GCA Newsletter

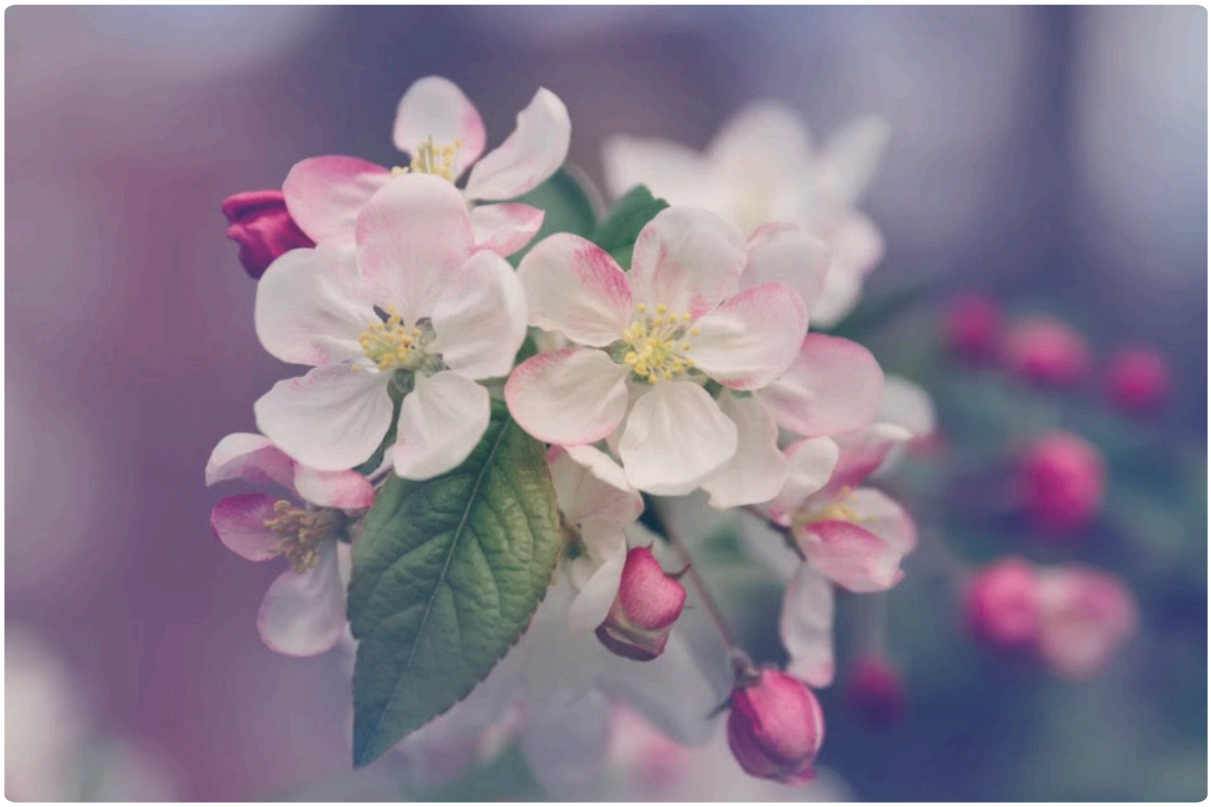
Gaucher Community Alliance <info@gauchercommunity.org>
Reply-To: Gaucher Community Alliance <info@gauchercommunity.org>
To: aviva@gauchercommunity.org

Tue, Apr 6, 2021 at 1:55 PM

DONATE NOW

GAUCHER COMMUNITY ALLIANCE

Photo by Marivi Pazos on Unsplash



Spring 2021
Gaucher Community Newsletter

Welcome to our spring newsletter! We offer you the below information with the hopes of looking forward to and welcoming better times for us all. 2020 proved to be a trying year for most of us, but we have stuck together and stayed strong through the worst of it. Many thanks to those of you who have reached out to us to let us know how you are doing and how we can help. Please continue to do so!

Spring represents rejuvenation and renewal, a time when we can start fresh and right all that may have gone wrong. As Lady Bird Johnson once said, "Where flowers bloom so does hope." We hope each day this spring and throughout the year is filled with flowers and hope for you.

Fondly,

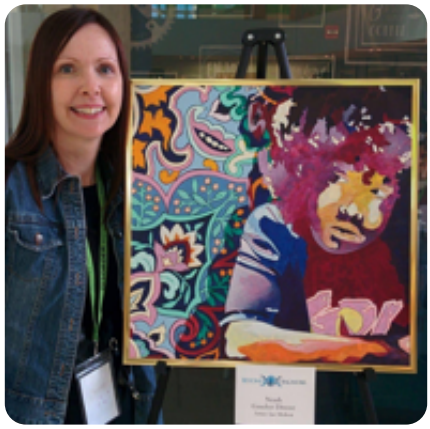
Cyndi Frank & Aviva Rosenberg,
Co-Founders and Co-Presidents

Our Lives with COVID-19

Dr. Deborah Barbouth

by Theresa Sweeny

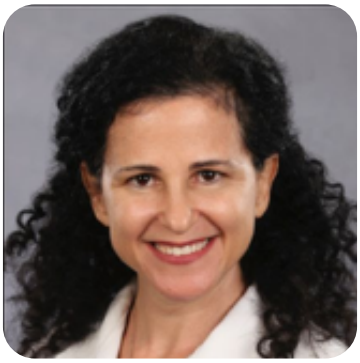
GCA Board of Directors



I found myself in every parent's nightmare….with my son lying in a hospital bed fighting for his life. There he was with tubes sticking out of his mouth and nose, keeping him attached to a ventilator. The incessant beeping of heart monitors intertwined with the Vader-esque whooshing of the vent, keeping 15-year-old Noah who has type 3 Gaucher alive. The scene continued day in and day out, hour in and hour out, for weeks. But it didn't happen during the past year; it happened in July 2011, nearly a decade before the current pandemic. It was not COVID-19 but Influenza A. And it scared me beyond words.

Every time an alarm sounded, which happened often, I jumped out of my skin. Noah was extremely difficult to keep sedated – he is a hyper-metabolizer, something the hospital staff did not seem to understand. I could not leave his side. One morning, while restlessly watching Noah very closely from the rock-hard sofa, I saw his hand twitch. Athough he was sedated, I asked him what he thought he was doing, and jokingly warned him no funny business. In the next

The Gaucher Providers



Dr. Deborah Barbouth has been an Associate Professor at the University of Miami Miller School of Medicine since 2005. She is Board certified in Pediatrics, Clinical Genetics and Clinical Biochemical Genetics. Dr. Barbouth earned her medical degree magna cum laude from Maimonides University in Buenos Aires, Argentina and completed postgraduate training in Pediatrics at Jackson Memorial Hospital at the University of Miami. She completed her clinical training in Medical Genetics at the Albert Einstein College of Medicine and the University of Miami where she received extra training in Clinical Biochemical Genetics.

Dr. Barbouth has presented numerous abstracts and platforms at scientific and professional meetings and published articles in prestigious journals. Her main interests include lysosomal storage disorders, Jewish genetic diseases, newborn screening and Fragile X syndrome.

In this issue, we would like to introduce Dr. Deborah Barbouth, our community's wonderful Gaucher specialist at the University of Miami in

instant, Noah's sedation wore off enough to cause him to panic, and he broke his soft restraints, ripped out his arterial line, and almost extubated himself. I had to flop on top of him to grab his wrist to stop the profuse bleeding and keep his other hand from pulling out his breathing tube. I felt like I was fighting with The Hulk and it seemed like an eternity until someone came into the room to help. I was breathless and trying to calm a very scared Noah. The doctor increased the sedation, and Noah went peacefully back into a deep sleep where he remained for five more days.

After those 10 long days, I could not imagine Noah undergoing intubation ever again, and neither could he. To this day he talks about how scary it was. He completed the book My Wishes, published by the National Institutes of Health to help young adults with end of life care decisions, and he had made his wishes clear.

And now, 10 years later, we find ourselves in another dire situation. We have done everything we can to keep Noah safe during the current pandemic, and we put into practice the lessons we learned in 2011: 1) use all the hand sanitizer; 2) do not touch anything you can use a key for (e.g., elevator and ATM buttons); 3) wear a mask in public, especially when travelling or in winter during cold and flu season; and 4) no unnecessary visitors. The rare times I have to go into a store, I wear head to toe PPE, eye everyone with suspicion, and treat people like Typhoid Mary. If someone coughs within two aisles of me, alarm bells ring in my head and my

Florida. Dr. Barbouth's interest in lysosomal storage diseases (LSDs) started while she was doing her clinical biochemical fellowship. She first got involved with mucopolysaccharidosis and subsequently with Fabry, Gaucher and Pompe. She had the idea of having a lysosomal storage disease center for many years once she got more experienced with these conditions. She truly enjoys meeting the families and LSD experts all around the world.

The most rewarding thing about working with the LSD community for Dr. Barbouth is the human contact. In genetics it is not unusual to be involved with the whole family in some sort of way. But in LSDs in particular, she gets to know and sometimes treat more than one individual in the family. Dr. Barbouth states that the LSD community is very caring, and she has been able to participate at international meetings with families and experts that have been very rewarding for her. As we all know though, it can sometimes be difficult to work within our healthcare system. There is a lot of paperwork, sometimes it is hard to find a good referral close to where the family lives, and it can also be difficult to get treatment approved, which many of us with Gaucher disease know well!

Dr. Barbouth has been coping well during the pandemic, and she is very grateful for what she has. She does yoga and meditation daily which helps her to stay focused and centered on good things during these unprecedented times. In her free time when she is not treating patients and staying healthy through yoga and meditation, Dr.

heart pounds. I double back and look for an empty aisle.

Noah and I have remained COVID-free, but my family has not been spared. My mother, who was living in a nursing home in Texas, contracted COVID and died in September 2020. Later that year my father, who already suffered serious health issues, also contracted the virus. It has damaged his quality of life, which was already in decline before the pandemic, but he is alive.

Noah has been extremely miserable for the last year. I often wonder if my anxiety levels will ever return to normal and cannot imagine how or when. Everytime I wipe something down, it amazes me there is a finish left. With every wipe, my steering wheel is becoming stickier and I ponder the difficulty of installing a new one (how many YouTube videos will I have to watch until I call my other kids?).

Noah and his “Uncle-Grandpa” (my brother Joe) went together for moral support to get their first and second doses of the COVID vaccine – masks, shields and six feet between them. Noah has talked every single day during the pandemic about getting his jab so he can once again have Monday night dinner with his “Uncle- Grandpa,” attend hockey games, and attend his cherished art therapy.

The warp speed with which the vaccines were created were a proof of concept, and also prove what we in the rare disease world already knew: science CAN progress at warp speed if the

Barbouth spends time with her family at her home in Miami Shores.

Thank you Dr. Barbouth for the wonderful work you do to help Gaucher patients and their families.

Gaucher Patient Advocacy Summit

For Patients, By Patients

It's been a super long time since we've all met in person, and we're planning for the GCA's first ever in-person patient conference! Vaccines are rolling out from state to state, and we're hoping to see everyone in the country vaccinated who wants to be vaccinated. If that happens, and if the CDC changes its guidelines on indoor gatherings, then the conference is on.

Gaucher Community Patient Conference
Austin, TX
November 7-9, 2021 (CDC permitting)

Day one is for patients, caregivers and family members only. It will be an opportunity for many of us to meet for the first time ever, get to know one another in person, put the face to the name we see on social media, and participate in small, personalized, interactive workshops where we

regulatory bodies don't inhibit that progress. We should do everything we can to compel the government to allow science to continue at this breakneck pace to benefit rare disease, as time is not a luxury in this space. I feel as though we are entering a second spring, a rebirth of sorts full of optimism. A chance for the world to spring forth from the earth and start again.

Online Learning Opportunities

Our online webinars are an opportunity to receive Gaucher community education from your own home. The more we know about the best ways to manage our disease, the more power we have to make educated decisions on what is best for us in our disease journey. Please feel free to view past webinars on our website here on a variety of subjects that can help you.

The Gaucher Community Alliance is an organization by the patient community for the patient community that encourages peer-to-peer support within the patient community. We would love to hear your thoughts and recommendations for future online learning opportunities. Email to let us know what subjects you'd like to see can gather the tools we need to live the best lives we can with our disease.

Day two is for other members of our Gaucher community who help us. Gaucher providers, medical experts, and industry professionals will be there to share with us information on the services they provide the patient and caregiver community. It is also an opportunity for patients and families to have a voice and share what works and doesn't work in their care and treatment options.

Please reach out to us via email to let us know what you'd like to see at the conference. Do you have a recommendation for a workshop? Would you like to lead that workshop? Are you able to volunteer at the event? Please let us know!

Current Research

by Nadine Henderson, MS, LCGC

Senior Genetic Counselor

Lysosomal Storage Disorders Program

UPMC Children's Hospital of Pittsburgh

NIH U.S. National Library of Medicine

ClinicalTrials.gov

The world of clinical trials is an exciting and constantly evolving area. For rare genetic diseases, participating in clinical trials is often where families can find researchers who are experts in these conditions. At clincaltrials.gov, anyone can search for studies using specific keywords. Studies listed on the site are either currently ongoing or have been closed or completed. Completed studies may have results

covered in our online webinar series.

Voicing the Patient Perspective at WORLD*Symposium* 2021

by Lori Ann Correia
AVROBIO

AVROBIO is a clinical stage lentiviral gene therapy company with headquarters in Cambridge, Massachusetts and an office in Toronto, Ontario. Our company's vision is to bring personalized gene therapy to the world. AVROBIO is currently studying the safety and efficacy of ex vivo lentiviral gene therapy in patients with Fabry disease, type 1 Gaucher disease and cystinosis, and we are planning for future programs in Hunter syndrome, type 3 Gaucher disease and Pompe disease. Our therapies have not yet been proven safe or effective or approved by the FDA or any other regulatory agency. For additional information about AVROBIO, our technology or our lysosomal disorders pipeline, please visit avrobio.com, and follow us on Twitter and LinkedIn.

AVROBIO supports the Gaucher community by working together to achieve shared goals such as raising awareness for Gaucher disease, educating and empowering those living with rare conditions and studying new treatment options. We also partner with the community to shine a light on the patient's voice as well as provide information on the latest research.

posted. Studies can be sponsored by a pharmaceutical company or may be initiated by an independent investigator or group of investigators who have found their own funding source.

There are a variety of purposes of the studies listed on this website, including observational and interventional. Observational studies study the natural history of a condition, evaluate new tests or procedures that might provide new data for improving outcomes, or monitor a patient on treatment over time. Interventional studies use some type of product (investigational drug, gene therapy, etc.) and evaluate if it is making a change in the patient's natural history trajectory resulting in a better outcome. Although this website is hosted by the National Institute of Health's US National Library of Medicine, it also lists studies that are outside the US. Some international studies accept subjects from other countries, and there is never any harm in asking, especially for rare diseases where there are few patients.

It is also important to note that the listing of studies on clincaltrials.gov does not mean that the study has been evaluated by the US Federal government. The sponsors and investigators of each and every study listed are the ones who are responsible for the safety and management of the study. You should always review the study with your health care team before participating. Since these are studies, you will be required to provide full informed consent prior to any procedures being done.

This partnership was illustrated at this year's WORLD*Symposium*, a conference bringing together the lysosomal disorders community and sharing its scientific advances. The patient's voice and information about Gaucher disease were well represented: we counted more than 70 platform presentations, posters and symposia covering topics related to Gaucher disease.

In "Unmet need in Gaucher Disease: *Future possibilities with ex-vivo lentiviral gene therapy,"* a symposium sponsored by AVROBIO, GCA's own **Cyndi Frank** spoke about the patient experience with type 1 Gaucher disease. Joining Cyndi were world-renowned Gaucher physicians **Dr. Timothy Cox**, **Dr. Roy Alcalay** and **Dr. Aneal Khan**. **Dr. Cox** spoke about unmet needs in Gaucher disease, while **Dr. Alcalay** spoke about the link between Gaucher disease and Parkinson's disease/Lewy body dementia. **Dr. Khan** described current treatment options and investigational treatment options including lentiviral gene therapy. He also provided an overview of **GuardOne**, a Phase 1/2 clinical trial evaluating an investigational lentiviral gene therapy called AVR-RD-02, which is currently enrolling adults with type 1 Gaucher disease. Find more information about the GuardOne trial here.

Patient advocate Meghan White of the National Gaucher Foundation of Canada collaborated with Fernanda Copeland and Lori Ann Correia from AVROBIO's patient advocacy and engagement team on the poster titled "Uncovering the burden

We recently searched the keywords "Gaucher disease" on clincaltrials.gov. We left blank the "other terms" and "country" fields and pulled up 126 Gaucher disease studies with the following statuses: recruiting, not yet recruiting, enrolling by invitation, active (not recruiting), suspended, terminated, completed, withdrawn or unknown status. Of these, we found 56 studies that are currently active and accepting Gaucher patients. As mentioned above, some studies listed are general observational studies of treated or untreated patients, screening for undiagnosed patients, drug pharmacokinetic studies, biomarker studies, immunologic evaluations, thrombocytopathy, new diagnostic techniques (MRI, QCSI, PET, elastography, Fibroscan), hepatitis and Gaucher interactions, genetic variation in Gaucher (especially related to Parkinson's), Gaucher in minority populations, digital health platforms, and cellular and molecular evaluations initiated by academic centers or registries/outcome surveys that are sponsored by industry. The remainder of the studies are those that are evaluating some type of intervention with investigational products or techniques such as: AVR-RD-02 (gene therapy), PR001 (gene therapy), venglustat in type 3 (US, Germany, Japan, UK), arimoclomol in Type 1 or 3 (India), ambroxol (Israel), stem cell transplant in neurologic Gaucher, or the use of other approved medications for off-label uses or in other countries outside of the US (imiglucerase, velaglucerase, elelyso, miglustat, and eliglustat).

Knowledge is power, and it provides a good

of Gaucher disease type 1: Patient perspectives on unaddressed symptoms, impact of disease, and the future of treatment." The poster reports results from a discussion with nine individuals living with type 1 Gaucher disease. This group of individuals described fatigue, bone pain, and bone crises as debilitating symptoms that limit personal and professional lives. Participants also helped to differentiate chronic, daily bone pain from a bone crisis. They expressed their concern for the association and increased risk of Parkinson's disease in individuals with mutations in the *GBA* gene and more research is needed to better understand this link.

Noting that existing therapies improve quality of life, this group of individuals also expressed the need for new treatment options that better manage unaddressed symptoms, including bone pain and fatigue. Patients have many questions about treatments being studied, such as gene therapy, including how it works, safety and efficacy, among others. As studies progress and regulatory agencies determine the safety and efficacy of gene therapy for the treatment of typed 1 Gaucher disease, patients will need information and education to make informed decisions.

World Rare Disease Day 2021

pathway for patients and caregivers to be strong advocates and make smart, educated decisions about your own care. Hopefully this introduction to available trials on clinicaltrials.gov will inspire you to search for new studies that may be relevant, interesting or beneficial to you. In combination with searching medical literature for new publications, your acquired knowledge will help you to advocate for yourself or your family member impacted by Gaucher disease.

Health Insurance Updates

Co-pay Accumulator Programs

In our Fall 2020 newsletter, we informed you about the development of large national insurance companies planning to implement copay accumulators. Historically, pharmaceutical companies have offered our community copay assistance which not only contributes toward the patient's copayment but also counts toward the patient's annual deductible. Due to the high cost of Gaucher treatments, these programs have been especially helpful since the patient is required to pay significantly high out-of-pocket costs and deductibles for treatments.

Health plans are increasingly implementing

Did you catch Bailey's heartwarming video on Rare Disease Day 2021?

This year, for Rare Disease Day, we celebrated 12-year-old Bailey Regalado. Bailey has type 3 Gaucher disease, and she and her family are passionate about raising awareness of Gaucher disease and its different types.

Bailey was diagnosed with type 3 Gaucher disease when she was just 10 months old. She has endured more in her 12 years than most adults do in a lifetime, yet she continues to stay positive and carry forward with love in her heart and a smile on her face.

If you haven't seen Bailey's heartening video yet, you can watch it here. And if you have seen it, watch it again. Bailey reminds us of the important things in life and how to stay positive, even when times get tough.

Type 3 Young Adults and Caregivers Needed for Study

IQVIA is looking for type 3 young adults and caregivers in the US and UK for a compensated study.

The purpose of this study is to provide feedback on a questionnaire that will be used in a clinical trial. It is designed to understand the daily activities of people living with type 3 Gaucher disease (referred to as “GD3” hereafter). The goals of this research are to determine:

copay accumulator programs which prevent copayment assistance from counting toward a plan enrollee’s deductible. However, patient assistance is not a bottomless well. Patients receive a finite amount each year. Once copayment assistance runs out and the burden to pay is on the patient, many patients can no longer afford their medications. This rule was intended to steer patients to less costly, generic medications when possible, but for Gaucher disease, there are no generic alternatives. This ends up placing Gaucher patients at risk for medication adherence issues, including skipping refills, rationing medications, or abandoning treatment altogether.

The GCA has joined with dozens of other patient advocacy organizations to fight these harmful programs. The fight must occur on two fronts: state level and federal. In addition to the six states that have already banned this practice, there are bills at various levels in the system working their way through 25 other states. When a bill is scheduled for a hearing, the GCA posts on social media requesting assistance with patient testimony from that state on the harmfulness of these accumulator practices. While the state level bills are moving along, state law only covers 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. Prior to 2020, there were certain federal protections that only allowed health plans to implement copay accumulators when both a brand and generic medication were available

- Whether the questionnaire content captures the most important aspects of the experience living with GD3 and its treatment
- Whether patients or people caring for individuals with GD3 who may answer these questions understand their meaning and how to provide responses that reflect their experience

To accomplish these goals, IQVIA is interested in speaking with YOU!

IQVIA is looking for pairs of caregivers/ adolescents to participate in this study. The study will consist of an approximately 45-minute, one-to-one telephone interview with the GD3 patient and one of our healthcare researchers. A separate one-to-one telephone interview will be scheduled for the caregiver.

The interviews will occur at a date and time convenient for you. To facilitate the interview process, online materials will be shared via a web-link which will be provided in advance of the interviews. The study is offering an honoraria of $90 USD to both the caregiver and the GD3 patient for participating in the interview.

If you are interested in participating, please send an email to Sully Kidwai at mkidwai@us.imshealth.com (the IQVIA representative coordinating this study) stating just your first name and a message indicating that you are interested in participating in the GD3 Research. At that point, IQVIA will send you a link to an online consent form to complete. Once you have consented, IQVIA will work with you to schedule the interview.

(NBPP 2020). However, the prior administration issued NBPP 2021 which explicitly allows health plans to implement copay accumulators in all instances. The GCA is working along with many partners to ask the current Biden administration to reopen NBPP and reinstate the 2020 protections.

The GCA has learned of several patients in our community who have already been affected by copay accumulators which is putting them under enormous financial hardship. If you have been affected by these harmful insurance programs, please let us know. The more patients we know of that have been affected by this, the more ammunition we have to argue for reenacting NBPP 2020. In addition, please watch social media for a call out to your state for you to get involved.

Shop on Amazon and Raise Funds for Gaucher Disease at the Same Time!

It's easy!

Support the Gaucher Community Alliance by starting your shopping at smile.amazon.com. 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.

DONATE NOW

Get Involved!

We would love to hear from you. Let us know what you need and work with us to make it happen. Visit our website to Volunteer, Donate, or Become a Member. Let us know how **you** would like to get involved!

Like us on Facebook!

For Patients, By Patients
Together We Can Make Change

Our mailing address is:
5255 Fair Oaks Street
Pittsburgh, PA. 15217

www.gauchercommunity.org

EIN# 83-4696980