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Research Update – December 2009

This has been a very exciting year – despite the overall poor economy, the Cure Tay-Sachs Foundation and the Tay-Sachs research community in general had a wonderful financial year. The CTSF ended 2009 with just over \$438,000 in donations received, we topped the \$1 million mark in donations back in October, and most importantly our primary research group (the Tay-Sachs Gene Therapy Consortium) won a four year, \$3.5 million NIH research grant. Your donations have created lots of momentum in the Tay-Sachs research world – and we cannot thank you enough!!

The Tay-Sachs Gene Therapy Consortium (<http://www.tsgtconsortium.com/>) remains the most promising research currently underway to stop the degenerative nature of Tay-Sachs disease. We have invested \$300,000 in support of this group's research and helped them win the \$3.5 million NIH grant in August 2009.

The most recent update highlighted the following items:

- The team has begun receiving NIH funds and studies are well under way. The treated GM2 mice – and I quote – “showing exceptionally positive results that meet or exceed our pre-defined success criteria”!!
- The key research objective for 2010 – and a critical step to advance to human clinical trials – is effectively treating an infected large animal. In our case a colony of GM2 cats has been invested in over the past two years and is paying dividends today. There have been 13 kittens born in the past month and eight more litters expected in the next four weeks. This is very exciting news for our cause!! We have declared 2010 to be “**The Year of the Cat**”. Even the Chinese calendar supports us as 2010 is the year of the Tiger!!
- In 2009 when available GM2 cats were very scarce, the few cats that could be treated showed a huge immune response to the AAV vectors. The remedy was to develop a vector from cat genes. This new vectors has been created and demonstrated the ability to generate large amounts of Hex A in cultured cells.



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- The Natural History study has received 145 responses from Tay-Sachs families. The data is being entered into a database. Of the total responses 62% are from infantile Tay-Sachs families and preliminary data is proving to be very helpful. They do still need more replies to create a statistically reliable database.

Additionally an imaging study (MRI) has been started to study the metabolic changes in the central nervous system (CNS) of late on-set Tay-Sachs (LOTS). There is also a study being done to study existing MRI results of infantile Tay-Sachs children. The team needs 12 MRI's but to date has only collected five. If you can help with MRI results from your Tay-Sachs child – we could use your help.

The new imaging technology and a developed clinical scoring systems will be used to match the disease progression in both LOTS and infantile patients and to validate biochemical markers. This is the kind of data needed to accurately gauge the impact of future gene therapy treatments. How can we know if we are helping if we don't have baseline data to compare results to?

The PYR (pyrimethamine) Chaperone Therapy clinical trials are underway!! We are still awaiting the studies first research progress update – but a number of patients are now using the drug and data is being collected.

A group of French researchers has successfully combined gene therapy and bone marrow transplant to halt the neurological degeneration in two ALD patients. The relevance here is if we can stop the neurological degeneration caused by one disease – it can create the roadmap to help other neurological diseases – like Tay-Sachs disease.

The National Tay-Sachs and Allied Disease (NTSAD) Association is requesting research funding proposals. This is the avenue we use to find project worthy of funding. As an affiliate of the NTSAD we use them as the avenue by which we connect with the research community. The NTSAD's scientific advisory board will



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vet the various research projects and recommend the most promising. We are hoping to have new, exciting research projects to fund by the end of March 2010. With the NIH taking over the funding requirements of the TSGT – we are well positioned to start some new projects. The more teams we have working on Tay-Sachs disease, the better our chance of success.

This update will be posted on the Cure Tay-Sachs website under quarterly updates. If you have any question or comments about this update I can be reached at ken.bihn@curetay-sachs.org or you can call the foundation offices at (216) 812-5855

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