National Tay-Sachs & Allied Diseases Association, Inc.

Friday, JULY 15, 2016

NTSAD Monthly Research Review

ANSWERS TO A FACEBOOK QUESTION

We decided to address a few questions that were posted in the NTSAD Facebook Family Support Group this week. The questions and the comments indicated there is an interest in these topics and we want to use this forum to answer them.

For the future, we always welcome questions and comments so please feel free to email or message any one in the office or on the board.

For past Research Reviews, click **here**.
For more info about the Research We Fund, click **here**.

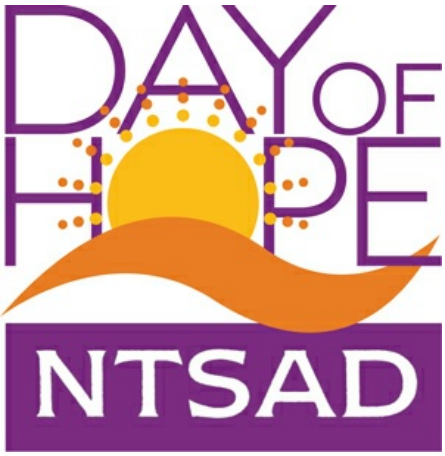
Why are some grant proposals submitted to NTSAD not funded?

NTSAD receives more than 20 research proposal applications every year. We award $300,000 to $400,000 per year in new research grants, with the maximum grant award of $80,000 for two years. So it is a very competitive process. Every proposal is reviewed by our members of our experienced and esteemed **Scientific Advisory Committee (SAC)** and other scientific experts, and with input from NTSAD's **Corporate Advisory Council**. Following are sample comments from reviewers when they don't think a project is a good fit:

- This program is very ambitious and I doubt that it is achievable in the time frame.
- Animal model suggested is not appropriate for our disease.
- Long term and still very basic. Worthy but not a priority. No immediacy to clinical practice.
- Long term as no ERT exists yet for disease so this refinement (focusing on delivery) is years away from reality.
- It is speculative whether knowledge gained through this project would lead us to translational progress.
- Translation to the human condition is likely to take many years.
- Aims 2 and 3 are dependent on the success of

Tay-Sachs Gene Therapy News

The IND-enabling studies had been delayed because of manufacturing challenges (which is not unique to our program). Those problems have been solved and the viral vectors will be delivered at the beginning of August, with the animal experiments using these vectors to being soon thereafter. These experiments will then provide data that will be used in the FDA regulatory filings. One step closer.



Rally & Raise Funds for Research September 17

The field of rare disease research is expanding. New

Aim 1. With the absence of any proof of concept with this or other fusion proteins from this investigator, this proposal comes with risk.

And, why are other grants funded?

Alternatively, here are comments from reviewers who approve the research proposals submitted.

- Addresses Clinical Trial Readiness; this could have immediate impact.
- The success of this imaginative project could lead to orally bioavailable CNS penetrant drug(s) to treat the gangliosidoses and is well worth doing.
- In light of unprecedented therapeutic efficacy of gene therapy, which has been demonstrated in animal models in recent years, there is an immediate need to establish validated biomarkers of disease progression that can also be used as secondary outcome measures in future human clinical trials.
- This investigator and this group have unsurpassed expertise in cell and gene therapy combination treatments for storage diseases. Encouraging them to apply their expertise to GM2 gangliosidosis is a major benefit.

What does NTSAD know about what research biotech companies are working on?

We are in contact with about 30 biotech companies who we periodically meet with to talk about our diseases and the potential to work with them within their research programs. These companies often insist on confidentiality in talking about their specific interests and programs, so we are not at liberty to discuss anything until they are ready to release the news.

However, when they do have something to share, we will let you know. For instance, when **Lysogene** announced their collaboration with UMass Medical and Auburn to work on developing gene therapy for GM1, we shared their press release immediately.

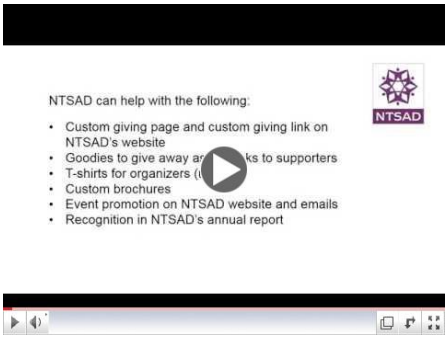
Where can I go to find the more current information on research?

Stay tuned to emails, Facebook and Twitter posts from NTSAD to learn the most current news about research.

technologies are developing. Really smart people are dedicated to hurdling the neurological roadblock so we can have treatments for these diseases. One big part of making it happen is...you guessed it...money.

All events held for **Day of Hope** go to NTSAD's Research Initiative Fund which allows NTSAD to make grants to those really smart people.

So...let's make this a big year.



2016 Day of Hope T-Shirt Campaign

Campaign is open until July 29th! Support Hope!

(74 t-shirts must be sold by July 29th before they are printed and sent out. We will open a second campaign if more are needed!)

Click **here**.

Another resource that can be interesting is the Internet News Feed on the NTSAD website. A link to that page can be found **here**.

IN-DISTRICT LOBBY DAYS

Deadline to Register is JULY 17th.

This legislation is critical to the rare disease community in that it will open more doors for rare disease research and improve access to therapies. Visit **rareadvocates.org** to learn more.

The first victory: Congress passed the 21st Century Cures Act (HR 6) last summer.

The challenge today: Get the Senate to approve the bill and include the provisions they left out for mandatory NIH/FDA funding and the OPEN ACT.

This bill needs co-sponsors and that is where you come into the picture. The RDLA and Advocacy Associates will take care of scheduling a meeting with your local legislators as long as you **register by Sunday, July 17th**.

What do you need to ask your legislators?

Mandatory Funding for the National Institutes of Health (NIH) and the Food & Drug Administration (FDA)
- Ask Senators to support robust funding
- The NIH supports basic and translational research, which is vital for laying the groundwork to understand diseases.
- The FDA plays a key role in reviewing applications for rare disease therapies while upholding high standards for safety and efficacy.

21st Century Cures Act
This act contains a wide variety of provisions critical to our community including:

- Advancing Targeted Therapies for Rare Diseases
- Advancing Neurological Diseases Act of 2015
- Advancing Hope Act of 2015
- Legislation Supporting Precision Medicine Initiative
- Patient Focused Impact Assessment Act
- NIH and FDA increase funding
- Orphan Product Extensions Now, Accelerating Cures & Treatments Act (OPEN Act) - Repurposing Incentive.

Read more details about the legislation **here**.

A HERD OF HOPE.

The Jacob Sheep are unique. The fact that they have Tay-Sachs makes them invaluable to researchers like the Tay-Sachs Gene Therapy Consortium group. However, while we need them, they need us to help feed them and care for their needs!

To see what's new with the herd and the opportunity to make a gift to support their care, follow them on their **Facebook page here**. Help them like they have helped us.

Are you on Facebook and Twitter?

Like **NTSAD's Facebook page** and follow **@susanrkahn** for new developments in therapies, science, genetic testing, patient advocacy, and more.

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