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Closing Care Gaps and Addressing Unmet Needs in SMA

Announcer:
You're listening to *NeuroFrontiers* on ReachMD. On this episode, we'll take a look at the unmet needs of patients with spinal muscular atrophy with Dr. Jennifer Kwon. Dr. Kwon is a Professor of Child Neurology at the University of Wisconsin School of Medicine and Public Health; she's also the Director of the Pediatric Neuromuscular Program at UW American Family Children's Hospital in Madison, Wisconsin. Here's Dr. Kwon now.

Dr. Kwon:
Spinal muscular atrophy occurs in roughly 1 in 10,000 births. That means that every year in the U.S., about 400 children are born with spinal muscular atrophy. Before the development of disease-modifying treatments, that would mean that those 400 children would grow up to develop progressive weakness in their life. Many of them would die very early usually before two years of age without intensive supportive care, such as chronic ventilation and artificial feeding.

We live in a time where spinal muscular atrophy is changing because of disease-modifying treatments. Depending on when children get these treatments and how weak they are when they receive them, their disease course will be different. And we don't fully understand how to help our adult pulmonology and neurology and rehabilitation colleagues care for them through their adult lives.

Another area of unmet need is that even though it's exciting that children can be treated for their spinal muscular atrophy, we really don't know how they're going to develop through their lifetime. We hope that they will have normal development, but they still need to be monitored closely for complications that frankly, we haven't really thought about. That means that a real unmet need in this population is the need to closely follow them, follow their development and interact with other specialists. So that calls for the need for a registry, so that we can enroll data about these patients and follow them over time and begin to understand what the effects of these disease-modifying treatments really are.

So, I would say those are the two greatest areas of unmet need. One is caring for our very weak patients as they grow into adulthood, and then caring for our stronger, treated patients as they grow in their life and maybe develop complications that we haven't thought about.

So in terms of the patients who are children but who did not have the benefit of disease-modifying treatment early in their childhood, but who are entering adulthood as very weak, often technologically dependent young adults, I think that we need to rely on those clinicians who have dedicated a large part of their career to studying what these patients need and how to best serve these needs as they enter into adulthood. So, these pediatric clinicians really need to talk with their adult counterparts.

I also think that we need to do a better job of interviewing families directly. We have had, again, some efforts at looking at families' quality of life and what's important to them. But I think that as these children grow into young adults, we really need to focus on them and try to figure out ways of accessing their preferences and their values. This can be difficult because many of these young adults have a very difficult time talking and communicating with others. Their families may understand them, and many of them live their lives through parents or caregivers who understand them well. So, I think we need to really think about how to better understand this population.

The second population of children who gratifyingly are treated early in life with disease-modifying treatments the issue of having a registry in place to follow these children as they grow and develop so that we better understand the needs and complications they may face as they grow up. That's an issue for the neuromuscular community and the families to sort of partner with and come into together. I think registry development is always the holy grail of natural history studies and understanding treatment outcomes. But they're just very hard to implement and put in place.

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Cure SMA has very ambitious ideas for how to collect medical data for their SMA registry. And I think that those ideas need to be supplemented by other efforts, for example, those in the newborn screening community to collect data on infants identified by newborn screening and how they do. I think that that is going to be more complicated to put in place, but I hope that the community will continue to have discussions about how to do this.

Announcer:
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