



## FOR IMMEDIATE RELEASE

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### **\$3.5 MILLION NIH GRANT ADVANCES BID TO CURE TAY-SACHS DISEASE**

#### *Families' donations crucial to start-up of research consortium*

Boston, Mass. (August 28, 2009) – The National Institutes of Health has awarded a \$3.5-million grant to the Tay-Sachs Gene Therapy Consortium to continue research that may halt the fatal genetic disorder.

The NIH award was eagerly awaited by Tay-Sachs families and their supporters, who raised nearly \$600,000 to assemble the international consortium of experts and help maintain its research agenda while scientists worked to secure federal funding. The NIH grant will help advance an experimental gene therapy for Tay-Sachs and Sandhoff diseases from animal tests to human clinical trials.

The consortium includes Douglas R. Martin, Ph.D., Auburn University; Thomas N. Seyfried, Ph.D., Boston College; Timothy M. Cox, M.D. and Begoña Cachón-González, Ph.D., University of Cambridge (UK); Florian S. Eichler, MD, Massachusetts General Hospital and Harvard Medical School; and Miguel Sena-Esteves, Ph.D., University of Massachusetts Medical School. All have considerable experience and proven track records in the fields of gene therapy and this class of diseases.

“The consortium has been held together with funds raised by people affected by this terrible disease,” said Thomas Seyfried, Ph.D. “The NIH grant is critical to our work and also for these families, who have lost children, who have a vested interest in this research.”

“This is a tremendous achievement,” said Susan Kahn, executive director of the Boston-based National Tay-Sachs and Allied Diseases Association (NTSAD), which made establishing the consortium a top research priority. “While we know much work lies ahead, the potential success of this gene therapy effort gives hope to our member families and may one day go beyond Tay-Sachs to other diseases that affect the brain.”

## About Tay-Sachs and Sandhoff Disease

Tay-Sachs is a fatal genetic disorder, historically known as a disease that affects Ashkenazi Jews. Those of Eastern European Jewish descent, Cajun, French-Canadian, and Irish descent are at higher risk for this devastating disease, although this disease can strike anyone. Approximately 25-30 individuals die from the disease annually, though genetic screening has greatly reduced deaths from Tay-Sachs. Infants can show signs of the disease as early as six months, cease meeting developmental milestones, and then begin to lose motor skills. Most children do not survive past age 5. Children affected by juvenile onset show signs after age three and quickly begin to regress physically and mentally. For adults afflicted by late onset Tay-Sachs, symptoms are often confused with mental illness or other neurodegenerative diagnoses.

Tay-Sachs and Sandhoff diseases are known collectively as GM2 gangliosidoses. Like Tay-Sachs disease, Sandhoff disease is a progressive neurological genetic disorder that is always fatal in children and can occur in all ethnic groups.

## About the Tay-Sachs Gene Therapy Consortium

The consortium was formed two years ago as scientists refined a way to deliver two human genes – known as HexA and HexB – which are deficient in Tay-Sachs and Sandhoff diseases, respectively. Injection of these genes in the brain has been shown in laboratory mice to spur production of normal enzyme at levels sufficient to correct the enzymatic deficiency throughout the entire brain of these subjects.

The consortium received grants through the National Tay-Sachs and Allied Diseases Association's Research Initiative, which raised the majority of the funds through family foundations: [Cameron and Hayden Lord Foundation](#), [Cure Tay-Sachs Foundation](#), Hoffman Family Fund, [Mathew Forbes Romer Foundation](#), Pesotchinsky Family Fund, Ungerleider Family Fund, and the NTSAD New York chapter.

## About National-Tay Sachs & Allied Diseases Association (NTSAD)

The mission of NTSAD is to lead the fight to treat and cure Tay-Sachs, Canavan and related genetic diseases and to support affected families and individuals in leading fuller lives. Founded in 1957, NTSAD was a pioneer in the development of community education about Tay-Sachs disease, carrier screening programs, and laboratory quality control programs. Today more than two million people have been screened for Tay-Sachs disease, thousands of Tay-Sachs carriers have been identified and thousands of healthy children have been born to high-risk couples. For more information, go to [www.NTSAD.org](http://www.NTSAD.org).

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