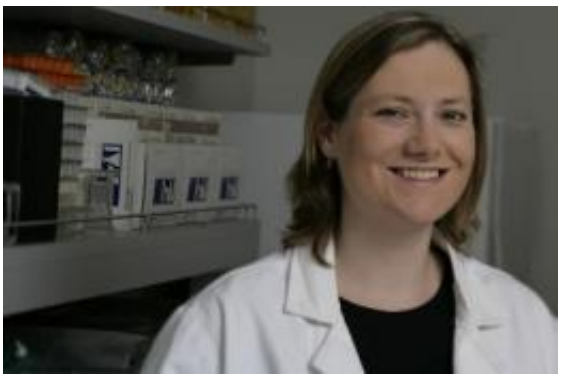
Research supports newborn screening and early treatment for rare genetic disorder, MPS I

December 1 2010



Patricia Dickson, MD, is an LA BioMed principal investigator and co-author of study supporting newborn screening and early treatment for rare genetic disorder, MPS I. Credit: LA BioMed

In a study that supports the need for newborn screening and early treatment for a rare genetic disorder, researchers at Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center (LA BioMed) and Iowa State University found enzyme replacement therapy beginning at birth eliminated almost all of the symptoms associated with mucopolysaccharidosis type I, or MPS I, in animal models.

The study, which was published today in the journal, Science Translational Medicine, is the first to show that early treatment with

enzyme replacement therapy for MPS I can significantly lessen disease symptoms in the heart, bones and brain. The researchers also found that treating at a higher dose, when begun at birth, significantly improved the response to enzyme replacement therapy, and that direct treatment into the spinal fluid resulted in a significant improvements in brain pathology.

"People with MPS I currently suffer from disease in the heart, bones, and brain, despite state-of-the-art care. Our research shows that with early intervention, these problems can be almost completely prevented," said Patricia Dickson, MD, a LA BioMed principal investigator and co-author of the study. "We found that treatment from birth with enzyme replacement therapy significantly improved outcomes, a finding that should be a huge boost to efforts to include MPS I in the diagnostic screening already performed on every newborn in the U.S."

MPS I is an inherited genetic disorder that affects many body systems. It is caused by a defect in the gene that makes an enzyme called alpha-L-iduronidase. Because of this defect, cells either produce the enzyme in low amounts or cannot produce it at all. The enzyme is needed to break down substances called "glycosaminoglycans," which are by-products of chemical reactions in the body's cells. If glycosaminoglycans are not broken down, they build up in the cell, eventually leading to cell, tissue and organ damage.

Currently, there is no cure for these disorders. But enzyme replacement therapy first developed at LA BioMed is able to give the missing enzyme back to the patient through an injection. In the study published today, the use of the alpha-L-iduronidase enzyme at birth in animal models with MPS I prevented nearly every aspect of MPS I disease, including in the brain. Previous studies performed on older patients were only able to incompletely reduce MPS I symptoms with enzyme replacement therapy.

The early treatment prevented the build-up of lysosomal substances in the liver, spleen, lungs, kidneys and heart, suggesting that enzyme replacement therapy could potentially eliminate the symptoms of MPS I in babies before the symptoms start.

"Most of our patients are diagnosed with MPS I only after they begin to manifest symptoms, so enzyme replacement therapy begins after they've already suffered damage from the disorder," said Dr. Dickson. "A test for identifying children with MPS I at birth already exists, and it will soon be possible to add the test to the newborn screening performed in hospitals around the country to identify other genetic disorders. While MPS I in itself is rare, our study implies that early treatment may have substantial benefit for the class of problems called lysosomal storage disorders, which – taken together – occur in one out of every 5,000 babies born in the U.S."

Provided by UCLA Medical Center