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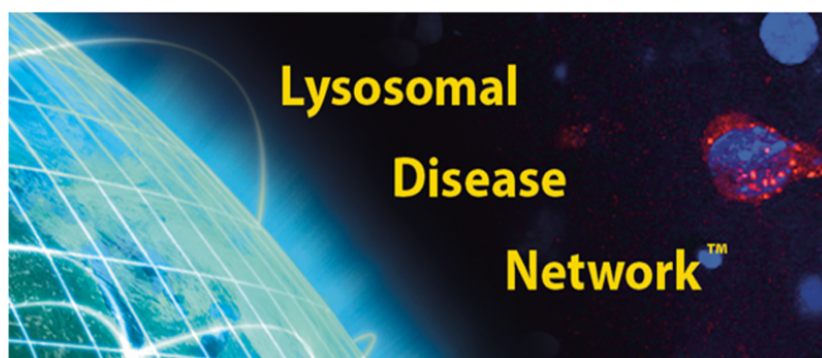


National Tay-Sachs & Allied
Diseases Association, Inc.

Friday, FEBRUARY 20, 2015
NTSAD Monthly Research Review

LYSOSOMAL DISEASE NETWORK (LDN) WORLD MEETING - FEB. 9 to FEB. 13, 2015

Highlights from the WORLD Meeting, Staci Kallish, DO



The **Lysosomal Disease Network** held their 11th annual *We're Organizing Research for Lysosomal Diseases (WORLD)* meeting last week in Orlando, Florida. **Scientific Advisory Committee (SAC)** members Bob Desnick, PhD, MD; Mark Haskins, PhD; Heather Lau, MD; and Steve Walkley, PhD were in attendance, as well as **NTSAD Board** members Staci Kallish, Jayne Gershkowitz, and Bradley Campbell, and NTSAD Executive Director, Sue Kahn. The meeting focuses on all aspects of current research for **lysosomal storage diseases (LSDs)**, from basic research, translational research currently involving animal studies, and ongoing clinical trials and brings together many participants from scientific research, healthcare, pharmaceutical industry, and patient advocacy.

More than any other year, there were a number of talks and abstracts about new therapies in development and in clinical trials for diseases that affect the central nervous system. The news that came out of the meeting was encouraging and truly offers hope for the future.

Below are summaries of a few talks that are relevant for the NTSAD community.

NATURAL HISTORY STUDIES and PATIENT FOCUSED DATA IN DRUG DEVELOPMENT

Strategic Collaboration for GM1 Research

Last week, LYSOGENE, a clinical gene therapy biotechnology company in Paris, announced a strategic collaboration with the University of Massachusetts Medical School (UMMS) and Auburn University (AU).

Through this partnership, LYSOGENE will work with Dr. Miguel Sena-Esteves from UMMS and Dr. Douglas Martin from AU to develop the pre-clinical studies needed to bring gene therapy for GM1 gangliosidosis to clinical trials.

"We are thrilled by our collaboration with University of Massachusetts Medical School and Auburn University, which constitutes a significant step towards the development of a treatment for patients affected with GM1-gangliosidosis, a severely debilitating disease. For each of these patients and their families, there is currently no option and an urgent need for a safe and effective therapy", said Karen Aiach, founding president and CEO of LYSOGENE.

Keynote Address by Laurie Muldowney, FDA

One of the keynote addresses was by Dr. Laurie Muldowney of the FDA, titled "The importance of natural history studies and patient focused data in drug development". Many NTSAD families have participated in natural history studies for **Tay-Sachs** and **Sandhoff** diseases and a new natural history study for Canavan disease was just announced. As the title implies, Dr. Muldowney discussed the importance of well-designed natural history studies for understanding ultra-rare diseases and for designing clinical trials. Natural history studies can provide insights into current standards of care even if the current standard is for supportive care only. Natural history studies can also provide information about appropriate clinical outcomes to be used in future clinical trials. Dr. Muldowney also noted that prospective studies (i.e., those studies collecting information on people in real time rather than looking back through medical records or relying on memory) yield better information because historical studies may include patients who have had different supportive care interventions, which can change rapidly and affect the course of the disease. Developing clinical trial outcome measures involves first understanding the disease, then conceptualizing treatment benefits, then developing and selecting the outcome measures to be used in a clinical trial.



NEW SUBSTRATE REDUCTION THERAPY A New Possible Treatment for Lysosomal Diseases

Dr. John Marshall from Genzyme gave a talk about a new **substrate reduction therapy** medication currently in development. This drug can cross the **blood brain barrier** to enter the brain and works by decreasing the amount of sphingolipids created in the brain so that less material can be stored. The drug was tested in mice with neurologic forms of Gaucher disease and showed that this treatment can delay onset of disease and increase lifespan of the mice by 40%. Mice also show reduced storage material. This drug is likely to be useful for other LSDs that affect the brain.



MODIFYING ENZYMES FOR TREATMENT Current Research in Japan Focuses on Sandhoff and Tay-Sachs Diseases

Dr. Keisuke Kitakaze from the University of Tokushima in Japan presented research focusing on modifying the **Hex B enzyme** deficient in **Sandhoff** disease to help it resist the protease that normally functions to break down proteins after they have fulfilled their role. This research studied several Hex B modifications which were directly injected into the brain of mice with Sandhoff disease. This is still early research looking at novel methods of delivering **enzyme replacement therapy** into the brain in rare diseases, and in this case, focusing on Tay-Sachs and Sandhoff diseases.

Read the full press release [here](#).

NTSAD on the Collaboration with Lysogene



NTSAD Executive Director, Sue Kahn, was interviewed by Rare Disease Report during the WORLD meeting in Orlando last week. See the video [here](#).

NTSAD Science Symposium

**April 16, 2015 - Hyatt Regency
Reston, VA ~ 8am-6pm**

NTSAD is hosting an invitation-only science symposium for researchers and healthcare professionals in Reston, VA on April 16, 2015 at the Hyatt Regency hotel. The co-chairs are Frances Platt, PhD, and Cynthia Tiff, MD, PhD. The meeting will cover advances and new avenues of research toward treatments for diseases affecting the CNS. Workshop topics include newborn screening, gene therapy, novel small molecules, and clinical trial readiness.

For more information please contact Sue Kahn, Executive Director at skahn@ntsad.org.



BIOMARKER STUDY AT UNIV. OF MINN. Update Presented on Ongoing Biomarker Study

Dr. Jeanine Utz, a pharmacist at the University of Minnesota, presented updated information from her ongoing **biomarker** study with Dr. Chet Whitley. They looked for differences in many chemicals in blood and cerebral spinal fluid (CSF) in children with infantile and juvenile gangliosidoses (Tay Sachs, Sandhoff, and GM1) and in children with different type of lysosomal storage diseases, the mucopolysaccharidoses (MPSs). The study included 8 children with infantile gangliosidoses and 4 with juvenile gangliosidoses and found 5 markers that were consistently elevated in the CSF of infantile patients. These chemicals are all associated with inflammation, which makes sense as inflammation is known to occur in the brains of children with these diseases. These chemicals may be useful as biomarkers of disease progression or improvement in future clinical trials.



ENZYME REPLACEMENT THERAPY Research at University of California, San Diego

Dr. Wenying Tong from the University of California in San Diego gave a talk titled "Intranasal **enzyme replacement therapy** in mice" on behalf of Dr. Jeffrey Esko. Their group is studying a method of altering enzymes to facilitate them crossing the blood brain barrier, similar to the research funded by **NTSAD's Research Initiative in 2014 by Dr. David Radin**. They are using a protein called GNeo to attach to the enzyme of interest with the hopes of aiding this enzyme entering the brain. Studying mice with MPS type I initially, they found that GNeo attached to IDUA (the enzyme deficient in MPS I) was not able to reach the brains of the mice when injected intravenously, but could reach the brain when given intranasally. This allows the enzyme to reach many parts of the brain. Their group is currently studying this technology in another type of MPS, MPS IIIA. This technology can also be applied to many LSDs that affect the brain.



NEW TECHNOLOGY FOR ENZYME DELIVERY



Penn Orphan Disease Center at University of Pennsylvania announces first grant recipients from the 2014 Million Dollar Bike Ride. NTSAD researchers Florian Eichler, MD and Annette Bley, PhD both received grants! Read the full press release [here](#).

The 2015 Ride is on May 9th, 2015 and we are currently looking for Team NTSAD riders!

Contact Sue at skahn@ntsad.org if you're interested in leading and/or riding for Team NTSAD!



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Work at Seattle-based Company, Immusoft

Another talk of interest to the NTSAD family was given by Dr. Eric Herbig of Immusoft, a company based in Seattle. His talk was focused on a new technology for enzyme delivery in which blood cells are removed from a person with a disease, genetically engineered to produce the enzyme missing in that disease, and then returned to the patient. The team is initially working with MPS I, but this technology could be applied to many different LSDs and is similar to the approach being studied by Dr. Gerhard Bauer at UC Davis. They are currently planning a Phase I study for this therapy.

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