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Study identifies patient's priorities in treating rare muscular dystrophy

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A new study of individuals with myotonic dystrophy type 2 (DM2) - a rare form of muscular dystrophy -has helped pinpoint the symptoms of the disease that are most important to patients. These findings, published today in the journal *Neurology*, could help create a roadmap for physicians to prioritize treatment of this complex, multi-system disease.

"This study represents the first large-scale attempt to obtain direct patient input to identify the most prevalent and life-altering symptoms of myotonic dystrophy type 2," said University of Rochester Medical Center (URMC) neurologist Chad Heatwole, M.D., the lead author of the study. "This information helps us to better understand the complexities of the disease from a patient's perspective, and may ultimately prove useful in the clinical management and early diagnosis of patients with this condition."

Myotonic dystrophy has been characterized as one of the most complex and diverse genetic diseases with a constellation of symptoms ranging from fatigue, muscle stiffness, muscle weakness, cognitive impairment, depression, difficulty sleeping, impaired vision, pain, difficulty swallowing, and gastrointestinal problems. The severity and onset of these symptoms vary from patient to patient.

Although DM2 shares many similar clinical features with its sister disease, myotonic dystrophy type 1, subtle differences exist such as prominent pain and a different pattern of weakness that more commonly originates in the upper arms, legs, and back muscles. Many clinicians,

including neurologists, are unfamiliar with the disease and it is often confused with DM1 and other conditions, such as fibromyalgia. As a result, individuals with the disease often go undiagnosed for a decade or more.

Even after the disease has been successfully identified, patients and physicians are often faced with a long list of symptoms to manage. The new study - called the Patient-Reported Impact of Symptoms in Myotonic Dystrophy Type 2 or PRISM-2 - represents the first effort to understand both the prevalence of the range of symptoms that patients experience and which ones had the greatest impact on their lives.

Participants were selected from a national database of muscular dystrophy patients developed by URMC. In all, 74 individuals with DM2 completed surveys that asked them to rate 310 identified symptoms of the disease.

The researchers found that the most common symptoms weren't necessary the same ones that the patients identified as priorities. For example, while "inability to do activities," "limitations with mobility and walking," and "hip, thigh, or knee weakness" were very prevalent symptoms, DM2 patients identified "fatigue" and "pain" along with an "inability to do activities" as the elements of the disease that had the greatest impact on their daily lives.

"These findings demonstrate that the symptoms that are the most prevalent in this population are not always the ones that are most important to the patient," said Heatwole. "This is an important distinction and understanding which symptoms matter most to patients will help physicians navigate the myriad of treatment options and develop strategies that reduce the burden of the disease."