Episode 3 - Down Syndrome Guidelines

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Sponsor: The Ohio Chapter of The American College of Physicians

Episode Summary:
In this episode of Everyday Medicine, Dr. Chris Chiu from The Ohio State University interviews Dr. Michael Neiger about the care of adults with Down Syndrome. Dr. Neiger discusses the importance of this topic, highlighting the increased life expectancy of this population and the associated medical challenges. They delve into recent guidelines published in JAMA, specific health concerns such as atlantoaxial instability, celiac disease, obesity, thyroid disease, osteoporosis, and mental health issues. Dr. Neiger also emphasizes the importance of specialized guidelines and resources provided by the Global Down Syndrome Foundation.

00:00 Introduction to Everyday Medicine Podcast
00:30 Meet Dr. Michael Neiger
01:02 Care of Adults with Down Syndrome
02:20 New Guidelines for Down Syndrome Care
02:57 Atlantoaxial Instability in Down Syndrome
05:02 Celiac Disease in Down Syndrome
06:02 Obesity and Endocrinological Concerns
09:30 Behavioral and Mental Health
14:25 Cardiovascular Screening and Stroke Prevention
16:26 Conclusion and Resources

Key Topics:

1. Background on Down Syndrome:
    - Michael Neiger's personal connection to Down Syndrome.
    - The evolution of life expectancy and medical care for individuals with Down Syndrome.

2. New Guidelines for Down Syndrome Care:
    - An overview of the important aspects from the 2020 JAMA guidelines.
    - Focus on evidence-based screening and treatment strategies.

3. Specific Health Concerns in Down Syndrome:
    - Addressing atlantoaxial instability, including screening recommendations.
    - The prevalence of celiac disease and approaches to screening.
    - Obesity management strategies and lifestyle interventions.
    - Endocrinological concerns, including thyroid disorders and osteoporosis.

4. Behavioral and Mental Health Considerations:
    - Discussing mental health stereotypes in Down Syndrome.
    - Tailoring screening and management for mental health disorders in Down Syndrome.

5. Cardiovascular Screening in Down Syndrome:
   - Exploring the prevalence of cardiovascular disease in Down Syndrome.
   - Recommendations for cardiovascular health and stroke prevention.

6. Key Takeaways:
   - Highlighting the importance of personalized and evolving care for adults with Down Syndrome.
   - The crucial role of interdisciplinary care and family involvement in health management.

Transcript:

Transcript has been edited for clarity

[00:00:00] **Christopher:** Welcome to Everyday Medicine, a podcast from the Division of General Internal Medicine at The Ohio State University. This podcast is focused on primary care and aims to provide current information to medical professionals from experts in Ohio. We're also graciously sponsored by collaboration with The Ohio Chapter of The American College of Physicians.

[00:00:18] I'm Chris Chiu, the Director of Education for our Division. I have a great doctor here with me today to talk about a very interesting topic that he recently presented to our division. I have Dr. Michael Neiger... how are things going with you today?

[00:00:33] **Michael:** So far so good. Residency is starting to come to an end, so I'm looking forward to it.

[00:00:36] **Christopher:** All right. So yeah, tell us a little bit about yourself.

[00:00:38] **Michael:** So I am a Columbus native and Columbus lifer. So I went to OSU for undergrad med school and then loved enough to stick around for combined med peds residency. So I am getting to the end of my fourth year and I'm very excited to be sticking around again. And I'll be with the general internal medicine department starting in the summer.

[00:00:53] **Christopher:** Awesome. And so, you recently did a presentation for our Faculty Development Series on an interesting topic. Do you want to explain a little bit on why you chose that topic and why it interests you?

[00:01:02] **Michael:** Yeah, the talk I gave was on care of adults with Down Syndrome. And it's a topic that's super important to me because I have an older brother with Down Syndrome. So growing up with him and being exposed to that population, it's just a really important group to me. And as I was researching this topic and coming up with more information, it's a significant gap in medical care currently.

[00:01:20] Back in the 1960s, the life expectancy of this population was only 10 years old. Even as recently as the 80s, the life expectancy was the mid 20s. Just 40 years later, that life expectancy has pretty much doubled to about 60 as of last estimate. These people are living longer and the general internist probably isn't really familiar with that population.

[00:01:39] With that lower life expectancy, these people probably didn't live long enough to see internists back in the 80s, maybe even 90s. So there wasn't necessarily much education or training in the population. And now as they've gotten older, they're having more comorbidities. The internist might not be

as comfortable with a population they never learned about or some of the congenital conditions associated with Down Syndrome that they're not familiar with.

[00:01:59] And the pediatrician probably isn't quite as interested as, in dealing with their hypertension, their diabetes, and some of their dementia. So I think Med Peds fills a nice gap in that spectrum where we have that training from both sides. And I'm just excited to share that information.

[00:02:13] **Christopher:** Excellent, excellent. I appreciate that background. Now, I think during your talk, you said that there's something there are newer guidelines that came out. Is that correct?

[00:02:20] **Michael:** In 2020, in JAMA, a group published new guidelines for the care of adults with Down Syndrome (*editor note: see also here*). And it was the first identified evidence based guidelines.

[00:02:27] There had been, prior to that, some other guidelines for care of adults with Down Syndrome, but for the most part, they were based on just expert opinion. And so the goal of this was identifying both screening that was done in the general population and seeing how it fit in the population of adults with Down Syndrome, as well as looking at specific comorbidities that are unique to Down Syndrome and seeing how best we go about screening for those and addressing them.

[00:02:50] **Christopher:** Can we first start talking about the ones that are specific just for Down Syndrome? These are things that you might screen that you may not do with anyone else.

[00:02:57] **Michael:** Absolutely. So one of the bigger things is atlantoaxial instability. So this was the classic med school teaching point, a classic question on the med school boards of this association of Down Syndrome and atlantoaxial instability.

[00:03:08] And the previous teaching and guidance on those previous guidelines had been routine screening to look for atlantoaxial instability with cervical spine x-rays. And when they looked at the evidence on this, they found that it actually wasn't that great. So the sensitivity of those x-rays to even identify atlantoaxial instability was pretty poor.

[00:03:25] And beyond that, even identifying it, there was no real good data on showing that. identified instability actually led to clinically significant spinal cord injuries or outcomes. So as we'll talk about later, obesity is a significant risk in this population as well. So in light of that risk and trying to encourage more physical activity, they found no real good evidence for this routine screening.

[00:03:45] So instead they recommended talking with the patients and screening them for signs or symptoms of cervical myelopathy, which would be the consequence of that atlantoaxial instability but didn't recommend routine screening for x-rays even before physical activity.

[00:03:59] **Christopher:** So what type of.. when you're saying, like asking them about consequences, like what types of things might they bring up?

[00:04:04] **Michael:** Primarily things like sensory changes or weakness in the upper extremities would be the main symptoms that you'd be looking for.

[00:04:10] **Christopher:** So you would ask them about that, like how they're handling things, but then also with like physical exam, is that?..

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[00:04:16] **Michael:** I'm not as entirely sure about the physical exam side of things. Obviously you could check for strength in the exam and see if that's changing over time. But, I don't think there was real specific guidance on the sensitivity of that for testing. I think it's more based on history.

[00:04:29] **Christopher:** So after history and it increases your pre-test probability, and then you would do x- rays?

[00:04:34] **Michael:** Yeah, so x-ray is still the best initial screening tool to look for atlantoaxial instability. But routine screening in an asymptomatic individual is no longer recommended.

[00:04:43] **Christopher:** What's expected after that? Do you then refer or what happens then?

[00:04:46] **Michael:** Referring. If you particularly have concerns for spinal cord pathology associated with it. If it's identifying the x-ray, I would assume that. Next steps would be referring into more advanced imaging, like an MRI, to confirm the diagnosis.

[00:04:58] **Christopher:** What...what other screening do we need to look for specifically in this population?

[00:05:02] **Michael:** So one area that's also unique to this population is celiac disease. Obviously, celiac exists in the general population as well, but the prevalence in Down Syndrome is estimated to be about 11%. There can be a pretty wide variety of manifestations of that. Obviously, the classic being the GI symptoms: diarrhea, bloating, things along those lines. But a lot of other you can have rashes, malabsorption, anemia. A whole host of other conditions that could be associated with it.

[00:05:26] There wasn't, unfortunately, very good data on routine screening in asymptomatic individuals, but essentially a very low threshold to screen for that in this population, with our normal screening tests, like a tissue transglutaminase just as initial screening tools, if you have any, if any of those concerns come up. And the Global Down Syndrome Foundation actually also came up with a very helpful checklist for families. So it's easily accessible on their website for free and it lists a lot of those, both the GI and extraintestinal manifestations, so families can review those, keep an eye on those, and bring it to their doctor's appointments if it's a concern that they have.

[00:05:58] **Christopher:** Gotcha, and this is a checklist screener specifically for celiac?

[00:06:02] Going to our next discussion, you said something about obesity. There are a bunch of endocrinological associations with Down Syndrome, so can you talk a little about obesity and then maybe some of those other things?

[00:06:11] **Michael:** Yeah, absolutely. From an obesity standpoint, it's certainly more common in adults with Down Syndrome than the general population. There wasn't a whole lot of specific guidelines for this article but just recommending a general lifestyle based approach including dietary and physical activity changes.

[00:06:26] Global Down Syndrome Foundation's website has links to a lot of local communities and resources, things like the Special Olympics, to try to help encourage physical activity. But there wasn't really a specific guideline for this population of one, one best approach.

[00:06:40] **Christopher:** Got you. You may not know the answer to this, but especially with the atlantoaxial things you talked about, do we have to be specific in guidelines about exercise for these patient populations?

[00:06:49] **Michael:** Not that they identified in the guidelines there. Actually, part of the atlantoaxial instability recommendations came from data from the Special Olympics that showed in their 30 to 40 years of operating they had no identified episodes of spinal cord injury associated with atlantoaxial instability.

[00:07:03] Obviously if you have a patient that has already had this screening prior to these guidelines coming out and has that identified, um, you would certainly want to have a risk benefit conversation with that family for more high contact sports but low impact activities, things like swimming, running should definitely be encouraged in all patients.

[00:07:19] **Christopher:** What other endocrinological things to worry about?

[00:07:21] **Michael:** One of the bigger things that's different in this population is thyroid disease. It's super common in the Down Syndrome population. Prevalence as high as 40 percent in 18 to 29 year olds and 50 percent in those greater than 30. Because of that and because of kind of the vague symptoms that can be associated with thyroid disorders, there's recommendations for routine screening with a TSH every one to two years regardless of symptoms.

[00:07:44] **Christopher:** Is there an age that you start when we see with our adolescents?

[00:07:47] **Michael:** Um, so this is actually starting down even in the pediatric population, so pretty much as soon as you start taking care of this patient as the internist, you can start screening them with the yearly TSH.

[00:07:57] **Christopher:** Excellent. Any other endo things we have to worry about?

[00:07:59] **Michael:** So one other topic that they addressed is osteoporosis and in this case they were trying to look at our general screening populations and seeing how that applies to the Down Syndrome population, and they really didn't find good evidence one way or the other. They had some interesting points about the bisphosphonates that we typically use for treatment of osteoporosis potentially not being as effective in the Down Syndrome population due to different mechanisms of how bone loss can occur.

[00:08:23] I'm not entirely sure of the specifics of that, but between that and differences in growth characteristics even screening DEXAs may not be quite as beneficial. So, the benefits of both screening and treatment are a little bit unclear in this population, but at the same time, there weren't significant risks associated with doing those things.

[00:08:38] So, the guideline just recommended a risk benefit discussion with the families of this is something we can assess, we can do, but we're not entirely sure the significance of these results or what the treatments will help with. On the other hand, if an adult with Down Syndrome does end up presenting with a fragility fracture, just like you would in an otherwise healthy adult, they would recommend the same screening for other conditions that could be associated with that.

[00:09:00] Particularly those that we already talked about, things like thyroid disorders, celiac disease that might cause malabsorption, and then other more general things like vitamin D deficiency,

hyperparathyroidism, and taking a good look at that medication list to see if there's any those medications that could be contributing to your osteoporosis.

[00:09:14] **Christopher:** So, basically shared decision making, some discussion, and then, maybe enlisting the help of your local endocrinologist, if there are some more issues that you're not quite sure how to handle next steps.

[00:09:24] **Michael:** That sounds like a perfect approach.

[00:09:26] **Christopher:** What other things do we need to be looking for in these patients? The big buckets we might have to be looking at?

[00:09:30] **Michael:** So in terms of things along the psychiatric approach different behavioral difficulties and mental health conditions. There's not really good estimates of how prevalent those are in this population, but it's certainly possible.

[00:09:41] There were old stereotypes that these patients were always happy all, and never had any, uh, concerns for depression or anxiety. Those have clearly been debunked. Depression and anxiety very much exist in this population. But it can be really hard to diagnose. Obviously there's a very wide spectrum of verbal skills, which can make the diagnosis challenging for someone that's not as familiar with this population.

[00:10:01] And depression and anxiety won't always manifest as they might in a typical patient. On that note the guidelines recommend that if you have a concern for a mental health disorder, referring to someone who is familiar with the Down Syndrome population, or at least a population with intellectual disabilities, so that they can get a more thorough screening and evaluation. Diagnosis is still recommended following criteria of DSM-V or the DM-ID-2, which specifically looks at behavior health conditions in populations with intellectual disabilities.

[00:10:30] **Christopher:** it's good to know that there's a different type of set of guidelines that may be more useful or validated in this patient population.

[00:10:36] **Michael:** Yeah, absolutely. So what these guidelines seem to recommend for the general internist, the primary care provider, is that each visit, particularly the annual physicals, doing a general screening of the patient's behavioral, functional, adaptive and psychosocial factors. This is actually another tool kit that the Global Down Syndrome Foundation has available on their website for families. It mentions different areas of functioning and changes that you could see in those areas. It helps families to identify changes that might be happening could pretty easily be overlooked, bring those concerns to the primary care provider so they can help screen for those things and then, as these guidelines recommend, referring out if the diagnosis is in question at all.

[00:11:15] **Christopher:** Now, does this toolkit have a provider answer sheet to help us interpret what these answers these are?

[00:11:20] **Michael:** It's more kind of like a list of symptoms and things to be on that on an eye out for. So for the provider that's not as familiar with this population, it could be helpful for them as well. But it seems like the main goal is really for families to help them identify things.

[00:11:33] **Christopher:** Gotcha. Are you aware of, here, especially here in the Columbus area, what type of resources we might have for these patients?

[00:11:38] **Michael:** So, here at Ohio State, we actually do have an adult Down Syndrome clinic. I believe it has a once monthly clinic for these patients with an in clinic psychologist. So, they're certainly able to help evaluate these patients from that perspective. And again, referring back to the Global Down Syndrome website, they actually have a list of Down Syndrome centers and facilities around the country. So people can hopefully find a location that can help with these next steps after they've identified a potential concern.

[00:12:06] **Christopher:** I know you said this is sort of where you might be referring to someone who may have a lot more experience as an internist or as a general practitioner... as a primary care provider. Would choices in management be pretty similar? Would it be a combination of things like CBT and SSRIs?

[00:12:20] **Michael:** Yeah, as, as far as

[00:12:21] I'm aware, once we get to the management steps, things are still pretty similar. If there's more problem behaviors, things like atypical antipsychotics could potentially be used sooner than they might in the more general population. But, as far as I'm aware, the CBT, SSRI, SNRI, still the first line in this population. But that wasn't something that was specifically addressed in these guidelines, at least.

[00:12:41] **Christopher:** Gotcha. There's increased prevalence of dementia in this population as well?

[00:12:44] **Michael:** Yep. So different studies will tell you different estimates on how prevalent it is and exactly when it starts developing. But, the limited studies that are available do seem to indicate that prevalence of dementia in adults with Down Syndrome below 40 is pretty much non-existent but with a sharp climb after age 40.

[00:13:00] Some of these studies estimated about 10% and those under 50, up to 30 percent by the age of 60, and some, one study even estimated up to 90 to 100% by the age 70. It certainly gets common later in life but one of the key points of this was to use pretty significant caution in diagnosing dementia as the cause of behavioral changes in adults with Down Syndrome that aren't even 40 yet. In those cases, we should be thinking about screening for some of those behavioral health conditions that we just talked about, or screening for some other medical comorbidities that might be presenting with these behavior changes.

[00:13:32] **Christopher:** And in thinking about some of these that you're talking about dementia, but also even beforehand. Many may have some sort of developmental delay or verbal difficulties. Can you explain a little bit about how we might approach advanced care directives, guardianship? Do you know much about that?

[00:13:46] **Michael:** So, it's not a topic that I'm as familiar with in terms of how to go about pursuing those things. It's something that I'm open to learn more about and pursue as I join the faculty here but it's something that I haven't had as much exposure to as a resident. I do know that our Down Syndrome clinic, I'm sure is familiar with some of these issues particularly for the non verbal patients that aren't really, is able to make those decisions for themselves. Guardianship is very often pursued. I'm sure that clinic has some familiarity with it, and some other clinicians within the Ohio State group that have familiarity with the population of intellectual disabilities can probably help provide some guidance as well.

[00:14:17] **Christopher:** Excellent. I think that's probably a good topic for the future because we probably have more than just outside the Down Syndrome patient population that this may be very applicable to.

[00:14:25] **Michael:** Absolutely.

[00:14:25] **Christopher:** So, I want to move on to something that probably most of us are much more comfortable with, like cardiovascular screenings, because I think this is something that we generally do for our adult populations, but may have different prevalence within our down syndrome population.

[00:14:37] **Michael:** Yeah, absolutely. So again, limited studies and limited data available on it. One of the studies actually seemed to indicate that ischemic heart disease is actually a little bit less common in the Down Syndrome population compared to the general population. So one of the few things we've talked about so far that's less common in this group but that estimate is about 8 percent versus 13 but given the kind of overall low quality of evidence and the benefits to screening and potential treatment, they still recommend pretty much the same thing as the general population. So, beginning at age 40 evaluating risk with an ASCVD calculator and then having a risk benefit discussion, recommending statins along with the same general risk guidelines as the general population.

[00:15:14] **Christopher:** Gotcha. So as far as we know, we can apply the same general risk calculators or pooled cohort to this population relatively reasonably.

[00:15:22] **Michael:** Relatively reasonably. Again, there's pretty low quality of evidence one way or the other but from just a risk benefit standpoint, the benefit seems to be in favor of treating those conditions.

[00:15:30] **Christopher:** And this is mostly for cardiovascular disease or can this be applied to stroke too?

[00:15:35] **Michael:** Similar for cardiovascular and stroke prevention. One caveat for stroke prevention is that congenital heart disease is very common in this population. So similarly to behavioral health, this is not something that the general internist is expected to do on their own. If there's a history of congenital heart disease this patient should really be seeing a cardiologist ideally an adult congenital cardiologist that's familiar with their underlying physiology. Stroke risk associated with that, and can have a more in depth, individualized discussion on the best stroke prevention approach in their specific anatomy.

[00:16:03] **Christopher:** And what one hopes is if I'm coming into a patient as an adult provider that some of these congenital diseases have already been identified and they've already been hooked up with some sort of provider that has good experience in that area.

[00:16:15] **Michael:** Absolutely. I would hope so.

[00:16:16] **Christopher:** Yeah. And I believe here at OSU we do have some good congenital heart disease physicians. Med-Peds trained as well.

[00:16:23] **Michael:** Absolutely. Another plug for that Med-Peds training.

[00:16:26] **Christopher:** I really appreciate the time we had today. Are there any other points you want us to know about before we leave?

[00:16:31] **Michael:** No, I think that was all my major points. Again, just throwing a shout out to that Global Down Syndrome Foundation website. That's the group that helped sponsor this article. And they have, as I've already mentioned, a lot of those different toolkits available for both providers and families. So they have this full guideline available in both original journal article format, as well as more of a kind of brochure format that looks at some of the, it gives you the specific studies and evidence that they used to make these recommendations, and they also modified it so it is more approachable for the patients with Down Syndrome themselves and their families along with these toolkits I've already mentioned.

[00:17:03] So, they have things looking at diabetes, celiac disease, behavioral health well as a nice handy checklist for families so that they can identify these things that should be evaluated and screened at each visit, bring them to their appointments, and make sure that all this this care being being addressed.

[00:17:18] **Christopher:** Fantastic. Do you have any other medical interests that you might have? We might have to bring you back on topic because I had a great time today.

[00:17:23] **Michael:** I'm very open. We'll see what comes up along my way once I start joining the faculty here.

[00:17:28] **Christopher:** Oh, I'm really excited to have you join our faculty and look forward to all the great things you're going to be able to do for us.

[00:17:33] **Michael:** I appreciate it. Thanks for having me.

[00:17:34] **Christopher:** Thank you for listening to another episode of everyday medicine, a podcast from the division of general internal medicine brought to you by the Ohio chapter of ACP. Please consider subscribing to our feed on your favorite podcasting platform. So you don't miss out. You can get all our show notes and transcripts soon from our division webpage at https://medicine.osu.edu/GIM. Have a great day. Bye!