Enzyme replacement therapy shows promising results in X-linked myotubular myopathy

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A collaborative research team including a Medical College of Wisconsin (MCW) pediatric neuropathologist successfully mitigated some of the effects of a muscular disease by using a new targeted enzyme replacement therapy strategy from 4s3 Bioscience.

The findings are published in the January edition of *Human and* Molecular Genetics.

X-linked myotubular myopathy (XLMTM) is a severe muscle disease caused by an absence of a protein called myotubularin. There is currently no treatment for this disorder, and most patients die in infancy or childhood. The overall incidence of myotubular myopathy is 1 in 50,000 live male births.

Michael W. Lawlor, M.D., Ph.D., assistant professor of pathology at MCW, researcher at the Children's Hospital of Wisconsin Research Institute, and director of the pediatric pathology neuromuscular laboratory in MCW's division of pediatric pathology, coordinated a study at Boston Children's Hospital and MCW that used targeted enzyme replacement therapy to deliver myotubularin to muscles of mice with XLMTM. After two weeks of treatment, the mice showed marked improvement in muscle function and pathology.

"These promising findings suggest that even low levels of myotubularin

protein replacement can not only improve weakness in patients, but also at least partially reverse the structural abnormalities seen in XLMTM," said Dr. Lawlor. "The next step is to determine appropriate dosage, and toxicity, before we venture into human trials," he continued.

More information:
http://hmg.oxfordjournals.org/content/early/2013/01/09/hmg.ddt003.full.pdf+html

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