Global Genes

Allies in Rare Disease

2024 Year In Review

IMPACT

The resulting effect, positive or negative, that the actions we take have to change the physical or mental state of others around us—and therefore have the power to change the future path of an individual or group.

Merriam-Webster Dictionary

2024 In Review

Impact is a powerful and meaningful term, especially when describing positive work to support others.

Global Genes has always viewed the work we do as important, needed and critical in helping build a positive trusted community for rare disease patients, families and advocates. We want to help support patient advocates on their journey with the resources and tools they need to change the future path forward. We want to positively impact them and their efforts.

While the rare disease world looks very different than it did 16 years ago when we started on this journey, what hasn't changed is the need for allies, for community, connection, learnings, answers, and guidance. Global Genes has always taken its role seriously, and we continue to passionately work to positively impact more patients, remove barriers to education and support, for advocates and their families.

A Special Thanks

We would be remiss if we didn't acknowledge the extraordinary Global Genes Board of Directors, a group that has helped guide Global Genes, provide expertise and insights, and help us continue to evolve while staying true to our mission.

A special thank you to Board Chair Walt Kowtoniuk, Co-Chair Katherine Maynard, Khrystal Davis, Ilana Fogelman, Simon Frost, Peter Goodhand, Christina Hartman, Hugh Hempel, Ram Iyer, Neil Kumar, Indu Navar, David Pearce, Morrie Ruffin, Alvin Shih, to our outgoing board members, Caroline Loewy, Betsy Bogard and John Reynders, and to our newest board member Kelly McVearry.

We are also so very fortunate to have a group of scientific advisors who help guide our research efforts and participate in helping support our Next Gen Advocates.

You can learn more about them here: https://globalgenes.org/team-and-advisory-boards/

Mission Driven Pillars

We would like to share highlights from our work and impact this past 2024, and also provide you with insights into what we will be embarking on in 2025!

SUPPORT EDUCATION RESEARCH

Our three mission driven pillars:are continuing to serve our community in ways that help them throughout the different stages of their rare disease journey. We design our programs around those three pillars.

SUPPORT

Global Advocacy Alliance

Serving 800+ Patient Advocacy Organizations through community, connection access to resources, tools and education.

- Dedicated Facebook Page for trusted connection, Q&A, Support, moderated by Global Genes program directors
- A dedicated Leadership Council: a newly nominated set of leaders that is comprised of 11 advocacy organization leaders,
- Leadership Council serves as Advocacy advisors to Global Genes and also serves as mentors to organizations part of the Alliance
- Quarterly meetings
- Access to educational resources, with new programming developed to tackle navigating insurance, IEP's, Grief through to Research Readiness
- 78% US, 22% Global

RARE Concierge (Patient Services)

- Serving 1400 patients and families, from 58 Countries, 16% inquiries from overseas, representing over 400 diseases and many undiagnosed families.
- Areas of Need: Support and Financial Resources, Connection to other patients and relevant communities, Clinical Trials/Research, Experts

Mission Driven Pillars

EDUCATION

Events

Week In Rare: Advocacy Summit and Health Equity Forum

- Serving over 500 attendees in person and over 200 virtually
- 3 Tracks [Living with a Rare Disease, Community Building, Research], 29 sessions and 25 posters
- 100 expert office hours, providing 1:1 support for patients more deeply in the areas of the three tracks

Rare Drug Development Symposium

- Serving over 200 patient advocates, representing ~101 disease communities
- Agenda Includes
  - 5 plenary sessions
  - 12 interactive workshops
  - 150+ expert office hours [providing 1:1 support]
  - 17 Abstracts presented

"RDDS provided us with an invaluable opportunity to connect face-to-face with other PAGs. Being in the same room allowed us to discuss possibilities for collaboration & share insights on research paths that did or didn't work. These interactions showed the importance of collaborative efforts in rare." - RDDS Participant

Queer Conversations

- 7 Speakers
- Topics addressed: [a sampling]
- When healthcare providers fail to consider the intersection of rare disease and LGBTQ+
- Navigating the intersection of being LGBTQ+ in the rare community and isolation of being rejected by both communities
- Raising a child while rare and queer

Rare Disease Curriculum for Community Health Workers

- In collaboration with Rare Disease Diversity Coalition [RDDC]
- Seven (7) modules developed:
  - Introduction to rare diseases, Unique challenges to diagnosis for minoritized communities, Family health history, Genetic diagnosis and testing, Newborn screening, Participation in research/clinical trials, Additional topics on living with a rare disease (long-term care, financial/disability, mental health, advocacy etc.)
- Additional Distribution Partners Identified: National Association of Community Health Workers, National Urban League, National Health Council.

Mission Driven Pillars

RESEARCH ENABLEMENT

RARE-X Research Program & Data Collection Platform

Rare-X has now become one of the largest research data platforms, serving the rare disease community.

- 83 Disease Areas and over 120 patient community partners leveraging the platform
- 8,248 patient enrollees
- 98 countries, 38% outside of the U.S. International participation
- Supporting 3 research consortiums – Sleep, Huntington's Disease, and Pompe Disease
- As a result of our 2024 platform investment in Genetic Data Curation
  - 1676 curated genetic tests uploaded and curated
  - First set of translations in Spanish and Portuguese

Research Readiness Fellowship Program

- Supported 4 Organizations with indepth analysis of their current state of research readiness in a dozen different categories
- The program was supported by several Scientific Advisory Board members and industry KOL's to help in the delivery of this valuable program
- At the conclusion of the program a comprehensive report was provided to each of the Fellows that illuminated:
  - gaps in community readiness for trials
  - prioritization of those gaps and what to pursue
- www.globalgenes.org/rare-research-readiness-fellowship/

"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."

Corporate Alliance

- A membership program that supports the intersectionality of our work between biopharma, patient advocacy organizations and researchers.
- Membership in 2024 included 45 biopharma partners
- 2024 Collective work included:
  - White Paper to support feasibility in trials and the increasing and critical role of patient advocacy organizations from a business value perspective
  - To download the White Paper – Early and Often:
  - https://globalgenes.org/report/announcing-early-and-often-reimagining-patient-community-engagement-to-improve-clinical-trials-feasibility/
  - With over 60 lit reviews, over 40 interviews, two roundtables and input from a Corporate Alliance Working Group, this collective work is significant for the evolving role of patients in helping accelerate the development of rare disease therapies

2025 Insights

Global Genes will be embarking on a Strategic Planning Process and Plan development January - June 2025. This process will include 4 Phases as well as interviews and business modeling.

OUR GOAL

A reimagined Global Genes, supporting patient advocates and their communities more effectively and in new ways, by meeting patients where they are. While remaining a trusted ally and central point for the growing rare disease ecosystem.

Thank you for your partnership, your support and your commitment to the work we do. We value your contributions of time, talent and treasure and recognize that we would not be here without it.

We are very optimistic about the future. Opportunities are growing, science is evolving, and with almost 11,000 rare diseases, there is no shortage of work that needs to be done.

We thank you in advance for staying the course with us, and we look forward to continuing to be a trusted partner and resource for those in need for years to come.

A life is not important except in the impact it has on other lives. Jackie Robinson

Always in hope,

Charlene Son Rigby
Chief Executive Officer

Nicole Boice
Founder, Chief Mission Officer

THANK YOU