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**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.*

<table>
<thead>
<tr>
<th>Genetic Syndrome</th>
<th>Physical Appearance or “Dysmorphic” Features</th>
<th>Risk for Medical Problems</th>
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</tr>
</thead>
<tbody>
<tr>
<td>Down syndrome</td>
<td>Short stature</td>
<td>Vision or hearing deficits</td>
<td>Anxiety and depression most common mental health challenges. May develop Alzheimer’s Disease early but check for depression!</td>
<td>Relative strengths in social-emotional development = often pleasant, affectionate very social “cheerful” more challenged with “means-ends” reasoning</td>
<td>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</td>
</tr>
<tr>
<td><em>Trisomy 21</em></td>
<td>Low muscle tone</td>
<td>Hypothyroidism</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3 copies of chromosome 21</td>
<td>Short neck, with excess skin at the back of the neck</td>
<td>Sleep apnea</td>
<td></td>
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<td></td>
</tr>
<tr>
<td><em>(1 extra)</em></td>
<td>Flattened facial profile and nose</td>
<td>Anemia Gastroesophageal reflux (GERD)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1 in 1200</td>
<td>Small head, ears, and mouth</td>
<td>Low muscle tone</td>
<td></td>
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<tr>
<td></td>
<td>Upward slanting eyes - Wide, short hands with short fingers</td>
<td>Constipation</td>
<td></td>
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<tr>
<td></td>
<td>A single, deep, crease across the palm of the hand</td>
<td>Osteoarthritis</td>
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<tr>
<td></td>
<td></td>
<td>Upper spine problem</td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Fragile X syndrome</td>
<td>Long face, prominent jaw, large ears</td>
<td>Connective tissue problems</td>
<td>ADHD</td>
<td>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</td>
<td>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</td>
</tr>
<tr>
<td><em>Fragile X</em></td>
<td></td>
<td>Ear infections</td>
<td>Anxiety</td>
<td></td>
<td></td>
</tr>
<tr>
<td><em>syndrome</em></td>
<td></td>
<td>Flat feet</td>
<td>Mood disorders</td>
<td></td>
<td></td>
</tr>
<tr>
<td><em>FXS, FRA-X</em></td>
<td></td>
<td>High arched palate (roof of mouth is unusually high)</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td><em>Repeats of genetic</em></td>
<td></td>
<td>Double-jointed fingers and hyper-flexible joints</td>
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<tr>
<td><em>sequences on X chromosome affecting the FMR gene</em></td>
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<td>Cornelia deLange Syndrome CdLS</td>
<td>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</td>
<td>Low level of growth hormones GERD is very common, Heart problems Vision problems Kidney abnormalities Sleep disturbances Seizures Highly variable from one affected person to the next!</td>
<td>Autism Spectrum Disorder, Anxiety “Low mood” or depression</td>
<td>High rates of ASD with significant sensory abnormalities, obsessive-compulsive features and compulsive self-injurious behavior</td>
<td>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</td>
</tr>
<tr>
<td>Prader Willi Syndrome PWS</td>
<td>Almond shaped eyes Thin upper lip Small downturned mouth, Small hands Nasal sounding speech</td>
<td>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</td>
<td>Obsessive Compulsive Disorder Mood disorders</td>
<td>Obsessive-compulsive features &gt;&gt; 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</td>
<td>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</td>
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<td>Smith Magenis Syndrome (SMS)</td>
<td>“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</td>
<td>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”</td>
<td>Impulse Control problems, ADHD Mood disorders as adults</td>
<td>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.</td>
<td></td>
</tr>
<tr>
<td>Small deletion at 17p11.2</td>
<td>1/25,000</td>
<td></td>
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<tr>
<td>Williams syndrome (WS)</td>
<td>Small upturned nose Long ridges in the skin that run from the nose to the upper lip Prominent</td>
<td>Cardiac problems Muscle tone problems Joint problems-stiffness Farsighted</td>
<td>ADHD Anxiety</td>
<td>Very friendly, trusting strangers, fearing loud sounds or physical contact, and being interested in</td>
<td>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</td>
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<td>22q11 deletion syndrome</td>
<td>Some have a long face and long tapered fingers (but this varies)</td>
<td>Cardiac problems are very common</td>
<td>Lots of variations but high risk for psychosis in adults, anxiety and depression in children and some ASD</td>
<td>Some increased risk of ASD has been reported.</td>
<td>When someone you support has 22q11 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.</td>
</tr>
<tr>
<td><em>Small deletion at 22q11</em></td>
<td>1 in 4,000 births</td>
<td></td>
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<tr>
<td>Tuberous Sclerosis Complex</td>
<td>Skin lesions are sometimes subtle and sometimes very evident Doctors use a special lamp to check for these</td>
<td>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</td>
<td>ADHD Depression Anxiety</td>
<td>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</td>
<td>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,</td>
</tr>
<tr>
<td><em>TSC</em></td>
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<td><em>Mutations</em></td>
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| (abnormalities) on either the TSC1 or TSC2 genes | 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 |