LIVING ULTRA-RARE: A PATIENT-FAMILY PERSPECTIVE

Mariah Gillaspie

Meet
Emma & Abby

- Living Undiagnosed
  - Support
  - Genetic Testing
  - Continuing the Search

- Living Rare
  - Finding a Community
  - Driving Research
  - Hope for Treatment


LIVING
UNDIAGNOSED

Support Resources

Podcasts

PODCAST
Once Upon A GENE
True stories of raising rare kiddos

RARE MAMAS Rising

Because WE ARE STRONG PODCAST

THE RARE LIFE

RARECAST
The GLOBAL GENES® Podcast

Networks

COURAGEOUS PARENTS NETWORK

Non-profit organization and educational platform that orients, empowers and accompanies families and providers caring for children with serious illness.

facebook

Parents of Children with an Undiagnosed Genetic Disorder

Private group · 1.3K members

Global Genes/NORD

Undiagnosed Rare Disease Registry

NORD®
National Organization for Rare Disorders

Global Genes®
Allies in Rare Disease

Gene-Based Diagnosis 101: How to Successfully Navigate the Diagnostic Journey

Genetic Testing

- Targeted Genetic Panels
- Whole Exome Sequencing
- Whole Genome Sequencing

Searching for a Diagnosis

- The Undiagnosed Diseases Network (UDN) is an NIH funded research study with a purpose to bring together clinical and research experts from across the United States to solve the most challenging medical mysteries using advanced technologies; and to help patients and families living with the burden of undiagnosed diseases.

- The Rare Genomes Project at the Broad Institute of MIT and Harvard is a team of researchers, physicians, software developers, genetic counselors, and study coordinators who believe that the latest advances in genomic sequencing are changing medicine and should be more broadly available to families with rare and undiagnosed conditions.

LIVING ULTRA-RARE

I HAVE A GENE NAME, NOW WHAT?

Power in numbers:
Find your patient community

1

Search social media, create social media groups, and mention your gene name in any Rare Disease/Genetic/ Undiagnosed group

2

Blanket search the internet (Blogs, research articles, etc.)

Set Google Alerts.

3

Become part of Match Matcher Exchange

The 'Matchmaker Exchange' project was launched in October 2013 to address this challenge and find genetic causes for patients with rare disease

Two Matchmaker Exchange connected partners, GeneMatcher and MyGene2 , support patient-led matching — encouraging individuals and families to share genetic and health information to identify other individuals with similar profiles.

Set yourself up for Research

Fundamentals of your Genetic Diagnosis

https://www.rareuniversity.com/cv-course/genetics-concepts-for-rare-disease-patients-families/

Global Genes® RARE University

Existing Research

Animal Models

IMPC
International Mouse Phenotyping Consortium

Cell Models

Coriell Institute for Medical Research

NIH National Institute of General Medical Sciences

Taking it to the next level...

- Launch a Patient Advocacy Foundation:

lightning and love FOUNDATION

- NORD RareLaunch

  RareLaunch® provides useful information and hands-on training and advice from leading nonprofit experts to help launch a rare disease nonprofit or take an existing one to the next level.

- Global Genes: RARE Entrepreneur Bootcamp

  This bootcamp is designed for patients and advocates who have started funding rare disease research and are looking to better coordinate and build structure around their efforts

- Open Treatments:

  Non-profit organization with a mission is to enable treatments for all genetic diseases regardless of rarity or geography…and to empower patients, families, or motivated individuals to create a treatment for a disease impacting their loved one.

- Perlara: Guided Cures

  Provides “Cure Sherpas” to guide families and research teams through the planning and development of the best cure and treatment paths that can synergistically lead to finding more potential cures for more diseases.

Explore Treatment Options

Drug Repurposing

Investigation of existing drugs for new therapeutic purposes.

Artificial Intelligence:

HealX, Medikanren, many, many, more…

Physical:

High-throughput Drug Screens

Rarebase

ASO

Antisense oligonucleotide therapy, or ASO, aims to alter RNA expression and thereby reduce, restore, or modify protein expression.

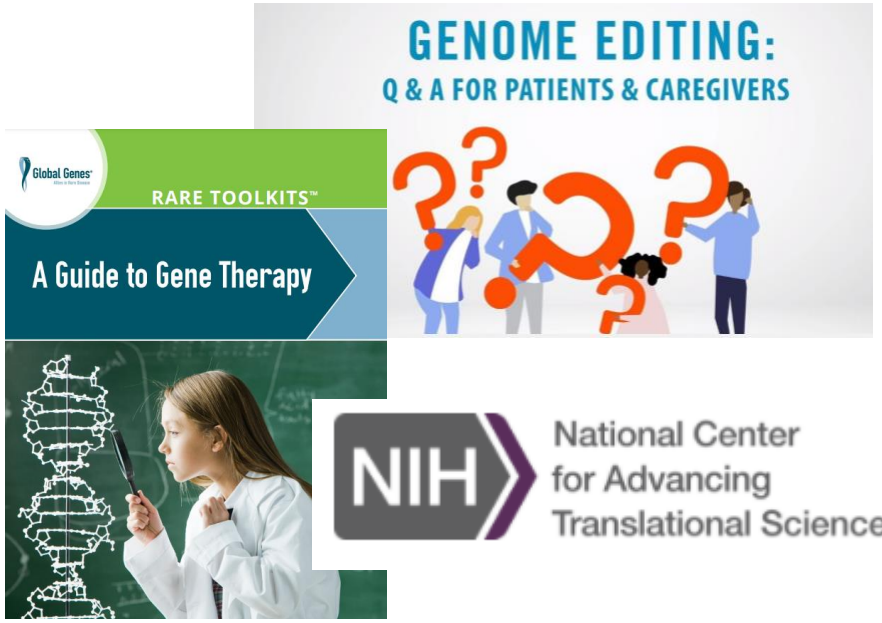
You may have heard of Mila, a little girl in Colorado who was the first person in the world to receive a drug customized for just one person.

n-lorem FOUNDATION

Gene Therapy

Most complex, most expensive, most time consuming, most risky…

But, possibly the most exciting!



NIH National Center for Advancing Translational Sciences

“The largest yet most neglected healthcare resource, worldwide, is the patient.”

Tracy Dixon-Salazar – LGS Foundation

Summary

- Living Undiagnosed can be a frustrating & helpless purgatory. But resources are available for support, and to give the best chance at finding a diagnosis. Science is moving fast, Don’t give up!

- Living Ultra-Rare can be just as frustrating, but with motivation and utilization of available resources, the patient family can be a driving force towards better disease understanding and a potential treatment.

- Despite having amazing medical care teams, Patients and patient-families are the only ones living with their rare disease. They are therefore often the experts and become the central driving force for research.

We are making a difference!

THANK YOU

- Mariah Gillaspie
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- www.lightningandlove.org

Resources

Once Upon a GENE
effieparks.com/podcast

findyourrare.buzzsprout.com/

RARECAST
The GLOBAL GENES® Podcast

globalgenes.org/rare-cast/

THE RARE LIFE

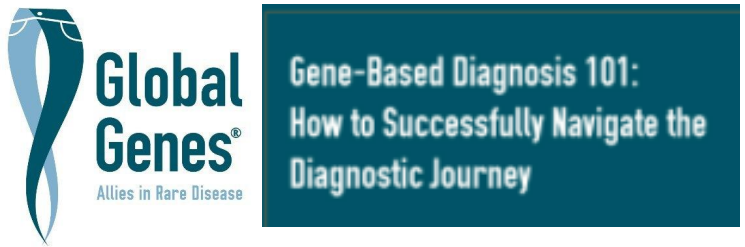
therarelifepodcast.com/

RARE MAMAS™
raremamas.com/podcast/

COURAGEOUS PARENTS NETWORK
courageousparentsnetwork.org/

Undiagnosed Rare Disease Registry
NORD® National Organization for Rare Disorders
undiagnosed.iamrare.org/

Global Genes®
Allies in Rare Disease



https://resource-hub.globalgenes.org/kb/article/298-gene-based-diagnosis-101/

https://youtu.be/eEdV2vwz160

Resources

Undiagnosed Diseases Network

undiagnosed.hms.harvard.edu

Global Genes® RARE University

Genetics Concepts For Rare Disease Patients

https://www.rareuniversity.com/cv-course/genetics-concepts-for-rare-disease-patients-families/

Coriell Institute for Medical Research

www.coriell.org/1/NIGMS

rare genomes project

raregenomes.org

lightning and love FOUNDATION

www.lightningandlove.org

Matchmaker Exchange

Genomic discovery through the exchange of phenotypic & genotypic profiles

www.matchmakerexchange.org

GeneMatcher

genematcher.org

MyGene2

www.mygene2.org

IMPC

International Mouse Phenotyping Consortium

www.mousephenotype.org/

RARE Entrepreneur Bootcamp

globalgenes.org/rare-entrepreneur-bootcamp/

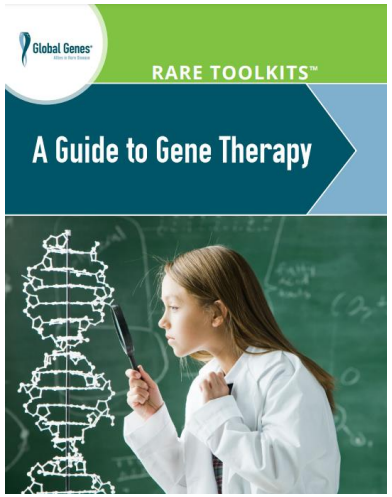
Resources

Open Treatments

www.opentreatments.org

Rarebase

www.rarebase.org/



https://globalgenes.org/wp-content/uploads/2018/11/Guide-to-Gene-Therapy_Toolkit_spread_DIGITAL-1.pdf

Perlara

www.perlara.com

n-lorem FOUNDATION

www.nlorem.org/

healx

healx.io

https://rarediseases.org/nords-new-genome-editing-videos-address-patient-caregiver-questions/

Medikanren

www.youtube.com/watch?v=vt70W55D58Q

NIH National Center for Advancing Translational Sciences

https://rarediseases.info.nih.gov/