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Evaluating Endocrine Complications in DMD Patients: Findings from a Survey

Dr. Turck:

This is *NeuroFrontiers* on ReachMD, and I'm your host, Dr. Charles Turck. Joining me to discuss the results of a survey that assessed endocrine complications and surveillance in patients with Duchenne muscular dystrophy, or DMD for short, are Drs. Nadia Merchant, Melody Shi, and Despoina Galetaki. Dr. Merchant is a pediatric endocrinologist and Assistant Professor at the University of Texas Southwestern Medical Center in Dallas. Dr. Merchant, thanks for being here today.

Dr. Merchant:

Thank you for having me.

Dr. Turck:

And Dr. Shi is a pediatric endocrinologist at Children's National Hospital in Washington, D.C. Dr. Shi, welcome to you.

Dr. Shi:

It's a pleasure to be here today.

Dr. Turck:

And also from the Children's National Hospital is Dr. Galetaki, who is a Pediatric Endocrinology Fellow. Dr. Galetaki, it's great to have you with us as well.

Dr. Galetaki:

Likewise.

Dr. Turck:

So let's start off with you, Dr. Merchant. Would you give us a brief overview and background on DMD and why steroid-induced endocrinopathies are so common in these patients?

Dr. Merchant:

So for Duchenne muscular dystrophy, one of the main treatments and standard of care is steroids. Studies have shown that it delays loss of ambulation in these children; it improves muscle function and strength and can also help from a cardiac standpoint. That is one of the main treatments. However, prolonged steroids has many endocrine side effects, and these include weight gain, insulin resistance, adrenal insufficiency, affecting growth, late puberty, and specifically, bone fragility, and so that is one of the main reasons why we thought this was important to address the side effects in these children who have steroid-induced endocrinopathy.

One of the primary complications is low bone mineral density and osteoporosis, which there's been a lot of work done compared to the other complications. And osteoporosis happens because the steroid itself leads to death, in a sense, of the osteoblastic cells, which decreases that bone formation and increases the osteoclastic activity. So basically, they are losing more bones, and the bones become weaker; and then, as they lose ambulation, they lose that muscle tension, and that affects it even more.

So the other side effects happened more because of the hypothalamic-pituitary-adrenal axis being suppressed, and so it affects like the

growth hormone pulsatile activity that we get along with affecting suppressing puberty. These all kind of go together, and many of these are treatable in some ways or manageable, and so I think having that awareness and knowing these side effects helps us so we can start thinking about them early and addressing them.

Dr. Turck:

Thank you for that background, Dr. Merchant. And if we turn to you now, Dr. Shi, and dive into your research, how was it designed, and what methods did you employ?

Dr. Shi:

Despite our understanding of these steroid-induced endocrinopathies, we don't really know how this data is being presented and perceived by the DMD community. I think, fortunately, there's been a bigger push to involve endocrine earlier on in the care of these patients, but we really wanted to hear from patients and families themselves how they like to receive information and what their experience has been in engaging in these sorts of conversations. So for our study, which was fortunately funded by the Foundation to Eradicate Duchenne, we designed a patient- and family-focused survey, you know, asking questions pertaining to the various complications from chronic steroid treatment. As Dr. Merchant alluded to, you know, questions on bone health, puberty, adrenal insufficiency, growth, obesity, etcetera. And we really wanted to keep the survey digestible, so we limited it to about 30 questions; for example, one of them being, "When did you first learn about adrenal insufficiency as a complication of steroid treatment? Was it at the start of your treatment? Or have you been on it for several years and then heard about it? Or during a hospitalization?" So really just getting at their lived experiences of these issues.

And we partnered with PPMD, which is the Parent Project Muscular Dystrophy group—which, if you haven't heard of them, is an incredible parent-run organization that not only provides support and education to families, but invests a tremendous amount of funding into research to fight for a cure for Duchenne's. So they helped disseminate our survey. In the end, we had 75 people, whether it's patients with Duchenne's or caregivers of patients with Duchenne's. They filled it out. We got a really good mix of children, teenagers, and young adults. And preliminarily, I would say that most of them were on steroids—about 95 percent of them, which is not surprising—and even though half of them were already being seen by an endocrinologist, almost a quarter of them actually said they never met with one before, suggesting that there may be some kind of educational gaps.

Dr. Turck:

And turning to you now, Dr. Galetaki, what were some of the other key findings in the study?

Dr. Galetaki:

Yeah, so there was a good mix of surprises in our results, actually. Starting off with bone health, this is one of the few domains where there seems to be the greatest success in evaluating and monitoring for complications, which is very encouraging. Almost 90 percent of our participants had discussions with their providers about the side effects of steroids on their bones, and a good amount of them were even receiving therapy, such as bisphosphonates. On the contrary though, as Dr. Merchant and Shi alluded to, adrenal is one of the domains that is very rarely discussed. Only half the participants have ever discussed the matter with their providers, and in few of these cases, that was even after an adrenal crisis or during a hospitalization. Only half of the participants had an emergency letter for adrenal insufficiency, and only a third had an emergency intramuscular rescue medication. So as you can imagine, this is extremely concerning given the life-threatening implications of an adrenal crisis and the fact that, essentially, all of these boys are considered to have adrenal insufficiency. On the other hand, close to 60 percent of the participants had discussed the matter of obesity and metabolic implications, and about 14 percent of them have been on some sort of oral medication for that. Still though, a quarter of patients reported that they have never had a discussion about obesity before, and they would really have liked to. And then lastly, about 50 percent of our participants were diagnosed with delayed puberty, and a little bit over 20 percent have been on hormone replacement therapy with testosterone.

And then, of course, one of the main things we also wanted to look at is how the caretakers and families feel about that and how can we do better. The overwhelming majority, close to 90 percent, reported that they want to learn more about the endocrine complications related to their disease and steroids, and they consider education as extremely important to them, which is something we need to take notice of.

Dr. Turck:

For those just tuning in, you're listening to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck, and I'm speaking with Drs. Nadia Merchant, Melody Shi, and Despoina Galetaki about a survey they conducted to learn more about endocrine complications and surveillance in patients with Duchenne muscular dystrophy.

So let's come back to you, Dr. Merchant. Given the results that Dr. Galetaki just described, what gaps currently exist in DMD care?

Dr. Merchant:

So Duchenne muscular dystrophy children are doing better, living longer, and going into adult life with all the new treatments coming through, and I think that this really shows us that we need to do better in improving the management of these complications. We expected more children and adults that had reported to have adrenal insufficiency education and to be started on treatment for delayed puberty, and so this really just proves to us that we could do better and really need to make sure we're doing what's best for these patients so they can live longer, healthier lives without having severe complications. So those are the gaps that I think this really illustrated to us.

Dr. Turck:

Thank you for that insight, Dr. Merchant. And now before we close, I'm curious what our other guests think we should keep in mind moving forward to help bridge these gaps and focus more on quality of life. Dr. Shi, care to start us off?

Dr. Shi:

I would say earlier involvement of endocrine to help families and neurologists monitor some of these complications. From our study, families living with DMD want to hear about these potential side effects just as much as they want to learn about the newest treatments coming down the pipeline, but we recognize that access to endocrine is still an issue across the world, so we still need to keep different avenues of education open and available as families also learn in different ways too, like webinars, maintaining a presence at conferences—such as the PPMD conference that's held annually that's specifically geared towards families—doing podcast shows like this and raising awareness of these complications, and certainly having printed materials in all sorts of different languages. And also from our survey, families want to hear directly from their neuromuscular teams who they have built up a trusting relationship with regarding these issues, so we need to continue to partner with the neuromuscular teams for early endocrine involvement.

Dr. Turck:

Thanks, Dr. Shi. And, Dr. Galetaki, I'll give you the final word. How can we better bridge these gaps in care?

Dr. Galetaki:

While much of these conversations we had today have been centered around these complications, we also need to be optimistic, especially given the many novel therapies coming down the pipeline, like the approved gene therapy access for all ages and vamorolone being accessible. We do need to continue thinking about how the field is growing, how this patient population is aging, and keep reassessing their needs and the family's needs while also keeping in mind the side effects of those new therapies. And even considering incorporating endocrine complications, for example, as a measure in clinical trials for the emerging therapies and treatments would continue to bring endocrine complications in the back of everyone's mind. Also, continue to try making that standard of care when caring for these patients is of great importance as well.

Dr. Turck:

These are great comments for us to consider as we come to the end of today's program, and I want to thank my guests, Drs. Nadia Merchant, Melody Shi, and Despoina Galetaki for joining me to discuss the results of their survey and how we can optimize patient care in the management of Duchenne muscular dystrophy. Dr. Merchant, Dr. Shi, Dr. Galetaki, it was great having you all on the program.

Dr. Merchant:

Thank you.

Dr. Shi:

This was great. Thanks for having us.

Dr. Galetaki:

Thank you so much.

Dr. Turck:

For ReachMD, I'm Dr. Charles Turck. To access this and other episodes in our series, visit ReachMD.com/ *NeuroFrontier*s, where you can Be Part of the Knowledge. Thanks for listening.