



Fall 2025

THE RAY

The Association for Glycogen Storage Disease



President's Message

AGSD Annual Conference 2025 – A Resounding Success!

This past June, the AGSD community gathered in Denver, CO for our annual conference — a vibrant and well-attended event that united researchers, individuals, families and friends in a shared mission of learning and connection. At the heart of AGSD is the belief that progress comes from collaboration and this year's conference embodied that spirit more than ever! Thanks to your feedback, we curated a dynamic mix of panels, discussions, workshops and lectures to meet diverse interests and needs. We were proud to introduce Continuing Medical Education (CME) credits for healthcare professionals and we've made the CME track lectures available on our YouTube channel— accessible to all ~ just two weeks post-conference. Our commitment to equitable access and lifelong learning continues to drive us forward. Each year, we strive to improve, innovate and expand the reach of AGSD's mission. Together, we are building momentum toward better treatments — and ultimately cures.

Keep the Momentum Going — stay Involved with AGSD!

The energy from our annual conference doesn't have to end in Denver. There are so many meaningful ways to stay connected and make a difference year-round:

- Participate in GSD Awareness Week! Help amplify our message by sharing stories, hosting events and/or simply spreading the word online.
 - Fundraise in your Community! Whether it's a bake sale, a 5K run, or a birthday fundraiser, every dollar helps fuel research and support.
 - Volunteer your grant writing skills! If you have experience with grants, your expertise can help us secure vital funding for programs and research.
 - Educate your children's classroom! Bring GSD awareness into schools with age-appropriate materials and presentations — empowering the next generation with knowledge and empathy.
- Together, we're building a stronger, more informed, and more compassionate community. Let's keep learning, sharing, and growing — every day, not just once a year!

Kim Burney

President, Association for Glycogen Storage Disease

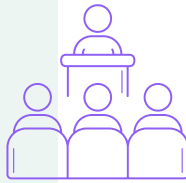
DID YOU KNOW?!



WE HAD **240** ATTENDEES
(OUR BIGGEST YET!)



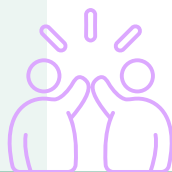
21 SPEAKERS



OVERALL, OUR PARTICIPANTS TRAVELED
FROM **37** DIFFERENT STATES,
AND **13** DIFFERENT COUNTRIES!



AND THERE WERE **7** DIFFERENT TYPES
OF GSD REPRESENTED



A Conference Perspective written by 2nd-time attendee - Joan Eisele-Cooper (Type 15)

The Association for Glycogen Storage Disease's Annual Conference this year was held in Denver, Colorado, from June 20-21, 2025. It was held at the Sheraton Denver Tech Center. In addition to comfortable rooms and amenities, the hotel had an accessible outdoor pool, which brought together sun worshipers and anyone wanting to cool off. This is the second year in a row that my daughter, Selena and I have attended.

The conference began on Friday morning with registration and vendor introductions. There were lots of informal get-togethers as people reconnected from all over the country. After lunch, the kids went off to the children's activity room, which was open all day, both days of the conference. All the adult attendees heard a keynote address from Dr. Patrick Ryan, MD, on all of the GSD types..

Attendees then had a choice of three breakout groups every hour from 2:30 pm to 5:30 pm. Some of the sessions included "Adult Management of GSD", "Glycogen Storage Disease Emergency Care", "Student Accommodations IEP/504 Plans", "Nutrition" for teens, and new this year, parent panels. Each session was led by an expert in the field who was available to meet with individuals afterwards.

In the evening after dinner, everyone went to a breakout room to mingle with families who had shared their specific GSD Type. We met with another family and enjoyed hearing their story and getting to know them and their son, while playing balloon volleyball.

On Saturday after breakfast, attendees had nine diverse choices of talks from 9:30 am until 2:30 pm. Some of the talks included "Gene Therapy and Type III", "GSD Biomarker", and "GSD value: Development of an International Consensus-Based Standard Outcome Set". Two parent panels included "GSD Exercise and Health" and a "Dietitian panel for GSD Types 1A and 1B". After lunch, five more choices were available including "Service Dogs for Hypoglycemia" and "Medicaid 101" from lawyer Josh Norris. Afterward we all gathered for our annual, really quick, drone group photo, in 103-degree weather!

Each year the conference planners organize a fun family event. This year we took a short bus trip to "Pindustry", a unique dining and entertainment venue, where we had dinner and then a choice of many activities such as bowling, darts, cornhole, ping pong, billiards, basketball and retro arcades.

In a quieter area outside I asked participants, "What is the benefit of attending the annual GSD conference?"

Their answers were:

- "I get to meet people - other adults, doctors, lawyers, families and kids in the GSD world."



- "I could see even the shy kids interacting with each other".
- "Every person plays a role in each other's journey. I got some great tidbits of information and advice from other parents".
- "My biggest takeaway was taking a look at what goes in my body."
- "It's amazing to meet other families. My 5-year-old daughter is so happy. It's wonderful to see everybody coming together. There is so much happiness and positivity. Children think they stand alone, but when they come here it's like family."
- "It's wonderful to have access to professionals in a friendly more personalized way. I was able to speak one-on-one to a doctor doing research on my specific GSD type."

We heard closing remarks from AGSD officers -- President, Kim Burney; Vice President, Katie Ward; Treasurer, Sochi Krikorian and Secretary, Steve Liu. Dr. David Weinstein, an internationally-recognized expert in Glycogen Storage Disease, gave some closing remarks on returning to Denver -- Reflections on 25 years of progress. He said, "Thirty-one years ago a GSD diagnosis was a death sentence. Today, look around, we see healthy people with GSD. There are less complications, new treatments on the horizon -- gene therapy, MRNA and gene editing. The future is bright!!"

A big thank you everyone who worked to make this year's conference a success. I hope to see you in Cleveland, Ohio at next year's conference in June of 2026!

Join the AGSD Community United for Progress in Glycogen Storage Disease

Whether you are a patient, a caregiver, a clinician, a researcher, or an advocate, the Association for Glycogen Storage Disease (AGSD) is here to support, connect, and empower you. Why become a member?

- **Affordable Dues:** Just \$25/year — because support should be accessible to all.
- **Scholarship Opportunities:** Members are eligible for post-secondary education scholarships.
- **Discounted Conference Registration:** Enjoy member rates for our annual AGSD Conference — a hub for learning, networking and community.
- **Your Voice Shapes Our Events:** We consider member locations when planning our annual conference to make it easier for you to attend.



Join AGSD today and be part of a community that understands, uplifts and advocates.



Donations

Thank You for Fueling Our Mission
The Association for Glycogen Storage Disease (AGSD) is a fully volunteer-run organization. Every program we offer — including our annual conference — is made possible through the generosity of donors like you. We simply couldn't do this work without your support. With heartfelt appreciation, we thank the following for their recent donations:

Deb Shuster

Richard & Marlene Mueller

Your contributions help us connect families, empower patients, educate clinicians, and advance research — all while keeping our events accessible and inclusive.

*Thank
You!*

Shop the AGSD Store

Get your fall swag, and keep a lookout for some new rare disease day merchandise coming soon!



www.agsdus.itemorder.com

Rachel's Picks



Submitted by her mom Barbara Hefter
(in honor of her daughter Rachel Zareski Walts (Type 1A))

Are you looking for a good book to read?? Check out the suggested book titles listed below:

For young adults (ages 8-12)

"Hello Universe: A Newbery Award Winner" by author Erin Entrada Kelly

This book is geared toward middle schoolers with the theme of friendship, diversity, self-discovery, the power of kindness and the connections that bind people together through fate and chance..

For adults

"The Mighty Red" by author Louise Erdrich

Is a 2024 novel set in the Red River Valley of North Dakota, focusing on the lives of a community, particularly a high school senior named Kismet, as they navigate the aftermath of the 2008 economic crisis. The story explores themes of love, family, heritage and the impact of powerful forces like big agriculture and climate change on the land and its people.

For adults (ages 22+)

"Wintering: The Power of Rest and Retreat in Difficult Times" by Katherine May

This is a memoir exploring the concept of "wintering," which refers to the involuntary, fallow periods in life that, like the winter season, are necessary for healing, rest and eventual renewal. A great read!!.

Conference Videos

Check out these NEW videos from the 2025 AGSD Conference!!

We have made great efforts this year to bring you some recordings from our speakers who presented at the 2025 AGSD conference in Denver, CO.

We aim to record key talks to bringing the most updated GSD news to those who were not able to attend. View them all now on our YouTube page!

www.youtube.com/@agsd-agsd



Trial Update: Ultragenyx (DTX401) For Glycogen Storage Disease IA
 Rebecca Riba-Wolman, MD
 AGSD Conference
 Date: Jun 27
 58:40

GSD Biomarkers
 Monica Boyer, CPN
 AGSD Conference
 Date: Jun 27
 27:57

GSD 101
 Patrick Ryan, MD
 AGSD Conference
 Date: Jun 27
 49:29

Drug Development Pre Clinical to Clinical
 Patrick Ryan, MD
 AGSD Conference
 Date: Jun 27
 32:28

Glycogen Storage A Hepatologist Perspective
 Kadalakal Radhakrishnan, MD, MRCP (UK), MRCPCH, FAAP
 AGSD Conference
 Date: Jun 27
 35:50

Adult Management of GSD
 Julieta Borwin Salgado, MD, MS
 AGSD Conference
 Date: Jun 27
 44:09

GSD Value: Development of an International Consensus-Based Standard Outcome Set
 Terry Derks, MD, PhD
 AGSD Conference
 Date: Jun 27
 41:04

Ketotic Hypoglycemia in GYGI - Related GSD
 Benjamin Coanougher, MD, PhD
 AGSD Conference
 Date: Jun 27
 41:13

Gene Therapy and Type III
 Giuseppe Ronzitti, PhD
 AGSD Conference
 Date: Jun 27
 1:01:59

Kidney Complications and Surveillance in GSD I
 David F Rodriguez-Burillo, MD
 AGSD Conference
 Date: Jun 27
 49:25

Check out our New Website!

We have worked hard to provide a new and easy to navigate website!

Our site is still a work in progress, but we hope you find it easier to manage and access the information you need. What we hope to achieve are guidelines and videos per GSD type that are easy to find, current news, articles and updates in the GSD field as well as a hub for yearly conference information. We have links to articles, patient advocacy sites, and information about membership, scholarships and so much more!!

www.AgsdUS.org



2026 Conference

Planning has already begun for our 2026 AGSD Conference and we hope you are as excited as we are about our location this next year! Next year our conference will be located in ...

Cleveland 
JUNE 19-20

Be sure to be on the look out soon for more information!



Drug Trial Updates

Ultragenyx

- In a recent news, Ultragenyx announced positive longer-term data from Phase 3 Study of DTX401 AAV Gene Therapy for the Treatment of Glycogen Storage Disease Type Ia (GSDIa)

Learn more here : <https://ir.ultragenyx.com/node/18036/pdf> 

- On August 18, 2025, Ultragenyx announced the initiation of a rolling submission of a Biologics License Applications (BLA) to the U.S. Food and Drug Administration (FDA) for DTX401, an investigational gene therapy for patients with glycogen storage disease Type Ia (GSDIa).
- A new Website was launched to help others understand and learn more about GSDIa <https://www.understandinggsdia.com/>

Beam Therapeutics

- Beam Therapeutics is continuing to enroll adult participants in a Phase 1/2 trial evaluating BEAM-301 for the potential treatment of GSD1a. There are three clinical study sites open across the United States. Please contact clinicalinfo@beamt.com for more information.

<https://www.clinicaltrials.gov/study/NCT06735755>

Visit our website under "News and Articles" for more up to date information.

In memoriam: Stephen G. Kahler, MD



Dr. Stephen Kahler was a pioneering geneticist renowned for his work on inborn errors of metabolism, especially glycogen storage diseases. His deep understanding of metabolic pathways led him to advocate for early diagnosis and treatment, notably through expanded newborn screening. He served for many years on the Scientific Advisory Board (SAB) of the Association for Glycogen Storage Diseases (AGSD), where his expertise and dedication were deeply valued. The AGSD Board and community are profoundly grateful for his enduring contributions. His legacy lives on in the lives he touched, the knowledge he shared, and the generations of clinicians he mentored. To access the full memoriam published in the *Journal of Molecular Genetics and Metabolism*, please access link below.

<https://doi.org/10.1016/j.ymgme.2025.109216>

Welcome Newest SAB Members

Monika Williams, MD

Dr. Monika Williams joined the AGSD SAB in September of 2025. She is a Clinical Assistant Professor at the University of North Carolina's Department of Pediatrics, Division of Medical Genetics. She earned a degree in Food Technology and Human Nutrition from the August Cieszkowski Agricultural University in Poznan, Poland, and her medical degree from Nicolaus Copernicus University, Ludwik Rydygier Collegium Medicum (Bydgoszcz, Poland) in 2014. Between medical school and her pediatric residency, Monika has worked in clinical research, involving e.g. clinical trials on extended-release cornstarch and gene therapy. She completed her pediatrics residency at Connecticut Children's (Hartford, CT) in 2022, a Medical Genetics residency at the University of North Carolina (Chapel Hill, NC) in 2024, and a Medical Biochemical Genetics fellowship at Duke University (Durham, NC) in 2025. She has clinical, research and personal interests in glycogen storage and hypoglycemia disorders (as well as other rare metabolic conditions). She is connected with the US and an international GSD community, published in medical journals on GSDs, and presented on multiple international and national conferences.



Julieta B. Bonvin Sallago, MD

Dr. Bonvin Sallago, is the Lead Clinical Research Associate at Connecticut Children's and an Assistant Professor of Pediatrics at UConn Health. After completing her pediatric training in Argentina, she earned master's degrees in Pediatric Neurology and Inherited Metabolic Diseases. Since 2019, Dr. Bonvin Sallago has coordinated clinical trials for rare diseases and glycogen storage disorders and is dedicated to advancing research, improving patient outcomes, and promoting advocacy. She serves on the advisory committee for the Living Rare Study at the National Organization for Rare Disorders, is a board member of the Hispanic Society for Rare Diseases, and collaborates with rare disease organizations internationally. Her work focuses on advancing rare disease research, fostering education, and supporting patient advocacy worldwide.



Member HIGHLIGHTS

GSD Awareness week this year was a big hit! With different themes for each day, everyone shared their stories, their journey, and all the awesome things they have accomplished in spite of their diagnosis. Here are some great pictures of our members highlighting the amazing things they've enjoyed doing!



#MoreThanGSD

#GSDstrong



Emily
An RN working in Womens Health



Ella
The dancer, builder, and occasional pirate princess.



Ira
Loves playing outside!



Dwayne
Loves music, sports and enjoying life!



Cason
Spends most days playing baseball.



Brayden
Is a freshman in college, works part time and has hobbies in fixing up vehicles, fishing, baseball and skateboarding.



Sophie
Soccer Star

