Study holds hope of a treatment for deadly genetic disease, MPS IIIB

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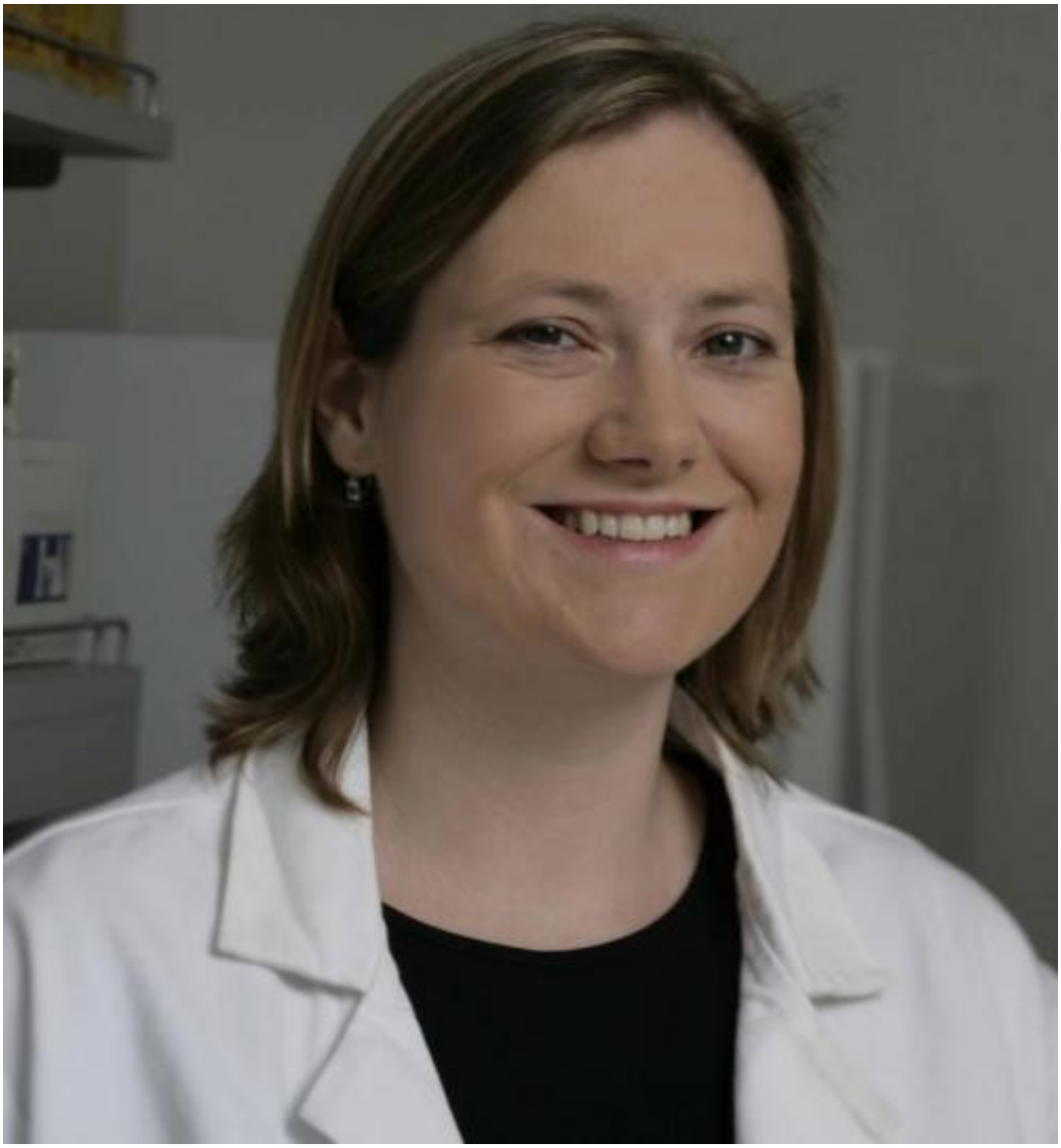

Dr. Patricia I. Dickson is the director of the LA BioMed MPS Research Lab where a new study holds hope for treating patients with MPSIIIB. Credit: LA

MPS IIIB is a devastating and currently untreatable disease that causes progressive damage to the brain, leading to profound intellectual disability, dementia and death—often before reaching adulthood.

Officially known as mucopolysaccharidosis type IIIB or Sanfilippo Syndrome type B, the disease causes the accumulation of waste products in the cells, leading to progressive damage to the brain. Patients with MPS IIIB lack a vital enzyme that is needed to break down long chains of sugars, known as mucopolysaccharides, leading these to accumulate in the cells.

Researchers reported today in the journal, *Proceedings of the National Academy of Sciences* of the United States of America, that they have found a way to replace the missing enzyme in the brains of disease models, opening the door to a potential treatment for MPS IIIB.

"Enzyme replacement therapy has been very successful in treating other forms of MPS but not MPS IIIB because the blood-brain barrier blocked delivery of the medication to the brain," said Patricia I. Dickson, MD, a Los Angeles Biomedical Research Institute (LA BioMed) lead researcher and one of the authors of the study. "By injecting a modified enzyme into the brain's left ventricle, we have found a way to bypass that barrier and deliver the needed enzyme to the brain. While more study is needed, this research holds great promise for the treatment of MPS IIIB."

Researchers at LA BioMed, the University of California, Los Angeles and BioMarin Pharmaceutical, Inc. administered a modified form of the missing enzyme, NAGLU, through the left ventricle of the brain. They

found the modified enzyme was "taken up avidly" by cells in both the brain and the liver. The researchers reported that the modified enzyme reduced the pathological accumulation of heparan sulfate, a mucopolysaccharide, and other metabolites to normal or near-normal levels.

More information: Delivery of an enzyme-IGFII fusion protein to the mouse brain is therapeutic for mucopolysaccharidosis type IIIB, *PNAS*, www.pnas.org/cgi/doi/10.1073/pnas.1416660111

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