

2021 Winter Newsletter

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Winter 2021 Gaucher Community Newsletter

A Look Back on 2021

At the end of each year we ask ourselves, was it a good year? What went well and what didn't? Am I happy where I am? Can I do more? At the Gaucher Community Alliance, we ask ourselves the same questions in providing needed services for Gaucher patients and families. And in order to continually provide the right type of support we ask ourselves these questions quite often.

We are in a very interesting and exciting time for Gaucher. Gene therapy is no longer something we hear might happen in our lifetime, but it is fast becoming a reality. That means we may finally have a treatment for types 2 and 3 neuronopathic Gaucher which could save lives. It also means there may be a one-time treatment for type 1 non-neuronopathic disease, and we won't have to have biweekly infusions or take daily pills to treat our disease.

One thing we have found is that as new treatments come to market and the world around us changes, our needs change as well. The GCA strives to adapt to these changes and provide accordingly. What we have found is that patient support continues to be necessary, so we recruit volunteers and community members to help and we are planning a conference to connect us together. We've found that education and awareness will always be relevant because information comes to us quickly and we need to find the best ways to inform our community. And finally, we've found that advocacy through community grassroots efforts is crucial, because we are rare and we need to have a voice so that people listen and we don't get overlooked.

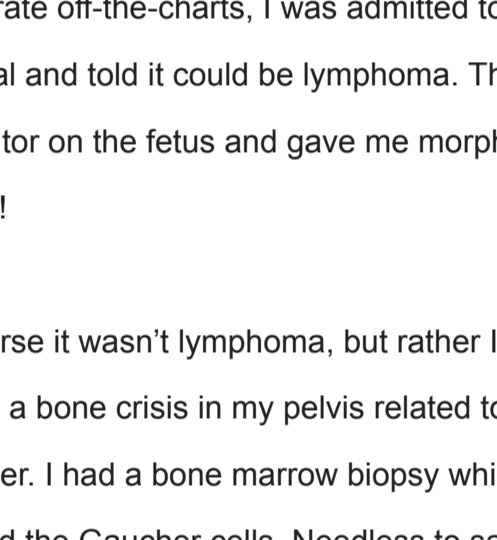
As you look forward to 2022 and want to make changes, do it with us! We would love to be in touch and work with you to better the lives of people with Gaucher. We're always here, you can reach out anytime!

Wishing you a wonderful, healthy and happy 2022!

Fondly,

Cyndi Frank & Aviva Rosenberg
Co-Founders/Co-Presidents

My Diagnostic Journey by Aviva Rosenberg GCA Co-Founder and Co-President



My diagnosis of Gaucher disease at age 27 came as a shock and a relief. I was newly pregnant with our first child and my husband and I decided to have the Ashkenazi Jewish Panel for genetic tests done since we are both of Ashkenazi Jewish ancestry. I was sitting in my law office when the geneticist at Tulane University called and said she would like me to come see her. Of course, I panicked. My first thought was that something was wrong with the baby. I was increasingly agitated and demanded that she tell me over the phone, and then she asked me if I knew I was homozygous for Gaucher disease. WHAT?!!! What is that?

Hence my entry into the lysosomal storage world began. But, what a relief that the symptoms I had my entire life finally made sense. Ever since I was a little girl I had constant bloody noses. My mother would take me to the ENT who would cauterize it and send me on my way with reminders to use saline spray to keep it moist. Some weeks I would have three or four bloody noses. As an adolescent I was diagnosed with anemia, so my pediatrician told me to take iron supplements. During law school, a routine blood test found very low platelets which resulted in a trip to the hematologist. This specialist simply explained that some people have low platelets and its no big deal.

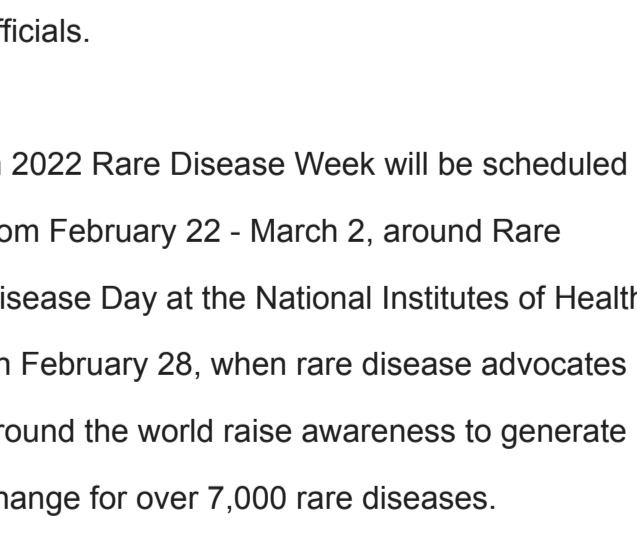
And the exhaustion. I couldn't understand how my roommates could stay out all hours of the night and then go to classes the next day. I needed to sleep—a lot!

After my diagnosis, my husband got tested and we learned he is a carrier for Gaucher. So we decided to check the fetus and fortunately, the baby was only a carrier. At 26 weeks pregnant, I could not move my left leg and the pain was incredible. I called the OB who explained it was sciatica and there was no treatment. I could not sleep, eat, bathe, or do anything. After a horrible week, I went to see an orthopedic surgeon who ordered MRIs and blood tests. Within hours, and a sed rate off-the-charts, I was admitted to the hospital and told it could be lymphoma. They put a monitor on the fetus and gave me morphine. Finally!

Of course it wasn't lymphoma, but rather I was having a bone crisis in my pelvis related to Gaucher. I had a bone marrow biopsy which showed the Gaucher cells. Needless to say, it was a very difficult pregnancy. By the time I went into labor at 38 weeks, my platelets were down to 30,000 and I had excessive bleeding during delivery, resulting in some 300 stitches. Within three months of having our beautiful son Jonah, I started on enzyme replacement therapy and things started to look up. I had a corrective surgery to remove scar tissue from the delivery and began to feel much better.

Seventeen years and two more sons later, my husband and I are enjoying a very active family life with our three boys. Our middle son Eli has Gaucher disease and is on treatment to ensure he doesn't have the complications I had as a child. Over the years I have switched to various ERTs and then enrolled in the FDA study for the oral treatment. In 2019, I transitioned from practicing law full time to founding the Gaucher Community Alliance with Cyndi Frank. It has been my honor to work with the Gaucher community and the amazing health care providers that work to keep us living our best lives.

We Want Your Video!



Gaucher Patient and Family Videos
What's a good way to raise awareness about Gaucher? By getting lots of video footage out there! To complement our This is Gaucher video project, we want to continually upload Gaucher and family videos to our YouTube Channel.

Tell us your stories: what type of Gaucher you or your family member has and how and when you were diagnosed, what treatment(s) you're on, how you're feeling, what your needs are, how your life is and how you cope, and anything else you'd like the world to know about living with Gaucher. We want to hear it!

Upload your video to any Dropbox and send us the link here. We will review and upload to our YouTube channel for you. Thanks for helping to raise awareness about Gaucher!

And make sure to subscribe to the GCA YouTube Channel to catch all the latest Gaucher videos here. We will be uploading video content as we receive it! The more content we have, the greater voice we'll have as a rare disease.

Patient Advocacy Update



The GCA continues its advocacy efforts on several fronts to give the Gaucher community a much-needed voice. We remain active with the national 'All Co-Pays Counts' coalition, continuing to press for state law prohibitions on co-pay accumulators used by insurance companies and pharmacy benefit managers to prohibit patients from the benefits of co-pay assistance funds. In 2021, Louisiana, Kentucky, North Carolina, Oklahoma, Arkansas and Connecticut joined a handful of other states in prohibiting co-pay accumulators. Michigan, Ohio and Pennsylvania have active bills. If you or a family member have experienced a co-pay accumulator related to Gaucher treatment, please notify us so that we can amplify your story to regulators.

We are also looking for patient and family experiences with newborn screening. If you live in one of the four states that offers Gaucher newborn screening – Illinois, Missouri, Kentucky and New Jersey – and you would like to share your story about the early diagnosis, please let us know. In addition, there is a pilot program in New York. The GCA would like to expand opportunities for early diagnosis of Gaucher, to shorten the diagnostic journey and decrease invasive testing on children affected symptoms. If newborn screening affecting your family, we would love to hear your story.

2022 Rare Disease Week



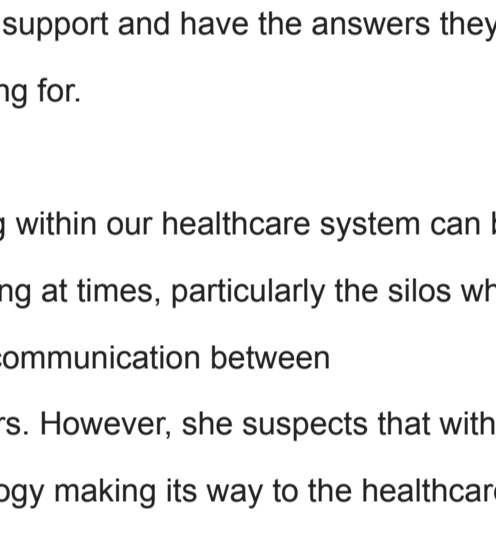
Two members of our Gaucher community will be featured at Rare Rare Disease Week on Capitol Hill in 2022!

Rare Disease Week brings together rare disease community members from all over the United States to be educated on federal legislative issues, meet other advocates, and share their unique stories with legislators. Advocacy matters so that we can bring our stories and needs to the attention of local, state, and federal government officials.

In 2022 Rare Disease Week will be scheduled from February 22 - March 2, around Rare Disease Day at the National Institutes of Health on February 28, when rare disease advocates around the world raise awareness to generate change for over 7,000 rare diseases.

Stay tuned for more information on 2022 Rare Disease Week. Let us know how you'd like to get involved!

Walla Al-Hertani, MD The Gaucher Providers



Dr. Walla Al-Hertani is a Medical Biochemical Geneticist and a Clinical Geneticist practicing at Boston Children's Hospital and an Assistant Professor of Pediatrics at Harvard Medical School. Dr. Al-Hertani received her Doctor of Medicine degree from Dalhousie University in Halifax, Nova Scotia, followed by the completion of her Clinical Genetics residency in Ottawa and a Biochemical Genetics fellowship at the Hospital for Sick Children and the University of Toronto. She is currently the Director of the Lysosomal Disorders and the Glycogen Storage Diseases Program as well as the Assistant Director of the Metabolism Program in the Division of Genetics and Genomics at Boston Children's Hospital. She is also actively involved with a number of clinical trials investigating innovative therapies for rare diseases.

Dr. Walla Al-Hertani is Director of the Lysosomal Disorders Program and the Glycogen Storage Diseases Program; Assistant Director of the Metabolism Program at Boston's Children Hospital and specialized in inborn errors of metabolism. When we asked her why she selected this line of work with lysosomal storage disorders and what part of her medical journey helped her to make this decision, she informed us that her work with patients and families with rare diseases is extremely rewarding and has been for many years. However, working with the lysosomal storage disorders community is especially rewarding.

Dr. Al-Hertani has been working with the LSD community since 2010 as a fellow. Since that time and because of how rewarding it is, she committed her career to helping patients with lysosomal storage disorders and their families to help find answers and get access to treatments. She particularly appreciates the connection and support the LSD family support groups facilitate to help patients who are newly diagnosed and their families who are struggling to deal with the new information they are receiving. She sees the relief many families feel when they are able to receive support and have the answers they were searching for.

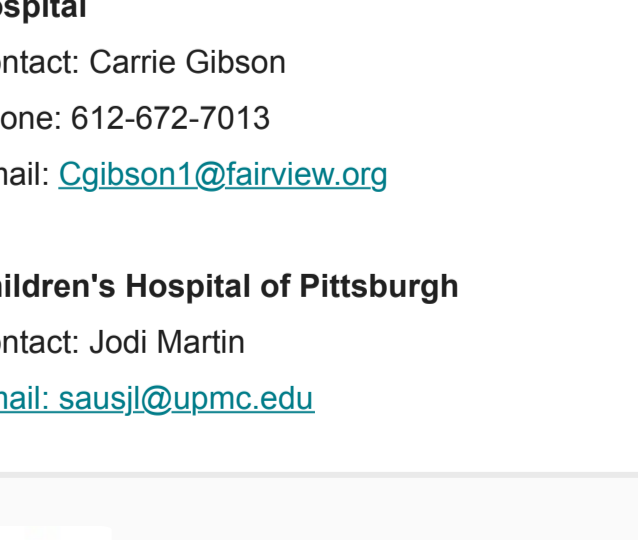
Working within our healthcare system can be frustrating at times, particularly the silos which impair communication between providers. However, she suspects that with new technology making its way to the healthcare sector, we will see significant disruption, which will improve the way teams practice medicine and improve the delivery of healthcare to patients and families.

We are in an exciting time right now for many genetic and rare diseases, and there are new technologies emerging. Gene and cellular therapies are the new frontier in terms of technology and novel treatments for our patients. Dr. Al-Hertani is confident that the natural history and long term outcomes in the next 10 years will be different from the previous 10 years, in the sense of being much better.

As almost all of us have, Dr. Al-Hertani's group at Boston Children's hospital has had to adjust to a pandemic world and shift the way they see and treat patients. Her group pivoted to a hybrid model of in-person and virtual delivery of healthcare. However, despite it being a successful model, virtual clinic visits still do not come close to in-person visits in terms of having the ability to connect with patients and families on a personal level. And it is this sort of connection that Dr. Al-Hertani appreciates and finds rewarding for both her and her patients and families. We hope someday we can move back to more in-person appointments and connection.

Thank you, Dr. Al-Hertani for the amazing work you do for the Gaucher patient community. We wish you a wonderful new year and hope that you take some time around the new year to do what you love to do -- go for long hikes with your golden doodle Van Gogh across the beautiful New England landscape!

Gaucher Community Patient & Family Conference



It's On!
The 2022 conference is confirmed! Please join us in Minneapolis for the Gaucher Community Patient & Family Conference from Sunday, October 23 to Tuesday, October 25, 2022. Join us to share, learn and laugh together and build the bonds between patient families that help us throughout the diagnostic journey and life living with Gaucher.

Sunday evening dinner and the Monday workshops will be for patients only, and Monday evening dinner and Tuesday conference sessions will include our doctors, researchers and industry partners. Keep a lookout for conference registration opening the spring. We will be offering patient stipends to help people attend the conference. If you are interested in volunteering at the event or running a workshop with information you think will of interest to other Gaucher patients and families, please reach out to us now! Email us here to let us know how you would like to be involved.

We're looking forward to seeing you in person. Finally!

Clinical Studies



AVROBIO
Phase 1/2 Lentiviral Vector Gene Therapy - The GuardOne Trial of AVR-RD-02 for subjects with type 1 Gaucher disease. 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, amongst other criteria:
• Be between the ages of 18 and 50 (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 this link to complete the pre-screener at https://www.gaucherclinicaltrials.com/ or call 1-877-330-5214.

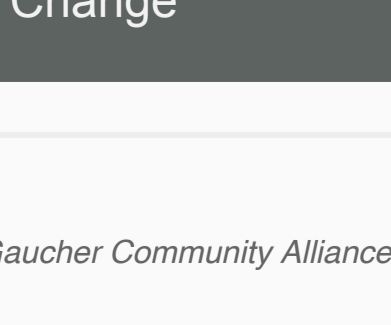
Prevail Therapeutics
Phase 1/2 Clinical Trial of PR001 in Infants With Type 2 Gaucher Disease (PROVIDE) - PROVIDE is a potentially disease-modifying, single-dose gene therapy for patients with neuronopathic type 2 Gaucher disease. For more information, visit the website here.

To enroll, participants must, 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
• Have neurological signs and/or symptoms consistent with diagnosis of type 2 Gaucher disease

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University of Minnesota Masonic Children's Hospital
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Children's Hospital of Pittsburgh
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For Patients, By Patients Together We Can Make Change

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