

Physical Therapy and CHARGE syndrome

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Physical Therapy

- Physical therapy is "a dynamic profession with an established theoretical and scientific base and widespread clinical applications and the restoration, maintenance, and promotion of optimal physical function."*

Role of a physical therapist

- Diagnose and manage movement dysfunction and enhance physical and functional abilities*
- Restore, maintain and promote, not only optimal physical function, but optimal wellness and fitness and optimal quality of life as it relates to movement and health*
- Prevent the onset, symptoms, and progression of impairments, functional limitations, and disabilities that may result from diseases, disorders, conditions, or injuries*

What are the physical challenges of individuals with CHARGE?

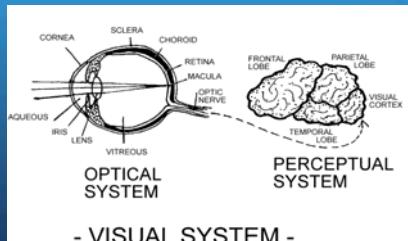
- Cardiac
- Respiratory
- Tone
- Range of motion
- Posture
- Balance
- Coordination
- Vision
- Hearing
- Cranial Nerve
- Other comorbidities:
 - Surgical intervention
 - Developmental delay
 - Medication side effects
 - Nutrition

The complex BALANCE system

- Balance is maintained through a complex interaction of the Central Nervous System (CNS) and the three primary sensory systems:
- VISION
- VESTIBULAR
- SOMATOSENSORY

Visual System

- A strong inhibitor of the other two systems (vestibular and somatosensory)



- VISUAL SYSTEM -

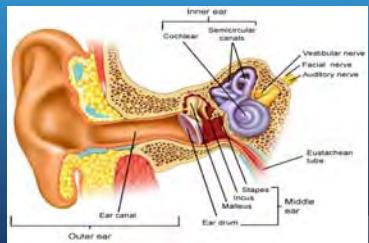
Somatosensory System

- The perception of sensory stimuli from the skin and internal organs of the body
- It is the fastest of the three sensory systems

- light touch
- proprioception
- deep pressure
- temperature
- kinesthesia
- vibration

Vestibular System

- The conflict resolver between the other two systems (vision and somatosensory)



Why do individuals with CHARGE have such a difficult time with balance?

- Vision**
 - Colobomas
 - Retinal Detachments
- Somatosensory**
 - Low tone (joint laxity and decreased proprioception)
 - Sensory processing issues (defensiveness, regulation)
- Vestibular**
 - Semi-circular canals
 - Cranial Nerve VIII impairment

If only it were so simple:

Neural connections
System coordination

Motor Control

- Area of study dealing with understanding of the neural, physical, and behavioral aspects of movement.

Motor Control

- Stimulus identification =>
- Response Selection (stimulus processing) =>
- Response Programming (motor output)

Balance Control

Sensory **Motor**

Where am I? **What am I going to do?**



Motor Learning

- The improvement of a given functional task resulting from practice or experience. In order for the motor learning to be meaningful, the performance of the new task must be reproducible over time, performed in multiple environments and generalized to similar activities

Principles of Motor Learning

- Amount of practice
- Learner needs to be actively involved
- Task conditions (speed, timing, and environmental conditions)
- Adaptability
- Reinforcement is required
 - Knowledge of results
 - Feedback can be decreased over time

CHARGE and Learning

How do you make a PT session successful with students with CHARGE syndrome?



UNDERSTANDING and KNOWING your student

- Mode of communication
- Behaviors and behavior plan
- Need for structure and routine
- Obsessive Compulsive Disorders
- Limitations



Methods of Developing a Relationship

- Team approach - Communicate with teacher, family members, speech therapist, occupational therapist, Orientation and Mobility specialist, Behaviorist
- Communicate with your student
 - Sign language (visual and tactile)
 - Photos
 - Verbal



Methods of Developing a Relationship



- Avoid power struggles
- Give lots of positive feedback
- Clear expectations
- Sandwiching non preferred activities in between preferred activities
- Choice making
- Mood induction

How should we approach our students?

- Be flexible
- Be patient
- Limit expectations
- Accept slow progress

How to justify physical therapy

- Need to have measurable and functional goals.
 - Need to show progress
- They need to relate to a “disability”
 - Unable to participate in a school sports team
 - Unable to safely play on playground equipment
 - Unable to use stairs for fire safety
 - Unable to walk outside without falling
 - Unable to negotiate curbs or ramps in the community
 - Strength and endurance limit the ability to participate in vocational work

Expanded Core Curriculum

- How does physical therapy relate to access to education?
- It's not just about being able to walk from one class to another or negotiating stairs!

Expanded Core Curriculum

- The National Advisory Council of the Nation Agenda states that blind and visually impaired students have two sets of essential educational experiences: (1) regular curriculum offered to all students-the core curriculum - and (2) learning experiences required because of vision loss - the expanded core curriculum.

ECC Components

- Compensatory, Functional Skills, Lang/Communication
- Career Education
- Social Interaction/Group
- Independent Living Skills
- Leisure and Recreation/APE
- Orientation and Mobility
- Assistive Technology
- Sensory Efficiency
- Self-Determination

How does the ECC relate to physical therapy?

- Compensatory, Functional Skills, Language/Communication
- Career Education
- Social Interaction/Group
- Orientation and Mobility
- Sensory Efficiency
- Self-Determination

How can physical therapists advocate for their students with CHARGE syndrome

- Understand the importance of functional activities
- Understand the importance of improving balance and coordination for safety in all environments
- Understanding the importance of improving and maintaining flexibility, strength and endurance

Recommendations:

- Exercises: strengthening and stretching
- Follow-up with an orthopedist and/or physiatrist for monitoring of scoliosis, hip integrity, kyphosis, and need for foot bracing.
- Proper seating: to decrease postural fatigue (armrests, feet on floor), prevent postural mal-alignments
- Changes in position for sensory input and to decrease postural fatigue

Balance training in a setting in which the individual can be an active learner. Give the individual with CHARGE syndrome the opportunities to explore, fall, progress and LEARN.