Jerome Lejeune FOUNDATION USA
Research Care Advocacy

JLF USA NEWSLETTER
Spring 2015

Committed to those with Down Syndrome and other genetic intellectual disabilities

> "Every patient is my brother."
>
> *-Dr. Jerome Lejeune*

We are happy to release this newsletter in honor of World Down Syndrome Day, March 21, a global awareness day set aside to raise awareness of what Down syndrome is, what it means to have Down syndrome, and how people with Down syndrome play a vital role in our lives and communities. In case the symbolism of the date is lost on some, 3/21 (March 21) was chosen because those living with Down syndrome have 3 copies of the 21st chromosome. This discovery, made by Jerome Lejeune in 1958, began the path of advocacy that has made this day's celebration possible.

Prior to Lejeune's discovery that Down syndrome is caused by an extra copy of the 21st chromosome, many thought that the disability was the fault of the parents. Some thought it was the result of a curse, and even that it was contagious, so people would cross the street when they saw a person with Down syndrome approaching because they were afraid they might catch it!

Thanks to Jerome Lejeune, we have known since 1958 that Down syndrome is not contagious, nor is it a curse. It is caused by a genetic event prior to conception. Rather than being *cursed*, families who have children with Down syndrome consider themselves blessed. Jerome Lejeune truly felt that each of his many patients was his brother or sister, and he loved them deeply. Twenty years after his death their affection for him and devotion to his memory remains strong.

We have some exciting events to report in this newsletter. We will tell you about a new memorial research fund, breif you on our participation in an event at the European Parliament, introduce you to the winner of the 2015 Sisley-Jerome Lejeune Prize, and more. Our lead article in this newsletter was written with World Down Syndrome Day in mind. It points out the clash of cultures caused by conflicting views on those living with Down syndrome.

We hope you enjoy this newsletter, and that you will share it with your friends.

JLF USA is proud to support World Down Syndrome Day!
MARCH 21, 2015

A Clash of Cultures


Dad Refuses to Give Up Newborn Son With Down Syndrome
Feb 5, 2015, 5:14 PM ET
By NICOLE PELLETIERE via GOOD MORNING AMERICA


Unique is special: Mother rejects newborn son with Down's syndrome
By Trishna Buch | February 19, 2015

In early February 2015 a story appeared that highlighted the radical difference in the way Down syndrome is perceived in various countries, or cultures, of the world. The story was so jarring that it evoked strong reactions from the public, similar to the way the Baby Gammy story had a few months earlier – that was the story of twins born to a surrogate in Thailand on behalf of an Australian couple. One of the twins, Gammy, was born with Down syndrome and was rejected by his Australian "parents," who wouldn't accept the child because of his disability.

In this newest story, a little boy named Leo was born on January 21 in Armenia to an Armenian mother and a father from New Zealand. The father described the birth by saying,

> *"This pediatrician walks out of the room with a little bundle -- that was Leo… She had his face covered up and hospital authorities wouldn't let me see him or my wife. When the doctor came out, he said 'there's a real problem with your son.'"*

The "problem" was that Leo was born with Down syndrome. For those not familiar with the story, the father claims that he wanted to keep the child and take him back to New Zealand, but his wife gave him an ultimatum: If he kept the child she would divorce him. The mother's story is a bit different so it's hard to know the true circumstances, but the details have little bearing on the point of the story: Baby Leo was caught in a conflict of cultures regarding how persons with Down syndrome are perceived, and whether they are accepted or rejected by the culture into which they are born.

The harshness of cultural attitudes toward children with Down syndrome is what captured the public's attention. Like other countries of the former Soviet block, in Armenia there is little acceptance and few support services for those with disabilities. It is typical that children born with Down syndrome are shuttled away from their families at birth and raised in poor conditions in orphanages. As the mother said,

> *"The first thing that came to my mind after the diagnosis was that I don't want my child to live in a country where certain stereotypes dominate the lives of people with DS and (they have) no opportunities at all."*

The doctors who assisted in Leo's birth reinforced this cultural prejudice when they presented Leo to his father with his face covered, and then asked the mother if she was going to keep him or not. For the father, the idea of abandoning his child was abhorrent. The mother, knowing Armenian culture, knew how difficult life for her child would be, and was willing to give up both the child AND her husband.

This story took place far from the United States, but tragically similar decisions are made here all the time. In spite of the overwhelming joy reported by families who have children with Down syndrome, a majority of the individuals conceived with Down syndrome in the U.S., the U.K., France, Australia, and many other countries, are never born because of lingering prejudicial attitudes against them.

Jerome Lejeune was renown for his compassion toward those with disabilities, and very aware of the attitudes against them held by his colleagues in the medical community. He wisely stated:

> *"Again and again we see this absolute misconception of trying to defeat a disease by eliminating the patient! It's ridiculous to stand beside a patient and solemnly say, "Who is this upstart who refuses to be cured? How dare he resist our art? Let's get rid of him!" Medicine becomes mad science when it attacks the patient instead of fighting the disease. We must always be on the patient's side, always."*

On World Down Syndrome Day, as we ponder the unique gifts those living with Down syndrome bring to the world, we might also consider another quote of Dr. Lejeune:

> *"I will go back to the Spartans, the only ones to eliminate newborns that they believed would be unable to bear arms or beget future soldiers. Sparta was the only Greek city to practice this kind of eugenics, this systematic elimination. And nothing remains of it: It has left us not a single poet, not a single musician, not even a ruin! Sparta is the only Greek city that contributed nothing to humanity! Is that a coincidence or is there a direct connection?*

Continued on page 2

A Clash of Cultures Cont.

Geneticists wonder, "Did they turn stupid because they killed their future thinkers and artists when they killed their less-than-beautiful children?"

World Down Syndrome Day is about those living with Down syndrome, but it is also a day that should serve as a mirror to reflect on how world cultures accept or reject those conceived with Down syndrome. In the U.S. and many other countries as well, the reflection we see is far from perfect. World Down Syndrome Day reminds us once again of an unbearable injustice that continues against those conceived with Down syndrome, and their families that are often torn apart when they face pressure to end a pregnancy following a prenatal diagnosis. This day sounds a call to heal and perfect the image we see so that all human cultures radiate love, acceptance, and support for those living with Down syndrome as well as other genetic differences.

To read the full story, visit www.lejeuneusa.org/culture-clash

Vander Woude Memorial Research Fund

Announcing the Thomas S. Vander Woude Memorial Fund for Down Syndrome Research

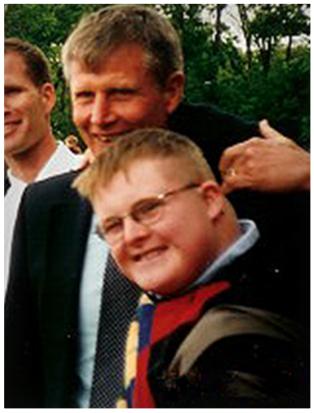
There is no greater love. . .

The Jerome Lejeune Foundation USA recently received a gift from Angels in Disguise to launch a research fund in memory of Thomas S. Vander Woude. Mr. Vander Woude was a devoted husband and the father of 7 sons, one of whom, Joseph, or "Josie," was born with Down syndrome.

On September 8, 2008, Mr. Vander Woude and Josie, then 20, were working together on the family farm in Virginia when Josie fell through the metal plate covering the farm's septic tank. Without a thought for his own safety, Mr. Vander Woude jumped into the 8-foot deep tank after his son and was holding him up above the sewage to be rescued when he himself drowned. Josie miraculously survived after recovering in the hospital from a coma and double pneumonia, but the family lost a heroic man, well-known for his deep love and generosity, and whose devotion to his family was constant to the very end.

The Jerome Lejeune Foundation is honored to have the opportunity to create this memorial to the heroic sacrifice of Thomas S. Vander Woude. We have created a perpetual fund in his memory to support research into treatments to improve the lives of those living with Down syndrome.

When many parents either refuse or question giving birth to a child who has been prenatally diagnosed with Down syndrome, Mr. Vander Woude's sacrifice shows how deep a parent's love can be for a child with a disability. There is indeed no greater love than for a father to give his own life to save his son.



To make a gift, visit www.lejeuneusa.org/vander-woude-memorial, or send checks to:

The Jerome Lejeune Foundation USA
6397 Drexel Rd
Philadelphia, PA 19151

Please write "VANDER WOUDE" on the memo line of the check.

Jerome Lejeune Foundation USA at the European Parliament

On March 10th, the Jerome Lejeune Foundation in Paris organized a major event at the European Parliament in Strasbourg, France to highlight progress being made in Down syndrome research.

Thierry de La Villejegu and Carlos Moedas with Robin Sevette, and Eleanor Laloux

U.S. Foundation president, Mark Bradford, was asked to attend the event in order to share with members of the European Parliament what the U.S. government is doing to support those living with Down syndrome.

The NIH is often criticized for the disproportionately low level of funding it provides to researchers working in Down syndrome. Funding for Down syndrome research has ranged between $18 and $20 million for the last several years, and at present averages only about $72 per person. However, in spite of poor funding, there is a strong commitment by the staff at the Eunice Kennedy Shriver National Institute for Child Health and Human Development (NICHD) to those living with Down syndrome.

The following are some of the NIH programs Mr. Bradford shared with the European Parliament that support the Down syndrome community in the U.S.:

- DS-Connect- An NIH-funded contact registry to gather un-identified medical data on those living with Down syndrome that will assist researchers in studying the disability.
- *Down Syndrome Directions*: 2014 NIH Research Plan on Down Syndrome- A blueprint for research drafted in coordination with researchers, families, and advocates that sets research goals for the future.
- The DS-Project- A private/public partnership that brings various Institutes and Centers of the NIH together with researchers and foundations to drive discoveries in Down syndrome research to treatment opportunities.
- NICHD Brain and Tissue Bank for Developmental Disorders- The NIH has designated certain sites across the U.S. to collect specimens from deceased individuals and to make them available to the research community.
- Jackson Labs Mouse Models contract- An arrangement with a major mouse lab to make mouse models of Down syndrome available to researchers.
- NICHD and National Institute on Aging jointly funded grants of up to $5 million/year for researchers to identify biomarkers (early indicators) for Alzheimer's disease in those with Down syndrome.
- A working group to establish outcome measures for evaluating the success of clinical trials on drugs to improve cognition in Down syndrome.
- The Trans-NIH Working Group on Down syndrome that consists of seven of the Institutes at the NIH that support Down syndrome research.
- They NIH Down Syndrome Consortium, a group of research and advocacy organizations that meet biennially with the Trans-NIH Working Group to discuss issues relevant to the DS community. The Consortium also helps administer DS-Connect. **The Jerome Lejeune Foundation USA** is a member of the NIH Down Syndrome Consortium.

L to R: Thierry de La Villejegu, Eleanor Laloux, Mrs. Lejeune, Mark Bradford, and Robin Sevette

There is no question that we need to continue to lobby for increased funding for research in Down syndrome, but it is important to also acknowledge the committed group of administrators at the NIH who are working as effectively and as strategically as they can with the limited resources allocated to them.

Advocacy

"Lejeune's Wall of heroes" and "Stories of Love and Hope"

In the United States and other countries of the west, we are living with an intolerable contradiction. On the one hand, we have research that shows how happy families are who have family members with Down syndrome (almost 100%), but on the other hand we are constantly faced with the horrifying reality that the majority of children who are conceived with Down syndrome are never given a chance to live. The Jerome Lejeune Foundation takes every opportunity to work toward solutions to this problem, and to emphasize the injustice also being committed against women and families.

One very effective advocacy tool is to make sure families are provided with good information following a prenatal or postnatal diagnosis. We are proud to have been a part of passing "Chloe's Law" in Pennsylvania last summer – Our state's Down Syndrome Prenatal and Postnatal Education Act – but these laws need to be supported by helpful resources that can be given to women following a prenatal or postnatal diagnosis.

What more encouraging support can we provide than to share the positive experiences families have, and the beautiful and happy faces of those who are so deeply loved by their families and friends. We have recently launched two special initiatives through our social media channels:

- **Stories of Love and Hope** is a collection of stories provided by families who are willing to share their experience of receiving either a prenatal or postnatal diagnosis of Down syndrome, or any other genetic intellectual disability.
- **Lejeune's Wall of Heroes** is a visual feast of faces and testimonies sent to us by families who want to pay tribute to their loved ones with Down syndrome, or another genetic intellectual disability.

In an article published in *Intellectual and Developmental Disabilities* in 2013 called "Receiving the Initial Down Syndrome Diagnosis: A Comparison of Prenatal and Postnatal Parent Group Experiences," the authors revealed that what gave parents the most encouragement after their diagnosis was meeting other families. One was quoted saying, "Talking to parents of kids with DS and meeting beautiful children with DS helped us to be comforted that our son would be just like any other child."

We hope that through our "Stories of Love and Hope" and "Lejeune's Wall of Heroes," parents who have received a prenatal or postnatal diagnosis will have the opportunity to meet families, at least virtually, and know that they, too, will fall in love with their child the first time they hold him or her in their arms.

The Jerome Lejeune Foundation has created a special video for World Down Syndrome Day 2015 called, "Being Different, it's Normal"

This video is a reminder of the discomfort that each of us feel when we find ourselves in a situation where we are noticeably different. It also suggests that acceptance of differences should be the only "norm" since the concept of abnormality and normality is largely a matter of context.

Individuals with Down syndrome cannot hide. As some have said, they carry their diagnosis on the face. In the spirit of World Down Syndrome Day, this video celebrates genetic diversity, and reverses roles to challenge its viewers to consider the feelings of others, and to always make them feel welcome and loved.

We invite you to enjoy the video, and to share it freely with your friends.

Watch the video at www.lejeuneusa.org/Being-Different

Sisley-Jerome Lejeune Prize Recipient, Randi Hagerman, MD, FAAP

On March 10, 2015 Randi Hagerman, MD, FAAP received the Sisley-Jerome Lejeune Prize at a ceremony in Strasbourg, France. Dr. Hagerman is an internationally recognized clinician and researcher. She is the director of the Fragile X Research and Treatment Center at the MIND Institute and holds an Endowed Chair in Fragile X Research at UC Davis. Dr. Hagerman has written over 300 peer-reviewed articles and numerous book chapters on neurodevelopmental disorders and several books on fragile X, including a *3rd Edition of Fragile X Syndrome: Diagnosis, Treatment, and Research (2002)*. She has also edited *Treatment of Neurodevelopmental Disorders: Targeting Neurobiological Mechanisms (2014)*.

Lejeune USA president, Mark Bradford, interviewed Dr. Hagerman on developments in fragile X research:

Mark: Congratulations on being selected to receive the Sisley-Jerome Lejeune Prize this year. I think you are the first researcher to receive this prize that is not working in the area of Down syndrome. Would you briefly explain what fragile X syndrome is?

Randi: Fragile X is the most common inherited cause of intellectual disability and is caused by an expansion of CGG repeats on the front end of the FMR-1 gene. In an individual with fragile X, the CGG repeat is over 200 and is frequent enough to cause the gene to be silenced. This causes a lack of a crucial protein that is critical for normal development of the brain and synaptic plasticity throughout life. Common features of fragile X, in addition to intellectual disability, are hyperactivity, poor eye contact, and unusual hand flapping. About 60% are also on the autism spectrum. In fact, this is the most common single genetic cause of autism.

Mark: When we last spoke in 2011, trials on targeted treatments were already underway and you expected results within two years. Can you give us a brief update on the progress being made in fragile X research?

Randi: Results of those initial trials have been mixed, but with some significant successes. A GABA-B agonist was investigated and showed no efficacy in adults or adolescents, but some effect on children 5 to 11 years old, including improvements in social interactions and parent stress, and some aspects of behavior. We also showed that a low dose of sertraline had some positive effect on receptive and expressive language in children 2 – 6 years old. Menocycline would also appear to have some promise for children, and we are currently looking at further treatments, including the use of ganaxolone to treat attentional problems. All targeted treatments demonstrate efficacy in mice, however efficacy may be more difficult in humans with fragile X syndrome.

Mark: I know researchers don't like to predict the future, but from the position of fragile X research today, what do you see emerging with regard to novel research targets or strategies, and also for the lives of those living with fragile X syndrome?

Randi: With the advent of targeted treatments, we know we will make a significant impact on cognitive deficits, but we can't just depend on targeted treatments. Medications need to be reinforced with intensive educational interventions. Reading is important throughout all age ranges. It should be boosted primarily because it expands one's ability to understand language, to carry out abstract reasoning, and to understand more about their environment and the world. Digital learning technologies can reduce the amount of time special education teachers work with individuals, but can never replace individual interventions. Children are starting to use digital learning aids earlier and earlier and this is a great thing. iPad apps are fabulous to boost language and reading abilities. Future treatments will consist of a combination of drug therapies and educational interventions.

To read the full interview and watch a 2011 interview with Dr. Hagerman, visit www.lejeuneusa.org/sisley-2015

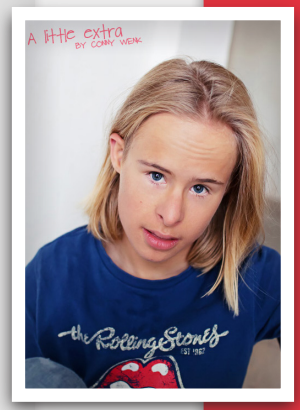
Ways YOU Can Help

Become a Donor
YOU are the Jerome Lejeune Foundation. Your gift supports our mission of research, care, and advocacy following the medical and ethical standards of Dr. Jerome Lejeune, the "father of modern genetics." Please make a tax-deductible contribution today.

Become a Volunteer
Contact us at contact@LejeuneUSA.org to join our network of volunteers and advocates.

Invite us to a Meeting
If you are a member of a local Down syndrome support group, we would love to learn more about your work and tell you about the work of the Foundation.

Spread the Word
Pass this newsletter on to families you know who might be interested in joining us in our exciting work.



Want to Learn More?
To learn more about the important research we fund, read stories about the care we provide and explore and join our network, visit our website at **www.LejeuneUSA.org**.

Giving to JLF USA
The Jerome Lejeune Foundation USA is a registered 501(c)(3) charitable organization. All contributions made to the Foundation in the U.S. are fully deductible from federal income tax.

YOU are the Jerome Lejeune Foundation. TOGETHER we can do amazing things!

Checks are welcomed at the address below, or you may go to the following link to contribute online: ***www.LejeuneUSA.org.***

Jerome Lejeune Foundation USA
6397 Drexel Road
Philadelphia, Pennsylvania 19151
267-403-2910

Pioneering Innovative Therapies

to improve the lives of those with genetic intellectual disabilities

Created in 1996 in Paris and in 2012 in the United States, the Jerome Lejeune Foundation is registered with the Internal Revenue Service as a 501(c)(3) nonprofit corporation serving those with genetic intellectual disabilities and their families...

Research in order to identify targeted treatments for patients affected by genetic intellectual disabilities. The Jerome Lejeune Foundation is the world's largest private funder of research on trisomy 21 (Down syndrome) and other disabilities. The Foundation funds projects in basic science, and also early clinical trials on drugs that offer hope that commercially available products will one day be available to improve the lives of those affected by genetic intellectual disabilities.

Care to provide specialized medical treatment and follow-up throughout the patient's life. The U.S. Foundation is involved in improving medical education and developing guidelines for care of individuals modeled after the work of the Lejeune Institute in Paris, a medical clinic which provides care to over 6,000 patients.

Advocacy for the fundamental human rights of persons with genetic intellectual disabilities. The Jerome Lejeune Foundation is committed to the inherent human dignity of all persons and the protection of life from conception to natural death.

Are You Connected?

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