Dr. Gerhard Bauer and His Work

Dr. Bauer and his team at UC Davis used the Sandhoff mouse model, which displays a disease very similar to Tay-Sachs disease, to establish proof of principle for a combined treatment approach using stem cells and gene therapy.

Stem cells are special cells that come from early development and become more specialized to make up all of the cell types in our bodies. The work is based on the idea that hematopoietic stem cells (HSCs) can be genetically engineered to contain functional copies of the Hex A and Hex B genes, so that all blood cells arising from these HSCs also contain these genes. If they could multiply and reach other parts of the body (in this case, the brain), they would then allow the gene-containing cells to produce the needed enzyme, which would lead to measurable therapeutic benefits.

In this study, mice received bone marrow (hematopoietic stem cell) transplants from other Sandhoff mice. Prior to transplant into the Sandhoff mice, the HSCs isolated from the bone marrow were genetically engineered with a lentivirus vector containing Hex A and Hex B genes.

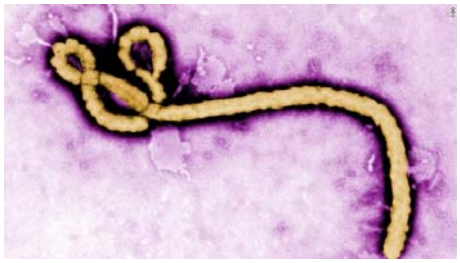
Summary of the Results

Edited by Allison Bradbury, PhD and Staci Kallish, DO

- Initial results in the mouse model are encouraging. The treated mice survived weeks longer than untreated mice, with

In this Issue

Rare Genetic Disease, Niemann Pick Type C, May Hold Answers in Preventing Ebola



A recent article in the *Wall Street Journal* and *The Scientist* addresses current research that suggests that's having **Niemann-Pick Type C disease** or being a carrier of Niemann-Pick Type C may make it harder for the Ebola virus to enter cells, offering some protection from disease.

Niemann-Pick disease refers to a group of lysosomal storage diseases called Type A, B and C. Type C is very different than Type A or B. It is characterized by inability to properly metabolize cholesterol and other lipids. There are about 500 known

many still currently alive. The treated mice do display symptoms, but their life span has clearly been extended. The living mice are still being followed and extensive analysis will be performed after their life span is exceeded.

- Dr. Bauer and his team are presently working on perfecting this transplant model. One of the major hurdles with bone marrow transplantation is achieving high levels of engraftment, or production of blood cells from the transplantated stem cells. The current engraftment level seen in mice is not as high as one would like; therefore, they are developing a new transplantation model into newborn mice that will increase the amount of engraftment. New transplants will follow in the next few weeks.

- Dr. Bauer has also received fibroblasts, or skin cells, from Tay-Sachs patients, which his group will try to genetically correct with the lentiviral vectors expressing the Hex genes. After this is accomplished, they can use these cells to make induced pluripotent stem cells (iPSCs), which are stem cells that have the ability to give rise to other cells types. iPSCs make early unspecialized stem cells from later, more specialized cells (in this case the fibroblasts). They hope to then differentiate, or develop, these stem cells into neurons. This experiment will give Dr. Bauer much needed information about the ability of this gene therapy vector to correct patient neurons.

Dr. Bauer and his team do not currently know how much enzyme will cross the blood brain barrier after HSC transplantation, however, this will be determined with the new mouse experiments.

This research project has been funded by the Cure Tay-Sachs Foundation. We send them best wishes for a successful event this evening in Cleveland, Ohio!

Who is Dr. Gerhard Bauer?

Dr. Gerhard Bauer, who spoke at the 2014 NTSAD Family Conference in Atlanta, was born in Austria, and attended college

cases worldwide but the incidence is believed to be higher due to challenges diagnosing Type C.

Read more of *The Scientist* article here. To read the *Wall Street Journal* article **online**, a subscription is needed.

NTSAD Awards Two-Year Grant to BioStrategies LC

NTSAD's 2014 Research Initiative grant was awarded to **BioStrategies LC**. It is a two year research grant to develop a central nervous system-targeted enzyme replacement therapy for GM1 gangliosidosis.

Read BioStrategies' full press release **here.**

Three More Jacob Sheep Treated with Gene Therapy

The sheep are thriving at Auburn University and continue to contribute to the research of the **Tay-Sachs Gene Therapy Consortium**.

The sheep, however, do need

and medical school in Vienna. He moved to the U.S. in the late 1980s to run the HIV research laboratory at the University of Maryland at Baltimore. A few years later he accepted a job at the Johns Hopkins University and started the development of clinical stem cell gene therapy for HIV.

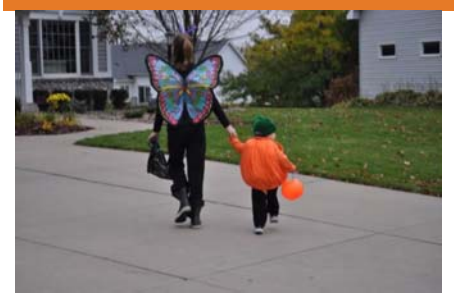
In 1995 he moved to Los Angeles to accept a position at the University of Southern California, Childrens Hospital Los Angeles (CHLA), where he was involved in the development of clinical grade stem cell gene therapy transduction and cell processing procedures. He performed all cell processing for the clinical trials of stem cell gene therapy for HIV, including the first child in the world treated with stem cell gene therapy for HIV. It was also at CHLA where he started with the development, design and implementation of academic Good Manufacturing Practice (GMP) facilities for cellular therapies.

In 2002 he was recruited to Washington University in St. Louis to build and direct a new GMP facility and center for cellular and gene therapy. The GMP facility there was soon to be named the best academic GMP facility in the United States at that time, and attracted many visitors from the US and abroad. In 2006 he was recruited back to California, to UC Davis, to be part of the new Stem Cell Program. Here he designed and now directs an improved GMP facility, which opened in February of 2010, again being called best academic GMP facility. It has gone through FDA review and sign-off, and has also received excellent reviews in the major facilities grant application to the California Institute for Regenerative Medicine (CIRM). CIRM awarded 20 million dollars to UC Davis to build the UC Davis Institute for Regenerative Cures, of which the GMP facility is an integral part. The state of the art setup he created at UC Davis allows moving laboratory research with human stem cells into clinical applications, and may therefore prove vital in developing cures and treatments for a multitude of currently incurable diseases.

your support as they graze the pastures in Auburn, Alabama.

Contact NTSAD's Director of Development, Joan Lawrence, at **joan@ntsad.org** for information on how you can support the sheep.

Read more about current and past grants funded by NTSAD on our website here.



Are you on Twitter?

Follow **@susanrkahn** for new developments in science, therapies, genetic testing, patient advocacy, and more.

Stay Connected to NTSAD!

"Everyone has some inner power that awaits discovery."

- Richard Paul Evans

national tay-sachs & allied diseases association
susan kahn, executive director (skahn@ntsad.org)
joan lawrence, director of development (joan@ntsad.org)
diana pangonis, communications manager (diana@ntsad.org)
ingrid miller, office manager (ingrid@ntsad.org)

2001 beacon street, suite 204
boston, ma 02135
(617) 277-4463
www.NTSAD.org