Transcript Details

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Diagnosing Duchenne Muscular Dystrophy: Recognizing Signs and Reducing Delays

Announcer:
Welcome to *NeuroFrontiers* on ReachMD. On this episode, we'll learn about screening and diagnostic strategies for Duchenne muscular dystrophy with Dr. Nancy Kuntz. Dr. Kuntz is an attending physician at the Ann and Robert H. Lurie Children's Hospital of Chicago and the Medical Director of the Mazza Foundation Neuromuscular Program. Let's hear from her now.

Dr. Kuntz:
Duchenne muscular dystrophy is something that, classically, has not been diagnosed in young children or infants. There was a study done by the CDC about 10 years ago and another one 30 years before that, and the average age at diagnosis was four and a half years. But the very interesting thing is that the families had already been concerned about the boys' motor development in each of those cases for several years. In fact, parents frequently report that—at least by the age of two—they begin to notice a difference between the boy who turns out to have Duchenne and his peers.

The primary symptom is a weakness in proximal muscles—the shoulders and hip girdles—so things like running, climbing, getting up from the floor, and trying to reach will show some of the earliest signs. The boys tend to have a gait with increased lordosis and a little bit of a waddle from side to side—because of the instability of the hip on one side—when they stand on one foot to advance the other. So in a fully developed case of Duchenne muscular dystrophy, the abnormal gait, difficulty getting up from the floor, and sometimes using the legs or knees to prop themselves onto their hands to stand in what's called a Gowers' maneuver are some of the very typical signs.

Once there's clinical suspicion of Duchenne muscular dystrophy, the most important thing to do is consider obtaining muscle enzymes, specifically a blood test of CPK. It would be very important for people to have a very low threshold for checking that in anybody—particularly boys—who have a motor delay and abnormal gait in their preschool or early school-age years. But in fact, it's important to recognize that Duchenne muscular dystrophy and dystrophin deficiency is something that also affects the nervous system, so there's a clear increase in the fraction of boys with Duchenne muscular dystrophy who have either mild cognitive delays, autism spectrum disorder, or executive dysfunction—for example, attention deficit.

So one of the things that I would encourage anybody evaluating boys for those kind of symptoms early in life is—in addition to any of the other tests they get—to add a CK in, because the CKs between a year and eight years of age are generally in the range of 15,000 to 25,000, which is massive elevation, making it very important to continue the diagnostic process. And if it comes back that high in a boy who has any weakness, then the next step would be to go directly toward confirmation with gene mutation analysis of the dystrophin gene.

It turns out there are a lot of families that have no prior history of Duchenne muscular dystrophy because the dystrophin gene is very large. It is 79 exons, and therefore, as such a large gene, it has a higher rate of spontaneous mutation. I would say that at least a third of the cases of Duchenne muscular dystrophy that we encounter are in a situation where there's no family history of such in the past—a de novo mutation—so just the absence of a family history shouldn't deter you from thinking about this as a possible diagnosis.

Announcer:
That was Dr. Nancy Kuntz discussing the importance of screening and diagnosing Duchenne muscular dystrophy. To access this and other episodes in our series, visit *NeuroFrontiers* on ReachMD.com, where you can Be Part of the Knowledge. Thanks for listening!