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Improving Spinal Muscular Atrophy Outcomes Through Early Intervention

Announcer:
You're listening to *NeuroFrontiers* on ReachMD, and this episode is brought to you by Biogen Inc. Here's your host, Dr. Charles Turck.

Dr. Turck:
Welcome to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck, and joining me to discuss the importance of early and sustained treatment in spinal muscular atrophy, or SMA for short, is Dr. Leslie Hayes. She's an Instructor in Neurology at Boston Children's Hospital and Harvard Medical School specializing in neuromuscular disorders. Dr. Hayes, thanks for being here today.

Dr. Hayes:
Thanks for having me.

Dr. Turck:
Now, so for some context, Dr. Hayes, why is timing such a critical factor in SMA care, especially in presymptomatic infants?

Dr. Hayes:
So I would actually say timing is sort of everything. We know that SMA, or the dysfunction of motor neurons that causes SMA, begins probably even in utero in some cases, particularly those with the most severe type, type 1 SMA, and that motor neuron loss begins before symptoms emerge. And so treating infants presymptomatically, or before we notice symptoms, as the parents or as neurologists, gives kids the best chance for preserving motor neurons and having the best long-term outcomes. We have amazing treatments for SMA, but unfortunately, those treatments really only stop the progression of the disease, and so we can't go back and salvage motor neurons that have been lost prior to treatment, either in utero or soon after birth. So treating earlier preserves the most motor neurons.

Dr. Turck:
Now, the NURTURE and SPR1NT trials showed that infants treated before symptom onset could achieve milestones like walking that were previously unthinkable in the natural history of the disease. In your opinion, why are these studies so pivotal? And how have they changed our clinical approach?

Dr. Hayes:
So I think they confirmed what we all thought before, which was that earlier treatment was probably better. When we looked at those studies as compared to the studies of kids who were treated symptomatically, kids treated presymptomatically did much, much better and achieved those milestones like walking, as well as had amazing gains in respiratory function and bulbar function. And that's really been sort of the motivating factor for the newborn screen and allowed us to advocate for SMA to be on the newborn screen now in all 50 states, which has allowed for the early treatment in SMA.

Dr. Turck:
Well, given those studies, it's clear that early intervention with the right treatment option is critical. So how do route of administration and age-related considerations shape therapeutic decisions across patient populations?

Dr. Hayes:
So they play a large role. Because at this point, many of the studies, as well as real-world experience, has shown that the differences between the treatments is minimal, or they're potentially even all the same. And so oftentimes, what patients are eligible for based on their age and the family's preferences around the route of administration, as well as side effect profile, end up being some of the drivers of the decision-making for which treatment a child gets.

So when we think about something like nusinersen, which is an intrathecal delivery of medication, there are some ages where that can be particularly challenging: the younger ages when they need anesthesia for that procedure, for example. And so there are factors that are sort of individualistic and occur at different points throughout their life.

Obviously, when we think about onasemnogene, that's a treatment that's only available for kids under two, so that has a more limited population that can receive that treatment. It's a one-time intravenous therapy.

Obviously, risdiplam is an oral daily medication. So that, on its surface, sounds the easiest, but some families would really prefer to have a treatment every four months and not have to remember to take a medication every day.

So these are all the discussions that we're having with families and really making sort of a recommendation based on the individual patient and the individual family's preferences.

Dr. Turck:
For those just tuning in, you're listening to *NeuroFrontiers* on ReachMD. I'm Dr. Charles Turck, and I'm speaking with Dr. Leslie Hayes about early intervention when caring for patients with spinal muscular atrophy, or SMA.

So Dr. Hayes, let's turn our attention now to another crucial part of SMA management, patient education. For the families of newly diagnosed patients, the decision to start an aggressive treatment can be daunting, so what can we do to effectively frame the urgency of early action without overwhelming them?

Dr. Hayes:
So I think this is crucial and really challenging. The first meeting with a family in those early days when their child's just been born is a really tough time to have these conversations. And it's really important that we go through everything thoroughly, using both visual education techniques as well as feedback from the families to make sure that they're grasping the concepts that we're illustrating. It starts often with describing the diagnosis and understanding the natural history of SMA, because I think that's really crucial to put into context when we then talk about treatment options. And then to lay out treatment options, emphasizing that early treatment is probably the most important thing, maybe even more important than which treatment they choose. And to present the different options, laying out all the risks, the benefits, the administration differences, but in a compassionate way, while also emphasizing that early treatment is going to improve outcomes and give their child the best chance for a maximal motor function.

Dr. Turck:
And is there anything else you can tell us about how providers can best incorporate information about the burden of treatments and long-term outcomes when educating patients and their families?

Dr. Hayes:
So I think, as I mentioned earlier, one of the important aspects around education is understanding the natural trajectory of what would happen without treatment. And so I think that helps families, even though these are really intense therapies and this is a difficult disease. But with treatment, the long-term outcomes look very different than what kids used to experience.

And I think once that's made very clear to families—yes, there are burdens of treatments, they're real, and they shouldn't be underestimated—ultimately, when they see how effective these treatments can be, I feel like most families come around to accepting that path and knowing that this is a chronic disease that will need to be managed, but the outcomes are quite good.

Dr. Turck:
And before we wrap up, Dr. Hayes, are there any final insights you'd like to share about early treatment decisions for patients with SMA?

Dr. Hayes:
I think I would just like to continue to advocate for urgent referrals and connection with an SMA care center that can provide these different treatments and can discuss them with families in a knowledgeable and compassionate way. And also to advocate for carrier screening, which would allow for even earlier diagnosis in the prenatal period and even more preparation for families to learn about the diagnosis and the treatment options so that both they and we are best prepared when the baby is born to treat early.

Dr. Turck:
Well, with those final thoughts in mind, I want to thank my guest, Dr. Leslie Hayes, for joining me to discuss how timely intervention can improve outcomes for patients with spinal muscular atrophy. Dr. Hayes, it was great speaking with you today.

Dr. Hayes:
Thank you.

Announcer:
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