Effective treatment for late infantile batten disease developed

February 11 2013

Batten disease is a rare, fatal genetic disorder that affects children. Currently, no effective treatment exists for the disease, which ultimately kills all who are affected. Dachshunds also suffer from Batten disease, and now researchers from the University of Missouri College of Veterinary Medicine and School of Medicine, in collaboration with BioMarin Pharmaceutical Inc., have developed a treatment for the disease that has significantly delayed the onset and progression of symptoms in the Dachshunds. The effectiveness of the treatment in the dogs has been so encouraging that plans are underway to initiate human trials of the therapy in children.

Batten disease affects the nervous system in both humans and dogs, causing progressive neural degeneration leading to loss of vision, cognitive and motor function, ability to communicate and, ultimately, death. A number of different forms of Batten disease exist. The treatment developed by MU and BioMarin researchers targets a type of the disease that first becomes evident in the late infantile stage of development. Symptoms for this type of Batten disease begin to appear in patients around the age of two.

Batten disease is caused by the absence of an important enzyme within cells of the neural system. This enzyme helps cells break down and eliminate waste proteins. Without this enzyme, cells accumulate waste and are unable to function properly. Martin Katz, professor of ophthalmology with dual appointments in the MU College of Veterinary Medicine and School of Medicine, along with MU researchers Joan

Coates, Fred Wininger, Dennis O'Brien, Gayle Johnson, Jacqueline Pearce and several postdoctoral fellows and students worked with Dachshunds affected with Batten disease in a similar way as humans. For their treatment, a therapeutic protein created by BioMarin Pharmaceuticals Inc., replaces the deficient enzyme and is directly administered into the spinal fluid once every two weeks. Untreated dogs ultimately succumbed to the disease around 10 to 11 months of age. Dogs treated with the enzyme replacement therapy showed significant delays in the onset of symptoms and survived ! substantially longer.

"This is an important step toward treating human patients with this debilitating disease," Katz said. "By introducing a replacement for the missing enzyme into the nervous system, we have been able to help the deficient cells eliminate their waste efficiently and slow the disease-related brain degeneration. We believe this treatment approach will be effective in humans as well. Based on research to date, this treatment does not appear to result in a complete cure for the disease, but it could extend the lives and improve the quality of life for those with this form of Batten disease."

"The researchers at MU have painstakingly characterized Late-Infantile Batten disease in these dogs, and their results indicate a striking similarity in the progression of the disease among dogs and humans," said Charles O'Neill, vice president of pharmacological sciences at BioMarin. "Treatment of the dogs with BioMarin's enzyme replacement therapy has characterized its safety and efficacy, and will enable accelerated clinical development of this potential treatment for this devastating disease."

Provided by University of Missouri-Columbia