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New research sheds light on childhood neuromuscular disease

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A study by scientists at the Motor Neuron Center at Columbia University Medical Center suggests that spinal muscular atrophy (SMA), a genetic neuromuscular disease in infants and children, results primarily from problems in the motor circuits that coordinate muscle movement. Previously, researchers thought that motor neurons or muscle cells were responsible.

In a second study, researchers at the Motor Neuron Center identified the molecular pathway in SMA that leads to problems with motor function. Findings from the studies could lead to therapies for the debilitating and often fatal neuromuscular disease.

"To our knowledge, this is the first clear demonstration that defects in the function of a neuronal circuit are the cause of a neurological disease," Dr. Brian McCabe, assistant professor of pathology and cell biology, said about the first study.

Both studies were published online Oct. 11 in the journal *Cell*.

SMA is a hereditary neuromuscular disease characterized by muscle atrophy and weakness. There is no treatment for SMA, which is estimated to affect as many as 10,000 to 25,000 children and adults in the United States and is the leading genetic cause of death in infants.

Based on the findings of McCabe and his colleagues, the SMA Clinical Research Center at CUMC launched a clinical trial last July of a

potassium channel blocker called dalfampridine for the treatment of patients with SMA. The drug is currently marketed under the brand name Ampyra for multiple sclerosis. "This drug is unlikely to be a cure for SMA, but we hope it will benefit patient symptoms," McCabe said.

Provided by Columbia University