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# Gene therapy slows progression of fatal neurodegenerative disease in children

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Gene therapy to replace the faulty CLN2 gene, which causes a neurodegenerative disease that is fatal by age 8-12 years, was able to slow significantly the rate of neurologic decline in treated children, according to a paper published online ahead of print in the May 2008 issue (Vol. 19 No. 5) of *Human Gene Therapy*, a peer-reviewed journal published by Mary Ann Liebert, Inc.

Late Infantile Neuronal Ceroid Lipofuscinosis (LINCL) is an autosomal recessive genetic disorder that causes degeneration of the central nervous system. It is a form of Batten disease, a group of lysosomal storage disease in which a lipofuscin-like material is not broken down and accumulates in neurons, causing cognitive impairment, visual failure, seizures, and progressive deterioration of motor function.

Ronald Crystal and colleagues from Weill Cornell Medical College (New York, NY), describe a study conducted in 10 children with LINCL who received gene therapy to replace the defective CLN2 gene via administration of human CLN2 carried in an adeno-associated virus (AAV).

In the paper entitled “Treatment of Late Infantile Neuronal Ceroid Lipofuscinosis with CNS Administration of a Serotype 2 Adeno-associated virus expressing the CLN2 cDNA,” the authors report that over an 18-month period, assessment using a neurologic rating scale demonstrated significant slowing of disease progression in the treated, compared to the untreated children. On the basis of these findings, the

authors proposed that additional studies to assess the safety and efficacy of AAV-mediated gene therapy for LINCL be pursued.

Although the treatment was associated with some serious adverse events in some patients, these were not unequivocally attributable to the gene therapy vector.

“This clinical trial is an important step toward the development of treatments for this group of underserved inherited neurodegenerative disorders,” says James M. Wilson, MD, PhD, Editor-in-Chief and Head of the Gene Therapy Program, Department of Pathology and Laboratory Medicine, at the University of Pennsylvania School of Medicine, in Philadelphia.

The paper is available free online at www.liebertpub.com/hum .

Source: Mary Ann Liebert, Inc./Genetic Engineering News