



Summer 2021 Gaucher Community Newsletter

It's Summer! We hope you are faring well as life gets back to a new normal and all things good open up again.

We are excited to announce that the first Gaucher Community Patient Conference will take place in Austin, TX this November 7-9! We are thrilled to see old friends in person and make new friends as many of us will meet for the first time. This conference will help to show you that you are never alone with Gaucher disease.

There is a lot of exciting research happening right now for all three types of Gaucher disease, something we have all wished for many years. At the conference you will be able to meet with the companies who are sponsoring these trials, ask questions, and learn more. This research has the potential to finally bring treatment to neuronopathic Gaucher and more treatment options to non-neuronopathic Gaucher. Please see below for some research studies that are now open throughout the US.

As always, we encourage you to reach out to us to let us know how you are doing and what you're needs are. Enjoy summer and please take care!

Fondly,

Gaucher for Both Mother And Son

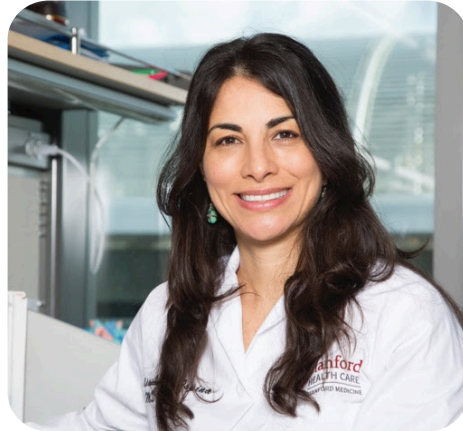
*by Melissa Steinman
GCA Board of Directors*



My name is Melissa Landau Steinman, and I'm an attorney who specializes in advertising, marketing and consumer product safety at the Venable law firm in Washington, D.C. I'm 52 years old, and I have two kids: Charlie (23), a PhD student at Columbia in NYC, and Jamie (19), a sophomore at Wesleyan in Connecticut. I was diagnosed with Gaucher disease at 29 years old, when I was five months pregnant with my oldest son Charlie. I had no symptoms at the time, but my then husband and I had asked for genetic testing after a cousin told us she had learned she was a carrier. We were shocked to learn that not only was my husband a carrier, but I had two N370S mutations and thus had Gaucher disease. We needed to decide quickly what to do regarding my pregnancy, since the baby could have Gaucher as well. Ultimately, we

Natalia Gomez-Ospina, MD, PhD

The Gaucher Providers



Dr. Natalia Gomez-Ospina is a Taube Pediatric Neurodegenerative Disease Scholar and Assistant Professor in the Department of Pediatrics, Division of Medical Genetics, and Division of Stem Cell and Regenerative Medicine at Stanford University, Lucile Packard Children's Hospital.

Born and raised in Medellin, Colombia, Dr. Gomez-Ospina is now a physician and scientist dedicated to diagnosing and treating genetic diseases. Her clinical practice focuses on lysosomal storage disorders (LSDs), the most common genetic cause of childhood neurodegeneration. She directs the enzyme replacement service at Lucile Packard Children's Hospital and the Program for Inherited Metabolic Disorders at Stanford, which aims to support the

decided not to risk an amniocentesis but to test the baby's cord blood instead. It turned out that my first-born did not have Gaucher—he's a carrier—but my second child, Jamie, does, and this article is about us.

After Charlie was born, I waited close to two years to have all the testing my geneticist recommended when we first learned I had Gaucher. I suppose I was in denial. I had blood testing during the pregnancy and things generally looked okay, but I did not have the MRIs, X-rays or other recommended tests done until I wanted to have a second baby. When those tests showed my liver and spleen were moderately enlarged, I started Cerezyme to “debulk” and make room for another pregnancy. After about six months, I got the okay to try to get pregnant again. I then went off Cerezyme to try for another baby, restarting it after the first trimester, and after a healthy pregnancy, Jamie was born.

Jamie, born in 2002, also has two N370S mutations. Raising a child with Gaucher disease when you have Gaucher yourself is a special challenge, particularly because the same genes or “genotype” (N370S-N370S) does not necessarily mean you have the same “phenotype,” i.e., that your disease will present the same way. Jamie is nonbinary, so I will use “they” pronouns in this article. So far, we both have a mild presentation of Gaucher disease. We have tracked Jamie closely since birth, first with regular blood testing, X-rays and appointments with their geneticist, and then adding MRIs once they were old enough to have them without

development of gene and cell-based therapies for LSDs.

We are grateful to the researchers and providers who dedicate their careers to serving the lysosomal storage disease (LSD) and rare disease communities. Dr. Gomez-Ospina specializes in Medical Genetics at Stanford University at the southern end of the San Francisco Bay Area and chose to work on LSDs during her training as a Medical Geneticist. Her goal to combine medicine and basic research helped her to decide how to apply her scientific skills to develop therapies for patients with genetic diseases. She works specifically on treatments that are either definitive or curative since many LSDs are amenable to the types of therapies she is interested in developing.

Dr. Gomez-Ospina is interested in all LSDs, including Gaucher, and she tries to keep up with all the therapy approaches in development. Specifically, she finds LSDs that have neurodegeneration as a feature interesting. In the laboratory, she focuses on the LSDs which are potentially addressable with hematopoietic stem cell transplantation. She is interested in the role that lysosomal dysfunction has in neurodegeneration. During her training and practice as a medical geneticist, the most challenging patients she has had the opportunity to care for have LSDs presenting with childhood-onset neurodegeneration.

sedation. Our goal has been to delay the need to start therapy as long as possible. As it turns out, now that puberty is over and Jamie has finished their growth spurt -- topping out at a healthy 6 feet 4 inches tall -- their blood levels have stabilized and other tests are looking good, so we have not yet started treatment.

I find the differences in making decisions and processing health issues as a parent vs. as a patient can be striking. As a patient with mild disease presentation and a far-too-busy work and personal schedule, I can sometimes be a bit too cavalier about my own health, delaying tests and appointments or forgetting to take my calcium and vitamin D. When it's my child, however, I tend to have a strong and even emotional response to test results that are even a little bit abnormal, and I've had to make peace with my own guilt for contributing the N370S mutation. Because of my perspective as a parent and a patient, I have made a strong commitment to contributing to the Gaucher community, which is one reason why serving on the Board of the Gaucher Community Alliance means so much to me. In the past, I've also participated in patient groups for Genzyme and Amicus and provided assistance to the National Gaucher Foundation. Moreover, as someone with milder Gaucher, I feel that I'm more able to participate in new drug trials, so I have signed up whenever possible. I was a subject in the Stage 3 FDA trials for both VPRIV and Cerdelga, and I also participated in a post-marketing (Stage 4) study on Zavesca. I've also tried to provide guidance when other moms or moms-to-be have contacted me with

Dr. Gomez-Ospina's laboratory develops therapies for LSDs combining stem cells and genome editing. Her lab championed the idea of hijacking the hematopoietic system to express proteins needed in other organs, including the brain. She established an adaptable platform for treating lysosomal enzyme deficiencies and performed first-of-its-kind preclinical studies to support the clinical development of autologous transplantation of genome-edited hematopoietic stem cells to treat patients with Mucopolysaccharidosis type I (Hurler syndrome) and Gaucher disease. Beyond delivering lysosomal enzymes, this platform has potential implications for providing many kinds of therapeutic proteins to the brain. In addition to therapy development, Dr. Gomez-Ospina led the discovery and characterization of a new infantile cholestasis syndrome caused by mutations in the bile acid receptor and participated in several multi-institutional collaborations that resulted in the discovery of multiple intellectual disability syndromes. Despite, her young career, she has been the lead author in research studies in the New England Journal of Medicine, Cell, Nature Communications, and American Journal of Medical Genetics.

There are many things Dr. Gomez-Ospina finds rewarding about working in the LSD community. Her goal is to improve individuals affected with these conditions, and having that clarity gives meaning and focus to her work. Some of her most challenging patients have LSDs, and Dr. Gomez-Ospina has learned immensely from them. She has learned not just about the clinical

questions.

But my most important role, I think, has been to model the role of a good patient to Jamie—someone who gets stuck with needles without complaint, who asks good questions at the doctor's office, who takes responsibility for their health, and who lives life to the fullest, even with a chronic disease. Jamie is a rising sophomore at Wesleyan University now, and last year, they co-founded a group for students with disabilities and chronic diseases to provide advocacy and support on campus during the pandemic (and beyond). It was a tough year for college students with chronic conditions, and I'm so proud to have raised a child who is ready to support and advocate for them. Because of the nature of our very rare disease, every Gaucher patient has to advocate for themselves on a regular basis, so if I can help Jamie to do that, and work with GCA to help the community do it too, I feel like I will have made a real contribution.

Clinical Studies



AVROBIO

**Phase 1/2 Lentiviral Vector Gene Therapy -
The GuardOne Trial of AVR-RD-02 for
Subjects With Type 1 Gaucher Disease**

aspects of their diseases but also about their lives. She sees how complex illnesses that usually manifest in a previously healthy child or person relentlessly progress and current interventions leave much to be desired.

Compared to other rare diseases, the LSDs have had some hope of treatment beginning with enzyme replacement therapy, and she's fortunate to have the opportunity to build on the fantastic work other people have done to this point. She feels this work is intellectually challenging, which is important to her.

Dr. Gomez-Ospina's lab is working on bringing genome editing to the clinic for patients with LSDs. It is the hope for truly precise and definitive therapies for these diseases. Bone marrow or hematopoietic stem cell transplantation (HSCT) has been used for decades to treat several genetic diseases whose hallmark symptom is rapid neurodegeneration. HSCT's effect is mediated in part from bone marrow-derived cells that migrate to the brain, engraft, and differentiate into microglia-like cells or central nervous system (CNS) macrophages where they can "cross-correct" affected neurons and glial cells. However, the widespread use of HSCT has been limited to a few severe LSDs because, as it is currently performed, it can cause significant morbidity.

One of the main drawbacks of HSCT is the use of allogeneic cells, which predisposes the patients to immunological complications such as graft-versus-host disease. The second is pre-

GuardOne is designed to evaluate the safety and efficacy of AVR-RD-02, an investigational gene therapy, in individuals with type 1 Gaucher disease.

To enroll, participants must be, amongst other criteria:

- Between the ages of 18 and 45 (depending on the region where you are participating) at the time of screening
- Have a confirmed case of type 1 Gaucher disease
- Have been stable on ERT for a minimum of 2 years OR have never received ERT or SRT or have not received ERT or SRT in the past 12 months

For more information, visit <https://www.avrobiogauchertrial.com/>, complete the pre-screener at <https://www.gaucherclinicaltrialus.com/> or call 1-877-330-5214.

Sites:

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Prevail Therapeutics

Phase 1/2 Clinical Trial of PR001 in Infants With Type 2 Gaucher Disease (PROVIDE)

PROVIDE is a potentially disease-modifying, single-dose gene therapy for patients with neuronopathic type 2 Gaucher disease.

transplantation conditioning that is required to deplete host hematopoietic stem cells but has significant side effects. Accordingly, to leverage the ability of hematopoietic cells to cross the blood-brain barrier (BBB) for therapeutic purposes, Dr. Gomez-Ospina's lab has been working on establishing efficient genetic modification strategies to enable autologous transplantation approaches and developing safe conditioning agents that act specifically on the brain and spare the bone marrow.

Dr. Gomez-Ospina's lab was first to demonstrate that ex vivo genome editing of human

hematopoietic stem cells could treat a model of an LSD with prominent neurodegeneration such as Mucopolysaccharidosis type 1 (MPS1).

Instead of targeting individual mutations of the affected locus, they used CCR5 as a safe harbor.

The use of a safe harbor enhances the therapeutic effect by allowing for supra-endogenous expression, constitutes a universal therapy for any given disease that is independent of mutations, and effectively establishes a platform for treating other LSDs, including Gaucher. The CCR5 platform is easily adaptable for expressing any secreted therapeutic protein and has also been used to target glucocerebrosidase expression cassettes as a curative therapy for Gaucher disease and a potential approach for Parkinson's disease with glucocerebrosidase mutations. The platform is currently being adapted to Krabbe and Progranulin deficiency.

With the amazing work on LSDs that Dr. Gomez-

For more information, visit the website [here](#).

To enroll, participants must be, amongst other criteria:

- Be 24 months or younger
- Have a confirmed case of type 2 Gaucher disease
- Bi-allelic GBA1 mutations consistent with a diagnosis of type 2 Gaucher disease confirmed by the central laboratory.
- Neurological signs and/or symptoms consistent with diagnosis of type 2 Gaucher disease

Sites:

University of Minnesota Masonic Children's Hospital

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Phone: 612-672-7013

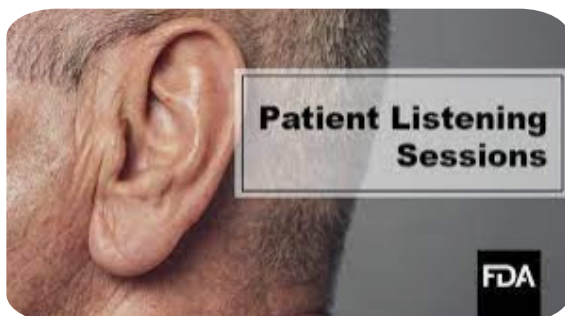
Email: Cgibson1@fairview.org

Children's Hospital of Pittsburgh

Contact: Jodi Martin sausjl@upmc.edu

Contact: Sarah Klotz klotzse@upmc.edu

FDA Listening Session



GCA Goes to the FDA

After many pandemic related delays, the neuronopathic Gaucher FDA Listening session is scheduled for this September 9th. Unfortunately it

Ospina is active in, it's hard to imagine she can fit in a personal life as well. But she is mother to a five-year-old daughter who loves to swim and bike just like her mother. When Dr. Gomez-Ospina was in graduate school, she used to do triathlons, and she continues to do all three sports recreationally to stay in shape.

Thank you, Dr. Gomez-Ospina, for keeping Gaucher patients in your line of research. We are grateful for the work you do, and we wish you the best of luck.

Gaucher Patient Advocacy Summit



The Conference Is On!!

The Gaucher Community Patient Conference for patients and their family members is confirmed! We can't wait for all of us to be together in Austin, TX.

We're still in the planning stages and would love to hear from you to understand if you are interested in coming and what your needs are to attend. Please fill out the questionnaire [here](#) to let us know your plans.

[Click HERE to complete conference survey.](#)

will be virtual, but we have no doubt that our three presenting families plus Dr. Goker-Alpan will do a fabulous job demonstrating to the regulators how many unmet needs the Type 2/3 Gaucher community faces. Stay tuned for a summary of the session in our fall newsletter.

Health Insurance Updates



Co-pay Accumulator Legislation

As we've reported in past newsletters, some national health plans throughout the US have implemented copay accumulators which prevent copayment assistance from counting toward a plan enrollee's deductible. This rule was intended to steer patients to less costly, generic medications when possible, but for Gaucher treatments, there are no generic alternatives. This financial burden then falls on the patient and ends up placing Gaucher patients at risk for medication adherence issues, including skipping refills, rationing medications, or abandoning treatment altogether. It is creating more difficult treatment access to Gaucher patients and families and putting our patient community at risk. For a complete understanding of this issue please see our previous newsletters [here](#).

Since our last newsletter, an additional five states

What the Conference Offers

The Gaucher Community Patient Conference is designed to connect, educate, and empower Gaucher patients and families by providing information and tools to best manage our disease and live a fuller life by connecting with and learning from our peers.

The conference will be two days, with the first day designed for patients only. This will establish an environment of sharing, openness and trust where participants can openly learn from one another and share their stories and tools in managing their disease. We will offer workshops led by the patient community who are experts in their fields and for the patient community.

Day two will be opened to industry sponsors, providers and researchers to discuss Gaucher research on potential treatments for non-neuronopathic and neuronopathic Gaucher disease.

We look forward to seeing you there!

Gaucher Community Patient Conference
November 7-9, 2021
Austin, TX

Gaucher Awareness Video

This is Gaucher

*For Patients, By Patients
Together we can make change*

Thank you for sending in your video footage!!

We are thrilled at how many people and families from around the world sent in video footage for the ***This is Gaucher*** video campaign. We received video uploads from Canada, Colombia, Guatemala, India, Mexico, Pakistan, Slovenia,

have joined the previous six in banning this practice. These legislative actions could not have been possible without the tireless efforts of patient advocacy groups and individual patients like yourselves who have called and are meeting with state legislatures. As of this newsletter, the list of states that have banned co-pay accumulators are: Arizona, Arkansas, Connecticut, Georgia, Illinois, Kentucky, Oklahoma, Tennessee, Virginia, West Virginia, and Puerto Rico. Bills continue to work their way through numerous other states including Pennsylvania, where GCA's Co-Founder/Co-President Aviva Rosenberg has published an op-ed asking legislators to act on the state bill. Please read the article [here](#) and consider sending in your own story to your newspapers.

Please be aware that state law covers only private insurance plans and not self-funded plans. Over 60% of beneficiaries with private insurance are covered by self-funded plans. These plans can only be governed by federal action, therefore we must keep this issue alive with our federal regulators as well.

If you have been affected by this bill, please know that we are working to reverse the legislation so that the Gaucher community can have better access to their treatments. We hear you and we are working to help.

Spain, the UK, and the US. With worldwide participation, we will be able to highlight the different types of Gaucher and the many faces, ages, ethnicities, languages, and cultures of people it affects.

Thank you for sharing your stories and experiences. Through this video we hope to raise awareness of Gaucher for easier diagnosis and empower the entire Gaucher community so no one feels alone with this disease. We are a strong patient community that is willing to support one another and we should be proud!

Save the date

The video will be released Saturday, October 2 at an online launch party (more information to come) and then will be distributed to patients and patient organizations worldwide, Gaucher treatment centers and centers of excellence, prescribing physicians, researchers and industry partners. We look forward to seeing you online to celebrate International Gaucher Awareness Day and Gaucher Awareness Month.



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It's easy! Just log onto to your normal Amazon account [here](#) and shop the way you normally do. Amazon will donate 0.5% of your purchases to the GCA.

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For Patients, By Patients
Together We Can Make Change

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