for Mental Health Disorders	Common Behavior Patterns, Temperament "Behavioral Phenotypes"	Key Implications for Direct Support and Daily Care
Down syndrome ***Trisomy 21 3 copies of chromosome 21 (1 extra)*** 1 in 1200	Short stature Low muscle tone Short neck, with excess skin at the back of the neck Flattened facial profile and nose Small head, ears, and mouth Upward slanting eyes - Wide, short hands with short fingers A single, deep, crease across the palm of the hand	Vision or hearing deficits Hypothyroidism Celiac disease Sleep apnea Anemia Gastroesophageal reflux (GERD) Low muscle tone Constipation Joint problems Osteoarthritis Upper spine problem	Anxiety and depression most common mental health challenges. May develop Alzheimer's Disease early but check for depression!	Relative strengths in social-emotional development = often pleasant, affectionate very social "cheerful" more challenged with "means-ends" reasoning	Know that health problems are the most common reason for changes in mood and behavior - like acid reflux, constipation, sleep apnea, hypothyroidism, osteoarthritis, joint problems. Bowel charting, and tracking physical signs can help. Also, send person to their medical doctor to be sure health issues are not causing challenging behaviors, especially of there are also changes in eating, drinking, sleeping, moving, alertness, bowel or bladder habits. Be patient, allow time for person to learn, repeat and reinforce! See "oppositional" behavior as communication of frustration and try to support person to better express needs – always give choices…even just between 2 options, helping people have control
Fragile X syndrome ***FXS, FRA-X Repeats of genetic sequences on X chromosome affecting the FMR gene***	Long face, prominent jaw, large ears	Connective tissue problems Ear infections Flat feet High arched palate (roof of mouth is unusually high) Double-jointed fingers and hyper-flexible joints	ADHD Anxiety Mood disorders	High rate of Autism Spectrum Disorder and anxiety, sensory problems (sensitive to noise, crowds, tough), poor eye contact, hand-biting risk for "fight or flight" aggression (anxiety based or panic driven aggressive behavior) May be social yet seeming shy with other ASD features (sensory, hand	Use tactics known to help with ASD features commonly seen such as helping person be less anxious about new things and transitions, provide reassurances versus "prompts" - recognize that being moody or aggressive or self-injurious is usually from anxiety and often related to sensory challenges and address sources of anxiety try to avoid noisy, crowded places and be sensitive to the individual's sensitivities! set up a predictable routine so person knows when transitions occur

1 in 5000 males				flapping, distressed about change, new things) rapid speech is usually anxiety based, not "manic"	give warnings, use timers and picture schedules or calendars, to show that a transition is coming
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Cornelia deLange Syndrome ***CdLS*** 1 in 10,000 to 30,000 births	Short Arched eyebrows that often meet in the middle Long eyelashes Low-set ears Small and widely spaced teeth Small upturned nose Some limb (hands, arms) abnormalities	Low level of growth hormones GERD is very common, Heart probvlems Vision problems Kidney abnormalities Sleep disturbances Seizures Highly variable from one affected person to the next!	Autism Spectrum Disorder, Anxiety "Low mood" or depression	High rates of ASD with significant sensory abnormalities, obsessive-compulsive features and compulsive self-injurious behavior	Note that large increases in challenging behaviors often caused by medical issues, especially GERD Swinging, therapeutic horse back riding, swimming activities help sensory challenges, deep pressure versus light touch Use visual aids/pictures to help with communication or technology (I-PAD etc) Help reduce anxiety – slow introduction to new things Use of a predictable, structured (visual) daily schedules and tactics listed above for people with Fragile X syndrome
Prader Willi Syndrome ***PWS*** 1 in 12,000 to 15,000 births	Almond shaped eyes Thin upper lip Small downturned mouth, Small hands Nasal sounding speech	Failure to thrive followed by obesity Short stature Hormonal deficiencies and hypogonadism (under developed genitals) Bone density problems Sleep apnea Other consequences of obesity in adults, i.e. high blood pressure, diabetes Decreased vomiting Seizures Hypotonia (Low muscle tone) Excessive food drive- appetite Sleep apnea	Obsessive Compulsive Disorder Mood disorders	Obsessive-compulsive features >> skin picking Some meet criteria for ASD High pain threshold Skin picking Very high food drive – even eating out of garbage, frozen foods obsessively Difficulty with changes in routine High rate of ASD features, social challenges	Need careful controlled diet because of severe health risks if not provided – be sensitive that person with PWS cannot help the desire for more food (their brains do not signal that they are full and they really feel hungry?) Psych meds can make this even worse at times… Use tactics that help with OCD and ASD like providing predictable routines, small healthy snacks more often, exercise is very important. Positive Behavior Supports can be very helpful with focus on strengths and interests to engage, have fun and divert focus from food or other obsessive tendencies like hording things

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Smith Magenis Syndrome ***SMS*** ***Small deletion at 17p11.2*** 1/25,000	"Cherubic" facial appearance Small jaw Rosy cheeks Deep set eyes – far apart Broad, square-shaped face Broad bridge of the nose Hair growth between eyebrows – looks like one long eyebrow Down-turned mouth Short, full-tipped nose Underdevelopment of middle aspect of face Head may seem disproportionately short	Low muscle tone Poor reflexes Early issues feeding Early onset of GERD Daytime fatigue as children Sleep problems are very common in children and adults and seem related to abnormal circadian rhythm Chronic middle and inner ear infections Sometimes later loss of hearing and even retinal detachment (can cause blindness) from head banging Frequent sinus infections Abnormalities of "voice box"	Impulse Control problems, ADHD Mood disorders as adults	High rate of ASD Head banging, self-hugs or squeezing self around upper body, pull out toenails and fingernails, insert objects into body orifices Other self injury, biting self, hitting self also common Aggression to others Sensitivity to loud noises, sounds Good sense of humor, and long term memory for faces and places, socially motivated	Important to have predictable schedule, avoid loud environments, and use multiple modes for communication (visual schedules and augmented communication) Recognize "attention seeking" is really a positive – as often there are some ASD symptoms but people with SMS are also socially motivated! Give lots of warm, positive interactions and use fun and humor to help teaching skills Sometimes challenging behaviors are due to one of the health problems listed here; always look for these and refer to medical providers of care. Positive Behavior Supports (with focus on strengths and interests) and teaching improved communication can be helpful.
Williams syndrome ***WS***	Small upturned nose Long ridges in the skin that run from the nose to the upper lip Prominent	Cardiac problems Muscle tone problems Joint problems-stiffness Farsighted	ADHD Anxiety	Very friendly, trusting strangers, fearing loud sounds or physical contact, and being interested in	Avoid places with loud noise, and be careful that people with WS may trust strangers when they should not because they are so friendly! Anxiety is usually at the core of any kind of

1 in 8,000	lips with an open mouth Skin that covers the inner corner of the eye (eye folds) Partially missing teeth, defective tooth enamel, or small, widely spaced teeth Sunken chest Short stature	High calcium in the blood		music Hypersensitive to loud sounds Speaking, conversing a big strength over other cognitive skills like math	challenging behaviors. Check on what might be causing anxiety, be supportive and reassuring. Music is a great way to promote wellness!
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22q11 deletion syndrome ***Small deletion at 22q11*** 1 in 4,000 births	Some have a long face and long tapered fingers (but this varies)	Cardiac problems are very common High arched palate (high roof of mouth) Thyroid problems Kidney Problems But.. "remarkable variability" Looks different in different people with the same syndrome!	Lots of variations but high risk for psychosis in adults, anxiety and depression in children and some ASD	Some increased risk of ASD has been reported.	When someone you support has 22 q11 deletion syndrome, always refer them to their medical doctors if they have unusual symptoms related to low energy, breathing problems, changes in bladder habits or drinking fluids more than usual. Though psychosis is more common in adults with the syndrome, sometimes people are depressed, and they need help to be active doing things that really engage them.
Tuberous Sclerosis Complex ***TSC*** ***Mutations***	Skin lesions are sometimes subtle and sometimes very evident Doctors use a special lamp to check for these	Skin lesions Tumors- usually non cancerous called "tubers" develop throughout brain and other parts of the body Heart problems common - May have kidney problems or	ADHD Depression Anxiety	When person has IDD, often have ASD May have self injurious behaviors and aggression related to health issues and ASD challenges, such as sensory and	If person develops challenging behaviors, these worsen or person is very irritable, fist have medical doctor and neurologist check if there may be problems from any anti-seizure medications or if one of the many health problems listed is developing or getting worse. Problems like constipation and being extra tired,

(abnormalities) on either the TSC1 or TSC2 genes 1/5,800		lung problems if there are tubers in these locations too Seizures are common Sleep problems		communication challenges, memory problems Problem arise from having to take seizure medications due to side effects like fatigue and sedation	or added mental confusion can occur from medications needed to control seizures. People with TSC may be embarrassed about their skin problems so it is important to support efforts to get help from a skin doctor and to be able to enjoy dressing in preferred clothes, using other things to take pride in personal appearance and improve self-esteem