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Delay in age of walking can herald muscular dystrophy in boys with cognitive delays

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The timing of a toddler's first steps is an important developmental milestone, but a slight delay in walking is typically not a cause of concern by itself.

Now a duo of Johns Hopkins researchers has found that when walking and cognitive delays occur in concert, the combination could comprise the earliest of signals heralding a rare but devastating disorder known as Duchenne muscular dystrophy (DMD).

The study, published ahead of print in *The Journal of Pediatrics* and conducted by a medical student and a pediatric neurologist, reveals that delays in the onset of walking—which should occur between 9 and 16 months of age—are common among boys with DMD and often happen alongside cognitive delays. That combination, the investigators say, can give pediatricians a critical early diagnostic clue and tip them off to the presence of DMD.

"Our review of patient records shows that delayed walking along with cognitive delays represents an ominous combination that should prompt pediatricians to conduct further testing and could speed up diagnosis and treatment," says Kara Mirski, a fourth-year medical student at the Johns Hopkins University School of Medicine. "Earlier diagnosis means that we can start treating these kids sooner and greatly improve their long-term outcomes."

DMD is caused by a defective muscle protein. It is marked by

progressive loss of muscle strength and function and, eventually, inability to walk at all. In its advanced forms, the condition can also compromise the function of the heart and breathing muscles. DMD, which almost exclusively affects males, is estimated to occur in one out of 3,500 boys.

Current guidelines from the American Academy of Neurology and the Child Neurology Society do not include DMD on the suspected diagnoses list for boys with developmental delays. While neither cognitive delays nor delayed walking by themselves are necessarily caused by DMD, when the two occur in tandem they should raise the index of suspicion and seriously narrow the range of diagnostic possibilities, the team says.

"The bottom line is that any delay in walking should lead to further probing, or at least vigilant monitoring, and when late walking occurs in the context of other developmental delays, it should put DMD on every pediatrician's radar as a possible cause," says study author Tom Crawford, M.D., a pediatric neurologist and muscular dystrophy expert at the Johns Hopkins Children's Center.

Once a physician suspects DMD, a child can be screened further with a cheap and widely available test that measures the blood levels of creatinine kinase (CK), a protein released as a result of muscle damage or muscle cell death. Normal CK levels rule out DMD.

Once diagnosis is made, treatment with steroids and physical therapy can halt or slow muscle damage and help preserve mobility and function, the researchers say. In addition, because most cases of DMD are inherited, earlier diagnosis would allow families to consult a genetic counselor who can help them make informed decisions about subsequent pregnancies.

DMD can be easily missed during the infant and toddler years, even

among children with developmental delays, Crawford notes. The condition's characteristic muscle weakness does not present at such an early age, and the absence of the disease's defining symptom can easily throw off pediatricians. This is why, Crawford says, any developmental delay should prompt pediatricians to probe deeper.

In addition, while most cases of DMD stem from inherited genetic defects, some genetic mutations can arise spontaneously in families without history of the disorder. In those cases, diagnosis can be delayed even further, until a child is 5 or 6 years old, the researchers say.

For the study, the investigators examined the clinical records of 107 children with DMD referred to the Johns Hopkins Children's Center between 1989 and 2012 for diagnosis or treatment. Nearly half (42 percent) had a history of delayed walking (age 16 months or later). Toddlers who started walking late were three times as likely to have cognitive delays as those who began walking on time. The link between the time of a child's first steps and cognitive delay persisted even when investigators eliminated other factors such as the speed and severity of muscle degeneration or age of diagnosis. The study also revealed that DMD patients who started walking late were not referred for diagnostic work-up any earlier than their counterparts who started walking at what is deemed a typical age. In other words, delayed walking did not emerge as the red flag it should have been, the investigators say.

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