



## Developing a Rapid Screening Method to Identify Potential Drugs for Metachromatic Leukodystrophy

Dr. Gustavo Maegawa, from the Johns Hopkins University School of Medicine, recently completed a two-year [Research Initiative](#) grant awarded in 2010 to develop a quick and effective method to screen potential therapeutic drugs for Metachromatic Leukodystrophy (MLD), a lysosomal disease related to Tay-Sachs. Like Tay-Sachs, it can be acute in children or mild in adults.

Small molecules may have the potential to cross the blood brain barrier and increase enzyme activity. This project developed a method to quickly screen known drugs for possible therapeutic benefit in MLD. Here is a summary of the results:

- Developed a method to quickly screen many known drugs in skin cells affected by MLD.
- The researchers are also working on creating nerve cells from these skin cells (called induced-neuronal, or iN, cells) to study the chemicals in nerves.
- The group then used this technology to create iN cells from several other lysosomal storage diseases, including neuronopathic Gaucher disease and GM2 (Tay-Sachs and Sandhoff diseases).
- Both the iN cells and the ability to screen for small chemicals affecting enzyme activity can be easily applied to many diseases, including Tay-Sachs, Sandhoff, Canavan and more!

Click [here](#) to download the full report and [here](#) to review a publication based on this project.

## For Researchers ...

The Center for Orphan Disease Research and Therapy at University of Pennsylvania is now soliciting [grant applications](#).

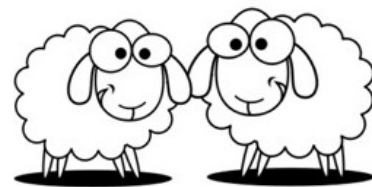
## You're all invited to ...

[FDA Patient Network Annual Meeting](#)

*Demystifying the FDA: An Exploration of Drug Development* in Washington, D.C. on September 10th.

More information is available on [the FDA web site](#).

## Sheep on the Move...



The [Jacob sheep](#) have moved 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.

NTSAD has awarded a grant to Auburn for \$39,000, which

## Baa baa Tay-Sachs sheep, have you any hope?

Dr. Doug Martin and the Tay-Sachs Gene Therapy Consortium continue to yield promising results in sheep treated with gene therapy. They just submitted their six month progress report for year 3 of the sheep studies. The highlights include:

- Jacob's sheep with Tay-Sachs disease continue to do very well after treatment with gene therapy. The oldest sheep are now around 15 months old (sheep must be put to sleep around 9 months of age without treatment).
- The sheep have had no side effects from the treatments and show HexA enzyme distributed throughout the brain.
- These sheep will continue to be followed to see how long they will benefit from the therapy.

### Stay Connected!



is the cost 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](mailto:joan@ntsad.org) or (617) 277-4463, or make a gift below.



### Day of Hope

Saturday, September 21st

Support NTSAD's Third Annual [Day of Hope](#) and Research Initiative with a gift today.

*Thank you for the support!*

Make a Difference  
Make a Gift

man can not discover new oceans unless he has the courage to  
lose sight of the shore. andre gide

#### **national tay-sachs & allied diseases association**

susan kahn, executive director ([skahn@ntsad.org](mailto:skahn@ntsad.org))

joan lawrence, director of donor relations and individual giving ([joan@ntsad.org](mailto:joan@ntsad.org))

kim kubilus, family services director ([kim@ntsad.org](mailto:kim@ntsad.org))

diana pangonis, communications manager ([diana@ntsad.org](mailto:diana@ntsad.org))

ingrid miller, office manager ([ingrid@ntsad.org](mailto:ingrid@ntsad.org))

[www.NTSAD.org](http://www.NTSAD.org)

2001 beacon street

suite 204

boston, ma 02135

(617) 277-4463

(800) 906-8723

[Forward this email](#)



This email was sent to [diana@ntsad.org](mailto:diana@ntsad.org) by [skahn@ntsad.org](mailto:skahn@ntsad.org) |  
[Update Profile/Email Address](#) | Instant removal with [SafeUnsubscribe™](#) | [Privacy Policy](#).

National Tav-Sachs & Allied Diseases Association | 2001 Beacon Street, #204 | Boston | MA | 02135