



DECEMBER 2022

# Rare Research Roadmap



# About **Global Genes**®

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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 care about has a rare disease or is searching for a diagnosis, you can *contact us by submitting our confidential form*. A Patient Services Guide will provide you with a personalized response within 2-3 business days that will include information, resources, and connections that address your specific needs.

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# Acknowledgments

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We are deeply grateful to all who contributed to this toolkit, as presenters at the 2022 Rare Drug Development Symposium, members of our Advisory Board, creators, and reviewers; from research, advocacy, regulatory and industry.

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Supported by generous  
charitable contributions  
from

**Horizon Therapeutics**

**Editas Medicine**

**Janssen Global**

**Jazz Pharmaceuticals**

**Travere Therapeutics**

**Alexion**

**Pfizer, Inc.**

**Regeneron**

**UCB**

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# Introduction: The Road From Bench To Bedside

To find the best therapies, scientists must understand the cause of the disorder and how it impacts your body and your life over time, then find safe and effective ways to change how the disorder progresses. **This journey, from discoveries about how our bodies work to finding diagnostic methods and therapies that doctors can use to help patients, is called translational research.**

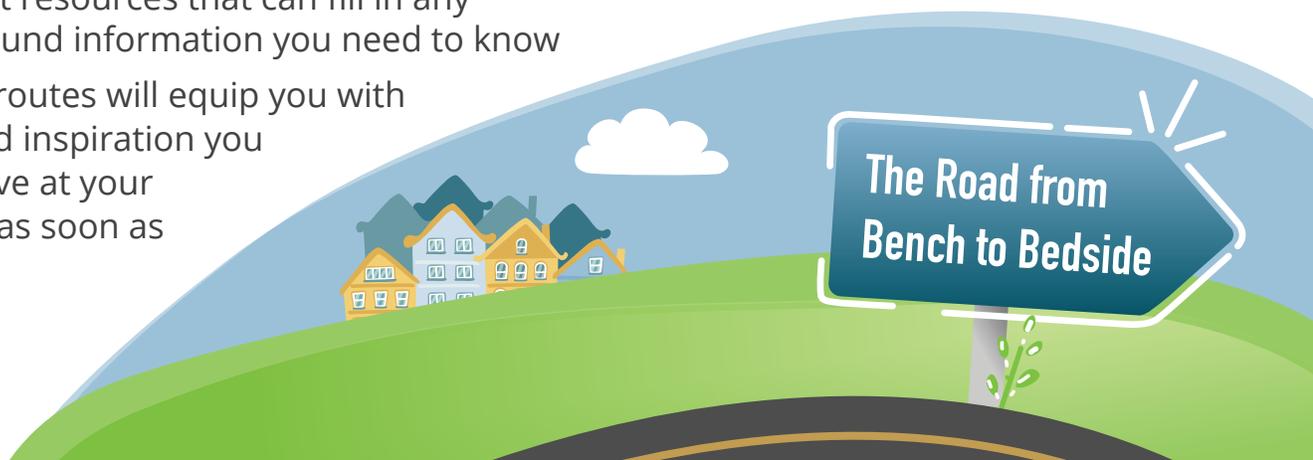
For rare diseases, the journey is seldom a straight path. You may encounter slow climbs, detours, and dead-ends that test your endurance. Sometimes you must backtrack and learn more about the disease or wait for new treatment approaches before you can move forward again. But there are also well-traveled paths and alternate routes that can help you get to your destination faster.

***The Rare Research Roadmap was designed to help you better understand some of the potential routes to treatment and some of the key concepts that are part of these processes.***

There are three ways you can use this resource:

- Follow the map to help understand the overall journey
- Access the additional information that is connected for an in-depth look at the options available to you and stories from those who have blazed a trail
- Stop in at the School of Rare Research for great resources that can fill in any background information you need to know

All of these routes will equip you with the facts and inspiration you need to arrive at your destination as soon as possible.





## Load Your Bus!



One thing is certain: you won't get far unless you know where you're going. To avoid delays, make sure you begin with clear, unified research goals for your community.

You can also count on making more progress if you bring others along. Don't go it alone. Instead:

- Organize scientific networks to help map research strategy
- Create research consortia and clinical networks
- Collaborate with researchers, biotechs, and related organizations

## You're in the driver's seat!

There are many ways that patient advocacy groups (PAGs) can help move research forward, including:

- Creating programs to award grants for research
- Supporting development of biomarkers, cell lines, animal models, etc.
- Launching registries
- Funding natural history studies and biobanks containing samples from individuals
- Connecting the patient community to clinical trial opportunities
- Contributing the patient perspective on desired therapies, access, etc.
- Gaining knowledge about the basics of data governance, the business of drug development, and intellectual property
- Driving forward requirements for open data sharing from any grant funded research, registry, or natural history data set

Ready to get started?  
**Let's go!**

# Basic Research



## What is basic research?

Basic research (often referred to as bench science) studies how a disease works at the molecular, cellular, and organism level, and in laboratory experiments. Understanding how the disease works helps researchers identify potential therapies and find ways to measure treatment response. Because so little is known about rare diseases, patient groups often play an important role in funding basic research in order to fill knowledge gaps. This work can be done with partners, such as researchers, clinicians, and biopharma, who help create models that reveal the underlying mechanism of the disorder. Biotech companies also have basic research pipelines related to the development of their products.

## Why is basic research important?

Find out what good basic research looks like, what medical advances have grown from this research, and the unanswered questions that could lead to future breakthroughs.

*Curiosity Creates Cure - The Value and Impact of Basic Research*

## Why do many rare disease basic researchers study genetics?

Because most rare diseases are caused by changes in genes, basic research often involves studying the genome – the complete set of genes or genetic material present in a person. The genome is like an “operating manual” that contains instructions that helped you develop from a single cell into the person you are today.

Advances in whole genome and exome sequencing, gene expression analysis, and cataloging of genetic variants have enabled rapid progress in understanding how relationships between genes affect the way your body develops and functions – and how changes in genes lead to disease. These areas of study are referred to as “-omics.”

- **genomics** - study of the complete set of genes or genetic material in a cell or organism
- **proteomics** - study of proteins that our genes make
- **transcriptomics** - study of all RNA molecules in a cell. RNA is copied from pieces of DNA and contains information to make proteins and perform other important functions
- **epigenomics** - the study of the set of chemical modifications to the DNA and DNA-associated proteins in the cell which alter gene expression

Other “-omics” include:

- **metabolomics** - study of small molecules produced during metabolism
- **glycomics** - the study of the structure and function of sugars in the body



Basic Research

## Need More Background on Genetics?

Here are two resources that can help you:

### *Genetics Concepts for Rare Disease Families*

This course from Rare University helps you understand concepts of genetics relevant to your rare disease.

### *Genomics 101*

Learn the basics in this series offered by the National Human Genome Research Institute.





## Fuel Up With Scientific Tools and Go!

One way that patient organizations can help advance research is by supporting the development of scientific tools and bringing them together so researchers can access them. The first step is finding out which tools exist and which will need to be created. Enlist the help of your scientific advisors and researchers who study your disorder in this process. This helps “de-risk” the project for investors who rely on data to choose which projects to pursue.

### Take An Inventory of Your Toolbox

Yael Weiss, CEO of Mahzi Therapeutics, *shares her recommendations* on how to build your research toolbox.

Here’s a list of tools that researchers use to understand a disease and identify therapies. The tools required will vary from one disease to another.

#### Animal Models

Non-human animals that are studied to gain understanding of how a disease or disorder may develop or progress in humans.

#### *What Advocates Need to Know About Cell and Animal Models*

How are cell and animal models created and what are the advantages and disadvantages of each type?

## Biobanks or Biospecimen Repositories

A collection of human biological samples (e.g., tissues, blood, cells, DNA) and related data (e.g., medical, family history, genetic) that researchers use in laboratory studies.

## Biomarkers

A biological substance (such as a disease-causing gene mutation or proteins in blood), that can be measured to determine the presence or progression of a disease or the response to a treatment.

## Cell lines

A population of cells that can grow and divide indefinitely in a lab. Scientists study these cells to understand the biology of the disease, identify new treatments and improve the efficacy of existing treatments.

## CRISPR Edited Cell Lines

Cell lines with a specific mutation created by gene editing methods.

### *Human Nature Film Trailer*

Watch an award-winning documentary from the Wonder Collaborative on CRISPR.

## Induced Pluripotent Stem Cell (iPS cell)

iPS cells are derived from skin or blood cells that have been

reprogrammed to act like embryonic stem cells. They can be “instructed” to create cells that are exact copies of those in your body. Researchers use these “models in a dish” to understand how the disease progresses and how cells may react to treatment.

### *iPS Cells as Avatars for Patients*

iPS cells are important in developing personalized medicines for an individual. But collections of these cells can also be used to develop treatments for larger populations. Find out how researchers use iPS cells to understand the way a disease works and search for cures.

## Patient Endpoints

An endpoint is a primary or secondary outcome (such as years of survival or blood levels of a biochemical substance that predicts disease progression) that will be measured in a clinical trial. Endpoints must be measured using validated tools that are reliable, tested, and acceptable to regulatory bodies responsible for approving drugs, such as the FDA. Understanding what a meaningful outcome of treatment would be from the patient’s point of view is also essential to the success of the trial. It’s never too early to consider endpoints and involve patients in the process.

## What Matters Most to Patients?

The patient perspective is needed throughout the research process. Here are some resources to help you find out how this information is gathered.

### *What is Patient-Focused Drug Development (PFDD)*

Read in-depth information on PFDD at the FDA.

### *Endpoint Selection and Use of Clinical Outcome Assessments in Rare Disease and Pediatric Trials*

Measuring improvement and deterioration from the patient's perspective

### *Putting Patients at the Center*

Scott Schliebner shares PRA Health Sciences toolkit, focused on patient-centric trial development.

## Patient Registries and Natural History Studies

Information about your community can be collected in many ways. Two of the most common are patient registries and natural history studies. Make sure you keep in mind the legal aspects of collecting and maintaining personal information, regardless of which path you follow.

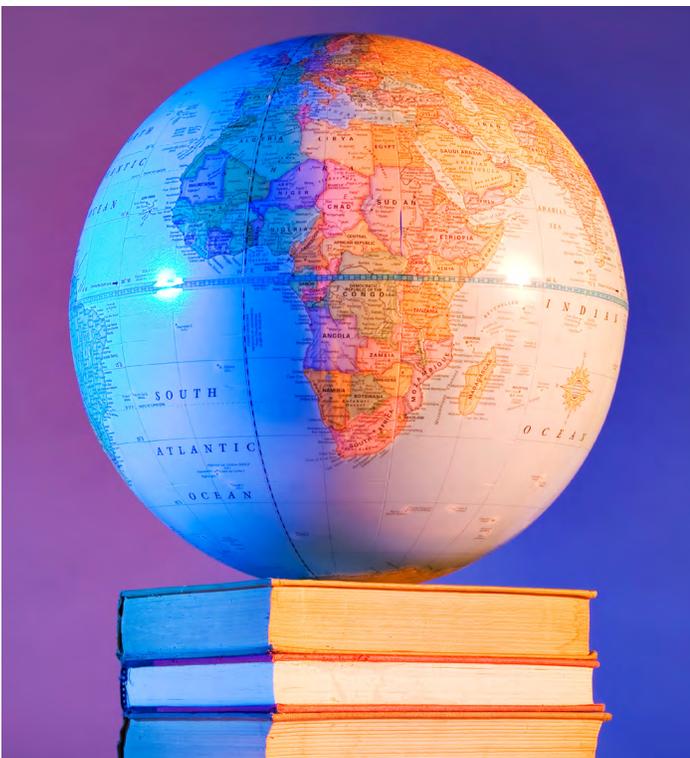
### **Patient Registries**

A collection of data from individuals with a particular disease or disorder. Registries vary widely but often include clinical history, genomic data, and laboratory test results.

- A contact registry is the simplest to create/collect; its purpose is to identify the patient population and can be used to facilitate communication. This data can be collected on any platform and the patient can provide their own information. Contact registries can be limited in their utility, however.
- For more advanced rare drug development programs, a contact registry could be used (under the proper regulatory oversight) to educate and recruit patients for participation in interventional clinical trials.

### **Natural History Studies**

A study that collects health



information and follows a group of people over time who have, or are at risk for developing a disease, to understand how it develops and can be treated. Natural history studies (NHS) may also be used to replace the placebo arm in clinical trials when small populations lead to challenges enrolling enough patients or there are ethical concerns with delaying treatment.

Natural History Studies have different implementation types:

- Prospective NHS is data captured longitudinally to chart out the progression of disease using both real world data (RWD), clinical encounters, and data contributed by the patient or caregiver. Data can be collected during the routine clinical management of the patient, and likely includes specialized tests, such as biological sampling and complex imaging, NOT typically collected during routine care.
- Retrospective NHS limits the data to those acquired during routine standard of care (SOC) visits, extracted from hospital/medical records (EHR/EMR). Though this approach has the potential to generate longitudinal data, this type of study may be missing key measures of disease that are not available in the data sources.

In both approaches, it is critical that natural history data is collected in a process with sufficient quality control that is expected under the guidelines of "GCP" (Good Clinical Practices) of FDA for capturing clinical trial data.

It's also important to be collaborative and, if possible, include patients across the globe. Natural History Studies can be expensive. It's best to make data available to all parties and avoid duplication.

*Designing Natural History Studies*  
Guidance from the FDA on creating and implementing natural history studies.





## How is Research Funded?

In rare disease, basic and translational researchers rely on several sources of funding, including:

**Grants from Federal and State Government Agencies:** In the U.S. the principal federal agency that funds basic and translational research in biological and biomedical sciences is the National Institutes of Health. The Congressionally Directed Medical Research Program (CDMRP) at the Department of Defense is another federal source of funding. The National Science Foundation also funds grants for biological basic research, but it does not fund research that explicitly tests a therapeutic or seeks to understand

the development of a disease. State governments also provide grants for biomedical research. Government agencies provide the majority of funding for basic and translational research.

**Grants from Patient Organizations:** Rare Disease Patient Organizations play an important role in raising funds for research, awarding grants, and supporting grant submissions. Your scientific advisors can help you create a research strategy and process for evaluating grant requests, including considerations such as the right to license, data sharing, and requirements for publishing within a certain timeline.

## Fundraising Tips

There are multiple ways to raise money to award for research. Crowdfunding, telling your story on social media or through the press, tabling at offline events, staging an event, writing grants to other philanthropic organizations, and cultivating major donors are a few of them. Global Genes offers toolkits and videos with fundraising tips.

### *Finding Your Fundraising Strategy*

Information on events, donors, sponsorships, grants and more.

### *Financing Research*

Hear the success stories of rare disease advocates.

### *How to Promote Your Story Through Social Media*

Advice on choosing the right platform and the right message

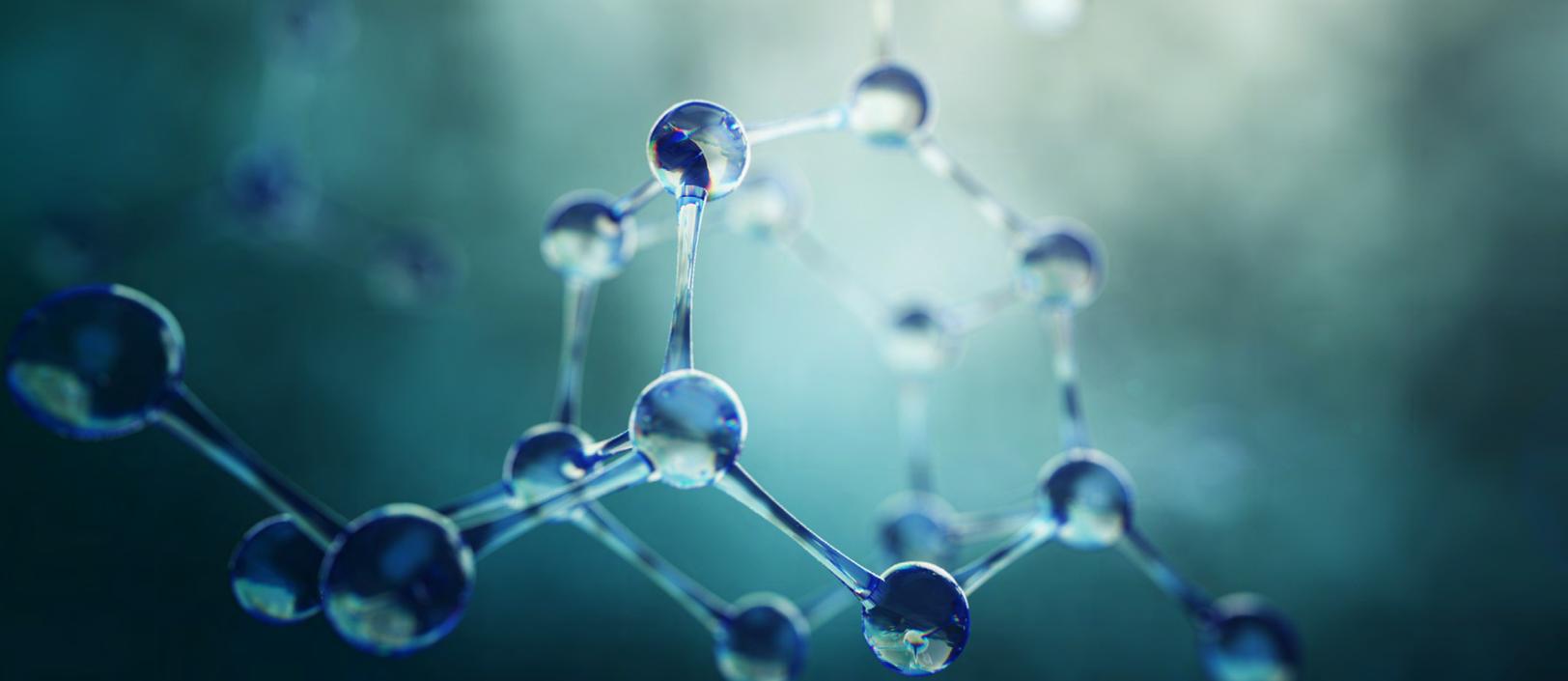
**Industry Funding:** Companies provide limited funding of grants to support registries and basic and translational researchers. However, most industry funding is targeted at preclinical and clinical research.

**Venture Philanthropy:** This is a new approach used by nonprofit groups that involves adopting tools and strategies of venture investment firms to philanthropy to support biomedical research. Organizations receive a predetermined ownership

stake with the plan to reinvest any profits into philanthropic efforts. They view their support as a contract, including monitoring the progress and evaluating the results against specified goals. These efforts are being credited with changing the R&D process to significantly accelerate research for organizations such as EB Research Partnership.

**Entrepreneurship:** A few rare disease communities have started their own for-profit companies. This is a challenging path, but has led to a few notable successes, such as GeneTx in the Angelman Syndrome community.





# Preclinical Research

Testing is carried out using cell or animal models of disease; samples of human or animal tissues; or computer-assisted simulations of drug, device, or diagnostic interactions within living systems.

## Is it Safe and Effective?

The goal of preclinical research is to show that a therapy is safe and effective before bringing it to trials in humans. There are two types of preclinical research:

- Animal research - may include basic research to understand how a disease works or safety testing to determine what amount of a therapy can be used
- Non-animal research - test tube research, computer models, and lab-based assays (chemical tests) used to understand disease pathways and design and test new therapies

You may also hear the terms *in vivo* (in a living organism), *in vitro* (in glass, such as a test tube or petri dish, with cultured cells), *ex vivo* (in cells taken from a living organism), or *in silico* (computer models).

This preclinical research involves many years of work by biologists, molecular biologists, chemists, biochemists, pharmacologists, toxicologists, computer scientists, and engineers.

## Drug Discovery

When the findings of basic research suggest a specific biological target (such as an enzyme, gene, RNA, protein, or biological pathway), researchers must check to see if affecting the target may affect the disease outcome.

Then the search for a therapy begins. In this leg of the journey researchers develop a hypothesis (an idea) about how inhibiting, activating, or knocking out (eliminating) the target will make a positive change in the disease.

Next they look for a therapy they believe will cause that change. For rare diseases, this could be a:

- small molecule - a drug made of chemical compounds
- biologic - a therapy made from living cells, for example, gene therapy or enzyme therapy
- RNA targeted oligonucleotide-based medicines (antisense, siRNA, mRNA, RNA editing, etc.)
- medical device - used in surgery, respiratory support, diagnosis, etc.

## Dive Deeper into Drug Discovery

Stanley T. Crooke, Founder, CEO, and Chairman of n-Lorem Foundation and former Chairman and CEO of Ionis Pharmaceuticals, a leader in RNA-targeted therapeutics, hosts a podcast on rare disease drug development including episodes on:

*The three platforms for drug discovery*

*What drugs do in the body and how scientists balance benefit and risk*

Here are some of the ways that researchers discover new therapies:

### High Throughput Screening (HTS)

This tool is used to comb through vast libraries of data and run automated biological and chemical tests on millions of molecules, looking for “hits” that show promise.

#### *Understanding HTS Assays*

Learn from Rick Monsma, PhD, who oversees research at the New York Stem Cell Foundation.

### Drug Repurposing

One of the fastest ways to bring therapies to patients who desperately need them is by identifying a treatment that has already been approved as safe and effective for a disease with a



Drug Discovery

similar pathway - or one that has unanticipated effects that may affect the target of interest.

### *How a Repurposed Drug is Saving Lives*

Dr. David Fajgenbaum explains how drugs developed and approved for other diseases are saving the lives of Castleman Disease patients.

### **Engineering New Therapies**

New technologies are allowing researchers to design therapies that target specific sites within the body or manipulate genetic material.

### *Using Artificial Intelligence to Design Treatments*

Chris Hart, PhD, CEO of Creyon Bio., explains how artificial intelligence (AI) reduces error in drug screening to accelerate treatment development.

### **Hit to Lead**

Once potential therapies are identified, computer simulations, cell models, and animal models are used to find out what effect each therapy has on the disease target, and identify any potential effects that could be a problem in humans. These tests, called assays, narrow the possibilities from a million possible molecules to a few hundred "leads."

## **What are Biological Assays?**

*Learn about the rigorous tests that measure the presence, amount, or activity of a molecule (i.e., a test that measures the effect of a drug on blocking a protein or how gene expression changes in a disease).*

### **Lead Optimization**

Lead therapies go through in-depth testing, focused on:

- how the body absorbs the therapy
- potential toxicity
- the dose necessary to achieve the desired effect
- method of administration
- interactions with other treatments
- effect on different populations
- ability to be produced or manufactured.

Only about half of the candidates will make the transition from "lead therapy" to "investigational drug" in a clinical trial.



# Clinical Trials

Once you have a lead candidate that has been tested, your investigational therapy must be tested in *patients* with your rare disease. This stage of research involves human volunteers and is conducted by doctors, nurses, and specialists under strict regulations that prevent any unnecessary harm to people participating. Because only 10% of rare diseases have an approved treatment option, for many patients, enrolling in a clinical trial may be the only option for treatment. Because these are investigational drugs, the risks and benefits are not known.

It can take many years to complete this stage. Research is conducted at trial sites and the work is coordinated by the Principal Investigator (PI) who ensures that all procedures are carefully followed.



## Terms to Know

- **Institutional Review Boards (IRBs)** approve, monitor and review biomedical and behavioral research involving humans to ensure that appropriate steps are taken to protect the rights and welfare of human participants.
- **Inclusion and Exclusion Criteria** specify which type of patients can participate in the study. It may specify what age, or biological sex, or physical abilities patients may have and if they must have any tests completed prior to the trial start date (like genetic tests).
- **Endpoints** are measures that show how a patient feels, functions, or survives that researchers look for to understand if the medicine works.
- **Controls-** a control is a group of clinical trial participants who don't receive the treatment that is being studied but instead receive the standard of care or a placebo,
- **Methods** are the exact procedures that researchers will use to select patients for participation and carry out the study.
- **Data Safety Monitoring Boards (DSMBs)** are independent groups of experts who monitor patient safety and treatment efficacy during clinical trials.

There are several phases in clinical trials. In rare diseases, these phases are sometimes combined to speed development of new drugs, such as phase I/II or II/III.

### Phase I

Small clinical trials usually involving 10-50 people designed to find out if a therapeutic is safe.

### Phase II

Clinical trials of a few hundred people. These trials continue to confirm safety and find out how effective a therapy is for a particular disease or condition.

### Phase III

Clinical trials of hundreds to thousands of people designed to continue to confirm safety and clearly show that a therapeutic is effective.

### Phase IV

Post-market research after the therapy is launched to track long-term safety issues, benefits, and clinical use of the therapeutic.

## Alternate Routes Available

In order to bring medicines to rare disease patients faster, the clinical trials process has been adapted for small populations.

- Rare disease clinical trials may include fewer patients than other clinical trials.
- Phase I trials in rare diseases are often combined together with

Phase II and carried out at the same time to help make faster progress.

- In some cases, researchers avoid using a control group that receives a placebo (sometimes using natural history studies for comparison).
- Expanded Access, also called compassionate use, is sometimes used to give patients with a life-threatening condition access to an investigational medicine outside of a trial, when there are no other options.
- New clinical trial models have been developed to meet the needs of small populations.

## Basket, Umbrella, Remote, and Decentralized Clinical Trials.

Craig Lipset, Advisor and Founder of Clinical Innovation Partners, *discusses innovative approaches* to clinical trials.

- What are some new clinical trial models?
- What new models hold the most promise for rare research?
- What are decentralized trials and why are they relevant to rare disease?
- What are adaptive trials and why are they useful in rare disease?

## A Road to Treatment for Individuals

For years, the number of rare diseases was thought to be “more than 7000.” Recent *studies* have shown that the number is likely more than 10,000. Some of these diseases are so novel that they have never been seen before, or they affect only a small handful of patients around the globe. Identifying these patients and their genetic variants means that others who suffer from similar conditions may at last be diagnosed and receive appropriate care. There is hope of treatment for some of these “n of 1” patients. Examples include:

- *N=1 Collaborative*, co-founded by Julia Vitarello, who worked with Boston Children’s Hospital researcher Timothy Yu to develop a customized antisense oligonucleotide to treat her daughter, Mila
- *n-Lorem Foundation* discovers, develops and provides experimental personalized antisense oligonucleotides (ASOs) to nano-rare patients who qualify and are accepted. *Watch a video* about one family’s journey.

## The critical role of patient communities in clinical trials

Rare Disease patient groups make valuable contributions to the clinical trial stage of therapy development by:

- Participating in creating the study protocols to help make sure that the relevant inclusion and exclusion criteria and endpoints are chosen;

- *Sharing knowledge of the disease, from their unique perspective*, to support efficient and effective research and payer approval;
- Finding patients who may be willing to participate in clinical trials through their own networks.

## Clinical Trial Resources

Finding information about relevant clinical trials can be challenging for patients. Here are some resources to help you help your community.

- *Getting Your Community Ready for Clinical Trials* Kari Luther Rosbeck and Gabrielle Rushing from TSC Alliance share information for your community when you are approaching clinical trials.
- *Global Genes Clinical Trial Database* A list of rare disease clinical trials posted by members of Global Genes Corporate Alliance.



Approved

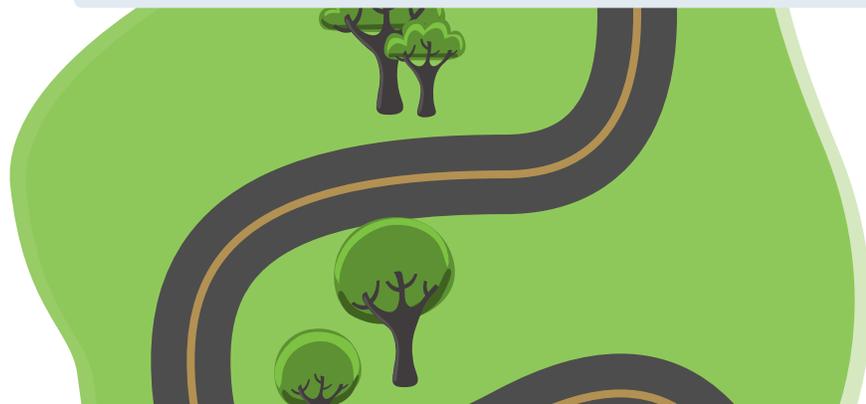
# Getting To the Finish Line: Regulators as Gatekeepers

When each phase of a clinical trial is complete, the data will be analyzed and submitted to government regulators to be approved as “safe and effective” before the next phase can begin. The job of regulators is to protect people in clinical trials and those who will one day receive approved drugs. This requires balancing the urgent need for new treatment with the ethical obligation to avoid undue harm.

Drugs already approved for one disease then studied in a different rare disease do not always require FDA approval to be used. It’s critical to do a clinical trial to prove that the drug works. But in many cases, especially if the drug is generic, there is no path forward that would lead to the FDA updating the label.

## Global Perspectives

Each country has its own system for reviewing and approving study data. Medicines that have been approved in the U.S. by the Food and Drug Administration (FDA) may or may not be approved by the European Medicines Agency (EMA), the Therapeutic Goods Administration (TGA) in Australia, or the National Medical Products Administration (NMPA) in China.



## Approval Applications

A complex set of regulations has been developed that determines how and when a new therapy is submitted and whether or not it will be approved. Here are some terms that will help you become familiar with the rules of the road. (International applications may have different names and are reviewed by country-specific agencies.)

- **Investigational New Drug (IND):** Application to allow clinical trials to begin for a drug based on preclinical test results.
- **New Drug Application (NDA):** Application to allow sales of a therapeutic made by a chemical process based on clinical trial results.
- **Biologic License Application (BLA):** Application to allow sales of a therapeutic made by a biological process (e.g., vaccines, gene therapy, antibodies) based on clinical trial results.

## Special Status that Speeds Regulatory Progress (in the U.S.)

Special status is possible if certain criteria are met regarding advantages of the therapeutic.

- **Abbreviated New Drug Application (ANDA):** Submitted for the review and potential approval of a generic drug product, to be used as a safe, effective, lower cost alternative to a brand-name drug.
- **Accelerated Approval:** Allows marketing approval based on

- intermediate effect or biomarker rather than the final effect.
- **Breakthrough Therapy:** Receives Fast Track designation and recognition that the therapeutic may be a large improvement over currently available treatments.
- **Fast Track:** Provides the company testing the therapeutic with more frequent meetings at the FDA for feedback on clinical trial design and may have rolling review of the final approval application. Therapeutics with Fast Track designation may also receive Accelerated Approval and Priority Review if qualified.
- **Orphan Drug Status:** Specifically for therapeutics to treat rare diseases that provides tax credits and other financial benefits related to the development of the therapeutic for the manufacturers/drug companies.
- **Priority Review:** Provides review of the New Drug Application (NDA) by the FDA within 6 months versus the standard 10 months.



## **Pediatric Priority Review Voucher**

**Program:** Upon approval of an NDA for a rare pediatric disease therapeutic, the sponsor receives a voucher that can be used to designate Priority Review for another therapeutic or may be transferred or sold to another sponsor to be used for an unrelated therapeutic. This program is not currently permanent.

## **Regenerative Medicine Advanced Therapy Designation (RMAT):**

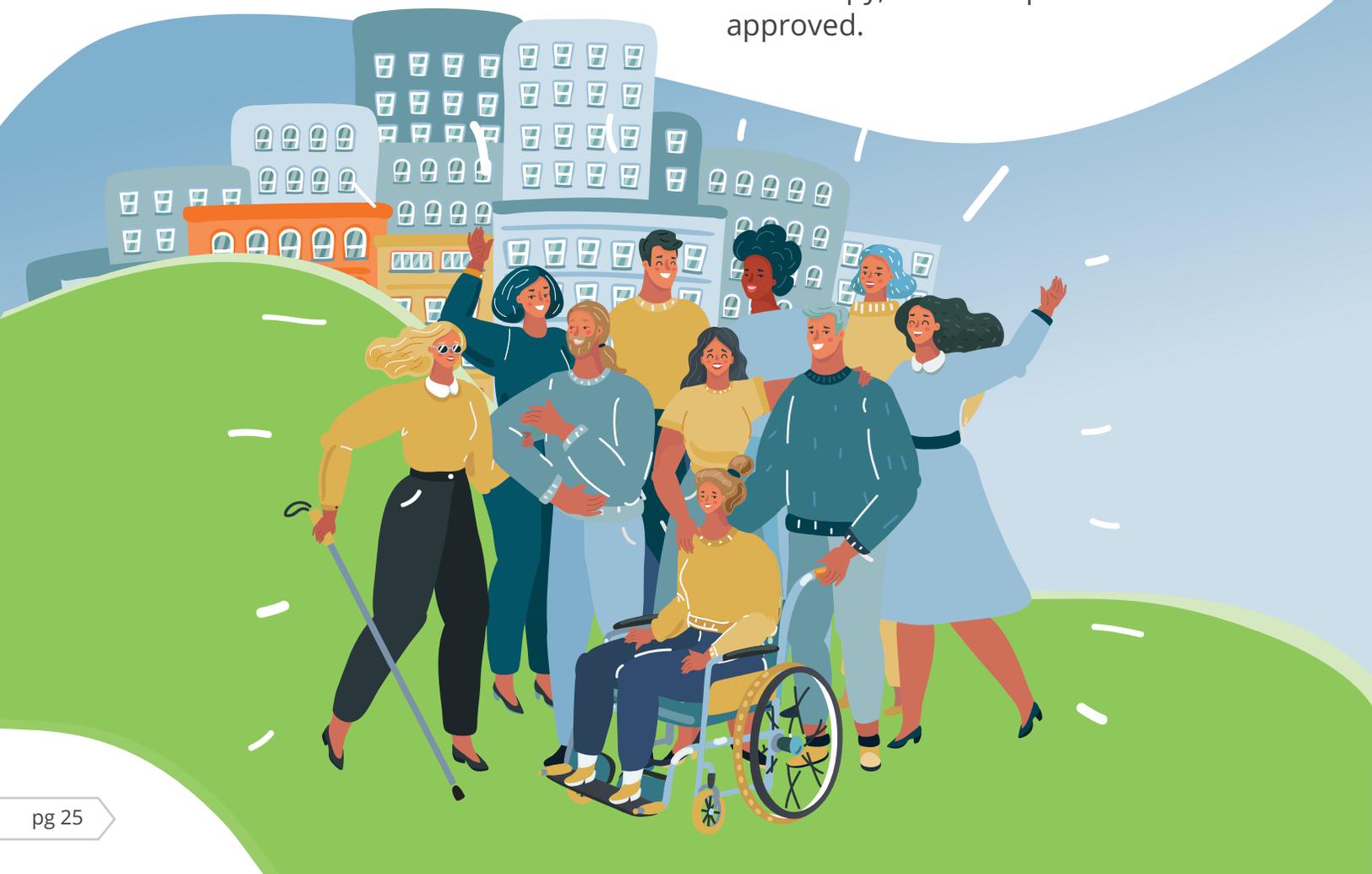
For regenerative medicines (e.g. cell therapy, therapeutic tissue engineering product, etc.) with the potential to treat, modify, reverse, or cure a serious or life-threatening disease or condition.

## **After Approval**

The research journey doesn't end after approval. Post-market (Phase IV) studies will likely be done to track safety and benefits in clinical practice.

Even if the therapeutic is approved, there may still be concerns about pricing and reimbursement. There have been cases internationally where government agencies have determined that the benefits of a therapeutic are not significant enough to justify the company's proposed cost. Input from the community is important at every stage of the process to demonstrate that therapies are making a real difference in patients' lives.

There are also sometimes criteria, such as age, that determine who is eligible for therapy, unless a special case is approved.



# Go Faster Together!

## Case Studies in Collaboration

### What Can We Do Together?

*Watch a panel discussion* on how collaboration accelerates rare disease research.

Every journey is smoother when there are others along for the ride. Whether your traveling companions are organizations that focus on diseases with similar pathways, pharmaceutical companies, federal agencies, or academic researchers, there are many ways to partner for the benefit of all. Here are some rare disease success stories to inspire you.

#### CASE STUDY 1

##### **Rare Diseases Clinical Research Network (RDCRN)**

Tiina Urv, PhD, who leads RDCRN, *explains the work* of this international network that joins 165 rare diseases into a research community with 20 consortia.

## CASE STUDY 2

### **The LouLou Foundation**

Dan Lavery, Chief Scientific Officer of the LouLou Foundation and Director of the CDKL5 Program of Excellence at the University of Pennsylvania Orphan Disease Center *discusses the pre-competitive collaboration* they are engaged in with seven pharmaceutical companies to develop clinical outcome measures for CDKL5.

## CASE STUDY 3

### **Rare Epilepsy Network and Ring14 USA**

Yssa De Woody, Co-Founder and Director of Research at Ring14 USA and Chair of the Coordinating Committee for the Rare Epilepsy Network (REN) talks about how collaboration has *helped advance research* in her community.

# The School of Rare Research

**Ready to dive deeper? Here are some reliable resources.**

## *The Business of Science*

Yael Weiss, Mahzi Therapeutics, provides tips on grants vs. sponsored research agreements and patent protections on drugs.

## *Ciscrp Webinar Series: Navigating Rare Disease and Clinical Research*

Patients and organization leaders describe clinical trials protocol, what it's like to participate, and how to prepare for when the clinical trial ends.

## *Data DIY*

Learn about the importance of aggregating patient data - and making it shareable - in this series of toolkits and videos from Global Genes.

## *Designing Natural History Studies*

Guidance from the FDA on creating and implementing natural history studies.

## *Drug Repurposing Course*

Inspired by his son who was born with a rare condition, Sanath Kumar Ramesh, founder and CEO of the Open Treatments Foundation created a Wiki for rare disease advocacy groups interested in repurposing. For more information, use the contact form on the Open Treatments website.

## *EveryLife's Guide to Patient Involvement in Rare Disease Therapy Development*

Information and action steps on patient-focused drug development.

## *How to Talk to Researchers*

Watch this video to find out how to engage and what questions to ask.

## *NCATS Toolkit for Patient-Focused Therapy Development*

The gold standard guide to all phases of therapeutic discovery.

## *N-lorem Foundation Podcast*

Interviews that teach core concepts about drugs. Host Dr. Stanley Crooke led the creation of antisense technology and launched a foundation to discover, develop, and provide personalized experimental antisense oligonucleotide medicines for nano-rare patients for free for life.

### *Prediction to Patient*

An infographic describing the drug discovery journey from HealX.

### *What are the Keys to Accelerating Rare Disease Research?*

RDDS 2022 keynote panel discusses how new trial models, data aggregation, artificial intelligence, and drug repurposing are helping overcome barriers in rare disease research.

### *What Does it Take To Become a Research-Ready Organization?*

Karsten Baumgaertel, Director of Translational Biology at Traverre Therapeutics discusses the role of advocates in gathering data, improving diagnosis, and endpoint selection.

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