

Key words that would trigger a call to the I/DD team in El Paso County

22q13 deletion syndrome

22q13.3 deletion syndrome, which is also commonly known as Phelan-McDermid syndrome, is a disorder caused by the loss of a small piece of chromosome 22. The deletion occurs near the end of the chromosome at a location designated q13.3.

The features of 22q13.3 deletion syndrome vary widely and involve many parts of the body. Characteristic signs and symptoms include developmental delay, moderate to profound intellectual disability, decreased muscle tone (hypotonia), and absent or delayed speech. Some people with this condition have autism or autistic-like behavior that affects communication and social interaction, such as poor eye contact, sensitivity to touch, and aggressive behaviors. They may also chew on non-food items such as clothing. Less frequently, people with this condition have seizures.

Individuals with 22q13.3 deletion syndrome tend to have a decreased sensitivity to pain. Many also have a reduced ability to sweat, which can lead to a greater risk of overheating and

dehydration. Some people with this condition have episodes of frequent vomiting and nausea (cyclic vomiting) and backflow of stomach acids into the esophagus (gastroesophageal reflux).

People with 22q13.3 deletion syndrome typically have distinctive facial features, including a long, narrow head; prominent ears; a pointed chin; droopy eyelids (ptosis); and deep-set eyes. Other physical features seen with this condition include large and fleshy hands and/or feet, a fusion of the second and third toes (syndactyly), and small or abnormal toenails. Some affected individuals have rapid (accelerated) growth.

Angelmans Syndrome

Angelman syndrome is a complex genetic disorder that primarily affects the nervous system. Characteristic features of this condition include delayed development, intellectual disability, severe speech impairment, and problems with movement and balance (ataxia). Most affected children also have recurrent seizures (epilepsy) and a small head size (microcephaly). Delayed development becomes noticeable by the age of 6 to 12 months, and other common signs and symptoms usually appear in early childhood.

Children with Angelman syndrome typically have a happy, excitable

demeanor with frequent smiling, laughter, and hand-flapping movements. Hyperactivity, a short attention span, and a fascination with water are common. Most affected children also have difficulty sleeping and need less sleep than usual.

With age, people with Angelman syndrome become less excitable, and the sleeping problems tend to improve. However, affected individuals continue to have intellectual disability, severe speech impairment, and seizures throughout their lives. Adults with Angelman syndrome have distinctive facial features that may be described as "coarse." Other common features include unusually fair skin with light-colored hair and an abnormal side-to-side curvature of the spine (scoliosis). The life expectancy of people with this condition appears to be nearly normal.

Asperger's/ Autism/ Autism Spectrum Disorder

Autism Spectrum Disorder (ASD) is a condition that appears very early in childhood development, varies in severity, and is characterized by impaired social skills, communication problems, and repetitive behaviors. These difficulties can interfere with affected individuals' ability to function in social, academic, and employment settings. People with ASD also have an increased risk of psychiatric problems such as anxiety, depression, obsessive-compulsive disorder, and eating disorders.

From as early as 1 to 2 years of age, people with ASD have an impaired ability to interact with other people; they

are often more comfortable dealing with objects. Affected individuals have difficulty understanding and using non-verbal social cues such as eye contact, facial expressions, gestures, and body language. Inability to recognize and use these cues makes it hard for affected individuals to understand the feelings of others or communicate their own feelings appropriately. Behavioral signs of ASD, such as reduced eye contact and social interaction, can sometimes be detected before age 2. However, the condition is usually diagnosed between ages 2 and 4, when more advanced communication and social skills, such as learning to play with others, typically begin to develop.

Repetitive behaviors in ASD can include simple actions such as rocking, hand-flapping, or repetition of words or noises (echolalia). Affected individuals often dwell on or repeatedly express particular thoughts; this behavior is called perseveration. People with ASD tend to be rigid about their established routines and may strongly resist disruptions such as changes in schedule. They may also have difficulty tolerating sensory stimuli such as loud noises or bright lights.

While social and communication difficulties and unusual behaviors define ASD, affected individuals can have a wide range of intellectual abilities and language skills. A majority of people with ASD have mild to moderate intellectual disability, while others have average to above-average intelligence. Some have particular cognitive abilities that greatly surpass their overall level of functioning, often in areas such as music, mathematics, or memory.

Some people with ASD do not speak at all, while others use language fluently. However, fluent speakers with ASD often have problems associated with verbal communication. They might speak in a monotone voice, have unusual vocal mannerisms, or choose unusual topics of conversation.

Several diagnoses that used to be classified as separate conditions are now grouped together under the diagnosis of ASD. For example, autistic disorder was a term that was used when affected individuals had limited or absent verbal communication, often in combination with intellectual disability. By contrast, Asperger syndrome was a diagnosis formerly applied to affected individuals of average or above-average intelligence who were not delayed in their language development. The broader diagnosis of ASD was established because many affected individuals fall outside of the strict definitions of the narrower diagnoses, and their intellectual and communication abilities may change over time. However, some individuals who were previously diagnosed with one of the subtypes now do not meet all the criteria of the new umbrella diagnosis.

Cerebral Palsy

Cerebral palsy is a disorder of movement, muscle tone or posture that is caused by damage that occurs to the immature, developing brain, most often before birth.

Signs and symptoms appear during infancy or preschool years. In general, cerebral palsy causes impaired movement associated with abnormal reflexes, floppiness or rigidity of the

limbs and trunk, abnormal posture, involuntary movements, unsteady walking, or some combination of these.

People with cerebral palsy may have problems swallowing and commonly have eye muscle imbalance, in which the eyes don't focus on the same object. People with cerebral palsy also may suffer reduced range of motion at various joints of their bodies due to muscle stiffness.

Cerebral palsy's effect on functional abilities varies greatly. Some affected people can walk while others can't. Some people show normal or near-normal intellectual capacity, but others may have intellectual disabilities. Epilepsy, blindness or deafness also may be present.

Cri-du-chat

Cri-du-chat (cat's cry) syndrome, also known as 5p- (5p minus) syndrome, is a chromosomal condition that results when a piece of chromosome 5 is missing. Infants with this condition often have a high-pitched cry that sounds like that of a cat. The disorder is characterized by intellectual disability and delayed development, small head size (microcephaly), low birth weight, and weak muscle tone (hypotonia) in infancy. Affected individuals also have distinctive facial features, including widely set eyes (hypertelorism), low-set ears, a small jaw, and a rounded face. Some children with cri-du-chat syndrome are born with a heart defect.

Dandy-Walker syndrome

Dandy-Walker malformation affects brain development, primarily development of the cerebellum, which is the part of the brain that coordinates movement. In individuals with this condition, various parts of the cerebellum develop abnormally, resulting in malformations that can be observed with medical imaging. The central part of the cerebellum (the vermis) is absent or very small and may be abnormally positioned. The right and left sides of the cerebellum may be small as well. In affected individuals, a fluid-filled cavity between the brainstem and the cerebellum (the fourth ventricle) and the part of the skull that contains the cerebellum and the brainstem (the posterior fossa) are abnormally large. These abnormalities often result in problems with movement, coordination, intellect, mood, and other neurological functions.

In the majority of individuals with Dandy-Walker malformation, signs and symptoms caused by abnormal brain development are present at birth or develop within the first year of life. Some children have a buildup of fluid in the brain (hydrocephalus) that may cause increased head size (macrocephaly). Up to half of affected individuals have intellectual disability that ranges from mild to severe, and those with normal intelligence may have learning disabilities. Children with Dandy-Walker malformation often have delayed development, particularly a delay in motor skills such as crawling, walking, and coordinating movements. People with Dandy-Walker malformation may experience muscle stiffness and partial paralysis of the lower limbs (spastic paraplegia), and they may also have seizures. While rare, hearing and vision

problems can be features of this condition.

Less commonly, other brain abnormalities have been reported in people with Dandy-Walker malformation. These abnormalities include an underdeveloped or absent tissue connecting the left and right halves of the brain (agenesis of the corpus callosum), a sac-like protrusion of the brain through an opening at the back of the skull (occipital encephalocele), or a failure of some nerve cells (neurons) to migrate to their proper location in the brain during development. These additional brain malformations are associated with more severe signs and symptoms.

Dandy-Walker malformation typically affects only the brain, but problems in other systems can include heart defects, malformations of the urogenital tract, extra fingers or toes (polydactyly) or fused fingers or toes (syndactyly), or abnormal facial features.

In 10 to 20 percent of people with Dandy-Walker malformation, signs and symptoms of the condition do not appear until late childhood or into adulthood. These individuals typically have a different range of features than those affected in infancy, including headaches, an unsteady walking gait, paralysis of facial muscles (facial palsy), increased muscle tone, muscle spasms, and mental and behavioral changes. Rarely, people with Dandy-Walker malformation have no health problems related to the condition.

Problems related to hydrocephalus or complications of its treatment are the

most common cause of death in people with Dandy-Walker malformation.

Deletion of the 5th Chromosome

Developmental Delay

Developmental disability Neurology A disability that affects a person's development, such as, mental retardation, epilepsy, autism, cerebral palsy or similar disability Pediatrics A lag in reaching developmental milestones by the expected age Types Biological—e.g., chromosomal defects or in utero infection, environmental—e.g., maternal mental malady or marital malaise, or 'transactional'—i.e. an interplay between biological and environmental factors Diagnosis Developmental screening tests to identify developmental red flags. The failure to meet certain developmental milestones, such as sitting, walking, and talking, at the average age. Developmental delay may indicate a problem in development of the central nervous system.

Developmental Disability

Developmental disability a substantial handicap in mental or physical functioning, with onset before the age of 18 and of indefinite duration. Examples are autism, cerebral palsy, uncontrolled epilepsy, certain other neuropathies, and mental retardation. FSIQ of less than 70

Down Syndrome

Down syndrome is a set of cognitive and physical symptoms that result from having an extra chromosome 21 or an extra piece of that chromosome. It is the most common chromosomal cause of mild to moderate intellectual disabilities. People with Down syndrome also have some distinct physical features, such as a flat-looking face, and they are at risk for a number of other health conditions.

Understanding Down syndrome and other intellectual and developmental disabilities is part of the reason the NICHD was established. Today, the Institute continues to lead research on the causes, progression, treatment, and management of Down syndrome, as well as on conditions and diseases that are associated with the syndrome.

Dysgraphia

Kids with dysgraphia have unclear, irregular, or inconsistent handwriting, often with different slants, shapes, upper- and lower-case letters, and cursive and print styles. They also tend to write or copy things slowly.

Parents or teachers may notice symptoms when the child first begins writing assignments in school. Other signs of dysgraphia to watch for include:

- Cramped grip, which may lead to a sore hand
- Difficulty spacing things out on paper or within margins (poor spatial planning)
- Frequent erasing
- Inconsistency in letter and word spacing

- Poor spelling, including unfinished words or missing words or letters
- Unusual wrist, body, or paper position while writing

This learning disability also makes it hard to write and think at the same time. Creative writing tasks are often especially hard.

Early Intervention (EI)

Colorado's Early Intervention program provides supports and services to children with developmental delays or disabilities and their families from birth until the child's third birthday. Early Intervention Colorado can help families learn ways to support and promote their child's development within their everyday routines and activities.

Research shows that the first three years of a child's life are the most important time for developing and learning. By providing needed services and supports during this time, families will be able to help their children with special needs develop to their full potential, and may decrease the need for additional help later in life.

The Early Intervention Colorado program bases its foundation of support on seven guiding key principles. They are a way to talk about how services are provided and delivered to the families we support. They include being family-centered, focusing on children's learning in their natural environment, adult learning and quality teaming.

Early intervention provides developmental supports and services to children birth through three years of

age, who have special developmental needs. It can help improve a child's ability to develop and learn. It can also help parents and family members learn ways to support and promote a child's development, within the family's activities and community life. Because an infant or toddler learns through his or her interactions with caregivers, parents and other caregivers are important members of the early intervention team.

Enuresis/Encopresis

Enuresis sometimes accompanies encopresis. Enuresis is a term used to describe children who urinate at the wrong place or the wrong time, either involuntarily or on purpose, for at least three months, twice a week, at age 5 or beyond. It is usually caused by a delay in the maturation in the part of the nervous system that controls bladder function. Sometimes enuresis can be caused by secondary stressful events in a child's life, such as a divorce, a move, a change in school, or an illness.

Encopresis (en-ko-PREE-sis), sometimes called fecal incontinence or soiling, is the repeated passing of stool (usually involuntarily) into clothing. Typically it happens when impacted stool collects in the colon and rectum: the colon becomes too full and liquid stool leaks around the retained stool, staining underwear. Eventually, stool retention can cause swelling (distention) of the bowels and loss of control over bowel movements.

Encopresis usually occurs after age 4, when the child has already learned to use a toilet. In most cases, soiling is a symptom of chronic constipation. Far

less frequently it occurs without constipation and may be the result of emotional issues.

Epilepsy

Epilepsy is a neurological condition affecting the nervous system. Epilepsy is also known as a seizure disorder. It is usually diagnosed after a person has had at two seizures, or one seizure with the likelihood or more, that were not caused by some known medical condition.

Fetal Alcohol Syndrome

Fetal alcohol spectrum disorders are a group of birth defects that can happen when a pregnant woman drinks alcohol. Fetal alcohol syndrome (FAS) is the most severe type of the disorder.

FAS and other spectrum disorders affect children differently. Symptoms can range from mild to severe. They can include:

- Problems with the heart, kidney, and/or bones
- Learning disabilities and low IQ
- Trouble with memory, coordination, and attention
- Hyperactivity
- Problems with sleep and suckling as an infant

The symptoms of FAS tend to get worse as a person grows up.

Fragile X Syndrome

Fragile X syndrome is a genetic condition that causes a range of developmental problems including

learning disabilities and cognitive impairment. Usually, males are more severely affected by this disorder than females.

Affected individuals usually have delayed development of speech and language by age 2. Most males with fragile X syndrome have mild to moderate intellectual disability, while about one-third of affected females are intellectually disabled. Children with fragile X syndrome may also have anxiety and hyperactive behavior such as fidgeting or impulsive actions. They may have attention deficit disorder (ADD), which includes an impaired ability to maintain attention and difficulty focusing on specific tasks. About one-third of individuals with fragile X syndrome have features of autism spectrum disorders that affect communication and social interaction. Seizures occur in about 15 percent of males and about 5 percent of females with fragile X syndrome.

Most males and about half of females with fragile X syndrome have characteristic physical features that become more apparent with age. These features include a long and narrow face, large ears, a prominent jaw and forehead, unusually flexible fingers, flat feet, and in males, enlarged testicles (macroorchidism) after puberty.

Hydrocephalus

Hydrocephalus is the buildup of fluid in the cavities (ventricles) deep within the brain. The excess fluid increases the size of the ventricles and puts pressure on the brain.

Cerebrospinal fluid normally flows through the ventricles and bathes the brain and spinal column. But the pressure of too much cerebrospinal fluid associated with hydrocephalus can damage brain tissues and cause a large spectrum of impairments in brain function.

Although hydrocephalus can occur at any age, it's more common among infants and older adults.

Surgical treatment for hydrocephalus can restore and maintain normal cerebrospinal fluid levels in the brain. A variety of interventions are often required to manage symptoms or functional impairments resulting from hydrocephalus.

IEP

Individual Education Plan is developed for children whose disabilities prevent them from accessing FAPE, Free and Appropriate Public Education. In an IEP a child has identified services and accommodations to assist them in receiving their educational instruction in the least restrictive setting possible.

Intellectual Disability

Intellectual disability, also known as mental retardation, is a term used when there are limits to a person's ability to learn at an expected level and function in daily life. Levels of intellectual disability vary greatly in children from a very slight problem to a very severe problem. Children with intellectual disability might have a hard time letting others know their wants and needs, and taking care of themselves. Intellectual disability could cause a child to learn and

develop more slowly than other children of the same age. It could take longer for a child with intellectual disability to learn to speak, walk, dress, or eat without help, and they could have trouble learning in school.

Learning Disability

Many children may struggle in school with some topics or skills from time to time. When children try hard and still struggle with a specific set of skills over time, it could be a sign of a learning disorder. Having a learning disorder means that a child has difficulty in one or more areas of learning, even when overall intelligence or motivation is not affected.

Some of the symptoms of learning disorders are

- Difficulty telling right from left
- Reversing letters, words, or numbers, after first or second grade
- Difficulties recognizing patterns or sorting items by size or shape
- Difficulty understanding and following instructions or staying organized
- Difficulty remembering what was just said or what was just read
- Lacking coordination when moving around
- Difficulty doing tasks with the hands, like writing, cutting, or drawing
- Difficulty understanding the concept of time

Examples of learning disorders include

- Dyslexia – difficulty with reading

- Dyscalculia – difficulty with math
- Dysgraphia – difficulty with writing

Children with learning disorders may feel frustrated that they cannot master a subject despite trying hard, and may act out, act helpless, or withdraw. Learning disorders can also be present with emotional or behavioral disorders, such as attention-deficit/hyperactivity disorder (ADHD), or anxiety. The combination of problems can make it particularly hard for a child to succeed in school. Properly diagnosing each disorder is crucial, so that the child can get the right kind of help for each.

Low IQ

Any FSIQ score two standard deviations below the mean (100). Anything below 70 is considered a low IQ and can be used to qualify for disability status for SSI and/or other federal and state programs.

Non Verbal

This means that a person does not use words to effectively communicate their needs and wants.

Pervasive Developmental Disorder

PDD-NOS is characterized by delays in the development of socialization and communication skills. Parents may notice associated behaviors as early as infancy. These may include delays in using and understanding language, difficulty relating to people, unusual play with toys and other objects, difficulty with changes in routine or surroundings

and repetitive body movements or behavior patterns.

Rett Syndrome

Rett syndrome is a childhood neurodevelopmental disorder that affects females almost exclusively. The child generally appears to grow and develop normally, before symptoms begin. Loss of muscle tone is usually the first symptom. Other early symptoms may include a slowing of development, problems crawling or walking, and diminished eye contact. As the syndrome progresses, a child will lose purposeful use of her hands and the ability to speak. Compulsive hand movements such as wringing and washing follow the loss of functional use of the hands. The inability to perform motor functions is perhaps the most severely disabling feature of Rett syndrome, interfering with every body movement, including eye gaze and speech.

Seizure disorder

The epilepsies are a spectrum of brain disorders ranging from severe, life-threatening and disabling, to ones that are much more benign. In epilepsy, the normal pattern of neuronal activity becomes disturbed, causing strange sensations, emotions, and behavior or sometimes convulsions, muscle spasms, and loss of consciousness. The epilepsies have many possible causes and there are several types of seizures. Anything that disturbs the normal pattern of neuron activity—from illness to brain damage to abnormal brain development—can lead to seizures.

Epilepsy may develop because of an abnormality in brain wiring, an imbalance of nerve signaling chemicals called neurotransmitters, changes in important features of brain cells called channels, or some combination of these and other factors. Having a single seizure as the result of a high fever (called febrile seizure) or head injury does not necessarily mean that a person has epilepsy. Only when a person has had two or more seizures is he or she considered to have epilepsy. A measurement of electrical activity in the brain and brain scans such as magnetic resonance imaging or computed tomography are common diagnostic tests for epilepsy.

Shaken Baby Syndrome

Shaken baby syndrome is a type of inflicted traumatic brain injury that happens when a baby is violently shaken. A baby has weak neck muscles and a large, heavy head. Shaking makes the fragile brain bounce back and forth inside the skull and causes bruising, swelling, and bleeding, which can lead to permanent, severe brain damage or death. The characteristic injuries of shaken baby syndrome are subdural hemorrhages (bleeding in the brain), retinal hemorrhages (bleeding in the retina), damage to the spinal cord and neck, and fractures of the ribs and bones. These injuries may not be immediately noticeable. Symptoms of shaken baby syndrome include extreme irritability, lethargy, poor feeding, breathing problems, convulsions, vomiting, and pale or bluish skin. Shaken baby injuries usually

occur in children younger than 2 years old.

Traumatic Brain Injury

Traumatic brain injury (TBI), a form of acquired brain injury, occurs when a sudden trauma causes damage to the brain. TBI can result when the head suddenly and violently hits an object, or when an object pierces the skull and enters brain tissue. Symptoms of a TBI can be mild, moderate, or severe, depending on the extent of the damage to the brain. A person with a mild TBI may remain conscious or may experience a loss of consciousness for a few seconds or minutes. Other symptoms of mild TBI include headache, confusion, lightheadedness, dizziness, blurred vision or tired eyes, ringing in the ears, bad taste in the mouth, fatigue or lethargy, a change in sleep patterns, behavioral or mood changes, and trouble with memory, concentration, attention, or thinking. A person with a moderate or severe TBI may show these same symptoms, but may also have a headache that gets worse or does not go away, repeated vomiting or nausea, convulsions or seizures, an inability to awaken from sleep, dilation of one or both pupils of the eyes, slurred speech, weakness or numbness in the extremities, loss of coordination, and increased confusion, restlessness, or agitation.

William's Syndrome

Williams Syndrome (WS) is a rare genetic disorder characterized by mild to moderate delays in cognitive development or learning difficulties, a

distinctive facial appearance, and a unique personality that combines over-friendliness and high levels of empathy with anxiety. The most significant medical problem associated with WS is cardiovascular disease caused by narrowed arteries. WS is also associated with elevated blood calcium levels in infancy. A random genetic mutation (deletion of a small piece of chromosome 7), rather than inheritance, most often causes the disorder. However, individuals who have WS have a 50 percent chance of passing it on if they decide to have children. The characteristic facial features of WS include puffiness around the eyes, a short nose with a broad nasal tip, wide mouth, full cheeks, full lips, and a small chin. People with WS are also likely to have a long neck, sloping shoulders, short stature, limited mobility in their joints, and curvature of the spine. Some individuals with WS have a star-like pattern in the iris of their eyes. Infants with WS are often irritable and colicky, with feeding problems that keep them from gaining weight. Chronic abdominal pain is common in adolescents and

adults. By age 30, the majority of individuals with WS have diabetes or pre-diabetes and mild to moderate sensorineural hearing loss (a form of deafness due to disturbed function of the auditory nerve). For some people, hearing loss may begin as early as late childhood. WS also is associated with a characteristic "cognitive profile" of mental strengths and weaknesses composed of strengths in verbal short-term memory and language, combined with severe weakness in visuospatial construction (the skills used to copy patterns, draw, or write). Within language, the strongest skills are typically in concrete, practical vocabulary, which in many cases is in the low average to average range for the general population. Abstract or conceptual-relational vocabulary is much more limited. Most older children and adults with WS speak fluently and use good grammar. More than 50% of children with WS have attention deficit disorders (ADD or ADHD), and about 50% have specific phobias, such as a fear of loud noises. The majority of individuals with WS worry excessively.