Orphan Drug Designation Approved!

NTSAD's orphan drug designation request for a Tay-Sachs gene therapy treatment has been granted by the U.S. Food and Drug Administration (FDA). We now await results for the same request for Sandhoff disease. The European orphan drug designation applications for Tay-Sachs and Sandhoff diseases will soon be filed.

Receiving an orphan drug designation is a positive step toward our efforts to bring hope to families affected by Tay-Sachs. This designation supports drug development for rare diseases, and provides substantial benefits including the potential of funding for certain clinical studies and study-design assistance. These benefits can also be transferred to companies who may want to help make the therapy available. NTSAD, along with its funding partners*, is committed to advancing the development of gene therapy by completing pre-clinical studies and preparing for human clinical trials.

NTSAD wishes to thank Sonali Talele, MD, MBS, and Tim Cote, MD, MPH of Cote Orphan Consulting, for their *pro bono* support in filing these applications.

*Cameron & Hayden Lord Foundation, Cure Tay-Sachs Foundation, Mathew Forbes Romer Foundation, NTSAD-NY Area chapter, Sophia Pesotchinsky, and many others.

Supporting Research and Scientists of Tomorrow

In the spirit of encouraging the scientists of tomorrow, NTSAD funded the cost of sending one junior scientist to the 2013 Gordon Research Conference on Lysosomal Diseases, a prestigious scientific conference.

Karen Finn, PhD, a junior researcher from Cardiff University in the UK, attended the meeting in mid-April. She wrote:

New Grant Awarded

We are pleased to announce that a **Research Initiative** grant award was made to **Alessandra d'Azzo, PhD, of St. Jude Children's Research Hospital** for her proposal, "Studies of the molecular and biochemical bases of neurodegeneration in sialidosis." The researcher and her group have a reputation for delivering high quality research. They have frequently published in the field of lysosomal storage disorders (LSDs), including in GM-1. This is a basic research project studying lysosomal biology and function of the lysosomes in a mouse model of sialidosis. Data gained from this could be important across many lysosomal diseases and may also apply to more common neurodegene-rative diseases.

Dr. d'Azzo submitted one of 16 grants that were received in NTSAD's annual Request for Proposal process.

The Sheep are Moving to Greener Pastures!

I would like to extend my sincere gratitude to NTSAD. Due to their generous support, I had the opportunity to attend and present my latest research on Tay-Sachs disease at the 2013 Gordon Research Conference (GRC) on Lysosomal Diseases in Tuscany, Italy.

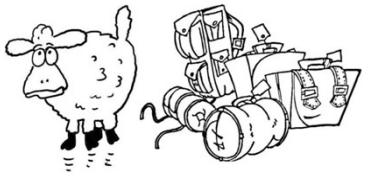
I presented a poster entitled "Inhibition of sarco/endoplasmic reticulum Ca^{2+}-ATPase (SERCA) activity by ganglioside GM2 contributes to cellular and behavioural pathophysiology in a mouse model of late onset Tay-Sachs disease." This research has identified a novel signalling defect in a cell culture-based model of Tay-Sachs disease, which we predict is an early event in disease pathogenesis.

The opportunity to network with world leading investigators in this burgeoning field and obtain valuable feedback on my research was immensely beneficial, and the exposure to such high quality science was a true learning experience. I find the highly translational and collaborative nature of this field to be incredibly motivating....It is my ambition to lead my own lysosomal disease research group in the future and it was extremely inspiring and motivating to hear about the different experiences of established scientitists and how they became successful principal investigators...Thank you.

Next Steps...

The **Tay-Sachs Gene Therapy Consortium** is diligently working on the next steps to clear the hurdles presented to the team this spring. Their commitment to the gene therapy clinical trials remains strong.

We will share news with you when we receive the next update.



The sheep are packed and ready to move to a new pasture!

The **Jacob sheep** are moving to a new pasture closer to the research conducted in Auburn, AL. Our many thanks to Joan and Fred Horak for shepherding them so lovingly over the years. Their contribution is invaluable and will make a difference in so many ways.

It will cost $35,000 for the sheep to move and for their care in the next year. If you're interested in naming a sheep for a gift of $1,000, or want to help fund their care over the next year, contact Joan Lawrence, Director of Individual Giving at **joan@ntsad.org** or (617) 277-4463, or make a gift below.

To help care for the Jacob sheep as they move to a new pasture, or to support NTSAD's Research Initiative, a gift can be made below. *Thank you for the support!*

Make a Difference
Make a Gift

"Dreamers are mocked as impractical. The truth is they are the most practical, as their innovations lead to progress and a better way of life for all of us."

~ Robin S. Sharma

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