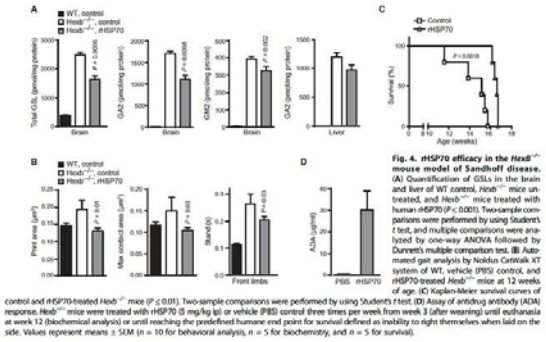
national tay-sachs & allied diseases association

RESEARCH REVIEW | August 14, 2017

FROM FACEBOOK: Orphazyme and Heat Shock Protein

There has been activity on Facebook relating to **Orphazyme** and their research on heat shock protein-based therapy which we thought we'd summarize based on a paper that was written. (Download **here**.) NTSAD's **Scientific Advisory Committee**Chair, Frances Platt, PhD is an author on the paper and is familiar with the therapy. We will be receiving an update from our contacts at Orphazyme on the latest news regarding this therapy and its potential to treat Sandhoff and related diseases. In the meantime, below is a summary of the paper that Allison Bradbury, Research Initiative Chair, wrote.



Download the full image **here**.

Summary of "Heat shock protein-based therapy as a potential candidate for treating the sphingolipidoses" *

- HexB-/- (Sandhoff) mice were treated by three weekly injections of rHSP70 (5 mg/kg) starting at 3 weeks of age.
- At endpoint, treatment was found to have significantly reduced the accumulation of storage products, including lipid gangliosides (GA2 and GM2), in the mouse brain and had a modest effect on the accumulation of the major storage metabolite GA2 in the liver (Fig. 4A).
- There was a reduction in ataxia as measured by quantitative gait analysis; however, the treatment failed to improve muscle strength as measured by bar crossing test (Fig. 4B).
- All of the rHSP70 treated mice lived beyond the age of the longest-living control animal (~16 weeks of age) but then all reached endpoint within a week of untreated animals (by ~ 17 weeks of age) (Fig. 4C).
- It was found that the treatment of HexB−/− (Sandhoff) mice with rHSP70 induced high levels of serum antidrug antibodies (Fig. 4D), which could explain the abrogated efficacy of the treatment.

* Authors: Thomas Kirkegaard, James Gray, David A. Priestman, Kerri-Lee Wallom, Jennifer Atkins, Ole Dines Olsen, Alexander Klein, Svetlana Drndarski, Nikolaj H.T. Petersen, Linda Ingemann, David A. Smith, Lauren Morris, Clause Bornæs, Signe Humle Jorgensen, Ian Williams, Anders Hinsby, Christoph Arenz, David Begley, Marja Jäätelä, Frances M. Platt

Research Grant Opportunities

The American Academy of Neurology (AAN) has launched an ambitious research program, furthering our commitment to make a profound difference in the lives of researchers and patients.

Five new awards are among the 2018 AAN Research Program opportunities, as well as three large-scale awards that mark the AAN's pledge to support all types of research across all career levels and discovery stages.

Applications are now open for the 2018 awards.

APPLY NOW
Please contact Kristin Roehl, Grants Program Manager, to learn more about the 2018 awards and ask specific questions.

Lysosomal Disease Symposium
Wednesday, October 11, 2017
held as a part of the
Individualizing Medicine Conference
October 9-10, 2017
Mayo Clinic, Rochester, MN

Join us in bringing attention to rare genetic diseases such as Lysosomal Storage Disorders and Tay-Sachs. Expert speakers will explain the integrated resources available for the diagnosis, management, and research relating to people with these disorders.

Register today and save $100 with discount code: CIM

http://individualizingmedicineconference.mayo.edu/

Awareness & Media

Sunday, October 29, 2017
12:00 p.m.

Carey Goldberg contacted NTSAD regarding the following CRISPR story for her broadcast on RadioBoston. Listen to her piece **here** as she has a thoughtful discussion about its implications.

From Radio Boston:

Scientists have successfully used CRISPR, a new technology for engineering genes, to correct a mutation that causes heart disease by editing the DNA of human embryos. The experiment correctly modified nearly two-thirds of the embryos and did not cause any other dangerous mutations in the DNA.

Our Heritage and Our Health - Ashkenazi Jewish Genetic Diseases and the Founder Effect

Learning about your heritage is a beginning. All around the world distinct ethnic groups have been identified as having increased risks for particular genetic diseases. In the Ashkenazi Jewish population, several such inherited diseases are known. These include Gaucher disease, cystic fibrosis, Canavan disease, Bloom syndrome and others. Gaucher disease is the most common Jewish genetic disorder and among Ashkenazi Jews, 1 person in 15 is a carrier for this disease, and ~1/850 has Gaucher disease. To learn more about genetic diseases among persons of Ashkenazi Jewish descent, please attend this complimentary presentation.

Location:
Temple Emanuel
7 Haggetts Pond Road
Andover MA

Guest Speaker
Gary S. Frohlich, MS, CGC
Senior Patient Education Liaison
Sanofi Genzyme

The study, led by scientist Shoukhrat Mitalipov in Oregon, could potentially improve the genetic makeup of families afflicted by genetic diseases

What are we to make over the eventual possibility of 'designer babies' and how should we begin the conversation around the ethical issues involved?

Guest
Carey Goldberg, host of WBUR's CommonHealth blog, which tweets @commonhealth.

Revisit the **Research Review** from 2016 that focused on CRISPR technology and download it **here**.

2017 Seventh Annual Day of Hope

Over 33 families across the country have joined the campaign to raise awareness and funds for research. From organized events to t-shirt campaigns, the families are on their way to having an impact on research. As of today, over $10,000 has been raised and they are just getting started!

Visit the 2017 Day of Hope page **here** to learn more and how you can support HOPE this year!

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