



Global Genes[®]

Allies in Rare Disease



Global Genes Overview 2018

Presented by: Global Genes

RARE Stats

RARE DISEASE TYPES



7,000+

distinct types of rare diseases exist, with more being discovered every day.

THE CAUSE

80%

of rare diseases are caused by faulty genes.



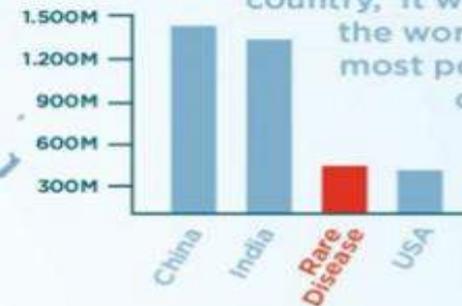
RARE DISEASE EFFECT

30 MILLION AMERICANS

350 MILLION WORLDWIDE



If all of the people with rare diseases lived in one country, it would be the world's 3rd most populous country.



THE IMPACT

Rare diseases impact more people than

CANCER AIDS
combined.



NO FDA APPROVED TREATMENT



95%

of rare diseases **do not** have a single FDA approved drug treatment.

RARE Stats

WHAT IS CONSIDERED "RARE"?



In the United States, a condition is considered "rare" if it affects fewer than **200,000** persons combined in a particular rare disease group.



International definitions on rare diseases vary. For example in the UK, a disease is considered rare if it affects fewer than **50,000** citizens per disease.

RARE DISEASE AFFECTS CHILDREN



Approximately **50%** of the people affected by rare diseases are children.



30% of children with rare disease will not live to see their 5th birthday.



Rare diseases are responsible for **35%** of deaths in the first year of life.

THE DIAGNOSIS



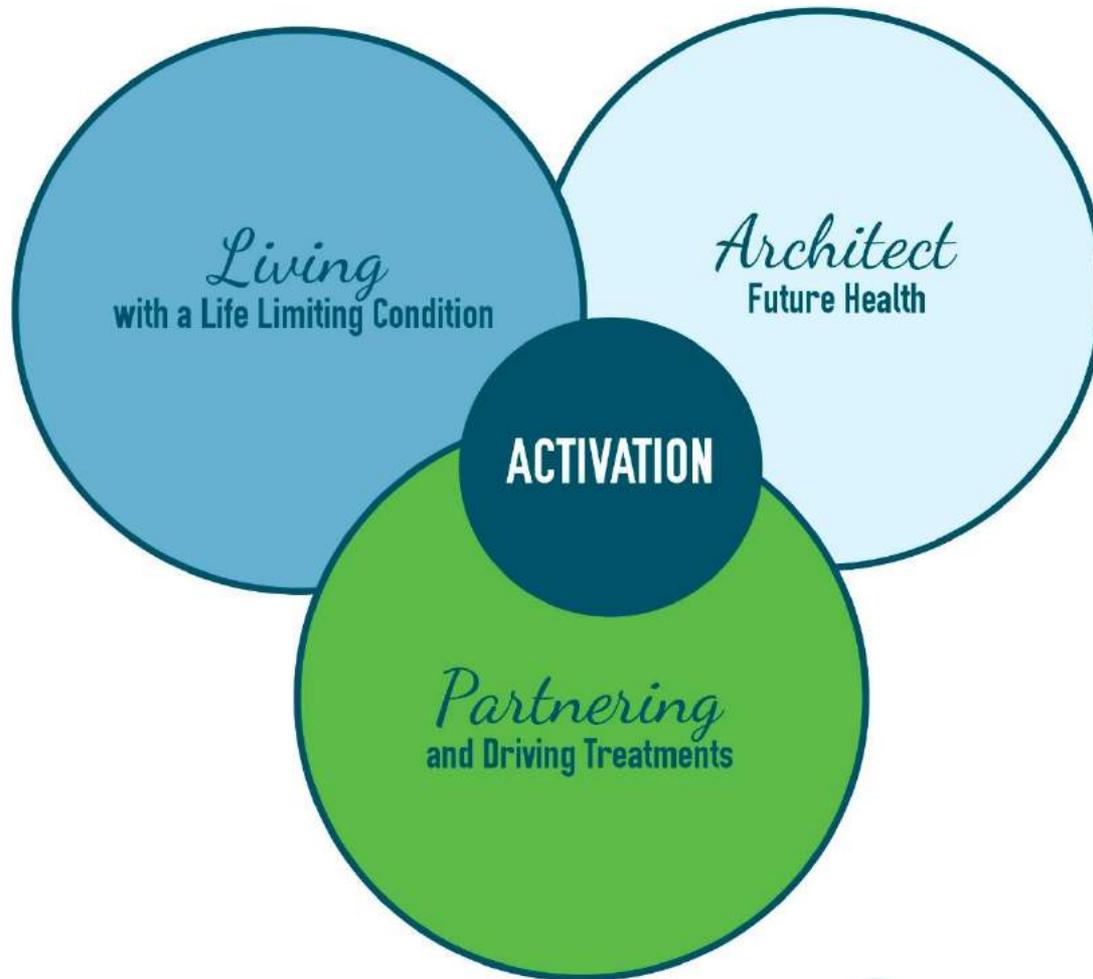
On average, it takes most rare disease patients **8 years** to receive an accurate diagnosis—within this time period, they have seen over **10 specialists** and have been misdiagnosed 3 times.

THE SUPPORT



Approximately **50%** of rare diseases do not have a disease specific foundation supporting or researching their rare disease.

Patients Responsible for More



Living with a Life-Limiting or Chronic Condition

Diagnosis
Living with
Life-
Altering
Condition

Become
Disease
Experts

Outreach
Community
Building &
Providing
Support

Need
Education
in Business,
Science,
Fundraising,
Policy, and
Legislation

Starting a
Non-Profit

Finding
and
Funding
Researchers

Partnering
with Biotech/
Pharma/
FDA/
NIH/EMEA

Patients as
Investors

Patients as
Biotech
Entrepreneurs

Legislative Advocacy

Architects of Future Health:
Individual & Community

Patients as Partners and Drivers

Patients in Drug Development

Basic Research

- Funding basic science
- Organizing patients
- Connecting to disease experts
- Meetings with FDA/NIH

Discovery & Preclinical

- Bio-specimen, natural history, health data collection
- Understanding and designing clinically meaningful endpoints
- Meetings with FDA/NIH

Clinical Trials

- Educating disease community about the clinical trial process & participation
- Disseminating clinical trial news and information
- Supporting and developing the clinical network
- Understanding clinically meaningful endpoints
- Informing protocol feasibility – Clinical Trial Guidelines and impact on family

Regulatory Approval

- Patient voice in the regulatory process
- Meetings with FDA/NIH

Commercial

- Improving disease awareness and diagnosis
- Informing clinicians
- Working with Bio/Pharma on marketing strategies

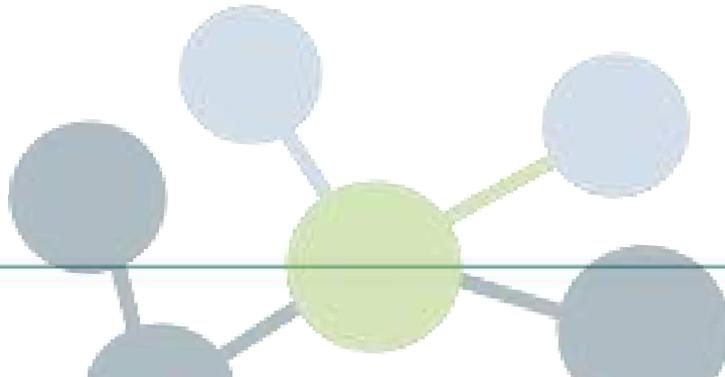


Patient Drivers



The New Norm

- Patients as Disease Detectives
- Patients Building Natural History and Registries
- Patients Seed Funding Basic Research
- Patients as disease educators - Clinicians
- Patients informing drug development process; meeting with FDA /Informing Clinical Trial Design
- Patients becoming investors
- Patients building biotech companies



THANK YOU!



Global Genes[®]

Allies in Rare Disease

www.globalgenes.org

it's in our genes.[®]