Global Genes®
Allies in Rare Disease

Annual Impact Report

2020

About **Global Genes®**

Global Genes is a 501(c)(3) non-profit organization dedicated to eliminating the burdens and challenges of rare diseases for patients and families globally. In pursuit of our mission we connect, empower, and inspire 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. We serve the more than 400 million people around the globe and nearly 1 in 10 Americans affected by rare diseases. If you or someone you love has a rare disease or are searching for a diagnosis, contact Global Genes at 949-248-RARE or visit our resource hub at www.globalgenes.org.

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Craig Martin

LETTER FROM THE CEO

Dear Friends and Colleagues,

This past year called upon Global Genes and the entire rare disease community to continue to have impact despite adversity, and to generate additional impact in response to adversity.

Impact despite adversity meant maintaining excellence and support for ongoing programs, partnerships and services while a pandemic shut down many of the more traditional pathways to engagement our community has come to rely on.

Virtual was the new reality. Yet we experienced unprecedented, global involvement in our Patient Advocacy Summit, and we saw increased participation, virtually, in the events and programs we were able to continue to offer in 2020.

We were also able to forge and launch a new partnership with Rady Children's Institute for Genomic Medicine designed to improve the diagnostic odyssey and increase access to rapid whole genome sequencing for rare disease families around the country.

Our Foundation Alliance grew from just over 500 foundations in 2019 to more than 750 by the end of 2020; a testament to the rare disease community's collaborative nature and a recognition of the importance of banding together in challenging times.

Adversity also teaches and often requires us to pivot and adapt to meet new needs. When COVID-19 and pandemic response efforts began to impact the rare disease community's access to critical services, trials and treatment, we responded, launching initiatives and working with other rare disease groups to identify and address pandemic-related issues affecting continuity of research and care.

Thanks to the committed, compassionate support of several of our Corporate Alliance members, Global Genes was able to take in requests from a wide variety of organizations from around the globe and issue more than $500,000 in RARE Impact grants, reaching more than 4,500 families and 40 foundations.

Our work in response to the pandemic, along with other developments, further highlighted inequities in access to clinical research, drug development, diagnostic tools, trials and treatment. We initiated efforts to assess and identify where communities of color may be disadvantaged or underserved in the diagnostic odyssey.

We're also proud to have worked with the Black Women's Health Imperative and joined 24 other patient and professional organizations in forming the Rare Disease Diversity Coalition. The Coalition is now working actively to address a range of equity, inclusiveness and diversity issues spanning the continuum from research to diagnosis to access to treatment and care.

While we won't look back fondly on 2020, we will at least acknowledge and appreciate the way in which the rare disease community, with help from our RARE Corporate and Foundation Alliance members and other valued partners, came together to face and address adversity. In so doing, we showed our strength and resilience, and built an even stronger foundation for progress in the future.

With gratefulness and hope for the future,

Craig Martin
Interim CEO
Global Genes

Rare Disease Landscape
by the Numbers

RARE Foundation Alliance Members
750+

RARE Corporate Alliance Members
100+

The number of rare diseases has **steadily increased** from **7,000** to an estimated **8,000** worldwide.

This increase continues to highlight the unmet needs and barriers to care for patients and caregivers living with the burdens of rare disease.

400M+ people worldwide, and nearly **1 out of 10** Americans have a rare disease

It takes an average of **7.3 years** to receive an **accurate diagnosis** of a rare disease

It takes an average of **7 clinicians** to **accurately diagnosis** a rare disease

Continuity of Care **& Research**

Addressing the Impact of COVID-19 on the Rare Disease Community

Global Genes is proud to have helped lead the way to ensure the continuity of care and research for the rare disease community during the onset of COVID-19. Through our efforts to convene members of our RARE Corporate Alliance, RARE Foundation Alliance and partner organizations, such as EveryLife Foundation, Haystack Project and the National Health Council, we focused on ensuring patient access to in-home infusions and nursing services, as well as critical access care for high-risk patients.

Thanks to your assistance, we have been able to make a significant impact on the lives of individuals living with CF and the families of children with CF who have been experiencing financial difficulty because of situtions caused by COVID-19.

-MilesforCysticFibrosis

In addition to clinical care, many rare disease patients rely on clinical trials as part of their regular care.

With 67.3% of clinical trials disrupted by COVID-19, and 18.4% of trials delayed, many rare disease patients were left without care or progress to find treatments.

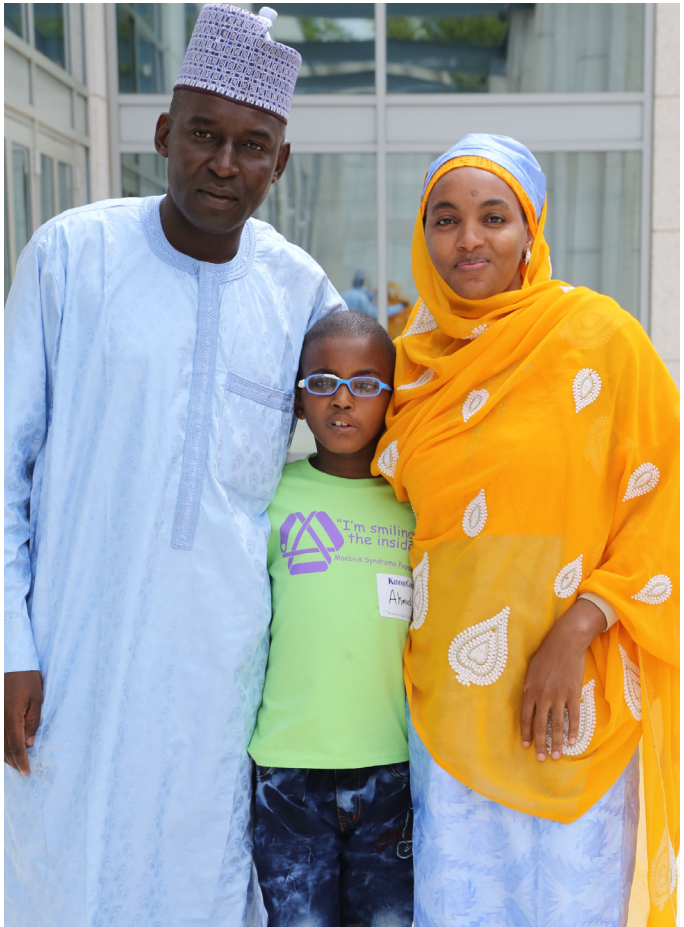
By continuing to connect and convene industry members, researchers, and patient advocates, we were able to help assess the impact of COVID-19 on late stage and human cell line research. Additionally, we fostered direct investments in research and development with innovative new non-profit models, supported FDA-approved pivots to de-centralized trials, and educated advocacy organizations on what to do next.

Continuity of Care RARE Patient Impact Grants:

40 grants awarded, $515,000 total funds distributed
More than 4,500 rare disease families supported

In 2020, the United States and other health systems were forced to undertake extreme measures to stop the spread of COVID-19 and prepare for a potential surge of demand on hospitals and health care providers. As these measures were implemented, and others contemplated, an increasing and alarming number of areas restricted, or denied entirely, access to care for rare disease patients.

We recognized the urgent need to identify where the gaps in care were occurring, and address and aid patients who were facing or would soon face limited or no access to care because of the current COVID-19 pandemic. We established the Continuity of Care RARE Patient Impact Grant to support rare disease families with access to care such as telemedicine, distance learning, educational resources, and access to psychosocial and mental health services. We were able to award a total amount of **$515,000** through the distribution of grants to **40** foundations and individuals. **Over 4,500** families in need were able to benefit from these efforts.



Shortening the **Diagnostic Odyssey**

Global Genes continues to address barriers and challenges of rare disease patients to obtain a timely and accurate diagnosis. In 2020, we partnered with the Child Neurology Foundation to better understand the journey through diagnosis from families and clinicians. This data helped to validate that families supported by Global Genes travel faster, fare better and have more confidence in the end results of their diagnostic odyssey.

31% of Global Genes families **got a diagnosis in less than a year**, vs 48% overall

None of the Global Genes families that responded **are still waiting for a diagnosis**

Global Genes respondents report **93%** have a high level of or some **confidence the current primary diagnosis is correct**, vs 85% overall

47% of Global Genes families **received at least 1 wrong diagnosis** vs. 36% overall

None of the Global Genes families reported having **more than 3 incorrect diagnosis**

52% of Global Genes families report spending **0 hours researching** to find diagnosis vs 30% overall

Full results can be found by visiting The Child Neurology Foundation

We have been able to help so many families and individuals that we would not have otherwise been able to help. We have been able to help them with access to care, access to activities they would not otherwise had the opportunity to do because of the support from this grant.

-ALS Ride for Life

Rady Children's Institute for Genomic Medicine Partnership:

As gene-based diagnostics are shortening the path to an accurate diagnosis, the risk of disparities in service and support have increased. To reduce those disparities, Global Genes was thrilled to announce a new partnership with Rady Children's Institute for Genomic Medicine (RCIGM) to develop a next-generation support network for families with gene-based diagnosed rare diseases.

Through this novel partnership, Global Genes will work with RCIGM to connect patients and caregivers to needed services, support, education, and resources regarding genetic testing. Together, RCIGM and Global Genes aim to reduce the time it takes to receive gene-based diagnoses and bring parents and patients the support they need, wherever they are in the country.

Health Equity, Inclusivity & Access

Access to Critical Therapies:

The Access to Critical Therapies (ACT) collaboration was founded in late 2018 based on evidence of significant unmet need and care variability in patients with rare diseases.

We wanted to ensure the rare disease patient perspective was fully considered as part of broader public discussions and policy formation that could impact and improve patient access to needed therapies, immediately and in the future.

In 2020, we published a report, entitled "*Guiding Principles of Rare Disease Care and Patient Access*," that characterizes 5 fundamental expectations for rare disease patient care that all health stakeholders should acknowledge and act upon. These expectations are:

1. **Timely and sustainable access to diagnostic testing that rapidly informs appropriate patient care and treatments**
2. **Timely and sustainable access to the highest quality care and most effective treatments that address underlying disease or key symptoms**
3. **Value assessment processes that provide timely and sustainable access to current and future therapies for which patient-centric benefit is the deciding factor**
4. **Fulfilling quality of life while lessening the disease burden for both patients and caregivers**
5. **Standards of care that reflect acceptance of each patient's uniqueness and equality for all patients regardless of disease rarity**

In normal times, our families struggle to meet the demands of their child with GRIN2B in addition to their other children, work, home, etc. Everyone is hanging on by a thread with schools being closed and losing access to therapies. With no end to this pandemic in sight, many families are losing hope. These grants gave 17 families a ray of hope and a chance to breathe. We're so happy we were able to lighten the load a bit for these families and give them something positive to focus on.

- GRIN2B Foundation

Through this and other partnership efforts, we will continue to advocate for patients and ensure that their perspective is front and center, and included in all decisions related to access to treatments and care.

Rare Disease Diversity Coalition:

The Black Women's Health Imperative launched the Rare Disease Diversity Coalition to address the extraordinary challenges faced by rare disease patients of color. To identify the most pressing problems—and the most promising potential solutions—the Steering Committee formed four dedicated working groups: Delays in Diagnosis; Government Regulation, Legislation, and Policy; Patient and Provider Education and Engagement; and Research and Clinical Trials. Global Genes is proud to participate alongside 24 other patient and healthcare professional organizations to help eliminate the burdens of rare disease by connecting, empowering and inspiring members of the community.

RARE Concierge:

RARE Concierge is an important component and driver of our efforts to serve those in the rare disease community who may be undiagnosed or are still relatively early in their journey. Additionally, RARE Concierge provides support for those who don't have a significant support system in place or know where to turn next for answers.

RARE Concierge provides patients and caregivers with a vital entry point into the rare disease ecosystem, direction on how to connect to or build a community, and guidance to help them find and access Global Genes and many other organizations' information and resources. Perhaps most importantly, it serves those who might otherwise be overlooked, under-represented and left to fend for themselves at the most critical and emotionally challenging point in their lives. In 2020 and into 2021 we are making great strides in enhancing our resources and technology to accelerate connecting patients with services and resources to help shorten their diagnostic odyssey and improve their access to quality care.

The COVID19 pandemic has brought substantial hardship to many members of the pediatric epilepsy surgery community. IN additional families struggling with basic living expenses, but distance learning is especially difficult for many children in our population in light of their multiple disabilities. This grant has made distance learning more accessible for many members of our community.

-The Brain Recovery Project

Where do I begin. Because of the support from Global Genes, we have been able to support so many of our families during this time of need. Many of our famlies have been desperate for support in virtual learning and telehealth as well as other ways to keep their child with AS engaged, active and involved. Many of these familie have shared that this has made such a huge impact in the overall quality of life for not just their child with AS, but them as well. One parent has three children with AS and they had no way to do their virutal learning becuase they had no computers or IPADS. Now each of them have one and they can conenct with their teachers, therapist and friends. Thank you Global Genes for showing up in a time that the need was so great.

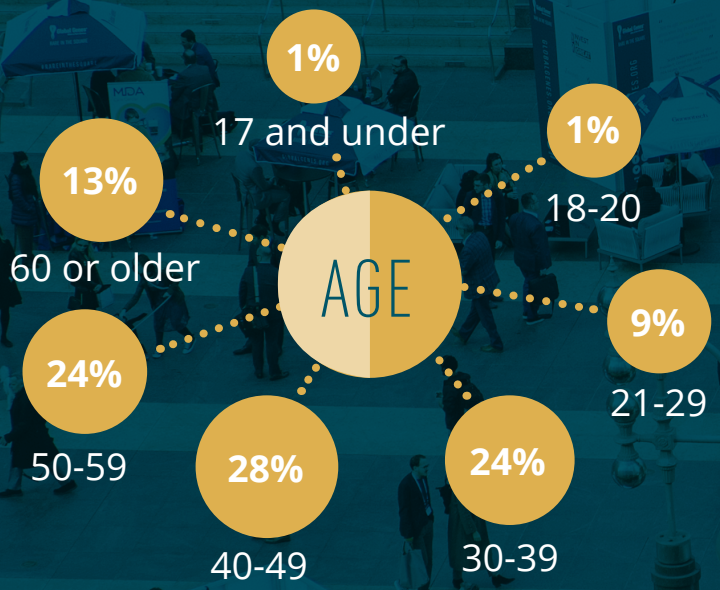
-Angelman Syndrome Foundation

GLOBAL GENES **EVENTS**

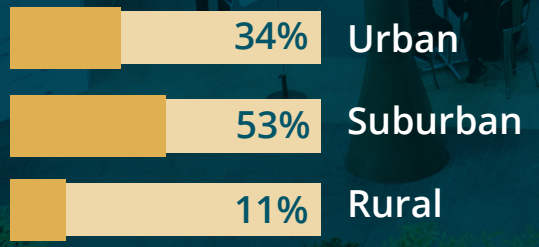
7 events

3,497 total registrants from over **40 countries**

650+ rare diseases represented



Region Type



RARE Leader Meetups:

In 2020, as responses to the COVID-19 pandemic continued to limit our ability to connect in person, we convened six virtual RARE Leader Meetups for our RARE Foundation Alliance members to continue to connect, empower and inspire the rare disease community. **Over 225 foundation alliance member organizations** participated in the Meetups, which included the following topics: Emotional Support in Crisis, Pivoting to Virtual Events, Creative Fundraising, Balancing School and Work from Home, Data Strategy: Driving RARE Research Forward, and Restarting Clinical Research.

Role

- **21%** Patient
- **16%** Caregiver
- **3%** Academic Researcher
- **1%** Government
- **31%** Foundation or Nonprofit Organization
- **11%** Industry: Non-Researcher
- **3%** Industry: Researcher
- **11%** Other

Global Genes **in the News**

Global Genes works to overcome barriers in rare-disease trials
– **March 31**

Outreach, Cultural Competency Key to Clinical Trial Diversity
– **April 6**

Designing Patient-Centered Clinical Trials Using Diversity, Inclusion
– **April 28**

Global Genes and Advocacy for Patients with Rare Diseases
– **May 20**

Future of Rare Diseases: Improving the Lives of Patients and Caregivers Living with Rare Diseases
- **June 26**

Sleuthing For Answers On Rare Diseases
- **July 6**

Diversity in Clinical Trials: Path to Achieving Health Equity
- August 19

Patients Are at the Heart of Cell and Gene Therapy
- August 13

Their lives are in other people's hands: Young people with immune conditions fear coronavirus - CNN
- August 19

COVID-19 Silver Linings
- **November 17**

Expanding the Spectrum of Clinical Trials
- **November 17**

Stories from the **Rare Disease Community**

My son, Ezra, was diagnosed with GRIN1 back in March, right at the very start of the Covid-19 pandemic.

It was an extremely hard time for us. We were given a phone call and told this diagnosis and then basically the whole country shut down. We were unable to go to follow up appointments, start therapies, etc. At the same exact time, my sister, who lives with us and is the only other person in my home to help with bills, lost her job. Like so many others, we were struggling emotionally and financially.

The grant we received from the CureGRIN Foundation was a lifesaver - literally. It helped us pay almost an entire month of rent which allowed us to catch up on our other bills and get back on track. Without it, who knows what would have happened. Ezra is safe and comfortable in his home as a result. We are
extremely grateful to have been able to receive this. Thank you to everyone who made this happen for us.

Thank you,
Natalia Gordon

We found out in 2016 that Juniper had Grin 1. We've been rolling with the punches ever since. The older June has gotten the harder it's been for us to tote her around, both in and out of the house. There's a lot of carrying involved on our end since she can't walk.

In February, we moved into our dream house, a one level ranch that would allow June much more room to get around on her own terms, as well as a more accommodating situation for storage of all of her equipment and easing the burden of carrying her up and down stairs.

After living here a month, COVID hit and threw us all for a loop. Not only did school become virtual which was in and of itself a challenge for a special needs child, I went from being an 18-year salaried employee to being paid an hourly freelance wage that significantly affected our income.

Trying to find additional income in the midst of a pandemic was next to impossible. Having the Continuity of Care Rare Patient Impact Grant awarded to us helped tremendously to be able to pay another month's worth of mortgage during a time when I didn't know if would even be able to stay where we are. We are very grateful for everything the Cure Grin Foundation is doing.

Abby and Mike Wood

Ramesh is trying to tackle another aspect that patient organizations hoping to develop gene therapies for ultra-rare diseases face. He's joined with several drug developers, contract research organizations, and other stakeholders to create an open-source system that can guide patient organizations through all the necessary steps for developing a gene therapy. He's raising money now for a pilot for three diseases including GPX4. The hope is that the system could be used to walk any organization through the process of developing a gene therapy and connect them to researchers, vendors, and other resources needed throughout the process.

"Patient foundations typically spend months trying to understand what to do, and then once they understand what to do, they spend a few more months deciding if this is the right investment for their money at this point in time," said Ramesh. "This will short circuit all of that. What used to take months will be shortened to hopefully days, if not hours, to make these decisions."

Global Genes®
Allies in Rare Disease

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