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Promising therapy for fatal genetic diseases in children nears human trials

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Researchers at University of Massachusetts Medical School and Auburn University College of Veterinary Medicine are nearing human clinical trials on a genetic therapy for two rare neurological diseases that are fatal to children.

The scientists are seeking approval from the U.S. Food and Drug Administration (FDA), to test a gene therapy treatment for Tay-Sachs and Sandhoff diseases, genetic disorders in a category known as lysosomal storage diseases.

Tay-Sachs and Sandhoff are inherited neurologic diseases that occur when genetic mutations prevent cells from producing enzymes needed to break down and recycle materials. Without these enzymes, the materials accumulate to toxic levels, slowly destroying the nervous system. The researchers are working on a gene therapy to correct the enzyme deficiency using adeno-associated virus, or AAV, vectors.

The average life expectancy for children with infantile Tay-Sachs or Sandhoff disease is only 3 to 5 years. There is currently no treatment. The gene therapy in development has shown promise in animal models of these diseases by extending lifespans by up to four times those of untreated animals.

"The proof-of-concept studies in affected animals are compelling, and the FDA provided a clear path of remaining experiments needed to seek approval for human clinical trials," said Douglas R. Martin, a professor

at Auburn University's College of Veterinary Medicine. "We now need the funding to complete the studies."

The animal phase of toxicity studies necessary to demonstrate the safety of the gene therapy for Tay-Sachs and Sandhoff diseases has been completed with the support of the National Tay-Sachs & Allied Disease Association and the Cure Tay-Sachs Foundation.

"Too many children with Tay-Sachs and Sandhoff have died since we started this project. The time has finally arrived to push back on these diseases," says Miguel Sena-Esteves, PhD, associate professor of neurology at UMass Medical School. "Our single-minded goal is to get a safe and potentially effective therapy to patients and their families as quickly as possible."

"Hopefully, once the news gets out that we are this close to human clinical trials, fundraising efforts will be sufficient so we can complete the IND-enabling studies and proceed to human clinical trials," said veterinarian Heather Gray-Edwards, an assistant professor at Auburn University College of Veterinary Medicine.

Additional funding of $1.2 million is being sought to complete the safety studies, fund the production of clinical grade AAV, and complete regulatory filings.

Provided by University of Massachusetts Medical School