Children with a wide variety of developmental needs and their families receive services and supports from early intervention providers. Some of the less common conditions that affect development and are seen in early intervention settings include genetic, congenital, acquired, autoimmune, or traumatic conditions. The following are short descriptions of some the less common conditions that may affect young children. As with all children with disabilities and delays in development, information and family support is very important and specific electronic support resources are listed with each condition.

**Genetic Conditions**

**Achondroplasia**
Achondroplasia is a genetic disorder that causes changes in bone growth. Cartilage at the growth plate of the long bones is slow to turn to bone. Individuals with achondroplasia have typical stature in their trunk but shortened limbs, large head with prominent forehead, and small hands and feet with shortened digits. Individuals with achondroplasia have typical cognitive function but are born with low muscle tone and are at risk for health conditions like hydrocephalus, dental problems, ear infections, hearing loss, spinal cord compression, and orthopedic issues including poor limb and spinal alignment. Motor and adaptive skill development is primarily affected but communication (if there is a hearing loss) and social and emotional areas can be affected as well. Assistive technology and environmental modifications are often important to maximize access to home, school and the community.

- Little People of America (LPA)
  [http://www.lpaonline.org/mc/page.do](http://www.lpaonline.org/mc/page.do)
- March of Dimes

**Fragile X Syndrome**
Fragile X syndrome is a genetic condition that is the most common cause of intellectual disabilities. The intellectual impact in children with fragile X syndrome ranges from learning disabilities to significant intellectual disabilities. The condition can be confused with autism spectrum disorder because of similar behaviors and delays in communication and social skills. Genetic testing is necessary to determine the diagnosis. Girls in general have fewer or milder
characteristics than the boys who are affected because females have two X chromosomes and only one X chromosome with the defect causes the more severe intellectual disabilities. All areas of development are affected especially cognition, communication, and social and emotional development but typically there are also delays in adaptive and motor skill development. Anxiety, attention, behavior challenges, depression, and sensory processing disorders are also common in children with fragile X.

- The National Fragile X Foundation  

**Limb Deficiencies/Amputations**

Limb deficiencies are reductions or absences in the digits of the hands and feet or in the limbs because of a variety of disruptions that occur sometime during fetal development. Genetic syndromes, vascular insufficiency, teratogenic exposure (medications, environmental toxins), infection or unknown congenital factors disrupt the digit or limb formation at critical periods during fetal development. The developmental impact of the reduction or absence of digits and limbs is primarily on motor skills. Prosthetic devices, assistive technology or environmental accommodations are needed to promote motor skills, self care, and adaptive skills. However, if a child’s amputation is caused by a genetic syndrome, his cognition, social, emotional, and communication skill development may also be a concern.

- American Amputee Foundation  
  http://www.americanamputee.org/default.aspx
- International Child Amputee Network  
  http://www.child-amputee.net/index.htm

**Muscular Dystrophy**

Muscular Dystrophy is a group of genetic diseases that affect the neuromuscular system that causes progressive degeneration of muscles. The most common form, Duchenne, affects mostly boys (girls are genetic carriers) and children with this form are missing the protein dystrophin critical to maintain muscle function. Symptoms develop in early childhood and progress through adolescence with a loss of mobility and self care and eventually compromised respiration. Becker muscular dystrophy is a similar but a less severe form because individuals have altered or less dystrophin rather than an absence therefore symptoms are not as severe. There are other forms that express symptoms later in adolescence and adulthood. The impact of the childhood onset muscular dystrophies is primarily to motor development, but children with muscular dystrophy may also have hydrocephalus and global developmental delays that require monitoring and an array of services. As the disease progresses, monitoring of health including cardiopulmonary health, joint contractures, and scoliosis formation become increasingly important. Assistive technology and environmental modifications become more critical to maintain or promote independent functioning at home, school, and in the community.

- Muscular Dystrophy Family Foundation  
  http://www.mdff.org/
- Muscular Dystrophy Association  
  http://www.mda.org/
**Osteogenesis Imperfecta (OI)**
Osteogenesis Imperfecta is a non-progressive genetic disease affecting collagen production causing the bones and teeth to be brittle and break easily in childhood but fractures decrease in frequency into adulthood. There are many forms of OI and the severity of the symptoms like bone fractures varies. Complications for children and individuals with OI include muscle weakness, bone deformities and fractures, bleeding and bruising easily, dental problems, hearing loss, scoliosis, joint laxity, respiratory complications, fragile skin integrity, and short stature. Developmental impacts are primarily in the motor and adaptive areas but communication can also be affected by hearing loss and jaw function. Pain management and monitoring pulmonary health are important even with young children. Assistive technology and environmental adaptations for mobility, self care, and care giver support are usually needed.

- Osteogenesis Imperfecta (OI) Foundation
  [http://www.oif.org/site/PageServer](http://www.oif.org/site/PageServer)
- Guide to Osteogenesis Imperfecta for Pediatricians and Family Practice Physicians
  [www.niams.nih.gov/bone](http://www.niams.nih.gov/bone)

**Spinal Muscular Atrophy (SMA)**
Spinal Muscular Atrophy is a progressive disease that causes muscle weakness and muscle wasting throughout the body. There are several forms of the disease. Type I, also known as Werdnig-Hoffman disease, is the most severe affecting infants prior to birth. Type II is usually identified in early childhood and Type III in adolescence or adulthood. Children with Type I usually die within the first two years of life but the life span is longer for children with Type II and III. Infants and young children with SMA fail to develop or lose global developmental skills, have low muscle tone, progressive weakness with limited movement and poor feeding and respiration. They are at risk for breathing problems, failure to thrive, orthopedic complications, and require assistive technology for self care, care giver support, communication and mobility. End of life care as part of early intervention services are critical to the child and care givers when working with children with SMA and their families.

- Spinal Muscular Atrophy Foundation

**Congenital, Acquired or Autoimmune**

**Arthrogryposis Multiplex Congenita**
Arthrogryposis is descriptive term for a group of disorders that cause fiberous contractures of and malformations around the joints. At least two joints are involved, but typically many joints are involved including the joints of the limbs, hands and feet and frequently the jaw. The characteristics of arthrogryposis include atypical muscle development or absences of some limb muscles, joint contractures and rigidity, joint dislocations (hips and knees) and fusions, bone changes in the limbs and skull, and structural malformations in the heart, urinary system, respiratory system, and skin. Early intervention services focus on promoting strength and motor development, mobility, self care, adaptive development, prevention and management of hip dislocation and joint contractures and communication if structures of the jaw and palate are
involved. Physical therapy, occupational therapy, and speech-language therapy are needed to facilitate the child’s development. Assistive technology and environmental accommodations are important to maximize development and function.

- Avenues: A National Support Group for Arthrogryposis Multiplex Congenita
  http://www.avenuesforamc.com/

**Plagiocephaly**
A condition that is characterized by an abnormal flattening or asymmetrical shape of the back side of the head. This condition can be caused by abnormal forces from positioning in utero, torticollis, or persistent positioning of back lying, extended time in car seats or other positions that infants are placed in that create extended pressure on the back of the head. Infants born prematurely are more susceptible to plagiocephaly because of the long term back positioning in the neonatal intensive care unit and importance of back lying for sleep to avoid Sudden Infant Death Syndrome. Treatment does not usually require surgery and includes varying infant positions and if severe enough, reshaping of the head with specialized helmets called orthotic cranioplasty. It is important to distinguish plagiocephaly from craniosynostosis which is a premature closure of the cranial bone connections or sutures. Craniosynostosis usually requires surgery and can effect brain growth.

- Plagiocephaly.info
  http://www.plagiocephaly.info/default.htm
- Neurosurgery Today
  http://www.neurosurgerytoday.org/what/patient_e/positional_plagiocephaly.asp

**Juvenile Rheumatoid Arthritis (JRA)**
Juvenile Rheumatoid Arthritis is an autoimmune disorder and chronic disease in children affecting the joints and sometimes the whole body system including internal organs. Children with JRA have episodes of joint inflammation, stiffness, and pain that lead to temporary muscle weakness and joint contractures. More permanent muscle weakness, joint contractures, joint damage and changes in growth are seen with frequent or long lasting episodes over time. The systemic form of JRA, or Still’s disease, causes fevers, skin rashes, and inflammation effecting functions of the heart, spleen and liver. The impact on development depends on the severity of the condition but can affect motor development, mobility, and adaptive development. Children with JRA may need support around pain management, assistive technology for mobility and self-care, and care giver support.

- Arthritis Foundation
  http://ww2.arthritis.org/conditions/DiseaseCenter/jra.asp

**Oral-Facial Clefts**
Incomplete closing of the oral facial structures during fetal development results in clefts to the lip and palate. Infants may be born with only a cleft lip, cleft palate (rare) or both the lip and palate may be involved. About 6,800 infants are born in the United States each year with oral-facial clefts. It affects infants of Asian ancestry and certain Native American groups followed by Caucasians and least frequently affecting African American infants. The cause of oral-facial
clefts is not well understood. Many genetic syndromes include oral-facial clefts, but not all children with this condition have genetic syndromes. Clefts that are not part of a syndrome are thought to be the result of genetic factors or fetal exposure to medications, infections, illicit drugs, smoking and alcohol. Infants with oral-facial clefts have difficulties in feeding, ear infections with hearing loss, speech and language delays, and dental problems. Clefts can be surgically repaired usually by timed, multiple procedures starting early in infancy.

- Cleft Palate Foundation  
- March of Dimes  
- Smile Train, United States  
  [http://medpro.smiletrain.org/medpro/partners/us/widesmiles.htm](http://medpro.smiletrain.org/medpro/partners/us/widesmiles.htm)

**Spina bifida**

Spina bifida, also called myelomeningocele or myelodysplasia, is a congenital disorder of the formation of the spinal cord and surrounding structures in fetal development. There are different forms effecting nerve function. A child may have a minor gap in a vertebra (occulta) or a bulge in the meninges or spinal cover (meningocele) that causes no disruption in spinal nerve function. If the formation was less complete, a protrusion of the spinal coverings and nerves outside the body in the low back is seen and is called a myelomeningocele. This condition causes varying degrees of spinal nerve and body system disruption. Children with myelomeningocele require surgery to close the open structures. Surgical repair of the myelomeningocele has no impact on improving function, but prevents further damage or infection. Most children with myelomeningocele have hydrocephalus and may have shunts placed to manage the fluid in the brain. The motor, adaptive and cognitive areas of development of young children are usually the most affected but communication and social emotional development can also be affected. Children with spina bifida have a high incidence of latex allergies, and development of spinal cord restrictions (tethering). They can also have learning disabilities, gastrointestinal disorders and skin breakdown. Assistive technology and environmental modifications are often needed to access home, community, and school as well as services to promote mobility and self care. Care giver support is also an important component of a comprehensive intervention approach.

- Spina Bifida Association  

**Torticollis**

Congenital Muscular Torticollis (CMT) is an asymmetrical posturing of the head in a tilted and rotated position because of the shortening of a major muscle group in the neck called the sternocleidomastoid muscle. Some infants also have deformations in the cranial and facial structures (plagiocephaly) and rarely, deformations of the outer and inner ear on the affected side. The reason for the shortening of the neck muscle is not always clear, but can be from positioning in the uterus, disruption in blood supply to the muscle, injury, infection or genetic factors. Other serious disorders can cause torticollis and it is important that other syndromes, vision problems, tumors, and brain injuries are ruled out as the cause before intervention begins. The impact on development is primarily in motor development but it can also affect vision

www.teachingei.org

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because of the asymmetry of the head and hearing if the ear is affected. Interventions include early exercises to stretch the muscles, the use of helmets to reshape the head, and rarely surgery are also used to treat the condition.

- Tummy Time Tools
- Deformational Plagiocephaly & Cranial Remolding in Infants
  http://www.pediatricapta.org/graphics/Plagiocephaly.pdf
- Torticollis Kids
  http://www.torticolliskids.org/

Trauma

**Brachial Plexus**
Birth trauma to the lower cervical and upper thoracic nerve root and coverings that innervate the arm cause brachial plexus injuries in newborn infants. The condition varies in severity from affecting parts of the arm and hand to paralysis in the whole arm. Brachial plexus injury usually follows a difficult birth where the arm and shoulder girdle became overstretched, tearing the nerve covering and causing nerve compression resulting in swelling and bleeding around the area of injury. An infant’s arm and shoulder are immobilized for the first week to ten days after birth to allow healing of the injury and then intervention begins to increase movement in the arm and promote arm use and motor development. Depending on the severity, typical arm function returns with early intervention and nerve regeneration within the first year of life. More severe involvement may require surgery and result in permanent reduction of arm and hand function.

- Brachial Plexus Palsy Foundation
  http://www.brachialplexuspalsyfoundation.org/index.html
- United Brachial Plexus Network
  http://www.ubpn.org/

**Spinal Cord Injury**
Bruising, compression, tearing or a complete break in the spinal cord causes loss of sensation, weakness, or paralysis in the limbs and body. The severity or the number of body structures and limbs involved depends where on the spinal cord the injury occurs. If the injury is high in the neck, breathing becomes compromised as well as paralysis of all the muscles below the shoulders. In young children, spinal cord injuries occur because of trauma (motor vehicle accidents, physical abuse, and falls), instability and developmental anomalies in the cervical vertebrae or blood supply, tumors, and infection. It is common for children to also have brain injuries because of the event that caused the spinal injury. Once the injury occurs the spine is stabilized with surgery and bracing. In young children all areas of development are at least initially affected because of the injury, the possibility of brain injury, and long hospital stays. An array of services is needed to promote developmental skills, mobility and self-care. Children with spinal cord injuries often use assistive technology such as wheelchairs and environmental modifications to access home, school and the community. Family members often need caregiver support.
• Christopher and Dana Reeve Foundation http://www.christopherreeve.org/

Traumatic Brain Injury (TBI)
Traumatic brain injury describes injury to the brain that occurs because of physical force and that is not due to birth, disease or infection. In young children, TBI occurs because of falls, motor vehicle accidents, physical abuse, or recreational related accidents. The brain is injured in more than one place because the initial forces to the head cause bleeding and swelling reactions that damage brain tissue more diffusely. Sometimes TBI is accompanied by spinal cord injuries because of the precipitating accident or abuse. Initial medical management includes monitoring of swelling and bleeding and might require surgery. The severity varies greatly and is determined by the structures and scope of the brain areas injured, but often all areas of development are affected by TBI. Vision, hearing and motor impairments may be present as well as difficulty with communication, memory, learning, planning and behavior. Children with TBI may have seizures, loss of balance and coordination, increased muscle tone (spasticity) in some of the limbs and attention problems. The amount of developmental delay and recovery from TBI also varies greatly with each child. Some recover fully; while others have life long deficits in brain function that require ongoing care. Brain Injury Resource Foundation, Children and Brain Injury http://www.birf.info/home/library/pediatrics/ped_chiltrau.html

Services and Support
Although these conditions may be seen less often in early intervention and early childhood settings, services and supports are important to help families feel less isolated and get support to promote their child’s development. Some aspects of service coordination and early intervention services that may be prioritized for families with children with these less common conditions include
• Connection to family support like support groups
• Education and information on the disorder and services
• Care coordination for medical aspects of condition
• Link to (or set up) medical home
• Information and coordination around assistive technology
References

Genetics Home Reference  

National Institutes of Neurological Disorders and Stroke  
http://www.ninds.nih.gov/


