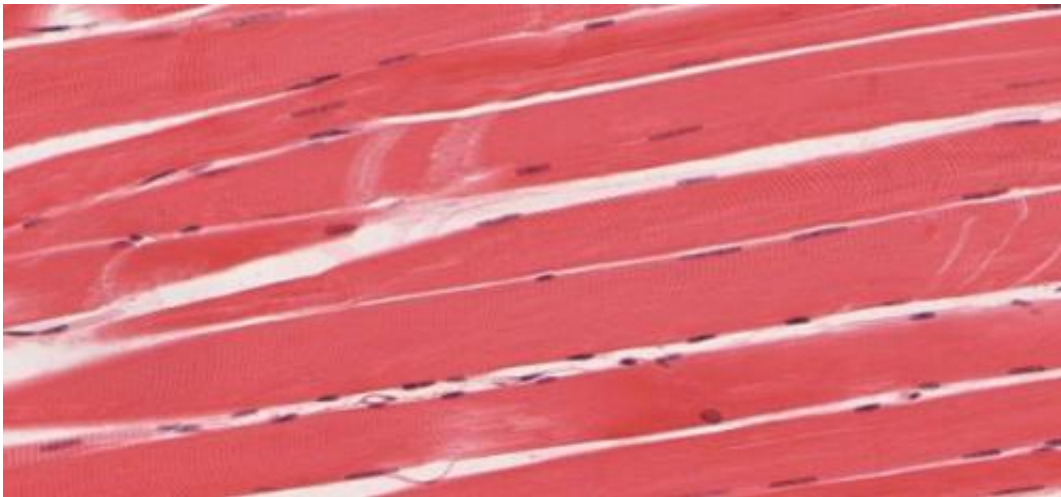


Muscle weakness studies suggest possible therapeutic strategies

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Skeletal muscle tissue. Credit: University of Michigan Medical School

A recently published study by a University of Colorado School of Medicine researcher and her colleagues suggests potential therapies for central core disease, a condition that can delay development of motor skills such as sitting, crawling and walking in affected infants.

Central core disease is most often caused by a mutation in the gene that makes a protein called ryanodine receptor type 1 (RyR1). RyR1 belongs to a family of proteins that create channels for the controlled release of calcium ions from stores within cells.

For [muscle cells](#) to contract, calcium ions must be released from these internal stores at the same time as potassium ions leave the cells. To relax the muscle cells, calcium ions are pumped back into the internal stores and potassium ions are taken back into the cell. Previous studies have established a role for RyR1 in the contraction of [skeletal muscle](#), but the precise molecular details are not clear.

In an article published this month in *eLife*, Lee Niswander, PhD, professor of cell and developmental biology and pediatrics, lead author M. Gartz Hanson, PhD, and colleagues studied mice that had symptoms of central core disease due to a mutation in the RyR1 gene.

The [muscle weakness](#) in these mice was caused by defects that hindered the release of [calcium ions](#) from internal stores and resulted in leakage of potassium ions from the muscle cells. The experiments reveal that a high-potassium diet alleviates the symptoms of disease in the mice by increasing the amount of potassium surrounding the muscle cells. Treatment with an existing drug called glibenclamide also reversed the disease symptoms by reducing the leakage of [potassium ions](#) from the cells.

The authors found several genes involved in controlling potassium ion levels in cells that could act as indicators of the presence of the disease. These findings suggest that therapies targeting the leak of potassium ion levels in muscle cells could minimize [muscle damage](#) in patients with central core disease.

Provided by University of Colorado Denver

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