

THE BOGGS CENTER ON DEVELOPMENTAL DISABILITIES

New Jersey's University Center for Excellence in Developmental Disabilities Education, Research, and Service New Jersey's Leadership Education in Neurodevelopmental and Related Disabilities Program

Care of the Adult with Down Syndrome

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Objectives:

- Understand some of the medical conditions that might accrue to adults with Down Syndrome
- Recognize the accommodations or supports providers may need to put in place when caring for adult patients with Down syndrome
- Become familiar with the Adult Health Care Guidelines for providing healthcare for adults with Down syndrome

GREEN HIGHLIGHTS GOOD ENCOUNTER STRATEGIES YELLOW HIGHLIGHTS KEY FINDINGS BLUE TEXT DISCUSSION POINTS

Case Review and Discussion

Part 1

Chart Review:

Lee is a 25 year-old man with Down syndrome. He has been a patient of your practice for the past year after his family moved to the area but you have only seen him once before for an upper respiratory infection. You take a quick look in the chart before going in.

<u>PMH/PSH</u>: VSD that closed by age 3 without any surgical intervention, Mitral valve prolapse, Conductive hearing loss from chronic serous otitis requiring 4 sets of myringotomy tubes b/l, Constipation, Tonsillectomy and adenoidectomy at 5 years-old for both recurrent adenoiditis and obstructive sleep apnea, developmental delay in gross and fine motor as well as speech with dysarthria.

Medications: None listed, Allergies: None

Labs: There are none on file in your EMR.

<u>Social History</u>: He lives with his parents and 18 year-old brother. He does not smoke or drink alcohol. He graduated high school at 21 and is <u>now working</u> part-time with a non-profit advocacy group organizing folders, files, and with community outreach.

Your nurse does intake and comes out and gives you the scoop:

"This is a downs kid here with his Mom who scheduled the appointment because she is concerned about a change in his behavior and functioning for the past 2 days. He seems fine. I couldn't really get any information from the kid. Here are his vitals . . ."

She hands you his blank med. list with some vitals jotted down on the back.

Vitals: Hgt: 5'7", Wght: 170, BP 120/72, HR 88, RR 15, Pain: 1

Take a moment to reflect thus far.

- 1. What are you concerns or thoughts about what may be going on entering the visit?
 - Certainly wide differential at this point adjustment disorder, infection, injury, pain?, behavioral, electrolyte/metabolic/thyroid, sensory decline (hearing/vision), cardiac (cardiomyopathy/valvular disease), seizures/neuro

- Be aware that patients may have limited expressive speech, and a tendency to manifest development of new disease as a change in behavior.
- 2. What is your plan for communication?
 - Direct questioning to the individual, make no assumptions about capabilities at this time, then certainly can also collect data from concerns of loved ones.
 - Intend to and provide the same level, type, and quality of care that you would provide any other patient.
 - Encourage the highest level of involvement of the patient in his or her care.
 - Ascertain the patients and caregivers preferred method of communication should there be evidence of increased challenge such as visuals, communication boards, PEC system.
- 3. How do you feel your office staff is doing in providing care? How could the patient's care have been improved and what sort of interventions may help with that?
 - Lack of people first/preferred language
 - Assumptions about his capabilities perhaps (after reviewing the social history seems surprising he is unable to provide any valuable info about wellbeing)
 - Seems to be making assumptions about his wellness.
 - Referred to this adult as a kid!
- 4. Other thoughts upon review of the information you have received?
 - Needs overall health update/preventative services
 - Would be ideal to have labs on file, getting old records may be useful
 - More regular visits may be warranted to establish patient physician relationship
 - Your office needs staff education on people first language and to treat people in age appropriate manner

Part 2

Upon entering the room you greet the patient, "Hello Lee," and he responds, "Hi." You ask the patient, "Who is this with you today?" as you point toward the other 2 people in the room and Lee says, "That's my mom and my brother." You introduce yourself to the patient's mom and brother and thank them for being there.

The patient then says, "My brother is going to college." You congratulate the patient's brother and do some brief small talk and learn his brother will be moving to Baltimore for college and will be leaving in a couple of months.

You turn to the patient and ask, "So, what brings you in today?" and he answers, "I come to see the doctor."

You then ask, "So tell me what is going on?" and he casually responds, "nothing."

Time to Reflect:

- 1. What has gone well so far?
 - Great pre-visit planning reviewing the chart prior to encounter
 - Nice introductions- first priority is the patient and then introduction to the parents as well
 - Evidence that small talk improves patient and family satisfaction so great job
 - Establishing rapport, assumed competence at play, patient opening up to you about some things that may be on his mind
- 2. What is not going well and what can you do differently?
 - Vague questions that may not be the most useful for collecting history (open ended questions are worth a try and often preferred in opening a medical encounter but he clearly is answering with routine and likely ingrained or standardized responses).
 - Modify your questions to be simple and straightforward
- 3. What do you want to know now and how will you gather more pertinent history?
 - Are you feeling sick?
 - Does anything hurt you?
 - Why did you come to see the doctor today?
 - Who decided he needed to come to the doctor today and why?
 - Ask if you can ask his mom and brother some questions as well . . . gather input?

Part 3

You realize that your questions are too vague and you decide to modify your technique moving forward but the mom starts to chime in at this point and says:

"We brought him in today because he just has not been himself lately for the past 2 days. He doesn't seem to want to get out of bed even to join us to eat meals, which is very unusual. He is resting much of the day or just wanting to lay around and watch TV. He will eat a bit when we bring him a plate but not as much as usual. He will get up to go to the bathroom but seems upset and uncomfortable to move around and at times he sits down on the floor half way there. He goes right back to the couch or bed as soon as he is done."

The brother adds:

"He was fine 2 days ago. We went out together to celebrate my high school graduation, to buy some stuff for my college dorm, and then we went to Sky Zone, which he loves! We were there for about an hour and everything was great then we went home and had dinner and went to bed. The next day he really was not himself."

- 1. Differential Diagnosis so far?
 - Injury/Trauma/Pain (MSK or AAD)
 - Infection
 - Adjustment disorder/depression
 - Metabolic, dehydration/electrolyte imbalance
- 2. What more do you want to know?
 - Fever, chills, pain, fatigue, respiratory symptoms, GI symptoms, urinary symptoms, MSK symptoms, neuro symptoms, full ROS
 - Anything else unusual happen while out the other day or since?
 - Any sick contacts?

Part 4

You continue to gather information from the patient and family by directing simple and clear questions to the patient but you invite the mom and brother to add in or help with clarification as necessary after getting your patient's permission to do so.

Review of Systems:

- General: No fever, chills, weight loss or weight gain. (+) fatigue, (+)malaise,
- HEENT: (-)rhinitis, (-) ear pain reported, (-) otorrhea, (-) sore throat reported, Rubs his head when you ask about if his head hurts and says yes
- Heart: (-) Pain in chest
- Lungs: (-) no apparent shortness of breath, Rare occasional cough
- Abdomen: (-) Diarrhea reported, Reports he is having bowel movements yesterday last,
 (-) abdominal pain
- MSK: Rubs his neck on the right side when you ask about pain in the neck, (-) back pain,
 Withdraws his right leg and says "I'm okay" when you ask if he has any pain in the legs
- Endocrine: (+) Decreased appetite, ??cold or heat sensitivity
- Uro: ??change in freq urination, (-) pain with urination
- Neuro: No change in gait reported though limiting, no change in speech reported though quieter and less talkative

You ask if he may have injured himself at all or if he ever reported any pain in these areas in the past?

Mom says sometimes he'll say he has a headache or pain in the neck though not often. "He doesn't complain much."

His brother says:

"I don't really think he got hurt, but toward the end of our time at Sky Zone he was jumping toward the edge and came down and landed with his foot on the side springs then ended up falling completely down and hitting his head into the side trampoline wall, the part that was padded I think. It happened so fast I can't be sure exactly what happened. He got a little jolted but said he was fine and didn't take long to get up. I asked him a bunch of times if he was okay and he said he was."

Patient says:

"I am okay on the trampoline, I can go. My brother's going to college soon, he won't be at home." His brother jokes, "maybe I'll actually sleep at night, I won't have to hear you snore all night." They both laugh.

- 1. Differential diagnosis?
 - MSK injury, pain (foot/ankle/leg/neck/head)
 - Concussion
 - Depression/adjustment disorder
 - Infection less likely
 - Dehydration/metabolic less likely
 - Cardiomyopathy/valvular as primary cause of change less likely in light of other finding as well
- 2. What Down Syndrome Specific Diagnoses need to be considered as well?
 - AAD/injury
 - Other things more common in adults with DS that should be considered as cause or contributing to fatigue sleep apnea, hypothyroidism though probably don't explain full picture of acute change
 - Dementia unlikely at this age though earlier dementia possible. again slower and more gradual presentation

Part 5

You let the patient know that now you want to examine him and you talk him through the whole process, letting him know each step of the way what you will be doing.

Physical Exam:

Vitals: Hgt: 5'7", Wght: 170, BP 120/72, HR 88, RR 15, Pain: 1

- General: quiet, appears withdrawn and tired, slow to answer questions
- HEENT: possible subtle faint swelling right frontal area just at hairline, says stop with palpation, ears small ear canal otherwise normal, PEERL, EOM intact, MMM, Absent tonsils, normal posterior pharynx
- Neck; no LAD, ROM seems limited with right rotation and flexion, no midline tenderness appreciated.
- Lungs: CTA b/l, no W/R/R
- HRT: RRR, midsystolic click at the apex and 2/6 diastolic murmur best heard over the 3rd left intercostal space
- Abd: Normal BS, soft, NT, ND
- Extr: no edema, no clubbing, normal Cap refill, normal DP,PT pulses
- MSK: FROM hip, knee intact without apparent tenderness, Left ankle, foot unremarkable, but pulls away and says "don't touch" when you attempt to examine the right foot. You gently reassure the patient that you will be careful and gentle. You notice some very mild swelling of the dorsal midfoot, no ecchymosis, pulls away with any attempts to palpation of the midfoot or ankle, pulls away for attempts on ROM of foot or ankle.
- Neuro: AAO to person, place and day of the week and year, CN intact, speech dysarthria, slow to answer questions, RAM lack precise coordination, grip strength symmetric but weak, no apparent limp though quickly sat down, refused to participate with tandem gait assessment

- 1. What do you think about the assessment of pain in light of your exam? Suggestions?
 - Pain scale was 1 on vitals not consistent with finding on exam
 - When assessing pain in people with Down Syndrome:
 - Avoid numeric pain scale
 - Avoid asking list of body parts and whether there is pain in the areas—tendency to say either yes, yes, yes or no, no, no
 - Avoid asking does it hurt here or there, or listing 2 options in questions. There is a tendency to state the second option.
 - Use simple and concrete questions such as "Can you point to where it hurts"?

- 2. What are your concerns? Differential Diagnosis top 3 concerns now?
 - Trauma to the foot/ankle
 - AAD
 - Concussion

Probably would want to clarify if mentation, orientation, dysarthria seem at baseline (other concerns after exam – should get echo at some point – though acutely doubt cardiomyopathy as main cause of symptoms acutely)

Part 6

You realize the numeric pain scale did not correlate to your exam findings and you want to learn more about the severity of the pain. You use the FACES scale.

- You say to the patient that it looks like you may have some pain in your head and your foot when I was examining you, can you show me how you feel by looking at these pictures? How bad is this pain? FACES scale was done and he circled 8.
- You ask him to point where it hurts the most? He points to his foot. He says "I am okay on the trampoline. I am fine."

Wong-Baker FACES® Pain Rating Scale

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Hurt

Hurts Little Bit



Hurts Little More



Hurts **Even More**



Hurts Whole Lot



Hurts Worst

- 1. What did you learn about pain perception and assessment in people with Down syndrome?
 - Numerical pain scale not as reliable for individuals with Down syndrome.
 - Use of FACES Scale seems to be better understood in both children and adults with Down Syndrome than the numerical pain scale
 - Look out for painful physical conditions in adults with Down syndrome.
 - Be mindful of possible delayed pain response, less obvious pain behavior, and difficulties with communication. all could lead to assumptions that people with Down syndrome have a higher pain threshold or lower pain sensitivity.
 - Encourage adults with Down syndrome to talk about pain
 - Trial of different pain scale modalities may be necessary to find which is best for the individual, family may be helpful
 - Input from the family on how patient normally expresses pain may be very valuable.
 - People with Down syndrome may exhibit decreased functioning/cognition when they are in pain. Examples are attention, memory, and the ability to regulate the own behavior.
- 2. Assuming you have access to labs and imaging in your center, what tests do you want to order?
 - X-ray ankle/foot
 - X-ray of the C-spine
 - CT head?
 - Labs? Not sure critical at this point but could consider what would you order?

Part 7

- CBC normal, BMP normal
- CT head Negative
- C-spine normal
- X-ray ankle normal
- X-ray foot



- 1. Diagnosis?
 - Base 5th metatarsal (Jones fracture)
 - Concussion still likely
 - Thankfully AAD ruled out and c-spine fracture ruled out
- 2. Plan?
 - Refer to ortho or podiatry for management of fracture
 - Put in place Concussion protocol/care plan
 - Plan close follow up
 - Plan for pain management
 - Still need additional work up and preventative services

DIVIDE CLASS INTO 3 GROUPS FOR FINAL PORTIONS OF THE CASE DISCUSSION

- 10 MINUTES TO PREPARE
- 5-10 MINUTES EACH TO PRESENT DISCUSSION POINTS

Case Review and Discussion TEAM 1 Part 8

You explain to the patient that he injured a bone in his foot and will need a cast, and if not improving he may need surgery. He says, "I don't want surgery." You explain that hopefully that will not be necessary but it gets you thinking, who would be the decision maker if that was needed or even for the casting? Who will be giving informed consent?

- 1. What is your plan for further treatment?
 - Refer to podiatry or ortho to arrange management and further discuss options of management
 - Tylenol for pain control, consider adding NSAID (?effects on bone healing but evidence not great that there are significant impacts and can help limit opiate use so short course if needed in addition to Tylenol likely appropriate)
- 2. What question(s) may you need to know from Mom regarding consent? What documents are necessary for another to make decision on behalf of a patient?
 - If mom has guardianship or limited guardianship, then I would suspect she would have brought that up, but it is reasonable and appropriate to ask if Lee has any legal documents in place with regards to medical decision making such as guardianship, or medical power of attorney (aka Healthcare proxy in NJ).
 - Medical POA (healthcare proxy) would go into effect if patient was deemed unable to make decisions at a point in time.
 - Healthcare proxy and medical POA are documents that the patient would have been considered competent to authorize and sign off on – meaning he would have chosen and designated a healthcare proxy understanding the rights of that individual.
 - Guardianship is granted in court when a patient is deemed to lack capacity to make decisions for himself. Evidence must be presented from 2 doctors that the patient lacks the capacity to make health care decisions.
- 3. If you determine the patient has no guardian for medical decisions, but rather a Healthcare proxy, what other questions may you ask the patient?
 - You first may just want to ask patient if he feels comfortable making this decision or he would like his mom to assist in making decisions about his treatment. Supported decision making is often the best strategy.

- You may want to assess decision-making capacity. Healthcare proxy goes into effect if
 the patient is unable to make decision on his own. The proxy should always take into
 consideration not only the medical facts and recommendations, but also the known
 patient wishes.
- After explaining in a simple and straightforward manner, you may ask patient to reiterate his understanding of the situation, the treatments being suggested and the consequences of not moving forward with the recommendations
- 4. How do you determine decision making capacity?

Technically, a patient remains competent until a court says otherwise. On the other hand, the determination of medical decision-making capacity can be made by the attending physician and does not typically require a court proceeding.

Medical capacity can be determined by the use of the four-point test, which assesses the following:

- 1. Understanding: The patient understands the nature of the intervention and consequences
- 2. Appreciation: The patient understands the consequences of the decision (especially refusal of treatment).
- 3. Communication: The patient is able to communicate his/her wishes.
- 4. Rationalization or Reasoning: Those wishes are compatible with the patient's known values

NJ STATUTE

26:2H-60 Determination of patient's capacity to make a health care decision.

(https://law.justia.com/codes/new-jersey/2014/title-26/section-26-2h-60/)

- "8. a. The attending physician shall determine whether the patient lacks capacity to make a particular health care decision. The determination shall be stated in writing, shall include the attending physician's opinion concerning the nature, cause, extent, and probable duration of the patient's incapacity, and shall be made a part of the patient's medical records.
- b. The attending physician's determination of a lack of decision making capacity shall be confirmed by one or more physicians. The opinion of the confirming physician shall be stated in writing and made a part of the patient's medical records in the same manner as that of the attending physician. Confirmation of a lack of decision making capacity is not required when the patient's lack of decision making capacity is clearly apparent, and the attending physician and the health care representative agree that confirmation is unnecessary.
- c .If the attending physician or the confirming physician determines that a patient lacks decision making capacity because of a mental or psychological impairment or a

developmental disability, and neither the attending physician or the confirming physician has specialized training or experience in diagnosing mental or psychological conditions or developmental disabilities of the same or similar nature, a determination of a lack of decision making capacity shall be confirmed by one or more physicians with appropriate specialized training or experience. The opinion of the confirming physician shall be stated in writing and made a part of the patient's medical records in the same manner as that of the attending physician.

- d. A physician designated by the patient's advance directive as a health care representative shall not make or confirm the determination of a lack of decision making capacity.
- e. The attending physician shall inform the patient, if the patient has any ability to comprehend that he has been determined to lack decision making capacity, and the health care representative that: (1) the patient has been determined to lack decision making capacity to make a particular health care decision; (2) each has the right to contest this determination; and (3) each may have recourse to the dispute resolution process established by the health care institution pursuant to section 14 of P.L.1991, c.201 (C.26:2H-66).

Notice to the patient and the health care representative shall be documented in the patient's medical records.

- f. A determination of lack of decision making capacity under this act is solely for the purpose of implementing an advance directive in accordance with the provisions of this act, and shall not be construed as a determination of a patient's incapacity for any other purpose.
- g. For purposes of this section, a determination that a patient lacks decision making capacity shall be based upon, but need not be limited to, evaluation of the patient's ability to understand and appreciate the nature and consequences of a particular health care decision, including the benefits and risks of, and alternatives to, the proposed health care, and to reach an informed decision."
- 5. What and when should parents think about with regards to guardianship? When is it appropriate?
 - These discussions and consideration should take place before turning 18 so that emergency guardianship does not need to be sought if that was necessary or alternatively so that proxy can be put in place to use if needed.

Part 8

The patient saw orthopedics and both he and mom agreed that the cast was a good idea and consented to treatment. He was treated with Motrin and Tylenol.

Within 3- 4 days he was acting more himself but still seemed a bit less energetic and parents are now concerned that he is fixated on his brother going to college. He also seems to be more forgetful, more easily fatigued, and less focused or able to complete tasks. In retrospect, mom states perhaps some of this has been going on longer but she had not really realized an issue until the severity of things when the foot issue hit. It may be months he has been less focused and more forgetful and perhaps even a bit less independent.

- 1. What other concerns do you have?
 - Depression likely, concussion could also be factor (though that would have been purely new) or may be further exacerbating
 - It has been noted that children and adults with Down syndrome and symptoms of depression often lose the ability to enjoy many activities they used to love, they lose skills and they become very withdrawn
 - Many studies have shown increased rates of depression in adults with Down syndrome compared to general population and under diagnosis (up to 30%)
 - thyroid possible
 - Poor sleep sleep apnea also should be considered
- 2. What do you think may be contributing to his symptoms?
 - Loss of social connections- now that out of school, brother likely key friend and social support, this anticipated separation is likely impacting mood and causing anxiety.
 - Also seeing brother move on to another stage that he is not able to be a part of or did not get opportunity for
 - Pseudodementia type picture?
- 3. What additional recommendations for treatment/care would you advise?
 - Check thyroid
 - Check sleep study, consider checking echo (easily fatigued murmur on exam suggestive of MR though no SOB, still needs to be checked so could consider)
 - Consider therapy with psych experienced with individuals with DD/Down syndrome
 - Socialization support groups
 - Consider some sort of college for patient- continuing education, classes
 - Consider medication SSRI
 - Increase exercise
 - assess what patient thinks will happen when brother goes to college- maybe not consistent with the reality

- Have a clear discussion with patient about what it means for brother to go to college and what he can expect – what will change and will not change and reassure that they will still be close and talk often and get to see each other
- 4. Would you advise any additional testing?
 - Thyroid test
 - Sleep study
 - Other labs can be considered b12
- 5. If he were 45 to 50 years-old, would that change your differential and recommended work up?
 - Dementia also in differential. (Results suggest that, compared to the general population for which short-term (episodic) memory loss is the most common indicator associated with the onset of AD, in people with DS, executive dysfunction and Behavioral and Psychological Symptoms of Dementia (BPSD) are commonly observed during pre-clinical and early stages and may precede memory loss. The review highlights the importance of using a broad spectrum of assessments in the context of heterogeneity of symptoms. Theoretical and practical implications are discussed, as well as the need for further research. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5359367/)
 - Recommend evaluation by neuropsych or dementia specialist there are several tools specifically utilized for assessing for dementia in people with ID/DD and specifically Down syndrome
 - Ideally having baseline cognitive functioning/functional assessment on hand starting at 35 for comparison
 - He was advised on cognitive and physical rest with the thought there may be a coexisting concussion as well

Part 8

The patient saw orthopedics and both he and mom agreed that the cast was a good idea and consented to treatment. He was treated with Motrin and Tylenol.

Within 3- 4 days you saw him back in follow up and he was acting more himself.

- 1. What specific things would you want to follow up with him in addition to how his foot is at this time?
 - Ability to function with the no weight bearing recommendations supports in place in home, fall precautions, headache and concussion follow up and compliance with protocol, neck pain follow up.
 - Mood- Depression
 - Need for routine labs, preventative services annually
 - Plan for repeat x-ray foot as per ortho and plan for ortho follow-up
 - Needs echo for heart (murmur)
- 2. What is your long term plan for health maintenance and follow up? What conditions may be more common in people with Down syndrome and how would you assess for or screen?
 - Annual health maintenance at least, allocate enough time for visits, recognize you may need closer follow-up since may take more time to accomplish goals of encounter then with some of your patients (also may take less)
 - Understand Common Chronic Conditions:
 - Thyroid Disease
 - Sleep Apnea
 - Obesity
 - o Diabetes
 - Depression/OCD
 - Hearing Loss/Vision Problems
 - AA Subluxation
 - o Alzheimer's Disease
 - Recognize need for Routine Screening but also screening for common conditions
 - Assess for signs/sxm spinal cord compression
 - Assess for dementia/memory consider referral for Neuropsych testing if concerns for individual familiar with screening for dementia in Down syndrome
 - Screen Vision and Hearing at least every other year
 - Screen annually for thyroid disease
 - Screen for Sleep apnea
 - Coordinate care with appropriate specialists as needed including any disease/condition specific but also ophthalmology, Audiology, Physical Therapy and Speech Therapy and Cardiology

- 3. What topics do you hope to discuss with him with regards to social history/health risk behavior?
 - Substance use update
 - Relationships/sexuality
 - Socialization, friendships
 - Exercise, weight (BMI) falls in overweight, diet
 - Supports
 - Abuse or neglect

(Do your full HEEADSSS Home, Education/Employment, Eating, Activities, Drugs, Sexuality, Suicide/Depression, and Safety.)

- 4. How may some of these conversations differ from the same topic with similar aged, typically developing young adults?
 - Explains things using pictures if needed, and in simple straightforward manner
 - Substance Abuse don't assume not involved, ask, explain risks
 - Exercise and nutrition encourage provide exercises sustainable and appropriate
 - Relationships/Sexuality: encourage healthy relationships
 - While a child's cognitive understanding of puberty and sexuality may be delayed for his or her age, the process of body maturation, hormonal changes, and sexual feelings usually is not — creating a mismatch that can be dangerous if ignored.
 - Sexuality and socialization should be discussed with teens and adults
 - Understanding and Interest in sex should be questioned
 - Recognize nearly all men with Down Syndrome are sterile (99%) and women tend to have reduced fertility.
 - An individual with Down syndrome has 50% of having a child with Down syndrome
 - LARC are typically preferred methods of contraception because of limited ability to consistently comply with pill taking or a barrier method.
 - Educate and screen for STD
 - Discuss ways to express affection physically and also ways to refuse inappropriate or undesired touching and report it.

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