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Down Syndrome 101 for Pediatric OTs

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About Cara:

Mother to two children who have autism, sensory processing disorder. Younger son has mitochondrial disease and Tourette’s disorder. He’s GJ tube fed.

Occupational Therapy Advisor & Contributing Author for AutismAsperger’s Digest, Asperkids, Autism File Magazine

Author of The Pocket Occupational Therapist for Caregivers of Children with Special Needs; The Special Needs SCHOOL Survival Guide book; and Weighted Blanket Guide; Sensorimotor Interventions, & Interoception: Sensing My World From the Inside

Founder of Aspire Pediatric Therapy, LLC; Route2Greatness, LLC; and The Pocket Occupational Therapist

OTD Candidate
Books

Cara’s Books
Objectives

After this course, participants will be able to:

1) Name the characteristics of Down syndrome including specific medical and functional concerns.
2) List accommodations and treatment strategies for those with Down syndrome.
3) Identify functional goals for children with Down syndrome across all settings.

“In every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Typically, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent. Down syndrome occurs when an individual has a full or partial extra copy of chromosome 21.”

“ This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes, and a single deep crease across the center of the palm – although each person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all.”

https://www.ndss.org/about-down-syndrome/own-syndrome/
Important to remember: HOPE

- Down vs. Down’s syndrome

- Down syndrome is named for the English physician John Langdon Down, who characterized the condition, but did not have it.

- While Down syndrome is listed in many dictionaries with both popular spellings (with or without an apostrophe s), the preferred usage in the United States is Down syndrome. The AP Stylebook recommends using “Down syndrome,” as well.

- ALWAYS use person-first language

- The word ‘retarded’ is NEVER appropriate!
  - Use new and accepted terminology such as: intellectual or cognitive disability

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Down syndrome fast facts

- The most common chromosomal condition diagnosed in America.
- 6,000 babies are born each year
- According to the Centers for Disease Control, Down syndrome occurs in about 1 out of every 700 babies. (Parker et al., 2010)

- Usually caused by an error in cell division called nondisjunction
- Occurs at conception and NOT related to actions of the mother.
- 80% of children with Down syndrome are born to women under 35 years of age

- Life expectancy for people with Down syndrome has increased dramatically in recent decades – from 25 in 1983 to 60 today.
  (National Down Syndrome Society, 2017)
About chromosome 21

- The smallest human chromosome, spanning about 48 million base pairs (the building blocks of DNA)
- It represents 1.5 to 2 percent of the total DNA in cells
- Was the second human chromosome to be fully sequenced
- According to Scientific American: “Down syndrome is associated with more than 80 physical and mental problems, including congenital heart disease, an increased risk for certain leukemias, and immunological deficiencies.”

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About Chromosome 21

- Mutations in 14 known genes on chromosome 21 play roles in one form of Alzheimer's disease, amyotrophic lateral sclerosis and progressive myoclonus epilepsy, among other diseases.
- Also on Chromosome 21 are genes that are as yet unidentified, but known to be responsible for five so-called monogenic disorders, including two forms of deafness and Usher and Knobloch’s syndromes.

- Scientific American
Occurrence specifics

- Occurs in people of all races and economic levels
- A 35 year old woman has about a one in 350 chance of conceiving a child with Down syndrome
- The chance increases gradually to 1 in 100 by age 40
- At age 45 the incidence becomes approximately 1 in 30. The age of the mother does not seem to be linked to the risk of translocation.

(National Down Syndrome Society, 2017)

Types of Down syndrome – caused by error in cell division called ‘nondisjunction’

- 95% of the time there is presence of a ‘free’ extra 21st chromosome
  - This is called ‘Trisomy 21’ Down syndrome is usually caused by an error in cell division called “nondisjunction.”
  - Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate
  - The extra chromosome is replicated in every cell of the body.

(American Academy of Pediatrics, 2011)
Types of Down syndrome – caused by an error in cell division called ‘nondisjunction’

- 3-4% is due to extra chromosomal unbalanced translocation of the 21st chromosome
  - A partial or full copy of chromosome 21 attaches to another chromosome
  - Usually chromosome 14

- 1-2% due to mix of two cell lines, some with the usual 46 chromosomes and the other with trisomy of 21st chromosome
  - MOSAICISM (or mosaic Down syndrome, may be more mildly affected)

(American Academy of Pediatrics, 2011)

Statistical information

- Once a woman has given birth to a baby with trisomy 21 (nondisjunction) or translocation, it is estimated that her chances of having another baby with trisomy 21 is 1 in 100 up until age 40.
  - Remember that the likelihood of the first child with the condition is 1 in 700 infants

- The risk of recurrence of translocation is about 3% if the father is the carrier and 10-15% if the mother is the carrier.

- Genetic counseling can determine the origin of translocation.

(National Down Syndrome Society, 2017)
How is it diagnosed?

- Prenatally
  - CVS first trimester between 9 and 14 weeks gestation
  - Amniocentesis during second trimester between 15 and 20 weeks gestation

- At birth
  - Identification of physical characteristics such as:
    flattened facial profile, upward slant to the eyes,
    single deep crease in palm of hand, low muscle tone
  - Completion of karyotype from blood sample
  - FISH test

At birth:

- Brain structure and function is NOT fixed at birth, it can be influenced by activity, social, early intervention, etc.

- We know children with Down syndrome will be delayed so use their individual baseline.

- Babies should have eye and ear tests

- Compassion and reassurance for the family

- Discuss brain plasticity and importance of early intervention with families
Co-morbid conditions

- GERD
- Celiac Disease
- Low tone
- 15% have underactive thyroid gland
- Vision problems
- Hearing problems
- Increased rate of leukemia
- Autism
- Half of the population with Down syndrome develop Alzheimer’s disease by age 50-60
- ADHD
- OCD

(National Down Syndrome Congress, 2017)

Cardiac considerations

- 40-50% have congenital heart disease
- Atrioventricular septal defect is most common
  - The atrioventricular (AV) valves are abnormal to varying degrees ranging from a cleft or gap in the mitral valve to severe malformation of the valves in which there is a single common valve entering both ventricles
- Ventricular septal defect second most common
  - If severe, damage to pulmonary arteries must be repaired early via surgery
- Mitral valve prolapse
- Arrhythmias

(Lane, 2005)
Medical Problems

- Hearing problems 75%
- Vision problems 60%
- Cataracts 15%
- Refractive errors 50%
- Obstructive sleep apnea 50–75%
- Otitis media 50–70%
- Congenital heart disease 40–50%
- Hypodontia and delayed dental eruption 23%
- Gastrointestinal atresia 12%
- Thyroid disease 4–18%
- Seizures 1–13%
- Hematologic problems:
  - Anemia 3%
  - Iron deficiency 10%
  - Transient myeloproliferative disorder 10%
  - Leukemia 1%
- Celiac disease 5%
- Atlantoaxial instability 1–2%
- Autism 1%
- Hirschsprung disease 1%

AAI (Atlanto-axial instability)

- United States, atlantoaxial instability (AAI) with or without subluxation has been reported in as many as 10-30% of individuals with Down syndrome (Alvarez et al., 2016)
- Either congenital absence or laxity of the transverse atlas ligament (which may be associated with congenital anomalies of the odontoid bone) must be considered
- Pain, stiff neck, torticollis, gait disorder, progressive paralysis
- Weakness can occur in legs first
- Compression of cervical roots and/or spinal cord
- Tingling, numbness, positive Babinski sign
## Developmental chart

<table>
<thead>
<tr>
<th>Area</th>
<th>Milestone</th>
<th>Range for Children with Down Syndrome</th>
<th>Typical Development</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gross motor</td>
<td>Sits alone</td>
<td>6-30 mo.</td>
<td>5-9 mo.</td>
</tr>
<tr>
<td></td>
<td>Crawls</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gross motor</td>
<td>Walks alone</td>
<td>1-4 years</td>
<td>9-18 mo.</td>
</tr>
<tr>
<td>Language</td>
<td>First words</td>
<td>1-4 years</td>
<td>9-18 mo.</td>
</tr>
<tr>
<td>Self-help</td>
<td>Finger feedings</td>
<td>10-24 mo.</td>
<td>7-14 mo.</td>
</tr>
<tr>
<td>Self-help</td>
<td>Bowel control</td>
<td>2-7 years</td>
<td>16-42 mo.</td>
</tr>
<tr>
<td>Self-help</td>
<td>Dresses self</td>
<td>3.5-8.5 years</td>
<td>3.25-5 years</td>
</tr>
<tr>
<td></td>
<td>unassisted</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Adapted from [www.nhs.org/resources/therapies-development/early-intervention](http://www.nhs.org/resources/therapies-development/early-intervention)

## Overall considerations:

- Good social interactive skills
- Sensitive to social cues *especially negativity
- Learn self-help and ADL skills
- Weak fine motor skills
- Weak expressive verbal skills
- Risk of vision and hearing impairments
- Speech and language delays relative to non-verbal mental abilities
Developmental considerations

- Delayed development especially apparent in early years
- Behavior issues
- Speech deficits in both receptive and expressive language
- Genetic causes of DS are associated with characteristic outcomes, such as relative strengths in visual-spatial skills and relative challenges in motor planning (Dunhauer & Fidler, 2011)

Why low tone/laxity?

- Collagen is a protein that makes up ligaments, tendons, cartilage, bone, support structure of skin
- Collagen type VI gene is encoded on chromosome 21
- Ligaments and laxity are greatly affected
- Looser joints, hip, knee, and others may require AFOs or braces
- Teeth come in later and may be pointed or incorrect order
Language

- Underpins social & cognitive development
- Remembering, thinking, reasoning
- Emotions, interoception, anxiety
- Social skills involve language

What IS talk?

- Communication = getting message across
- Gestures = non-verbal, requests, facial expressions
- Vocabulary = making meaning out of words
- Grammar = stringing words together to learning and complex meaning/communication
- Speech = how we express ourselves
- Understanding is ahead of expression
- Make themselves understood
- Deficits in speech production, sounding words
Cognitive function

- Delays become apparent by school-age
- Nonverbal skills can be a strength
- Memory skills strong
- Attention deficits
- Executive function difficulties
  - Metacognition
- Concrete thinkers so avoid ‘gray’ or abstract concepts when teaching
- Numbers and abstract concept learning may be delayed

Unique pattern of development across the lifespan

- Characteristic features associated with DS (cheerful, social nature) are personality assets.
- Children are at a lower risk for psychopathology compared to other children with ID; families report lower levels of stress and a more positive outlook.
- In youth, externalizing behaviors may be problematic, whereas a shift toward internalizing behaviors emerges with maturity.
- Changes in emotional/behavioral functioning in adulthood are typically associated with neurodegeneration and individuals with DS are higher risk for dementia of the Alzheimer’s type.
- Possess many unique strengths and weaknesses that should be appreciated as they develop across the lifespan.

(Grieco, et al., 2015)
Behavioral concerns

- Decreased communication
- Decreased interoception
- Increased tendency to show behavioral outbursts as children
- Use of visual schedules
- Consistency across settings with clear rules and expectations
OT’s role

- ADLs: feeding, dressing, bathing
- Motor skills (both gross and fine motor)
- Play
- Infant’s feeding
  - Hypotonia and muscles in mouth, cheeks, lips, tongue, and be affected
- Positioning and adaptations for successful ADLs
- Leisure
- Fine motor in the classroom
- Sensory needs**
- Job training
- Advocacy and transition

(Bruni, 2001)

OT’s role

- Praxis including difficulty selecting motor responses
- Decreased motor function on tests such as the Vineland Adaptive Motor Scales
- “Not using perceptual information in order to plan their reaching strategy as effectively as children with other types of developmental disabilities,” (Fidler, et al., 2005)
  - errorless learning techniques
  - targeting other areas of strength (i.e., visual-spatial processing and social functioning) in order to prevent task abandonment in Down syndrome

- Toileting (Dolva et al., 2004)
Feeding

- Stability
  - Low tone
  - Positional recommendations
    - Swaddling in early infancy and working to build skills at midline in later infancy

- Floppy or low tone
  - Difficulty latching on
  - Alertness for feeding
  - Swallowing air
  - Texture transition and tolerance

- Protruding tongues
  - *NOT big tongues
  - Small mouths and low tone

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Feeding

- Appropriate oral-motor toys

- Aspiration is common (Comrie, 2015)
  - Silent aspiration
  - Decreased cough
  - Increased risk of infection after aspiration

- Suck, swallow, breathe coordination

- Sensory concerns when initiating feeding

- Reflux can be traumatic (behavior as communication)
Sensory processing

- Massage to gums and oral musculature
- Oral-defensiveness
  - Mouth stuffing
  - Drooling
  - Not used to new sensations, tastes, and textures
- Co-morbid sensory processing disorder

Vision

- More than half have visual deficits
- Cataracts (even at birth)
- Crossed eyes
- Strabismus (squint)
- May need bifocals
- Upward slanting of the eyelids
- Folds of skin between eye and nose
  (Woodhouse, 2017)

- Decreased accommodation, decreased acuity, contrast sensitivity (may not see pencil lines on paper) (Watt, et al., 2015)
Fine motor

- Bruni’s ‘House’ model:
  - Building blocks (ground) are: stability, motor coordination, sensation
  - Top floor: dexterity and daily living skills
    - School, self-help, household tasks, leisure activities

- Hands of children are often smaller and the fingers shorter and stubbier
- Thumb is often set lower down.
- Some children may not have all of the usual wrist bones. These factors will inevitably affect the ability to hold and manipulate objects. (Alton, 2005)

- Stability, hand exercises, strengthening, positional changes

Work and IADLs

- Provide a consistent routine
- Outline clear expectations and directions
- Use visual charts and instructions
- Learn the individual’s strengths and weaknesses and build on them
- Meet frequently
- Encourage inclusion and cooperation from ALL other staff
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References:


References

- http://pediatrics.aappublications.org/content/pediatrics/128/2/393.full.pdf

References

- https://www.ndsccenter.org/programs-resources/health-care/
- Lane, J. R., M.D., F.A.C.C., Director, Adult Congenital Heart Service, Akron Children’s Hospital, Akron, Ohio, 2005
Resources for therapists and caregivers

- Down Syndrome Research Foundation
- Down’s Syndrome Association
- Global Down Syndrome Foundation
- National Association for Down Syndrome
- National Down Syndrome Society
- National Institute of Health’s Genetics Home Reference

Job and workplace inclusion:

- APSE: http://apse.org/
- JAN: https://askjan.org/
- NDSS helpline: https://www.ndss.org/rescat_lifespan/tax/education/
- NCWD: http://www.ncwd-youth.info/