Gaucher Teen and Young Adult DC Advocacy Program Focuses on Unmet Needs and Policy Concerns Affecting Rare Disease

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Introduction

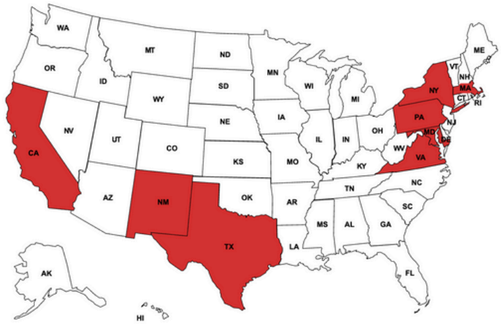
- Gaucher disease (GD) is a rare autosomal recessive lysosomal storage disorder (LSD) caused by mutations in the GBA1 gene.
- GD has a heterogeneous presentation from mild non-neuronopathic Type 1 to severe neuronopathic GD (nGD).
- Type 1 GD accounts for ≥90% of cases in the United States with symptoms beginning from 6 months of age into adulthood.
- nGD patients develop severe neurological impairments and symptoms in infancy, generally before 6 months of age.
- Symptoms of all GD may include splenomegaly, hepatomegaly, bone and hematological abnormalities, and cardiovascular issues. nGD involves the central nervous system and also includes progressive myoclonus epilepsy, cerebellar ataxia or spasticity, pulmonary issues and dementia, and behavioral changes.
- FDA-approved enzyme replacement therapies and substrate reduction therapies are available to treat the visceral symptoms of GD.




Background

- The Gaucher Community Alliance (GCA) hosted its first **Teen and Young Adult Advocacy Retreat** in Washington, DC from June 24–26, 2025.
- All US-based Gaucher teens, young adults, and their families were invited to Capitol Hill to share their personal stories and advocate for the needs of the Gaucher community.
- More than 30 young community members attended and learned how to effectively communicate with Senators and Representatives regarding issues directly affecting people with Gaucher disease.
- Key legislative topics included:
  - Newborn screening policies and modernization
  - Medicare home infusion coverage
  - NIH research funding cuts
  - Medicaid cutbacks
- For many participants, this marked their first advocacy experience and first visit to the nation's capital.

Patient Representation at DC Program



- Maryland
- Virginia
- Pennsylvania
- California
- Texas
- New Mexico
- Massachusetts
- Puerto Rico
- New York

Program Design

- The program began with advocacy training led by experienced Capitol Hill lobbyists and teen peer advocates who frequently visit DC to meet with legislators.
- Participants practiced telling their personal stories and shaped clear "asks" in small group sessions.
- On Hill Day, participants were organized into groups representing 2 to 3 states, each scheduled for 5 to 6 congressional meetings with offices from their home delegations.
- Each group was led by an experienced advocate to support transitions between buildings, help guide conversations, and ensure accommodations for mobility-impaired participants.
- The program emphasized hands-on learning, confidence building, teamwork, and peer support.



Participant Thoughts

"Our representatives being able to meet me and some members of the Gaucher community made me feel seen and gave them a window into my health struggles in a very personal way that felt hard to ignore. It also felt like I could actually talk to these people who make our laws, and that helped it feel like what I was doing mattered -- almost that it held them a little more accountable. We introduced a rare disease to them and asked for help which is scary and odd, but we are still real people who need real help with our real problems."

– Rebecca, she/her, 16, Los Angeles, CA

"I had the opportunity to visit the office of the Resident Commissioner of Puerto Rico, Pablo José Hernández Rivera, and I was able to speak to his staff about some of the struggles that the patient community has to face on a daily basis. My experience in DC was incredible; I felt a part of a welcoming community, I got to meet awesome people, I visited some beautiful museums, and I left the capital with hope and the desire to continue advocating for rare disease patients in Puerto Rico and the United States."

– Laura, San Juan, Puerto Rico

Outcomes

Participants and staff completed over 20 congressional meetings across the House and Senate.

As a full group, participants also met with:

- Representative Debbie Wasserman Schultz (D-FL)
- Congressman Eugene Vindman (D-VA)
- Congresswoman Jan Schakowsky (D-IL)

Legislative staff expressed interest in:

- Exploring policy options for Medicare home infusion coverage
- Learning more about current issues affecting federal newborn screening guidelines

Beyond policy impacts, the program provided:

- Practical advocacy, leadership and communication skills
- Increased understanding of government processes
- Community bonding with peers who share similar health and life-stage experiences
- Strong positive feedback from parents and caregivers, who described the experience as empowering, life-changing, and long overdue for young people with Gaucher disease

Since the program, several teens have already participated in local and state advocacy efforts with their new knowledge and empowerment.

Acknowledgments

We extend our sincere gratitude to the participating teens and families, our advocacy trainers, congressional offices, and all supporters who made this program possible.