The Implications of Down Syndrome on Health Support Needs

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What is Down Syndrome?
Common Physical Traits of Down Syndrome

- low muscle tone
- small stature
- an upward slant to the eyes
- a single deep crease across the center of the palm
- Seen at birth but confirmed with karyotype
How Common is Down Syndrome?

- One in every 691 babies in the United States is born with Down Syndrome, making Down Syndrome the most common genetic condition.
- Approximately 400,000 Americans have Down Syndrome
- About 6,000 babies with Down Syndrome are born in the United States each year.
Trisomy 21 (Nondisjunction)

Typical Cell Division

Trisomy 21 (Nondisjunction) Cell Division
MOSAICISM

Diagram showing the process of fertilization and the resulting mosaic chromosome pattern.
An unbalanced translocation with the arrows pointing to the three copies of chromosome 21
Causes of Down Syndrome

- Risk for nondisjunction increases with age.
- Due to higher birth rates in younger women, 80% of children with Down syndrome are born to women under 35 years of age.
- The additional partial or full copy of the 21st chromosome which causes Down syndrome can originate from either the father or the mother. Approximately 5% of the cases have been traced to the father.
Causes of Down Syndrome

- Occurs in people of all races and economic levels.
- In one third of cases of Down syndrome resulting from translocation, there is a hereditary component - accounting for about 1% of all cases of Down syndrome.
- The age of the mother does not seem to be linked to the risk of translocation. Most cases are sporadic - chance – events. However, in about one third of cases, one parent is a carrier of a translocated chromosome.
General Health

- Life expectancy for people with Down syndrome has increased dramatically in recent decades - from 25 in 1983 to 60 today.
- Multiple co-morbid conditions
  - ADHD
  - Autism
  - Mental health disorders
  - Others
Alzheimer’s Disease

- Not typical of the aging process
- Increased risk because of the shared chromosome
- Screen beginning at age 40 or if there is a cognitive change
- Maximize the independence and quality of life
- Loss of short term memory and inability to learn and recall new information
The Span of Alzheimer’s Disease: Early Stage

- Short term memory loss (difficulty recalling recent events, learning and remembering names and keeping track of the day or date; asking repeated questions or telling the same story repeatedly)
- Difficulty learning and retrieving new information
- Expressive language changes (word finding difficulties, smaller vocabulary, shorter phrases, less spontaneous speech)
- Receptive language changes (difficulty understanding language and verbal instructions)
The Span of Alzheimer’s Disease: Early Stage (cont.)

- Worsened ability to plan and sequence familiar tasks
- Behavior changes
- Personality changes
- Spatial disorientation (difficulty navigating familiar areas)
- Worsened fine motor control
- Decline in work productivity
- Difficulty doing complex tasks requiring multiple steps (including household chores and other daily activities)
- Depressed mood
The Span of Alzheimer’s Disease: Middle Stage

- Decreased ability performing everyday tasks and self-care skills
- Worsened short-term memory with generally preserved long-term memory
- Increased disorientation to time and place
- Worsened ability to express and understand language (vocabulary shrinks even further, communicates in short phrases or single words)
The Span of Alzheimer’s Disease: Middle Stage (cont’d)

- Difficulty recognizing familiar people and objects
- Poor judgment and worsened attention to personal safety
- Mood and behavior fluctuations (anxiety, paranoia, hallucinations, restlessness, agitation, wandering)
- Physical changes related to progression of the disease including: new onset seizures, urinary incontinence and possible fecal incontinence, swallowing dysfunction, mobility changes (difficulty with walking and poor depth perception)
The Span of Alzheimer’s Disease: Late Stage

- Significant memory impairment (loss of short term and long term memory, loss of recognition of family members and familiar faces)
- Dependency on others for all personal care tasks (bathing, dressing, toileting, and eventually, eating)
- Increased immobility with eventual dependence on a wheelchair or bed
- Profound loss of speech (minimal words or sounds)
- Loss of mechanics of chewing and swallowing, leading to aspiration events and pneumonias
- Full incontinence (both urinary and fecal)
Health Care Guidelines

- TSH and T4-Thyroid Function Test (annual)
- Auditory testing (every 2 years)
- Cervical spine x-rays (as needed for sports); check for atlanto-axial dislocation
- Ophthalmologic exam, looking especially for keratoconus & cataracts (every 2 years)

Health Care Guidelines

- Clinical evaluation of the heart to rule out mitral/aortic valve problems.
- Echocardiogram-ECHO (as indicated).
- Reinforce the need for sub-acute bacterial endocarditis prophylaxis (SBE) in susceptible adults with cardiac disease.
- Baseline Mammography (40 years; follow up every other year until 50, then annual).

Health Care Guidelines

- Pap smear and pelvic exam (every 1-3 yrs. after first intercourse). If not sexually active, single finger bimanual exam with finger-directed cytology exam. If unable to perform, screen pelvic ultrasound (every 2-3 years)

- Breast exam (annually)

- General physical/neurological exam

- Routine adult care

- Clinical evaluation for sleep apnea

Health Care Guidelines

- Low calorie, high-fiber diet
- Regular exercise
- Monitor for obesity
- Health, abuse-prevention and sexuality education
- Smoking, drug and alcohol education
- Clinical evaluation of functional abilities (consider accelerated aging); monitor loss of independent living skills

Health Care Guidelines

- Neurological referral for early symptoms of dementia: decline in function, memory loss, ataxia, seizures and incontinence of urine and/or stool
- Monitor for behavior/emotional/mental health. Psych referral (as needed)
- Continue speech and language therapy (as indicated)

*Down Syndrome Health Care Guidelines for Individuals with Down Syndrome as published in Down Syndrome Quarterly (Volume 4, Number 3, September, 1999, pp. 1-16).*
Anesthesia Considerations

- Behavior/communication
- Airway obstruction/sleep apnea
- Bradyarrhythmias
- Gastroesophageal reflux
- Atlantoaxial instability (a potentially unstable joint between the top two vertebrae of the neck)
- Airway size
Atlantoaxial Instability

- A condition marked by the distance on X-ray between the atlas (1st vertebra) and odontoid process (2nd vertebra) of more than 4.5 millimeters (mm).
- If detected, participation in sports activities needs to be restricted.
- If not observed on initial x-ray, there is no reason to repeat.
Approximately 15% of youth with Down Syndrome have AAI. Almost all are asymptomatic.

Neurologic manifestations of symptomatic AAI include easy fatigability, difficulties in walking, abnormal gait, neck pain, limited neck mobility, torticollis (head tilt), incoordination and clumsiness, sensory deficits, spasticity, hyperreflexia...and (other spinal cord) signs and symptoms.
Hematologic Disorders

- **Leukopenia**: low white blood cells. May account for increase in infections

- **Myelodysplastic syndrome (MDS)** is a pre-cancerous condition that originates in the bone marrow in which there are cells that are abnormal. If untreated will progress to leukemia.

- **Leukemia**: 10 to 20-fold increase
Oral Health

- Teeth tend to be smaller and with shorter roots
- Large tongues, or average tongues within a small lower jaw
- Small jaw may result in crowding of the teeth and impaired bite
- Increased periodontal disease
ENT Problems

- Chronic rhinitis and sinusitis
- Sleep apnea
- High risk for conductive hearing loss re. excessive ear wax
Hypothyroidism

- Incidence between 13% and 50%
- Can occur at any time
- Symptoms
  - fatigue,
  - mental sluggishness
  - weight fluctuations
  - irritability
Behavioral Approaches

- Traditional methods of offering incentives or rewards become counterproductive.
- Do not negotiate using logic or reason.
- Non-verbal communication is critical.
- What is it that he or she is really trying to say?
General Verbal Communication Tips

- Use short, simple words and sentences
- Give one-step directions and ask one question at a time
- Patiently wait for a response
- Avoid open-ended questions.
- Provide choices or suggestions: For example, instead of “What do you want for breakfast?” Say, “Do you want oatmeal or toast?”
General Verbal Communication Tips

- Expect to repeat information or questions.
- Turn negative statements into positive statements. For example, instead of “Don’t go into the kitchen.” Say, “Come with me, I need your help with something.”
- Make statements rather than asking questions. For example, instead of: “Do you want to go?” Say, “Let’s go!”
Common Triggers for Behavioral Problems

- Communication problems (misunderstanding what is being said)
- Frustration due to tasks that are too difficult or overwhelming
- Environmental stressors (loud sounds, including loud voices, poor lighting, disruptive housemate)
- Personal upheaval (family illness, death of loved one, change in staff member)
- Medical status (physical pain, discomfort, illness)
- Stress of the caregiver or environment
Techniques for Modifying Triggers

- Assess for change in health status
- Provide reassurance and, if appropriate, a gentle touch
- Use redirection techniques or distraction to something pleasurable
- Keep in mind that different approaches work at different times
- Be patient and flexible
Nursing Responsibility

- Prevention of secondary and tertiary complications
- Continual assessment
- Support coordination
Life expectancy for individuals with Down Syndrome has increased dramatically in recent years, with the average life expectancy approaching that of peers without Down Syndrome.
Resources

National Down Syndrome Society
https://www.ndss.org/

National Association for Down Syndrome
http://www.nads.org/pages_new/resources.html