Transcript Details

This is a transcript of an educational program. Details about the program and additional media formats for the program are accessible by visiting: https://reachmd.com/programs/neurofrontiers/continuum-of-care-managing-dmd-in-pediatric-and-adult-patients/26382/

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Continuum of Care: Managing DMD in Pediatric and Adult Patients

Announcer:

This is *NeuroFrontiers* on ReachMD, and on this episode, we'll discuss how we can best treat and support patients with Duchenne muscular dystrophy throughout their entire care journey with Dr. John Brandsema. He's a pediatric neurologist and the Neuromuscular Section Head at the Children's Hospital of Philadelphia. Let's hear from Dr. Brandsema now.

Dr. Brandsema:

One thing we've struggled with in the Duchenne community is a diagnostic delay, so it is quite well documented that the symptoms and signs of Duchenne start before the time that a child is engaged with a neuromuscular specialist, on average anywhere from six months to up to two or three years of seeing multiple specialists before the patient presents to neurology. What we tend to hear from families is about difficulty with motor function and specifically, things that take a lot of coordination and strength, so that would be things like rising from the ground or going up and down the stairs. Parents start to notice that their child falls frequently compared to other children, and this is usually around the age of 2 or 3 years that this becomes evident, but there's been delays in milestones previously when you suss it out with families. It's important to recognize that about a third of our patients also have speech delay and cognitive differences. Some can be on the autism spectrum or have issues with ADHD and cognitive delay, so this can also be a presenting symptom but is not usually the first thing that comes to mind when addressing that in the pediatric clinic, and so it can lead to further delays in diagnosis if someone's very severely cognitively affected also.

So the young patients need careful attention to some of the musculoskeletal aspects. Often the heel cords will become contracted early on. We see pseudohypertrophy of the calves in many of the children, and night braces and physical therapy are helpful for that. We need to do screening to make sure that there's not early cardiac involvement, but that's rare. And then we have to think about getting people on targeted therapies because we have many different options to choose from now, depending on the person's genotype or what kind of dystrophin mutation they have and also their age. You know, the standard of care is corticosteroid therapy with either deflazacort or prednisone. We also have vamorolone as an alternative steroid now, and then we have givinostat for those over 6 years of age of treatment, and then we have genetically targeted treatments, exon skipping for those who have skippable mutations of either 51, 53, or 45 in the dystrophin gene thus far what's on the market, and then gene transfer with delandistrogene moxeparvovec currently in clinic.

As this disease progresses, there is a significant limitation in function for patients. So the average young adult living with Duchenne is non-ambulatory, is dependent on a wheelchair for ambulation, and has impaired arm function that can range from barely being able to move the fingers and use a joystick in the early 20s, but that would be uncommon. Most people still have a significant amount of arm function in their 20s, and it tends to get worse more around their 30s.

The issues with the transition to adulthood in this disease are many. You know, going from a school-based system where often many of the therapies that they are receiving, such as physical and occupational therapy and sometimes speech therapy, are part of the school setup now switches to an environment where they may be pursuing secondary education or employment, or they may be unemployed, and in that situation, having insurance coverage in this country for therapy is more challenging to navigate. Also issues with the cardiorespiratory failure that happens in this disease start to become more prominent for most young adults. Some have been living with this even earlier on, but that's uncommon. Usually, it's in late teens and early 20s that you first start needing BiPAP support when you're sleeping, when you have significant enough heart failure that you're on multiple medications to try and ameliorate your heart failure, and you can have flares of either acute respiratory illness or cardiac-related disease that can lead to hospitalizations and symptom burden. These are all important to be aware of when you're dealing with the older individual with Duchenne.

Announcer:

That was Dr. John Brandsema discussing DMD care throughout the entire patient journey. To access this and other episodes on our series, visit *NeuroFrontiers* on ReachMD dot com, where you can Be Part of the Knowledge. Thanks for listening!