

CHARGE Syndrome and Physical Therapy

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A successful physical therapist creates a relationship of mutual trust. This respect is essential when working with children and young adults with CHARGE syndrome. The goal of physical therapy is to help these children gain confidence so they can participate in life activities through adaptation and compensation for their physical and sensory challenges. David Brown (2003) states, "Children with CHARGE are also likely to be amongst the most truly 'multi sensory impaired' people you meet, having difficulties not just with vision and hearing but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell" (p. 1). Sensory and motor challenges are the result of abnormal physical structures, impaired sensory pathways, and underdeveloped processing centers. These impairments lead to deficits in balance and development of motor skills, and therefore negatively impact participation in social, academic, and vocational activities.

Balance

The visual system is the primary component for balance and movement (Haibach & Lieberman, 2013). It alerts us to our environment for anticipation and adaptation, and gives awareness of the head and body in the environment. Individuals with CHARGE syndrome have been documented to have colobomas of the eyes and impairments in the ability to process visual information (Blake & Prasad, 2006).

The vestibular system consists of structures in the inner ear, otoliths and semicircular canals, that take in information about head position and movement (Haibach & Lieberman, 2013). Individuals with CHARGE syndrome often have underdeveloped or missing semicircular canals, involvement of cranial nerve VIII, and difficulty processing this information (Brown, 2003).

The somatosensory system is composed of various sensors or input mechanisms through touch, pressure and stretch of the joints, muscles and tendons. Proprioception is part of the somatosensory system that provides information when an individual is working against resistance or against gravity (Brown, 2006). Individuals with CHARGE syndrome often have low tone, decreased strength due to impaired muscle length and muscle imbalances, and abnormal (too much or too little) joint mobility.

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Information from the sensory systems is gathered and processed in the brain stem, resulting in appropriate motor responses (Hall, Umphred & West, 2007). An appropriate balance response could be a small ankle movement, a large hip movement, or taking a step to prevent a fall. An accurate motor response starts with accurate sensory information. When these systems are impaired, balance is a daily struggle.

Movement facilitates growth and development. From birth onwards, movement experiences stimulate the vestibular and somatosensory systems (Benabib, 2004). The infant moves its head from side to side to clear its airway. Proprioceptive input occurs during kicking against the surface while it's resting on a parent's chest or in its carrier. Visual and hearing impairments limit the child's curiosity to explore his or her environment, and therefore, decreases the motivation to move. Additionally, children with CHARGE syndrome are deprived of early movement due to long hospitalizations for medical issues.

Motor Learning and Balance

As children with CHARGE syndrome develop, it is important that their balance is challenged so that they have opportunities to enhance their sensory experience and practice motor responses. Movement and balance experiences will allow these children to know the strengths and limits of their own visual, vestibular, and somatosensory systems (Benabib, 2004). It will also allow them to learn how to adapt and compensate for compromised sensory systems. Motor learning will not occur without motor challenges (Gordon & Magill, 2012).

A typically developing child who is learning to walk will rock side to side, attempt to let go of supportive surfaces and fall. They will repeat this, and in the process make slight adaptations to be more successful (Gordon & Magill, 2012). The child uses his eyes to explore the environment, including changes in the surface of the ground, or will learn that the sensation in their feet will tell them how to adjust to obstacles. This process is the same for children with CHARGE syndrome. The most important aspect is that the more motor experiences these children have, the more effective they will become in using their residual vision, vestibular sense, and proprioception.

Parents, caregivers, and clinicians may observe certain 'behaviors' as the child is developing due to anxiety or insecurity related to balance. A child who twists his legs around himself might be seeking proprioceptive information as to feel more secure about where his body is in space (Brown, 2006). A child lying on the ground, might be trying to stabilize himself as to be able to use his eyes more effectively (Benabib, 2004). A student who has a hard time standing still might be shifting his body and head to gain as much proprioceptive and vestibular information as possible (Brown, 2006).

Balance activities may not be rewarding or fun when they are unreasonably challenging. With the goal in mind of helping these individuals to improve motor skills, the physical therapist needs to create motivating and rewarding opportunities. The

following recommendations will guide physical therapists in thinking about how to approach a child with CHARGE syndrome.

Recommendations

- 1) Evaluate and determine the sensory needs of the child. A sensory program or sensory routine may need to be the first step in the session. A consult by an experienced professional, such as an occupational therapist, is a valuable tool.
- 2) Know and use the child's most reliable communication method: photos, tactile symbols, sign language, tactile sign, or speech. Speaking with the family, classroom teacher and/or speech therapist will assist in knowing this information. This will allow the child to develop a relationship with his physical therapist, and gain trust.
- 3) Everyone working with the child should know the specific behavior program as well as the academic and motor goals. Consistency and a team approach are key components to guiding the child toward success.
- 4) Create a consistent session routine with clear expectations. A schedule of the activities within the session can be helpful. It will also give the child the opportunity to anticipate the activity. The child will also need a consistent routine for transitioning to and from the physical therapy sessions.
- 5) Allow the child to explore within safe parameters. Constant physical and verbal cues can overwhelm the child. Provide one clear instruction or goal, allowing the child to focus, and give him time to react.
- 6) Give the child the opportunity to participate in activities that will allow adaptation and multiple repetitions to be successful (Gordon & Magill, 2012). For example, walking on grass, roller skating, or using a standing scooter will allow repetition of weight shifting and balance responses. Girardi (2011) *shows various functional activities that challenge the balance system.*
- 7) The child needs to know that they are successful. Using praise and providing activities that give innate feedback can be very successful for children with CHARGE syndrome. The goal is to instill comfort and confidence in the child. Maintain reasonable expectations and allow for slow progress.
- 8) Due to poor vision, low tone, decreased postural strength and endurance, children and young adults with CHARGE syndrome are at risk for orthopedic issues. The child's seating in the classroom and home should be assessed. A supportive foot surface and armrests provide a good base of support to improve postural alignment. Visual materials should be presented in the student's visual field. Slanted or elevated surfaces can be beneficial in preventing a collapsed forward posture.
- 9) Provide the child with the opportunity to change positions (i.e. standing, walking, sitting on a ball, etc.) every 15-20 minutes if they appear to be having difficulty focusing on the activity or task. This will change the child's arousal level, and give opportunity for different postural muscles to activate.

10) Yoga provides a child who has low tone, balance, strength and coordination deficits, with proprioceptive input through various poses that can improve body awareness and strength. It also gives the child a routine to follow that is enjoyable, and can be done as a group to encourage socialization.

11) Many individuals with CHARGE syndrome develop a flexed forward posture. Activities that involve weight bearing through the arms for upper back, abdominal, and arm strengthening should be initiated early in an attempt to prevent postural weakness that may occur. The child should have regular follow-up with an orthopedist or physiatrist to monitor skeletal structure, especially in the spine, hips, ankle and feet.

Like many typical children, there are multiple factors including medical complications and guarded lives that limit a child with CHARGE syndrome to experience the world. They should be given opportunities to explore, make mistakes, and learn from those mistakes. As the child with CHARGE syndrome gains skills, he or she will be able to participate more at home, school, and the community with confidence.

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