10 Rare Diseases found in Pediatrics

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| **Disorder** | **Inheritance/General Info** | **Common Characteristics/Impairments** | **Treatment Considerations** |
| Angelman’s Syndrome  [www.angelman.org](http://www.angelman.org) | A genetic disorder in which a random deletion occurs that affects the nervous system. | Children have delayed development, including problems with movement and balance. Many present with small and/or abnormal head shapes so it will be important to measure and then suggest more accurate cranial measurements for potential banding. Children exhibit intellectual disability and severe speech impairment. Signs and symptoms become noticeable between 6-12 months old. | Children generally have happy, excitable behavior with short attention spans and have difficulty sleeping. As they get older they are less excitable but improve their ability to sleep. These children will need simplified directions and may need strong behavior management techniques to keep them focused on the activity. |
| Arthrogryposis  [www.amcsupport.org](http://www.amcsupport.org) | A sporadic genetic condition that is often seen in other family members.  When one area of the body is affected it is referred to as an isolated congenital contracture. When two or more areas of the body are affected it is referred to as arthrogryposis multiplex congenita (AMC) | Patients display with congenital joint contractures of the hands and feet, most commonly overlapping fingers, all fingers angled towards the 5th finger, and club foot. | Most children have normal intelligence so they will be able to work with a therapist to develop unique adaptations in order to be more functional. |
| Batten’s Disease (CLN3)  [www.battens.org.au](http://www.battens.org.au)  [www.beyondbatten.org](http://www.beyondbatten.org) | Inherited disorder that affects the nervous system. There are several different types, CLN3 is the most common. | Children develop normally and then after 4-6 years they begin to display signs of regression, most notably vision impairments, intellectual disability, movement problems, speech difficulties, and seizures. These regressions become worse over time as most people do not survive past early adulthood. | Because this is a progressive disease, there is very little research on the effect of PT interventions. PTs and OTs serve to maintain the function the patient has to slow the progression of the disease. Children will gradually lose their ability to swallow and will need a speech therapist for feeding therapies along with their speech difficulties. PTs, OTs, and SLP will be greatly involved in the families obtaining adaptive equipment and instructing them how to use the equipment. It will be important to educate the families on how to safely transfer their child. |
| CHARGE Syndrome  [www.chargesyndrome.org/](http://www.chargesyndrome.org/) | Caused by a mutation of a gene. Not a hereditary condition | Named for common features of the disorder  C=coloboma (gap or hole in the structures of the eye)  H=heart defects  A=atresia choanae (a block in the back of the nasal passage)  R=retardation of growth  G=genital abnormalities  E=ear abnormalities  Patients may have visual impairments depending on the size and location of the coloboma, difficulties with breathing, hearing difficulties, and speech problems. May also display cranial nerve abnormalities such as controlling muscle movement, transmitting sensory information, problems with swallowing, and facial paralysis | With children with this diagnosis, it will be important to figure out the best way to communicate with them. If they have seen or are currently seeing a speech therapist, they would be a good resource for understanding the best way to communicate treatments with the child. Be sure to monitor their breathing for any changes with interventions as well as understanding what kind of sensory information they are trying to seek. Due to their visual impairments, a vision therapist may be warranted so the child may better participate in other therapy services. |
| DiGeorge Syndrome  [www.22q.org/](http://www.22q.org/) | Now more commonly known under the spectrum disorder referred to as 22q11.2 deletion syndrome | Patients can have many signs and symptoms, most commonly heart abnormalities, cleft palate, and some distinct facial features. They are at greater risk to have ADHD and autism, as well as mental illnesses (schizophrenia, anxiety, depression, bipolar disorder). They can suffer from recurrent infections due to problems with their immune system | Children with DiGeorge syndrome exhibit developmental delays, growth and speech development delays, and learning disabilities. Some patients may have breathing problems, hearing loss, and suffer from seizures. With these children, it will be important to establish efficient communication and to make sure to monitor their respiratory rate and effort with breathing during interventions. |
| Dravet Syndrome  [www.dravetfoundation.org](http://www.dravetfoundation.org) | A severe form of epilepsy that appears in the first year of life. | Children will have severe full body convulsion seizures or hemiconvulsions in the first year, then they experience atypical absence seizures, myoclonic seizures, or other partial seizures. Children begin showing signs of intellectual developmental deterioration around 2 years old. Children display poor coordination, poor language development, hyperactivity, and difficulty relating to others. | Because a seizure could happen at any time without warning, children with Dravet should always be closely monitored for potential drop attacks, even if they are excelling at a task independently. Children with any form of epilepsy will experience setbacks with their development, but children with Dravet Syndrome will experience even greater setbacks when seizures occur because of their intensity. It is important to always reassess the child after they suffer a seizure episode since it is not uncommon for them to regress. It is also important to be aware of behavioral disorders these children may have been born with or may develop. |
| Mitochondrial Disease (a.k.a. mitochondrial genetic disorders)  [www.umdf.org](http://www.umdf.org)  [www.mitoaction.org](http://www.mitoaction.org) | Group of conditions that affect the mitochondria, which are responsible for making energy for the body to use. Can be a mutation and can be passed down to offspring | The most common organs and tissues affected are the brain, heart, muscles, liver, eyes, ears, nerves, and kidneys. The most common symptoms include poor growth, loss of muscle coordination, muscle weakness, seizures, autism, problems with vision and/or hearing, developmental delay, and learning disabilities. | Energy levels with these children are important to consider as they may tire more quickly than other children. It is important to give them adequate rest breaks and find a balance of challenging them, but at the same time not completely exhausting them so they cannot make it through the rest of the day. |
| Osteogenesis Imperfecta  [www.oif.org/site/PageServer](http://www.oif.org/site/PageServer) | Also, known as Brittle Bone Disease is a mutation genetic disorder. This disease can be passed down from a parent or it can occur sporadically. There are 8 types, type I being the mildest form and type II being the most severe, but all are distinguished by their signs and symptoms. | People with this condition have fragile bones that can break easily. Fractures can range from a few over their lifespan for milder cases to many fractures per year with no apparent cause for more severe cases. Some can suffer fractures from sneezing or coughing. It is expected to reach normal height, or close to it, for those with milder cases, but those with more severe cases will exhibit decreased growth. Patients can also display muscle weakness, loose joints, and skeletal malformations. | Some patients can have respiratory problems, potentially due to underdeveloped lungs, and can develop hearing loss in adulthood. It is important to monitor these patients for breathing difficulties during exercise activities. Although, hearing loss does not typically develop until adulthood, it is still something to keep in mind when working with older children. Hearing loss can be caused by deformities in the ossicles and inner ear, or can be caused by loud noises, head injuries, and infection. Aquatic therapy can be a safe environment for younger children to develop strength because their risk of injury due to falling will be reduced. |
| Phelan-McDermid  [www.22q13.org/j15/](http://www.22q13.org/j15/) | It is a 22q13.3 deletion syndrome. Typically not a disorder that runs in the family | Common characteristics include developmental delay, hypotonia, intellectual disability, and absent or delayed speech. Some patients have autism or autism-like behaviors. | These patients may have a decreased sensitivity to pain so it is important to check them if they fall or bump into something. They also can have a reduced ability to sweat so it is important to give them adequate rest and water breaks to avoid overheating and dehydration. Some can have frequent episodes of nausea and vomiting and suffer from reflux. |
| Spina Bifida  [www.spinabifidaassociation.org](http://www.spinabifidaassociation.org) | A type of neural tube defect in which bones in the spinal column do not completely form and leave a part of the spinal cord exposed. If the spinal cord sticks out and is within a fluid-filled sac on the person’s back covered by skin it is called a myelomeningocele and they will suffer permanent nerve damage. | Signs and symptoms vary depending on the severity and the level at which the spinal cord is exposed and sticks out. Patients can display weakness or paralysis and/or loss of sensation of the legs or feet. Bowel and bladder functions could be compromised. | Some patients can have a backup of excess spinal fluid that causes hydrocephalus which can lead to the patient having learning problems. It is important to monitor for symptoms of hydrocephalus. For infants, this includes a rapid increase in head circumference, vomiting, sleepiness, irritability, downward deviation of the eyes, and seizures. For children symptoms include headache, vomiting, blurred or double vision, downward deviation of the eyes, urinary incontinence, balance, coordination, and gait disturbances, lethargy, or irritability. Tethered cord is another complication that could occur. Symptoms include low back pain with activity, leg pain or numbness, gait disturbances, foot and spinal deformities, and bowel and bladder difficulties |
| Tetrasomy 18  [www.chromosome18.org](http://www.chromosome18.org) | A chromosomal disorder that affects many parts of the body | Children commonly have changes in muscle tone. Some can display with hypotonia, while others can display with hypertonia and spasticity. This contributes to a developmental delay of their motor skills. Children can exhibit abnormal curvatures of the spine and a shortage of growth hormone. | Babies have trouble feeding and vomit frequently, which makes it very difficult for them to gain weight, and they can also have breathing problems. Children can suffer seizures, have vision issues, hearing loss, and psychiatric conditions such as ADHD, anxiety, and social and behavioral challenges. |

Resources

<https://ghr.nlm.nih.gov> – Genetics Home Reference

<https://rarediseases.info.nih.gov> – GARD (Genetic And Rare Diseases) information center

<https://rarediseases.org> – NORD (National Organization for Rare Disorders)