Care of the Adult with Down Syndrome

Case Review & Discussion

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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

Building Capacity to Improve Adult Health Care for Patients with I/DD
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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.

PMH/PSH: 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.

Social History: He lives with his parents and 18 year-old brother. He does not smoke or drink alcohol. He graduated high school with IEP diploma and is now working 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?

2. What is your plan for communication?

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?

4. Other thoughts upon review of the information you have received?

Case Review and Discussion

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?

2. What is not going well and what can you do differently?

3. What do you want to know now and how will you gather more pertinent history?

Case Review and Discussion

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."*

Reflection:

1. Differential Diagnosis so far?

2. What more do you want to know?

Case Review and Discussion

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.

Reflection:

1. Differential diagnosis?

2. What Down Syndrome Specific Diagnoses need to be considered as well?

Case Review and Discussion

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

Reflection:

1. What do you think about the assessment of pain in light of your exam? Suggestions?

2. What are your concerns? Differential Diagnosis top 3 concerns now?

Case Review and Discussion

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."*

Reflection:

Assuming you have access to labs and imaging in your center.

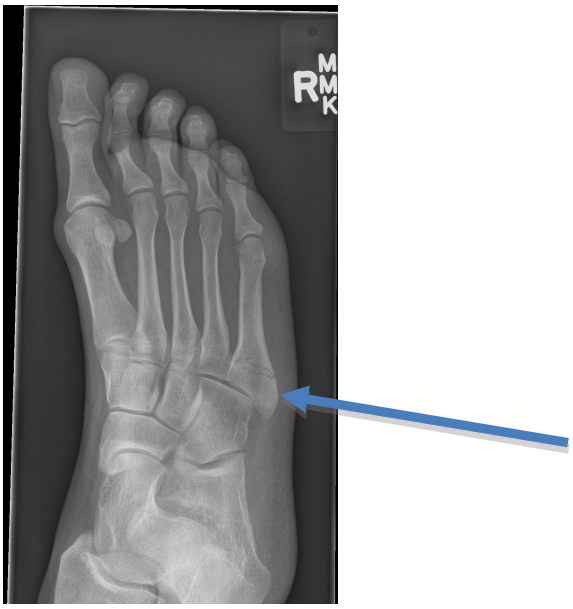
1. What did you learn about pain perception and assessment in people with Down syndrome?

2. What tests do you want to order?

Case Review and Discussion

Part 7

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



Reflection:

1. Diagnosis?

2. Plan?

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?

Reflections:

1. What is your plan for further treatment?

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?

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?

4. How do you determine decision making capacity?

5. What and when should parents think about with regards to guardianship? When is it appropriate?

Case Review and Discussion
Team 2
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?

2. What do you think may be contributing to his symptoms?

3. What additional recommendations for treatment/care would you advise?

4. Would you advise any additional testing?

5. If he were 45 to 50 years-old, would that change your Differential and recommended work up?

Case Review and Discussion
Team 3
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?

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?

3. What topics do you hope to discuss with him with regards to social history/health risk behavior?

4. How may some of these conversations differ from the same topic with similar aged, typically developing young adults?

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