



Global Genes[®]

Allies in Rare Disease

2025

Year In Review

Letter from the CEO



Dear Friends and Partners,

2025 was a defining year for the rare disease community. Scientific progress continued at an extraordinary pace — from advances in gene and cell therapies to growing momentum in individualized and precision medicine approaches. At the same time, the community faced increasing complexity: evolving regulatory and policy environments, funding pressures, access challenges, and the urgent need for stronger collaboration across the entire ecosystem.

Like much of the rare disease field itself, Global Genes entered a period of intentional evolution this year. We took time to listen deeply — to patient advocates, families, researchers, clinicians, industry leaders, and partners across the community — to better understand where the needs are greatest and how we can create the most meaningful impact in the years ahead.

That work resulted not only in a renewed mission and vision, but in the creation of a new strategic direction for Global Genes — including the strategies, goals, and priorities that will guide our future. As we move into 2026, we are committed to co-creating the execution of that vision alongside patients, caregivers, advocacy organizations, researchers, clinicians, industry, policymakers, and partners across the ecosystem.

The future of rare disease progress will depend on connected communities, shared responsibility, and bold collaboration. We are dedicated to helping build that future through adaptive programs, stronger partnerships, leadership development, and research acceleration initiatives designed to support the entire rare disease community.

This work is only possible through partnership. To our community, sponsors, researchers, advocates, industry supporters, and philanthropic partners: thank you for your trust, collaboration, and shared commitment to transforming the future of rare disease.

Together, we look ahead to 2026 with urgency, optimism, and resolve.



Charlene Son Rigby
Chief Executive Officer

2025 IMPACT BY THE NUMBERS

Eliminating the burdens and challenges of rare diseases for patients, their families and disease communities globally.

THE GLOBAL RARE DISEASE LANDSCAPE

1 in 10

AMERICANS
ARE AFFECTED
BY RARE DISEASE

400M

PEOPLE SUFFER
FROM RARE
DISEASE GLOBALLY

8 in 10

RARE
DISEASES
ARE GENETIC

95%

RARE DISEASES
LACKING APPROVED
FDA TREATMENT

10,000+

DISTINCT
RARE & GENETIC
DISEASES

7 in 10

RARE DISEASES
START IN
CHILDHOOD

6 YEARS

AVERAGE TIME
TO DIAGNOSIS FOR
RARE PATIENTS

Our Strategic Direction and Intended Impact

The path toward every rare disease treatment begins with patient leadership.

Through our programs, by **2035** we will:

EQUIP

TO CHANGE THE LIVES OF

5,000

PATIENT ADVOCATES

5 Million

RARE DISEASE PATIENTS



4-Phase Strategic
Planning Process
Completed in
2025



New Mission,
Vision & Strategy
Created



Developing
Rare Leaders
Together



GLOBAL ADVOCACY ALLIANCE

833 Member
Organizations

50 New Members
Joined in 2025

2,007 Members in
Facebook
Community

11 Member
Leadership
Council

78% U.S. Members

22% International
Members



Quarterly Meetings + Year-Round Programming
On leadership, fundraising, IEPs, grief, insurance,
research education and more



CORPORATE ALLIANCE

39 Biopharma Partner
Members in 2025

Collaborative, Peer-Reviewed Publication
Developed and Informed by

40+
Stakeholder
Interviews

2
Strategic
Roundtables



RARE CONCIERGE [PATIENT SERVICES]



1,590 Patient & Caregiver Inquiries
Supported in 2025



70+ Countries Reached,
Including 48 U.S. States



500+ Distinct Rare Diseases
Supported



Serving a Global Community of **400M+**
PEOPLE



RARE DISEASE CURRICULUM

7 Educational
Learning Modules
Developed



Submitted for
Accreditation
through PIM



Freely available to the
rare disease community!



Launching June 2026!



QUICK GUIDE SERIES

10 New Quick Guides Released in 2025
Addressing critical challenges faced by the rare disease
community in a short, resource-forward format



EDUCATIONAL CONFERENCES & EVENTS



RARE ADVOCACY EXCHANGE

1,976 Unique Registrants

968 Views of Session
Recordings

9 Live, Interactive
Virtual Sessions

86% Met / Exceeded Expectations

81% Left with Actionable Skills
& Next Steps

RAD BRAIN WORKSHOP

25 Advocacy Groups

29 Lectures Across 3 Days

1 Research Roadmap
Session per PAG

100% Increased Knowledge

81% Increased Confidence
in Presentation Skills

RARE DRUG DEVELOPMENT SYMPOSIUM

336 Total
Attendees

2 PAG Pitches
(New in 2025)

224 Rare Diseases
Represented

84% More Confident Addressing Research Goals

72% Left with Actionable Next Steps for Research

242 In-person
Registrants

2 Interactive
Workshops

72 Expert Office
Hour Sessions



RARE-X RESEARCH PROGRAM

89 Rare
Diseases

130 Patient
Advocacy
Orgs

38% International
Participants

98 Unique
Countries

189 Data
Requests
Fulfilled

91 Data Usage
Agreements
Executed

76% Communities
w/ Genetic
Testing Info

9,040
Participants
Enrolled



RESEARCH ACCELERATION

4

Fellowship Reports Completed in 2025

4

Additional PAGs Selected for Next Cohort



Program Expanded Through Partnership
with the University of Notre Dame



PUBLICATIONS & THOUGHT LEADERSHIP

2 Peer Reviewed
Papers Published

2 Papers Submitted
for Publication

3 Scientific Posters
Presented

15+

Talks Given at Major
Scientific, Regulatory
& Industry Meetings

TOGETHER, WE ARE BUILDING A FUTURE WHERE EVERY RARE PATIENT THRIVES.

We are grateful to our community, partners, advocates, researchers, sponsors and supporters for your trust, collaboration and commitment to transforming the future of rare disease.

Our Theory of Change



Global Genes underwent a Strategic Planning Process in 2025.

The process included 4 phases: discovery interviews, modeling and plan development, stakeholder engagement and plan finalization. This deliberate process yielded our updated Vision and Mission and a new Theory of Change, charting a course for our future.

OUR VISION

Every rare patient deserves a future where the burdens of rare disease are eliminated.

OUR MISSION

Grow the ranks of patient leaders who will drive the next generation of rare disease breakthroughs. We equip rare disease advocates with the **tools, training, and trust to lead** — to activate communities, advance research, and widen the drug development pipeline - while **expanding and strengthening the rare disease ecosystem.**

THE NEED

Under-resourced patient advocates are leading and driving rare therapy development out of inescapable urgency — yet most don't have the training, tools or networks they need.

INTENDED IMPACT

The path toward every rare disease treatment begins with patient leadership. Science accelerates when patients lead. Systems change when the lived experience is central. Breakthroughs happen faster when advocates drive the process.

Through our programs, by 2035 we will equip 5000 patient advocates to change the lives of 5 million rare disease patients.

WHAT'S CHANGED

Global Genes was founded in 2008 to help build and unite the rare disease community. For almost two decades, Global Genes has been dedicated to connecting, empowering and inspiring the rare disease community.

Today's patient advocates are leading in ways that weren't possible even 10 years ago. Advances in therapy development, precision medicine and social media have laid the foundation for this sea change. Patient advocates are leading because of urgency - for themselves, for their loved ones, for their community. Global Genes' updated mission is to equip these heroic patient advocates, and help them develop vital ecosystems to achieve critical progress.

Programmatic Focus



EDUCATION EVENTS

RARE Advocacy Exchange;
RDDS; RAD Workshop



TOOLS & RESOURCES

RARE Concierge; RARE-X
platform; Content Library



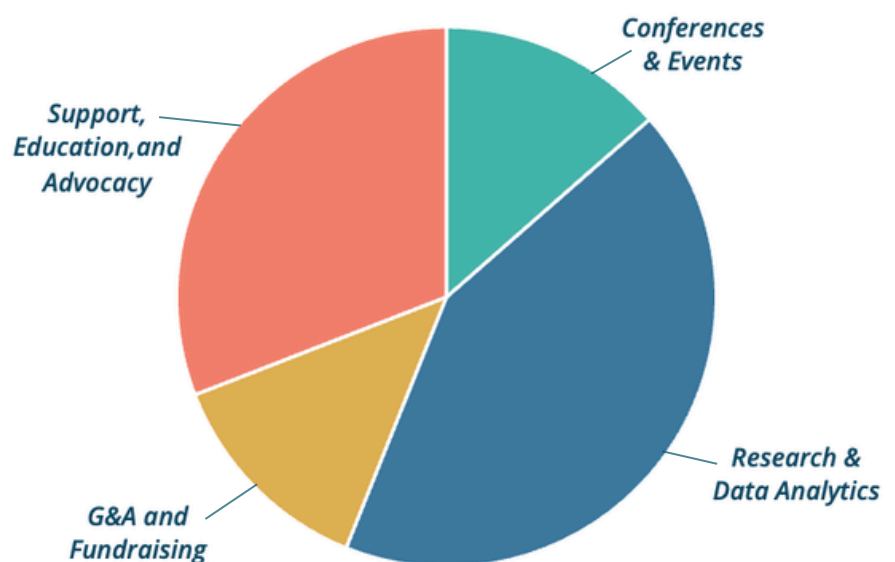
NETWORK BUILDING

Corporate Alliance; Global
Advocacy Alliance; Events



2025 Program Expense Breakdown

Total Expense: \$3,753,406



Rare disease advocates are doing extraordinary work - often without roadmaps, without research networks, and without the practical tools to move their communities forward at the speed their loved ones need.

That gap is the reason our programs exist.

Through education, direct patient support, network building, and research acceleration, Global Genes equips advocates with what they need to lead - not just to survive the diagnosis, but to drive the progress. In 2025, we served 1,590 patients and caregivers through RARE Concierge, equipped nearly 2,000 advocates through our virtual programming, and advanced research across 89 rare diseases through RARE-X. Every program we run is designed around a single commitment: that patient leadership is not a nice-to-have, it is the lever that makes breakthroughs possible.

Network Building



Global Advocacy Alliance

The Global Advocacy Alliance (GAA) is a community of non profit organizations and support groups who are committed to changing the way the world tackles rare diseases. Together, more than 825 organizations from around the world are working to build awareness for rare disease, create community and support networks, drive toward treatments and cures, provide direct patient assistance programs, educate healthcare professionals, and more.

GAA by the Numbers: 2025

- **833** total members
- **50** new advocacy organizations joined in 2025
- **2,007** members in a dedicated FB community
- **11**-member Leadership Council mentoring Alliance organizations
- **No cost membership** for any PAG or support group
- **Global membership footprint:**
 - 78% U.S. | 22% Non-U.S.
- **Quarterly meetings** plus year-round programming on leadership, fundraising, IEPs, grief, insurance, research education, and more

<https://globalgenes.org/about-us/global-advocacy-alliance/>



Corporate Alliance

The Corporate Alliance (CA) is Global Genes' collaborative leadership community designed to strengthen the rare disease ecosystem through co-creation, shared learning, and cross-sector partnership. Bringing together biopharma leaders, patient advocacy organizations, researchers, and innovators, the Corporate Alliance exists to elevate the practice of patient advocacy and accelerate more patient-centered approaches to research and drug development.

Through quarterly convenings and annual in-person collaboration, the Alliance fosters strategic dialogue, knowledge exchange, and collective action across the rare disease landscape. In 2025, the Corporate Alliance included **39 biopharma partners** committed to advancing more connected, community-informed therapeutic development.

<https://globalgenes.org/about-us/rare-corporate-alliance/>

2025 Collaborative Work: Peer-Reviewed Paper

'Beyond Engagement: Integrating Insights from Patients and Advocates in Rare Disease Drug Development' (paper submitted to peer-reviewed journal)

- The publication is the collaborative output of our Corporate Alliance and Global Advocacy Alliance. The paper makes the case for embedding patients and patient advocacy organizations earlier and more strategically throughout the drug development lifecycle - demonstrating how patient leadership can improve clinical trial feasibility, strengthen research readiness, accelerate therapeutic innovation, and create more patient-centered development pathways.
- Incorporates over **40** interviews and **2** roundtables, as well as a comprehensive literature review
- The submitted paper builds on the ***'Early and Often' White Paper*** written in 2024

Tools & Resources



RARE Concierge

Global Genes' RARE Concierge directs patients, caregivers, advocates, and healthcare providers to information, education, and support services they need. A one-on-one navigation service providing personalized support and trusted resources, RARE Concierge is our trusted entry point into the rare disease ecosystem for the 400M+ people living with a rare disease worldwide, as well as their caregivers and advocates seeking answers and connection.

Concierge by the Numbers: 2025

- **1,590** total inquiries in 2025
- **70+** countries reached, including 48 U.S. states
- **500+** distinct rare diseases supported
- **Areas of need:** support & financial resources, connection to patient communities, clinical trials, research, expert referrals

<https://globalgenes.org/rare-disease-patient-services/>

“Harlow starts treatments this year. We wouldn't be here without what you guys do at Global Genes. Because of the support and resources you have, my kid has a chance. It's not definite, right? But my kid has a chance, and it's because of you guys.”

— Daphne Graskewicz-Prado, Founder Kinslow Foundation, Harlow's Mom



Rare Disease Curriculum

The Rare Disease Curriculum was developed to address a gap in educational content and formalized programming to support knowledge and upskilling in rare disease. Developed in collaboration with the Rare Disease Diversity Coalition, the Rare Disease Curriculum provides foundational knowledge to Healthcare Professionals, Allied Health Professionals, Caseworkers and Case Managers, as well as anyone associated with rare disease, including Patients, Caregivers, Family Members and Advocates.

Key Features of the Curriculum:

- **7 Learning Modules** developed
 1. What is a Rare Disease?
 2. Unique Challenges to Diagnosis for Marginalized Communities
 3. Family Health History
 4. Genetic Diagnosis
 5. Newborn Screening
 6. Participation in Research and Clinical Trials
 7. Additional Topics on Living with a Rare Disease
- Learning Modules incorporate **Knowledge Checks, videos, and additional learning resources**
- Submitted for **accreditation through PIM** (Postgraduate Institute for Medicine)



Launching via PIM June 2026!



Freely available to the rare disease community!

Tools & Resources



QUICK GUIDE SERIES *for Rare Disease*

In 2025, we released a new series of 10 Quick Guides for the rare disease community.

These guides address some of the most pressing challenges faced by those affected by rare disease to:

- Empower individuals affected by rare or undiagnosed diseases with knowledge and actionable next steps to help navigate the complexities of a rare disease diagnosis, treatment options, and emotional well being
- Equip patient advocacy organizations with critical capacity building resources to better serve their communities

Global Genes 2025 Quick Guide Series responds to common needs we have heard for more information in the rare community. Think of them as the answer to frequently asked questions or pain points we have heard from our Rare Concierge, our online and in person communities and our Global Advocacy Alliance. Each one is complete with links to additional related resources across various media types, both those that are authored by Global Genes and from outside sources as well.

<https://globalgenes.org/press/global-genes-2025-quick-guide-series/>

Next Generation
Drug
Development
Resources

Interacting with
FDA Through
PFDD and Listening
Sessions

Medical
Gaslighting

Strategies for
Improving
Cooperation
Across PAGs



Guide to Creating
a Comprehensive
Fundraising
Strategic
Document

Knowing the Rights
of Your RARE
Child, (or Yourself)
– A Checklist

The Importance
of Getting
a Genetic
Diagnosis

How to Start a Rare
Disease Patient
Advocacy
Organization

Guide to
Fundraising



Securing a State
Proclamation
for Rare Disease
Awareness

Educational Events



RARE Advocacy EXCHANGE

A year-long virtual patient summit

March -
October
2025

The RARE Advocacy Exchange was a year-long virtual series that equipped advocates with the leadership skills, fundraising playbooks, regulatory know-how and peer connections they need to lead rare disease communities. Eight live sessions covered critical topics from values-driven leadership and innovative fundraising to engaging with the FDA's PFDD program and navigating ambiguous grief, and underscored the need for digital programming. The topics in this series were developed out of a needs assessment from within our Global Advocacy Alliance of >800 organizations.

<https://globalgenes.org/rare-advocacy-exchange/>

1,976

Unique
Registrants
Across All
Sessions

1,304

Live Attendees

9

Live Sessions
March-
October 2025

86%

of Attendees said
Sessions Met or
Exceeded
Expectations

81%

of Attendees said
they can apply
learnings to their
advocacy and
leadership
efforts

“ This session was **MANY** things. I **got the most from brainstorming**, and as the speakers talked, **I came up with three ideas to fundraise!** Such an impressive panel and the flow of the presentation was great. ”

“ Global Genes does it again!! They featured speakers where you walked away with action points. **It has been a very long time to feel this empowered.** ”

RAD RARE ADVOCATE
DEVELOPMENT

Brain Workshop

January 28-30, 2025
New York, NY

The RAD Brain Workshop is a three-day intensive meeting focused on the development of CNS therapies. This small-group format educational meeting is designed for rare disease patient advocates who are accelerating research in their neurological condition, and have already started funding and engaging directly in research projects.

This program was delivered in collaboration with the Rare Epilepsy Network (REN) and Mahzi Therapeutics.

25

Advocacy Groups
from Rare Epilepsies

29

Lectures
Over 3 Days

1

Facilitated Research
Roadmap Session with
EACH Patient
Advocacy Group

81%

of Attendees felt more
Confident in their
Presentation Skills
after the Presentation
Skills Session

100%

of Attendees felt more
knowledgeable about
Rare Research, Drug
Development and
Strategies for Patient
Advocates to Engage

Educational Events



RARE Drug Development SYMPOSIUM

 Advocates Powering Research

September 3 - 4, 2025
Boston, MA + Virtual

The RARE Drug Development Symposium brought patient advocates, scientists, regulators and biopharma leaders together to advance rare disease therapeutics, with a focus on help patients and advocates upskill their knowledge on the R&D process. In its first year in Boston, the agenda included 6 general sessions, 2 interactive workshops and 72 expert office hour sessions — plus the RARE Rounds program with the Termeer Institute, an exclusive bonus session pairing advocates with senior biotech leaders on biomarkers, regulatory strategy, commercial viability and research roadmaps. We are grateful for our collaboration with the Termeer Institute and Boston Children's, which provide critical connection and partnered content for these advocacy leaders driving research.

Watch sessions from 2025 here:

youtube.com/playlist?list=PLaA365_pZgMlOlB2Cgx6C-9mkEkd2tZhF&feature=shared

Plenary Sessions

- Patient Advocates Transforming the Landscape of their Disease
- Navigating the Drug Development Roadmap – An Overview
- Therapeutic Modalities and their relevance to your disease
- Pitch Perfect: De-Risking Your Disease to Drive Investment
- Developing Data Assets to Support Therapeutic Development
- Clinical Trials - Early and Often Practical Framework

84% of Attendees Left Feeling More Confident
Addressing Research Goals

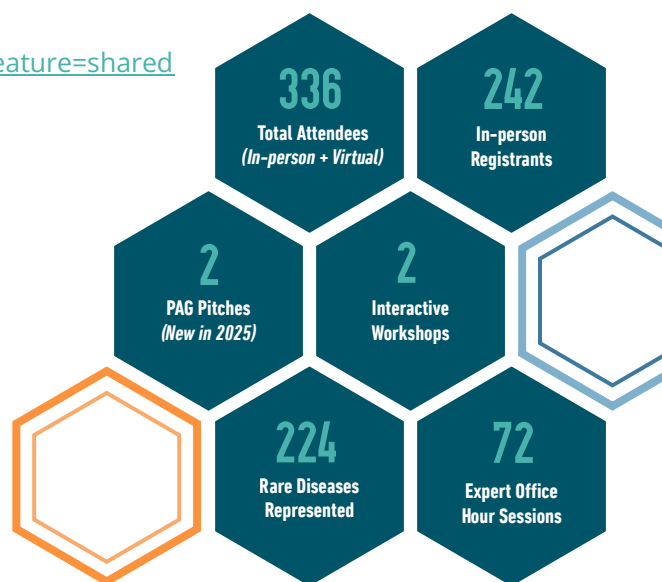
72% of Attendees Left with Actionable Next
Steps for Research

“ My colleague and I thought the Symposium was excellent, providing insight on relevant topics, thought provoking discussion and the opportunity to connect with leaders in the rare disease arena. **We left fully charged to address the work that lies ahead.**

- Anne Lenzin, President & Founder, US MEF2C Foundation

“ The symposium was an incredible opportunity to learn, connect, and collaborate, with a valuable mix of scientific, regulatory, and patient perspectives. I was especially pleased by how **every Global Genes staff member was welcoming, supportive, and eager to connect us with the right people, making advocates feel truly seen and empowered as essential partners in rare disease research and drug development.**

”



Research Enablement



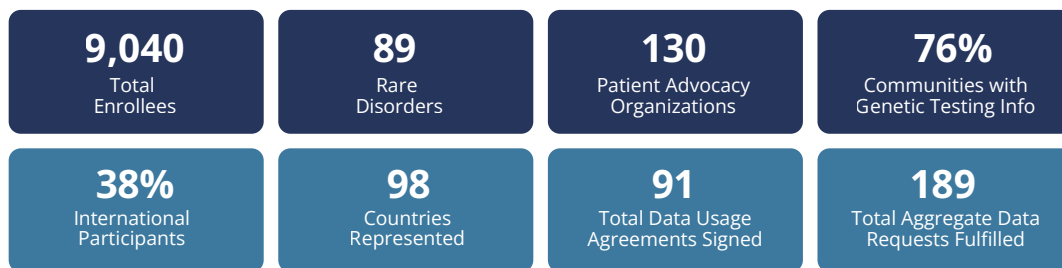
Research and Data Collection Program

The RARE-X research program has experienced tremendous growth, collecting vital patient data across 89 rare diseases in collaboration with 130 Patient Advocacy Groups. The RARE-X platform provides the foundation for robust rare disease research, with its standardized patient-reported and patient-provided data collection.

The research program also advanced work on sponsored projects across a number of areas including Homocystinuria, central disorders of hypersomnolence (Sleep Consortium), rare vision disorders (Vision Consortium), and natural history studies for STXBP1 and SYNGAP1.

With the support of a \$1.5 million grant from the Chan Zuckerberg Initiative, we are enhancing the RARE-X analytics capabilities through AI integration and the implementation of a large-language model for patient-reported data. Our vision is to democratize data analysis, extending access beyond data scientists to patient advocacy leaders and biologists working directly with affected communities.

<https://globalgenes.org/about-us/about-rare-x/>



RESEARCH ACCELERATION Fellowship Program

The Research Acceleration Program is an advisory program that helps identify needs in a community's clinical trial readiness, and – with input from clinical experts and drug developers – prioritize the gaps for investment. Using a structured assessment methodology, Global Genes guides patient advocacy groups, their key researchers, and scientific mentors through a process to develop a roadmap to advance research in their disease, and identify and evaluate projects. The advisory program runs for one year.

This year, reports for four Research Readiness Fellowships who were selected from Global Advocacy Alliance applicants in 2024 were completed: CureARS, MED13L Foundation, KARES Foundation, and the Rory Belle Foundation. For 2025, four additional patient advocacy groups were selected for the Research Acceleration Program: SHANK2 Foundation, MSL3 Syndrome Foundation, KIF1A.org and CureEDMD.

The University of Notre Dame has now joined Global Genes to advance this program in partnership.

“The fellowship has profoundly reshaped our organization’s research strategy by challenging the way we engage with KOLs. . . Moving forward, we are committed to fostering a more visionary, boundary-pushing research strategy, one that encourages KOLs to think beyond their comfort zones and aims for breakthroughs, not just progress. This shift in perspective will be pivotal in driving our future research endeavors.”

Thought Leadership



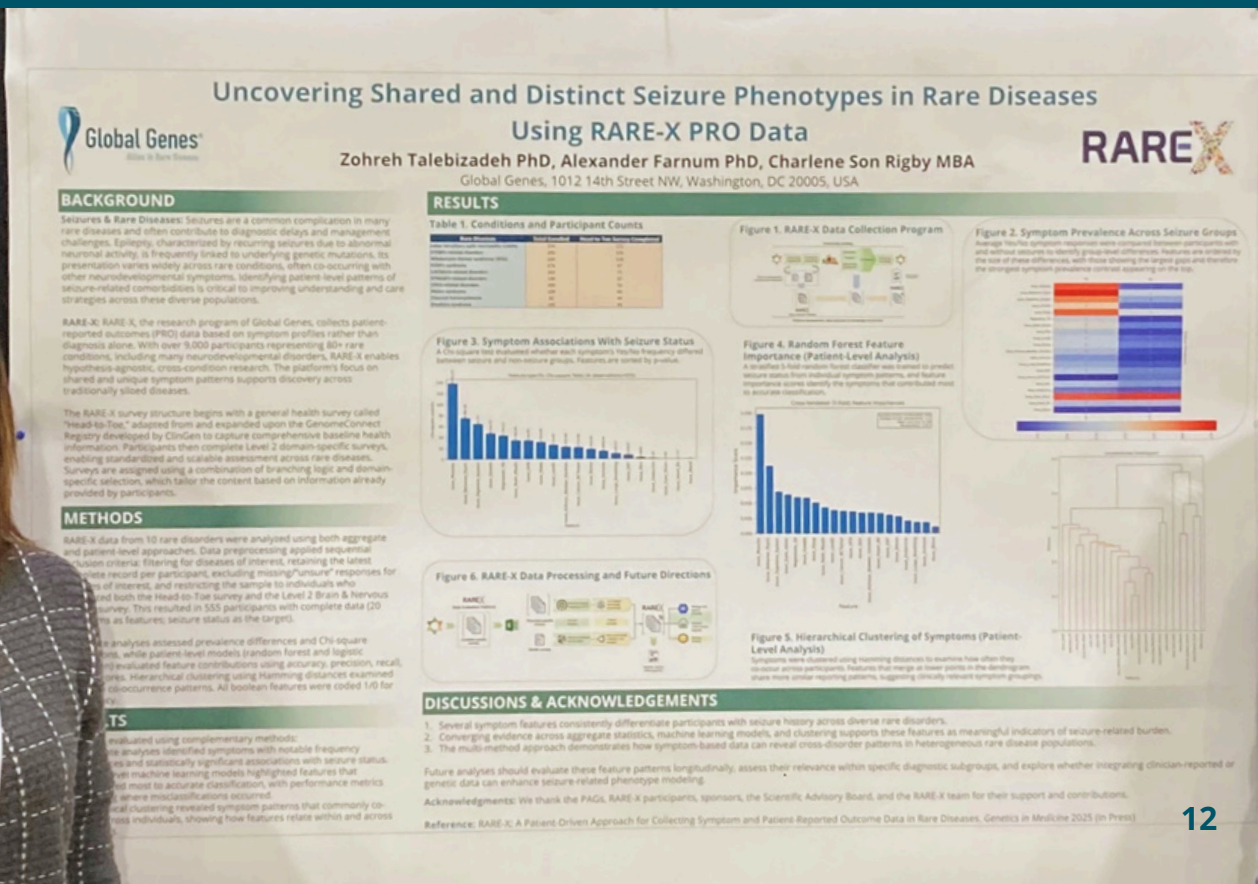
Publications

In 2025, two papers were published and two additional papers were submitted for publication. Further, three posters featuring RARE-X data were presented at scientific conferences.

Publications are a new focus for Global Genes and critical to advance durable patient engagement and patient-centered research work in the rare disease ecosystem.

- "Breaking barriers in rare disease research: The RARE-X Open Science Data Challenge as a model for collaborative innovation and community partnership" published in **Human Genetics and Genomics Advances** (July 2025)
- "RARE-X: A Patient-Driven Approach for Collecting Symptom and Patient-Reported Outcome Data in Rare Disease" published in **Genetics in Medicine** (December 2025)
- "Beyond Engagement: Integrating Insights from Patients and Advocates in Rare Disease Drug Development" – a collaborative output of our Corporate Alliance and Global Advocacy Alliance makes the case for early patient involvement in biopharma drug development (submitted December 2025)
- "Patient-reported outcome measures in central disorders of hypersomnolence: consensus of a sleep consortium/RARE-X expert working group", Trzupek, K., Wylds-Wright, C., Kuan, C., Sleep Consortium Expert Working Group, Jesteadt, L. (2026). SLEEP Advances, 7(1), 100462. (submitted November 2025)

<https://globalgenes.org/publication/>



Thought Leadership



In 2025, Global Genes continued its thought leadership work with presentations at various meetings including:

- Feb 2025** • **Bio-Neuroscience:** Charlene Son Rigby, CEO, spoke on *"Patient Driven Innovation"*
- March 2025** • **KARES Foundation - KDM5C Virtual Research Symposium:** Zohreh Talebizadeh, Sr. Director, Research, spoke on *"KDM5C Data Collection & RARE-X Updates"*
- March 2025** • **Autism Across the Life Span Conference:** Zohreh Talebizadeh, Sr. Director, Research, spoke on *"NDDs in Rare Diseases: A Symptom-Based Cross-Disease Analysis Using RARE-X Data"*
- April 2025** • **Health Evolution Summit:** Charlene Son Rigby, CEO, spoke on *"Investing in Rare Disease Treatments and Exploring Sustainable Business Models for Gene Therapies"*
- June 2025** • **Beyond Sleepy Conference:** Karmen Trzupek, Sr. Director, Scientific Programs spoke on *"DREAMS Portal: A Collaboration between Sleep Consortium and Global Genes"*
- June 2025** • **United Mitochondrial Disease Foundation Annual Conference:** Karmen Trzupek, Sr. Director, Scientific Programs, spoke on *"LHON Science: Moving Forward"*
- July 2025** • **Orphan Drug Summit:** Barry Weber, CISO/DPO, spoke on *"Navigating the Intersections: Privacy, Security, and Ethics in Rare Disease Data"*
- July 2025** • **Chicago Monogenic Epilepsy Summit:** Zohreh Talebizadeh, Sr. Director, Research, provided a *"RARE-X Overview"*
- Sept 2025** • Rare Disease Innovation, Science, and Exploration (RISE) Workshop Series sponsored by the FDA Rare Disease Innovation Hub:
 - **Workshop 1: On the RISE: Controls in Rare Disease Clinical Trials for Small and Diminishing Populations:** Karmen Trzupek, Sr. Director, Scientific Programs, spoke at *"Considerations for Control Decisions"* session
- Sept 2025** • **Notre Dame Science & Patient Advocacy Workshop:** Karmen Trzupek, Sr. Director, Scientific Programs, spoke on *"Scalability for Rare Disease Research"*
- Oct 2025** • **Alliance for Regenerative Medicine's Cell & Gene Meeting on the Mesa:** Charlene Son Rigby, CEO, led session on *"How Patient Groups are Driving Innovation and Research in the Cell and Gene Therapy Sector"*
- Nov 2025** • Rare Disease Innovation, Science, and Exploration (RISE) Workshop Series sponsored by the FDA Rare Disease Innovation Hub
 - **Workshop 2: Individualized Therapies on the RISE:** Charlene Son Rigby, CEO, spoke at *"Generating Evidence from Clinical Experience and Integrating Existing Data"* session
- Nov 2025** • **Alliance for Regenerative Medicine workshop on the Evolution of the Cell & Gene Therapy Sector:** Charlene Son Rigby, CEO, spoke on *"Evolution of the Patient Participation in Cell & Gene Therapies"*
- Nov 2025** • **National Society of Genetic Counselors:** Karmen Trzupek, Sr. Director, Scientific Programs, spoke on *"The Next Phase: The Evolving Role of Genetic Counselors in Clinical Trials and Research"*
- Dec 2025** • **Health and Human Services' 1st Rare Disease Meeting:** Secretary Kennedy announced expansion of the RUSP newborn screening panel for DMD and MLD - Charlene Son Rigby, CEO, represented Global Genes

A Special Thanks



To Our Board of Directors

We extend our deepest gratitude to the extraordinary members of our Board of Directors. These dedicated leaders provide invaluable guidance, expertise, and vision that help advance our mission while strengthening and empowering the global rare disease community.

We also extend our sincere appreciation to outgoing board members **Peter Goodhand** (GA4GH) and **Ilana Fogelman, M.D.** (C2N Diagnostics), for their many meaningful contributions and service.

Walt Kowtoniuk, Ph.D., Third Rock Ventures - *Board Chair*
Katherine Maynard, PWR - *Board Vice Chair*
Simon Frost, Oxbridge Capital Group - *Board Treasurer*
Christina Hartman, Orchard Therapeutics - *Board Secretary*
Nicole Boice, SEE RARE; Global Genes
Khrystal Davis, J.D., Texas Rare Alliance
Wendy Erler, Sarepta Therapeutics
Hugh Hempel, N=1 Collaborative
Ram Iyer, Enterprise Architect; FDA
Neil Kumar, Ph.D., BridgeBio
Kelly McVearry, Ph.D., BentoBio
Indu Navar, EverythingALS
David Pearce, Ph.D., University of South Dakota; IRDiRC
Morrie Ruffin, Adjuvant Partners
Alvin Shih, M.D., Imbria Pharmaceuticals
Charlene Son Rigby, M.B.A., Global Genes

To Our Scientific Advisory Board

We are fortunate to collaborate with an exceptional Scientific Advisory Board whose expertise helps guide our research initiatives, accelerate innovation, and support the development of our Next Gen Advocates, and broader rare disease programs.

Dan Chung, D.O., Beacon Therapeutics
Wendy Chung, M.D., Ph.D., Boston Children's Hospital
Scott Demarest, M.D., Children's Hospital Colorado
Ilana Fogelman, M.D., C2N Diagnostics
Rachel Groth, Ph.D., Neuren Pharmaceuticals Ltd.
Jeanette McCarthy, Ph.D., Genetic Epidemiologist
Tim Miller, Ph.D., Sangrail Biologics
David Pearce, Ph.D., University of South Dakota; IRDiRC
Steve Rodems, Ph.D., Travers Therapeutics
Eric Sid, M.D., NIH / NCATS
Alison Skrinar, Ph.D., Ultragenyx
Audrey Thurm, Ph.D., Boston Children's Hospital
Ashley Winslow, Ph.D., Odylia Therapeutics

To Our Global Advocacy Alliance Leadership Council

We are equally grateful to the members of the Global Advocacy Alliance Leadership Council, who serve as a vital voice for advocacy organizations around the world. Their leadership, collaboration, and commitment to patient communities help shape strategic initiatives that strengthen the global rare disease ecosystem.

Jonathan Bracey, MVA Society
Amber Denton, NBIA Disorders Association
Sarita Edwards, The E.WE Foundation
Barby Ingle, International Pain Foundation
Connie Lee, Psy.D., Alliance to Cure Cavernous Malformation
Megan Maack, Childhood Dementia Initiative
Lakeia Nard, Melanin Children Matter, Inc.
Christina O'Keeffe, Wiedemann-Steiner Syndrome Foundation
Isaac Okello, Raising Hope International Friends
Cheryl Petruk, Heal Canada
Michele Wise Wright, Ph.D., National Organization of African Americans with Cystic Fibrosis (NOAACF)

Looking Forward



As we move into 2026, Global Genes is evolving our programming to deliver on our new strategic plan and the bold goal at its heart: **equipping thousands of patient advocates to change the lives of millions of rare disease patients, and building the vibrant ecosystems needed to accelerate their work.**

We believe:

- *The path toward every rare disease treatment begins with patient leadership*
- *Science accelerates when patients lead*
- *Systems change when lived experience is central; and*
- *Breakthroughs happen faster when advocates drive the process*



To meet this moment, we are sharpening and expanding the programs the community relies on. Our leadership development work — anchored by the new Leadership Lab, the RARE Drug Development Symposium (in its 10th year), and the RAD Workshop — is being designed as a connected learning pathway that meets advocates wherever they are in their journey, from foundation start-up through clinical trial readiness. Our network-building work through the Global Advocacy Alliance and Corporate Alliance will deepen peer mentorship and cross-sector collaboration. Our tools and resources, including RARE Concierge, the Quick Guide Series, the Rare Disease Curriculum, and our content library, are being expanded to reach more patients, advocates, and providers — including underserved and international communities. And our research acceleration work, including the RARE-X platform's new AI-enabled analytics and the Research Acceleration Program, will give more patient communities a faster, more democratized path from data to discovery to therapy.

None of this is possible without partnership. To our donors, sponsors, biopharma partners, scientific advisors, Board of Directors, and the patient advocates who trust us with their stories and their leadership journeys, thank you.

Together, we continue to change what it means to live with a rare disease.



“ Having experienced Global Genes as a parent, a program participant, and now its leader, I am guided by a perspective shaped by every stage of the journey.

In 2025, our team and partners delivered remarkable impact, and I remain deeply committed to building on that progress and advancing our mission in 2026 and beyond.

- Charlene Son-Rigby, CEO, Global Genes; STXBP1 Mom

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THANK YOU