MedicalXpress

# Gene therapy treatment extends lives of mice with fatal disease

July 16 2012

A team of University of Missouri researchers has found that introducing a missing gene into the central nervous system could help extend the lives of patients with Spinal Muscular Atrophy (SMA) – the leading genetic cause of infantile death in the world.

SMA is a rare genetic disease that is inherited by one in 6,000 children who often die young because there is no cure. Children who inherit SMA are missing a gene that produces a protein which directs nerves in the spine to give commands to muscles.

The MU team, led by Christian Lorson, professor in the Department of Veterinary Pathobiology and the Department of Molecular Microbiology and Immunology, introduced the missing gene into mice born with SMA through two different methods: intravenously and directly into the mice's central nervous systems. While both methods were effective in extending the lives of the mice, Lorson found that introducing the missing gene directly into the central nervous system extended the lives of the mice longer.

"Typically, mice born with SMA only live five or six days, but by introducing the missing SMN gene into the mice's central nervous systems, we were able to extend their lives 10-25 days longer than SMA mice who go untreated," said Lorson, who works in the MU Bond Life Sciences Center and the College of Veterinary Medicine. "While this system is still not perfect, what our study did show is that the direct administration of the missing gene into the central nervous system

provides some degree of rescue and a profound extension of survival."

There are several different types of SMA that appear in humans, depending on the age that symptoms begin to appear. Lorson believes that introducing the missing gene through the central nervous system is a way to potentially treat humans regardless of what SMA type they have.

"This is a treatment method that is very close to being a reality for human patients," Lorson said. "Clinical trials of SMA treatment using gene therapy are likely to begin in next 12-18 months, barring any unforeseen problems."

**More information:** The study, "Direct central nervous system delivery provides enhanced protection following vector mediated gene replacement in a severe model of Spinal Muscular Atrophy", was published in *Biochemical and Biophysical Research Communications*.

Provided by University of Missouri-Columbia