Global Genes®
Allies in Rare Disease

Annual Impact Report

2022

ABOUT GLOBAL GENES

Global Genes is dedicated to eliminating the burdens and challenges of rare diseases for patients and families globally.

In pursuit of that mission, Global Genes connects, empowers, and inspires the rare disease community to stand up, stand out, and become more effective on their own behalf — helping to spur innovation, meet essential needs, build capacity and knowledge, and drive progress within and across rare diseases.

Together with our partners, we are here to help the more than 400 million people worldwide living with rare diseases by:

- Activating, inspiring, supporting, and equipping communities to drive progress
- Collaborating and connecting people to resources and communities based on their needs
- Sharing insights and knowledge to support global innovation, care, and access
- Encouraging and enabling equitable participation in research, development, and advocacy
- Reducing diagnostic odysseys and accelerating progress toward treatments and cures

400M+
People suffer from a rare disease globally
1 in 10 Americans

There are
10,000+
Distinct types of rare diseases

~7 years
The average time it takes to receive an accurate diagnosis for a rare disease

8 out of 10
rare diseases are genetic

3 out of 10
children with a rare disease won't live to see their 5th birthday

95%
of rare diseases lack FDA approved treatment

MEMBERSHIPS

Global Advocacy Alliance

Established in March 2022, the Global Alliance is a global community of non-profit organizationss and support groups who are committed to changing the way the world tackles rare diseases by collaborating to realize positive change.

380+
rare disease organizations

28
countries represented

340+
diseases served

22
skill building sessions

> *I've connected with peers all over the world, and the challenges I face with my daughter's rare disease they face on the other end of the world as well. While the solutions may look different, the problems are the same. Being a part of the Global Advocacy Alliance helps to call attention to those needs, and work together with a group of advocates to solve those challenges – that is why I wanted to be part of the Alliance*
>
> *~ Global Advocacy Alliance Member*

Learn more about Global Advocacy Alliance

RARE Corporate Alliance

The RARE Corporate Alliance is a partnership between rare disease industry stakeholders committed to forging and supporting collaboration between industry and patient communities.

80+
industry partners

> *One of the most important contributions Global Genes delivers is in bringing the rare disease communities and pharmaceutical companies together to advance shared goals. The Global Genes Corporate Alliance enables those of us working in rare disease to learn from each other, make connections and identify areas of opportunity to collaborate. Together, we will be able to do more for all of the families impacted by rare disease.*
>
> *~ Wendy Erler, Alexion, Co-Chair of RARE Corporate Alliance*

Learn more about RARE Corporate Alliance

EDUCATIONAL EVENTS

Global Genes events connect stakeholders from the rare disease community and facilitate collaboration among patients, caregivers, researchers, and industry professionals. In 2022, we held 4 events that impacted thousands of people, both in person and virtually.

RARE Patient Advocacy Summit

Global Genes convenes one of the world's largest gatherings of rare disease patients, caregivers, advocates, healthcare professionals, researchers, partners, and allies at the RARE Patient Advocacy Summit.

> *Connecting with people who share the same passion for the rare community as I do reignites my drive to continue to do great work. I was beyond inspired by the people, stories and dedication and ultimately I left there feeling like I need to do more. It was a tremendous opportunity and I am so grateful to have been a part of it. Thank you.*
>
> *~ Erin Moriarty Wade*

647
attendees

350K
total social reach

91%
made important connections with others

See some more stats from last year's event

Champions of Hope

The RARE Champions of Hope Awards honors and recognizes champions in the rare disease community. In 2022, we recognized champions in 7 categories:

- Adrian Goretzki, RARE Champion in Advocacy – Individual
- Lysosomal Storage Disorders Support Society – India, RARE Champion in Advocacy - Foundation
- Dr. Yuriy Stepanovskiyy, MD, PhD, RARE Champion in Medical Care and Treatment
- Arthur A. Levin, PhD, RARE Champion in Industry
- Nell Choi, RARE Rising Star
- Richard Horgan, RARE Champion in Science and Technology
- Cynthia and Joe Lang, RARE Founder's Award

333
attendees

196
advocates and organizations in 7 categories

RARE Drug Development Symposium

Global Genes, in partnership with the Orphan Drug Center of the University of Pennsylvania, hosted the RARE Drug Development Symposium to give patient advocates and organizations an opportunity to learn about the drug discovery and development process, how they can cost-effectively advance research, attract partners, and use innovative approaches to accelerate progress. Sessions focused on:

- Accelerated clinical trials
- The use of AI-driven screening platforms
- Emerging models and partnerships, and
- Fostering successful connections between rare disease stakeholders

140
attendees

192K
total social reach

92%
of respondents were able to reach specific and actionable solutions to address the challenges laid out in the workshops

See some more stats from last year's event

RARE Health Equity Summit

The RARE Health Equity Summit, hosted in partnership with the Rare Disease Diversity Coalition (RDDC) convened stakeholders in the rare disease community to identify collaborative efforts to address inequities in the care of patients with rare diseases, specifically focusing on reducing disparities, discrimination, and biases felt by patient communities based on factors including socioeconomic status, ability, geographic location, religion, race, ethnicity, gender identity and/or language.

279
attendees

84K
total social reach

86%
of respondents felt this event equipped them with a greater understanding of health equity practices and methods

See some more stats from last year's event

> *"This whole event was amazing and so well organized. Usually there are a couple of sessions I'm not interested in but I never wanted to leave because every session was so good and interesting!"*
>
> *~ Nicole Kressin*
> *Rare Diseases Clinical Research Network*

PROGRAMS AND REPORTS

Global Genes programs encourage patients and advocates to connect around the globe. Through these programs, members of all disease communities engage with each other, share resources, get involved, and are well on their way to becoming effective advocates on their own behalf.

RARE Concierge

Our Patient Services arm, RARE Concierge, provides information, resources, and connections for those on the rare disease journey. Individuals ask unique rare disease-related questions and get information relevant to their needs at no cost, including:

- Identifying clinical trials/research studies
- Offering disability, financial, and mental health resources
- Connection to resources, including finding a specialist, center of excellence, or patient advocacy organization
- Finding support while getting a diagnosis, including genetic testing & counseling services

In 2022:

1,400+
were assisted through RARE Concierge

40%
requests came from undiagnosed families

>450
diseases supported

inquiries came from
44
countries and 44 U.S. states

> *Global Genes was able to complete a deep dive into the gene in days which in turn may have saved [his] life. He needs further genetic testing, but there are ongoing trials for potentially curative approaches. [He] probably would have accepted the standard of care which is to do nothing until you are really sick. We are all so grateful that you have devoted yourself to selfless and meaningful lives.*
>
> *~ Friend of a patient (U.S.)*

RARE Portal

The RARE Portal is a private online community launched in 2022 for patients, caregivers, members of non-profit patient advocacy organizations, and academia meant to make meaningful connections with others. Participants share inspiring stories, resources from their own organizations, and browse discussion topics including financial advocacy, diversity and health equity, mental health, gene-based diagnosis, and more.

> *I never knew how good it would feel to be part of this community, where everyone has been through so much for so long alone or thinking we were alone when all the while there is this humongous group all over the world going through the exact same struggles with only the name of the disease changed between us.*
>
> *~ RARE Portal parent, Founder of The Healthcare Navigation Report*

3,000+
posts created

8,000+
views on resource library

48%
open rate for community news

RARE Compassion Program

The RARE Compassion Program provides an opportunity for medical students to learn about the unique needs and challenges individuals and their families face living with an undiagnosed or rare disease. The program offers students from around the world a chance to build compassion, understanding, and empathetic communication skills, and gain interest in specializing in fields most relevant to rare disease.

> *The discussions really made what I was learning in school feel real. It's easy in medical school to get stuck behind books memorizing the details of 'rare' conditions, but there are indeed people living with these diseases. This knowledge now that I am learning could help make a rare disease diagnosis in the future.*
>
> *~ Erin Kim, 2022 medical school participant*

698
students and rare individual/family

861
rare families

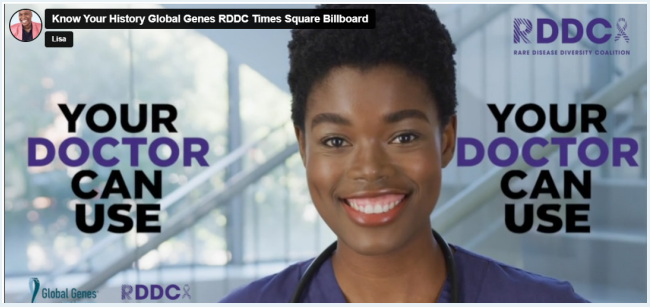
206
medical students

>1,300
hours spent together

Know Your Family History

The Know Your Family History program, through a partnership with the RDDC, engages and educates underserved communities of color, particularly Black and Hispanic, about the importance of talking to their family about their health history and assessing whether genetic testing is right for them.

Explore the educational resources in English or Spanish



> *Underserved populations have a longer diagnosis process. If we can help the underserved population get diagnosed sooner, then treatments can happen quicker, and hopefully that can have an impact on their quality of life.*
>
> *- Jenifer Waldrop, Executive Director, RDDC*

Know Your History Global Genes RDDC Times Square Billboard

RARE Patient Impact Grants

Global Genes provides RARE Patient Impact Grants to rare disease patient advocacy organizations to make specific, tangible differences for the communities they serve. These funds support initiatives to address health equity within the rare community, make in-person meeting and community building opportunities possible, empower people through financial advocacy and resources, and provide urgent relief for rare disease patients and families impacted or displaced by the war in Ukraine.

- RARE Meet-Ups
- Health Equity in RARE
- Financial Advocacy
- RARE Relief

29
grants

$415,483.50
awarded

177,000+
reached

Reports

Global Genes creates these reports to provide a summary of activities for the coming year, and to recap events and programs of the past year.

NEXT Report 2022
The NEXT Report forecasts the developments, trends, and the progress being made across the rare disease landscape.

RARE Drug Development Symposium Report 2022
This report summarizes conversations from the 2022 RARE Drug Development Symposium. Topics include accelerating rare disease research, drug repurposing, innovating clinical trials, and more.

Health Equity Report 2022
The Health Equity Report includes insights and challenges from stakeholders from across the rare disease community, highlighting issues that need to be addressed for underrepresented people in the rare disease space.

PIE4CNS Report 2022
The PIE4CNS Report summarizes the two phases of the Patient Identification, Inclusion & Engagement for Central Nervous System Conditions (PIE4CNS) Initiative, which aims to address key gaps in identifying, diagnosing, and developing treatments for rare neurological/ central nervouse system conditions.

LOOKING FORWARD

Global Genes and RARE-X merged their organizations on December 31, 2022. The new, combined organization provides patient advocates with a complete and integrated set of essential services to continue their work towards support and awareness, and accelerates them towards drug development through a collaborative approach of biopharma, researchers and funders, along with patient health data collection as a central core.

In 2023, the shared mission and vision of Global Genes and RARE-X focuses on next generation advocacy, supporting individuals who can utilize the tools and resources needed to accelerate advocacy efforts and also support opportunities to drive research and therapy development.

Next generation advocacy requires a different and expanded type of support. Global Genes is meeting the needs of advocates through expanded tools, relevant connections, and by providing more resources to support the increased opportunities for advocates as research drivers. To simplify how our services and capabilities will be built and organized we have identified three pillars:

- **Support** - for patients and developing communities
- **Education** - robust tools, events and educational opportunities
- **Research and Development** - focusing on research and data enablement, with platform services to support these critical efforts.

Global Genes®
Allies in Rare Disease

+ RAREX

Powering Next Generation Advocacy

28 Argonaut, Ste. 150
Aliso Viejo, CA 92656
+1-949-248-RARE (7273)

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www.GlobalGenes.org
@GlobalGenes