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Practical Perspectives in Myasthenia Gravis # 1: Diagnostic Logistics—Navigating Complexity Across Ages

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Welcome to CE on ReachMD. This activity, titled “Practical Perspectives in Myasthenia Gravis # 1: Diagnostic Logistics: Navigating Complexity Across Ages” is provided by Prova Education.

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Dr. Strober:
Welcome. I'm Dr. Jonathan Strober, and this is CE on ReachMD. Today, I'm joined by my colleague, Dr. Diana Castro. We're exploring the diagnostic journey in generalized myasthenia gravis. While recognition and logistics are similar, they do differ between children and adults.

Dr. Castro:
Good to see you. And yes, you're right that the problem is that the immunopathology is shared between adults and kids, but the way the patients get diagnosed, the way the patients present to the clinic, it's a little bit different. And the things that we can do in pediatrics, for example, I'm always looking for genetic causes. So this is something that maybe on the adult side, we don't do as often, but it's definitely something that, once you learn the difference, I think it helps you moving forward to identify pediatric patients versus something else.

Dr. Strober:
Yeah, I agree. I think in adults, I know that delayed recognition can come because a lot of people will just say, “Oh, they're just fatigued or aging.” So what does the delay look like in children?

Dr. Castro:
I mean, the good thing with children is that most of the patients I have seen over the years present with eye symptoms, with unilateral ptosis, most of them, and then it can progress to go through other side. So those patients, many times, will go first to the ophthalmologist. And the ophthalmologist will be the one looking at the nerve. Optic nerve is fine. Everything else is fine, but the patient has ptosis. So the next thing they will recommend is to see a neurologist.

So in that case, I think it's helpful. And later we'll talk a little bit more about adults, but the patient usually doesn't present with a straightforward fluctuating fatigue, for example. So I will say eyes. Keep an eye on the eye.

Dr. Strober:
Yeah, I agree. I think that strabismus is so common in the pediatric population that oftentimes a little bit of extraocular muscle weakness gets diagnosed as strabismus in a child, especially the younger kids.

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So let's talk about those early red flags. What's usually your first indication that you might be dealing with myasthenia rather than another neuromuscular or ocular disorder?

Dr. Castro:
So like I said, the ocular symptoms go first. And you just reminded me of something, because the first symptom I see is ptosis, like I say, unilateral, and they will move through the other, but also eye movement, the limitation of the eye movements. That's one of the things that will yield for—you will start looking for a central condition, like brain conditions, if you have the eye movement limitation. But let's say you have the eye movement and you have ptosis, that's a really good way to get a patient diagnosed.

With older patients, what I see is then they can express, "I'm fatigued. I'm tired. I wake up with energy, but then I'm tired and get tired, tired over the day." The weakness, it's key to ask if it fluctuates throughout the day, because this is typical for myasthenia gravis that we don't have in other conditions. So the patients tell you, "I feel good, but then I went to a game, and after the game, I was done for the day," right?

Weakness not only affecting the muscles of the face, sometimes obviously muscles to be able to speak. So the speech may sound different. But then proximal weakness, so shoulder weakness, hip weakness, can be another thing that we find. Sensory deficits should not be present. Atrophy should not be present, because this is more a subacute than acute disease, right? And reflexes are something that I always tell people, normal reflexes with any kind of facial weakness, most of the time, is going to end up in myasthenia.

Dr. Strober:
Yeah, I agree. I think the fatigue thing is also something that gets missed a lot in the pediatric age group. A lot of parents will come in and say, "My kid loves to go to the playground," but then they're sitting on the side and they can't really keep up with their kid, and so we have to kind of decipher what the parents are telling us, and kind of go, "Oh, that must be fatigue."

Dr. Castro:
Before I forget something, I have seen a handful of patients that present with inability of drinking from a straw. That's very interesting.

Dr. Strober:
Yeah, I think that reminds me also of a lot of our adolescent patients that get diagnosed, right? Either the ptosis, people think that they're on drugs because they always have their eyelids closed. Or they don't smile. I've had parents say, "They never smile for pictures," right? There's always some other excuses as to what is going on that kind of makes you not think about myasthenia. But hearing all those clues, I think, really helps you go, "Wait, there's something going on here that we need to kind of get a better handle on."

So once you've raised your suspicion clinically, the next big step, of course, is confirming it, which is probably a little bit easier in adults than in kids. How do you address this challenge?

Dr. Castro:
Yeah, so in children, I'm going to be honest, I try as best as I can to avoid electrophysiologic testing because it is painful and it's difficult; the patient has to cooperate. Like you said, an adult will hold on much better than a pediatric patient. So the first thing I do is that if I have that high suspicion, I will do antibody testing.

So right now, my practice is sending the acetylcholine receptor testing and then they reflex to MuSK and LRP4 testing, in case acetylcholine receptors are negative, right? And if I have negative on all of them—acetylcholine receptor, MuSK, and LRP4 negative—I think about, in pediatrics, for example, I do genetic testing because genetic testing is something that is easier to get in the pediatric population and can give us some other differential diagnoses. For example, congenital myasthenic syndromes, right? They can present very similar. Usually they are more chronic in time compared to MG, but it can happen.

I have done—for some of my seronegative patients, I have done also a cell-based assay because those are cases that I know they have myasthenia, so I keep trying to test and see what else. And then we may do single-fiber for the older patients and obviously the repetitive nerve stimulation testing.

Dr. Strober:
Yeah, I find that the repetitive nerve stimulation testing is not just hard but also kind of has a low likelihood of picking up those kids that

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are seronegative, although sometimes we do. The single-fiber is a little bit more sensitive but really hard to do, especially in the younger kids since they do have to really cooperate to do it.

One of the other things in the old days, as I like to say, we used to do Tensilon [edrophonium] testing, right? We would bring the kids into the office and give them Tensilon and see if they improved. But nowadays, I'll sometimes, if I really suspect it, I'll put them on Mestinon [pyridostigmine] and see if we get some improvement. That just gives me another clue that we're dealing with myasthenia. So it's kind of that triad of antibodies, some sort of medication response, and electrophysiology if you can get it there.

For those just tuning in, you're listening to CE on ReachMD. I'm Dr. Jonathan Strober, and here with me today is Dr. Diana Castro. We're discussing practical perspectives in myasthenia gravis.

Dr. Castro:
Yeah. But talking about how easy is to get it in adults compared to children is, yeah, it's key in terms of electrophysiology.

Dr. Strober:
What makes it so complicated in pediatrics?

Dr. Castro:
So I think, I mean, obviously cooperation. They don't want to stay still for somebody that is giving them electric impulses in their muscles, so they don't cooperate with that part. And then really, like you said, if you want to do a good repetitive nerve stimulation, you have to be very still.

So I, in the past—I'm talking about 10 years ago, 8 years ago—I was doing a lot of studies under sedation. So for example, the patient was going to get a muscle biopsy because we knew there was a congenital—some kind of myopathy, but we wanted to make sure that we're covering the spectrum of myasthenia gravis, so we'll put the patient under sedation. Under general anesthesia, they would get the biopsy. And then after that, he would leave, then I will stay and do the repetitive nerve stimulation. I think that was the best way I could do it.

I saw a lot of cases of botulism, so I did a lot of babies in the NICU, which I had to do. But you can hold a newborn, a little kid, better than you can hold a 5-year-old who is not going to let you do anything.

So I think that is the difference, the challenges that we find with electrophysiologic testing. So that's why I try to do first the other things we were talking about.

Dr. Strober:
Yeah, and I think, then, if that's negative and you still like to do single-fiber, besides it being really hard in the kids because they do have to cooperate, is finding the people who can do single-fiber in kids. It's hard enough to find it in adults, but I know that there's very few people who actually know how to do a single-fiber and can do it in a pediatric patient. So that just makes it a lot harder to kind of utilize in our tool kit, unfortunately.

So if you had one takeaway for clinicians, regardless of the patient's age, what would it be?

Dr. Castro:
Yeah, you're asking me, regardless of the patient age. I wish you did not, because I would say in children, I think the first thing you have to look for is the ptosis in the eyes. So the eyelid weakness, the limitations of the eye movement. I will say those two are going to be some of the first things you're going to see in the kids.

If you think about an older—kind of like teenagers, like you said, plus they have this facial weakness, they may have difficulties, for example—the example about the straw—they may have difficulties using a straw. So I think the clinical picture is not as complicated. I will say that with adults, it gets more complicated. It's not as complicated in kids as it is to me in adults, because in adults, there are so many other comorbidities and other things that can happen and can cause the problems.

And I have seen, unfortunately, many women, they call them, “Oh, it's just anxiety. It's just depression. You don't have anything.” And

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the poor woman is completely fatigued and weak. I have had patients that I met that the first encounter was, in full, they couldn't swallow or they couldn't breathe, and they had to run to the ER. But they had been in that ER 5 times already, right?

So I think that's the difference. That's why I say I don't like when you ask me for the children and the adults at the same time.

Dr. Strober:
Well, it does make me think in the adolescent population, I think, functional symptoms, sometimes people think adolescents are kind of—not making it up, but they're undergoing a lot of stress, and maybe that's bringing things out that aren't real.

I think, to me, the one key takeaway is to think about myasthenia. You talk about facial weakness, like I think of FSH in muscular dystrophy, which can have unilateral symptoms, facial weakness, can present with some facial weakness. We talk a lot about proximal muscle weakness, which looks like a myopathy, hence why you do EMGs in those kids that are getting their muscle biopsies.

So I think it has to be on the table. I mean, I've heard people say kids don't get myasthenia. Right? But it's so important you've got to at least have it on your list, and you've got to be thinking about it, because otherwise you could miss it, because it's really hard to diagnose too. So if you're going down the myopathy route and you're not really getting there, at least kind of think about that as another possibility.

So in this program, we focused on getting the diagnosis right. In another discussion in this series, we're going to pick up where this conversation leaves off. Stick with us to learn more about designing truly individualized treatment plans and blend established and novel therapies that are guided by antibody profiles, age, and patient-specific needs.

Thank you for joining us for practical perspectives. See you next time.

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