Muscular dystrophy diagnosis delayed almost 2.5 years in boys

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Boys show signs of Duchenne Muscular Dystrophy (DMD) for 2 ½ years before they obtain a diagnosis and disease-specific treatment, about the same length of delay children have endured for the past 20 years despite advances in genetic testing and treatment. A simple and inexpensive blood test for any boy with symptoms and signs of motor delays and abnormalities could speed up the process while pilot studies on newborn screening are conducted.

Recent University of Rochester Medical Center research published in the *Journal of Pediatrics* shows that boys who are eventually diagnosed with DMD show signs of the disease for more than a year before families bring it to the attention of a health care provider. It takes another year before these children are screened with a serum CK test - a simple and inexpensive blood test for creatine kinase, an enzyme that leaks out of damaged muscle.

"The CK test is an easily available and cheap test," said Emma Ciafaloni, M.D., associate professor of Neurology at the University of Rochester Medical Center and author of the paper. "If they get the test and the diagnosis earlier, they can start treatment earlier and access the best care in the appropriate clinics and the best available services in their school. Early diagnosis will avoid unnecessary and costly tests and numerous unnecessary referrals to the wrong specialists. Parents and maternal relatives can also seek genetic counseling before they plan to have more children."

DMD, the most common muscular dystrophy in children is a particularly devastating form of the disease that affects 1 in 3,500 boys. It is an X-linked recessive genetic disease with onset of symptoms in boys between 2 and 6 years old. It progresses rapidly, rendering patients wheelchair bound by 10 or 11 years old. Most patients die in their mid-late 20s.

The Centers for Disease Control and Prevention-funded study analyzed medical records of 453 boys born since 1982 with DMD or Becker Muscular Dystrophy in the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARnet). Of those, 156 boys had no known family history of muscular dystrophy. The first signs of the disease in those boys were seen at an average of 2 ½ years old, but the average age when families brought the signs to the attention of a health care provider was 3 ½ years old. The average age for children to receive the CK test or to see a neurologist was more than 4 ½ years old.

"We need to educate families to bring delays or abnormalities in motor skill - such as frequent falls, difficulty jumping, running or claiming stairs - to the attention of their health care providers as soon as they see them. And we need to educate pediatricians, family practitioners and all providers involved in the care of young children to recognize the early signs of DMD and to order a CK test if they see any motor delays or abnormalities," Ciafaloni said. "The sooner we start treatment, the more potential we have for delaying the disease's progression.

Source: University of Rochester Medical Center (news : web)