



Research Exploration Toolkit:

Your Guide to Getting Started in Rare Disease Research

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Please use this Table of Contents to navigate to the sections you're most interested in by using Ctrl + click.

TABLE OF CONTENTS

Welcome to Rare Disease Research	3
What is rare disease research?	3
Why does it matter?	3
Why are rare diseases scientifically interesting?	4
EXPLORE	4
Step 1: Start Exploring Where You Might Fit	4
A Few Ways to Explore Any Area of Research	4
Lab & Molecular Research	5
Computational and Data Science Research	7
Clinical & Translational Research	10
See Rare Disease Research in Action	13
PREPARE	15
Step 2: Build the Right Foundation	15
Step 3: Start Getting Experience	16
CONNECT	18
Step 4: Find Opportunities.....	18
How to contact a researcher you don't know.....	19
Step 5: Find a Mentor.....	20
Step 6: Participate in Conferences and Communities.....	21
TAKE YOUR NEXT STEP	22
Step 7: Find Programs and Funding That Match Your Stage	22
Undergraduate Students.....	23
Medical Students	25
Graduate Students	27
Gap Year and Post-Baccalaureate Trainees.....	29
Opportunities that Cut Across Stages	31
Advice from Rare Disease Researchers	32
Your Rare Research Launchpad: The Next 30 Days	33
Research Opportunity Search Planner	35
Research Opportunity Comparison Tool.....	36
Mentor Map: Building Your Network.....	37

Your Guide to Getting Started in Rare Disease Research

Welcome to Rare Disease Research

Welcome! You belong here. If you're curious about a future career in research, whether you're just beginning to explore or already have some research experience, this guide is for you. The field of rare disease research is not a field where everyone follows the same path. Whether you're interested in genetics, molecular biology, medicine, bioinformatics, computer science, engineering, psychology, public health, or data science, there are meaningful ways for you to contribute to rare disease research. There are so many unanswered questions waiting for curious people to help solve them. The goal of this guide is to help you take your next steps.

What is rare disease research?

Rare disease research involves the scientific study of medical conditions that affect small populations of people. Rare disease is defined in the U.S. as a disease that affects fewer than 200,000 individuals.¹ However, collectively, people living with rare diseases are estimated to be as much as 10% of the population in the United States.² Rare disease research often focuses on finding genetic causes, tracking disease progress, and finding or creating safe and effective treatments where few or none currently exist.

Why does it matter?

More than 300 million people worldwide are living with one of more than 10,000 known rare diseases. While the patient populations for each rare disease are small, collectively, rare diseases affect millions of children and adults. All too commonly, people living with rare disease experience delayed diagnoses (sometimes for years), limited treatment options, or no approved therapies. Rare disease research has the power to change that.

Researchers around the world are working to understand the underlying causes of these rare conditions, to develop better diagnostic tools, and to discover and develop new therapies to extend and improve the lives of those living with rare diseases. Every scientific discovery, whether it happens in a laboratory (lab) or through data analysis or a clinical trial, has the potential to move the field forward. And behind every research project is a family waiting for answers and holding out hope.

¹ <https://ncats.nih.gov/research/our-impact/our-impact-rare-diseases>

² <https://fda.gov/patients/rare-diseases-fda>

Why are rare diseases scientifically interesting?

Rare disease research provides unique insights into how the human body works. Because many rare diseases are caused by a variant in a single gene or biological pathway, by understanding what goes wrong in rare diseases, researchers frequently find discoveries that improve our understanding of biology as a whole. In fact, rare disease research has helped advance knowledge in genetics, immunology, metabolism, neuroscience, and gene therapy. Treatments that were originally developed for rare diseases have also influenced treatments for more common conditions.

Rare disease research is challenging, but that also makes it exciting. Many important research questions remain unanswered, creating opportunities for students and trainees like you to contribute to discoveries that could shape the future of medicine.

EXPLORE

Step 1: Start Exploring Where You Might Fit

There isn't just one kind of rare disease researcher. Rare disease research needs people with many different interests and skills. Some researchers spend much of their time in a lab studying cells, genes, or proteins. Others work with large sets of health or genetic data. Some work directly with patients through clinical studies. There are many researchers who work across more than one of these areas.

Rare disease researchers share all kinds of ways that they became interested in rare disease. Sometimes it's through a particular condition, or a scientific question, a mentor of theirs, a patient community they encounter, or another opportunity they didn't anticipate.

You do not need to know exactly which path you might choose right now. See which of the following broad categories of research in this field interest you and choose the starting point that fits you best.

New to research? Start with [Explore It](#) in each section of this guide. We'll explain what this kind of research looks like, introduce some terms you may hear, and show you how to begin getting exposure.

Already have some research experience? Jump to [Build on It](#) in each section for ideas, experiences, and connections that can help you take your next step.

A Few Ways to Explore Any Area of Research

No matter what type of research interests you, there are a few strategies you can use to start exploring:

- **Search by the science that interests you.** Look at university, hospital, academic medical center, and research institute websites. Search for a disease, biological process, technique, or research area (not only the words "rare disease"). Look for sections like "Research," "Clinical Research,"

“Clinical Trials,” or “Research Programs.” Departments such as genetics, neurology, pediatrics, hematology, immunology, and other specialties may also have their own research pages.

- **Look at researchers’ work.** Read faculty and lab pages and skim a recent publication, project description, or news story to see what questions they’re asking. Then, check whether they have opportunities for students or trainees. If nothing is listed, consider reaching out directly. See [*“How to Contact a Researcher You Don’t Know”*](#) in Step 4 for some advice.
- **Look beyond your own institution.** Nearby universities, hospitals, independent research institutes, and NORD Rare Disease Centers of Excellence may have research you won’t find through your own school’s website.
- **Ask people for connections.** Professors, advisors, research offices, career centers, graduate students, clinicians, and other researchers may know about people or opportunities that aren’t easy to find online – especially at nearby hospitals and universities.

Keep these strategies in mind as you explore the research areas below. Each section also includes ideas specific to that type of research.

Lab & Molecular Research

Have you ever wondered, “But what is actually happening inside a cell?” If so, lab and molecular research may be very interesting to you.

Lab and molecular researchers investigate what is happening biologically when someone has a disease. They may study genes, cells, proteins, or other biological processes to understand why a disease occurs and what might be done to treat it.

For example, a researcher might grow cells in a lab and compare cells affected by a genetic condition with unaffected cells. Another might study what happens when a specific gene isn’t working properly. Others may test whether an experimental treatment changes what happens in those cells.

You might enjoy this type of research if you:

- Like biology and understanding how things work
- Enjoy hands-on experiments
- Like solving problems when the answer isn’t obvious
- Are curious about genetics, cells, or disease mechanisms

Explore It

“But I’ve never worked in a lab.” That’s okay! You aren’t expected to walk into your first lab research experience knowing how to grow cells, analyze proteins, or use gene editing tools. These are techniques people learn through coursework, training, and working alongside more experienced researchers. **Your goal right now is to find a place where you can begin learning.**

How do I get exposure to this?

Look at the webpages of biology, genetics, neuroscience, biochemistry, or biomedical research departments at your school. Find faculty whose research sounds interesting, even if you don't understand all of it yet. Read the description of their lab and look for one recent paper or news story about their work. Then, consider reaching out to ask whether there are opportunities for students to get involved.

What might you actually do when you're starting out in a first lab experience?

Your first responsibilities might include preparing solutions, labeling samples, maintaining lab records, helping care for cell cultures or model organisms, observing experiments, entering data, or learning one technique alongside a more experienced researcher. You may begin by watching and assisting before conducting parts of an experiment yourself. The goal of a first experience is to learn how research actually works and gradually build skills and independence.

Some terms you may come across:

- **Animal models:** Non-human living organisms used in medical research to study biological processes or human diseases. Key types include mice, rats, and zebrafish.³
- **Assay:** A test or experiment used to measure something – for example, whether a protein is present or whether a treatment changes how cells behave
- **Cell culture:** Growing and studying cells under controlled conditions in a lab
- **Gene editing:** Tools that allow researchers to make targeted changes to DNA so they can study genes or potentially correct disease-causing changes.
- **Protein analysis:** Methods researchers use to study proteins, including how much of a protein is present or whether it is working normally

Build on It

If you already have some lab experience, think about where you could deepen it or broaden it. Experts in [NORD's Rare Disease Centers of Excellence Network](#) who contributed to this toolkit highlighted experience with **cell culture, assay development, protein analysis, animal models, gene therapy, and gene-editing techniques** as useful areas of exposure.

You don't need experience in all of these areas. Instead, ask yourself: ***What technique would help me answer questions I'm interested in, and where could I learn it?*** Then, talk with your principal investigator (PI), mentor, or another researcher about opportunities to learn a new technique or contribute to a different project.

Look for opportunities to take on more responsibility and ownership. If you already know how to perform a technique, ask whether you can begin understanding or contributing to the decisions behind the experiment. Could you help plan an experiment, choose appropriate controls, troubleshoot a result

³ <https://genome.gov/genetics-glossary/Animal-Model>

that didn't turn out as expected, analyze or interpret the data, or present your findings at a lab meeting?

Learn a technique because it helps answer a question. As you gain experience, try not to think of lab skills as a checklist. Cell culture, protein analysis, assay development, gene editing, and other techniques are tools researchers use to answer questions. Ask your mentor, **“What question are we trying to answer, why are we using this technique, and what will the results tell us?”**

Learn from a lab that does something yours doesn't. If you're interested in a technique or approach your current lab doesn't use, ask whether your PI or mentor collaborates with another research group that does. You might be able to observe experiments, attend another group's lab meeting, contribute to a collaborative project, or spend time learning a method from someone with different expertise.

Connect the skills you're building to rare disease. Your current lab does **not** need to study a rare disease for the experience to still be relevant. Skills in molecular biology, cell biology, genetics, protein biology, neuroscience, immunology, pharmacology, developmental biology, and many other areas can be applied to rare disease questions later. Ask yourself: ***what am I learning how to do here, and what other kinds of scientific questions could I use that skill to investigate?***

Follow a potential treatment from idea to experiment. Lab researchers may investigate many types of potential treatments, including small-molecule drugs, antibody-based therapies, RNA-based approaches, and gene therapies. If your lab studies a potential treatment, ask how researchers decide what to test, how they determine whether it has an effect, and what evidence would be needed before the research could move to another stage.

Computational and Data Science Research

Do you like finding patterns or asking what a collection of information can tell us?

Rare disease researchers don't only work at lab benches. They also work with data.

Imagine researchers have health information from 500 people living with the same rare disease. Why did some develop symptoms earlier than others? Are certain genetic variants associated with different symptoms? Can researchers identify patterns that might help predict how the disease progresses?

Computational and data science researchers use statistics, computer programming, and specialized tools to investigate questions like these.

You might enjoy this type of research if you:

- Like finding patterns and solving puzzles
- Enjoy working with numbers, data, or computers
- Are curious about what large amounts of information can tell us
- Like working on biological or medical questions from an analytical perspective

Explore It

“But I don’t know how to code.” That’s okay! **You don’t need to be an expert coder to start exploring this area.** If you’re completely new to it, an introductory course in statistics, programming, computer science, or data analysis is a legitimate first step. Even basic familiarity can help you understand what interests you and prepare you to learn more.

How do I get exposure to this?

See what is available at your school. Look beyond programs with “data science” in the name. Faculty in biology, genetics, computer science, statistics, mathematics, epidemiology, psychology, or public health may use data in their research. Read faculty profiles and lab or research pages to see what kinds of questions they study and whether they work with biological, genetic, clinical, or health data. If the work interests you, ask what skills they recommend for students who want to get involved.

Look beyond your own institution. Explore universities, academic medical centers, research institutes,

No data science program at your school? You can still start.

Computational research skills can be developed through coursework, online training and publicly available datasets. Your first experience doesn’t need to involve a rare disease or a formal research position. Learning to work with data and developing strong statistical or computational skills can prepare you to apply them to rare disease questions later.

and hospitals in your area. Or there may be organizations that offer summer or remote research opportunities. Search for terms like “undergraduate bioinformatics research,” “computational biology summer research,” “genomic data internship,” or “biomedical data science undergraduate research.”

Practice with publicly available data. Many researchers learn by working with datasets that are available for education and research. A class project or independent project using biological or health data can give you something concrete to practice with even if you don’t yet have access to a research lab. Free, guided online tutorials can walk you through bioinformatics tools and datasets step by step. For example:

- **New to bioinformatics?** [EMBL-EBI Introductory Bioinformatics](#) offers a free, self-paced introduction for learners with no prior bioinformatics skills. It includes activities, quizzes, and introductions to resources for exploring genes, gene expression, proteins, and other biological data.
- **Ready to work with data?** [Galaxy Training Network](#) provides free, hands-on tutorials for analyzing biological data. Learners can start with the [Introduction to Galaxy and Sequence Analysis](#) learning pathway, which walks through the basics step by step.

- **Want to explore the databases researchers use?** Try the [National Library of Medicine's NLM Navigators to NCBI Biology Databases](#). Start with the NCBI Gene tutorials to practice finding a gene, exploring its location and reference sequences, examining gene expression information, and looking at genetic variants associated with disease.

Ask researchers what they recommend. If you find someone's work interesting, you don't have to begin by asking for a position. You can ask, "I'm interested in getting experience with computational biology/genomic data, but I'm very early in my training. What skills would you recommend I build first, and how?"

Some terms you may come across:

- **AI applications:** Software programs that use artificial intelligence like machine learning, natural language processing, or computer vision to complete tasks that usually need human thought
- **Bioinformatics:** Using computers and data analysis to answer biological questions
- **Coding:** Writing instructions in a programming language that tell a computer what to do. Researchers may use coding to organize, analyze, and visualize data or automate repetitive tasks.
- **Data analysis:** The practice of collecting, cleaning, and interpreting data to identify patterns, draw conclusions, and support informed decision-making
- **Genomic data:** Information about DNA and genetic variation
- **Machine learning:** Computer methods that can identify patterns in large amounts of data or make predictions
- **Patient registry:** An organized collection of information about people who share a disease or condition

Build on It

Already have some experience with statistics, coding, or data analysis? Your next step may be to put those skills to work on real research questions or learn to work with new kinds of data.

Look for ways to apply what you already know. Look for research teams working with **genomic data, patient registries, natural history data, clinical trial data, or electronic health record data**. Ask whether your existing skills could be useful to an ongoing project. Search faculty and lab webpages using the scientific area that interests you and terms like "bioinformatics," "computational biology," "biostatistics," "biomedical data science," "genomics," or "clinical data science."

Add a skill because you need it (not just because it sounds impressive). Explore opportunities to learn how to analyze genomic data or strengthen your skills in bioinformatics, statistical programming, data visualization, machine learning, or a new computational method. It can be tempting to think you need to learn everything (R, Python, machine learning, AI, data visualization, etc.), but you don't. Instead,

start with a research question. Ask yourself: ***What kind of data or analysis could help me answer questions I'm interested in?*** Then, talk with a professor, principal investigator (PI), or researcher about what new skill will help you get there.

Learn from people who have the expertise you don't yet have. Computational research is collaborative. If your lab or research group doesn't have the expertise you want to develop, ask your PI or mentor whether there is a biostatistician, data scientist, computational biologist, or collaborating research group you could learn from. You might be able to contribute to a collaborative project, attend another group's lab meetings, take a workshop they recommend, or work with a different type of dataset.

Look for opportunities to take more ownership. If you've already contributed to a research project, consider what the next level of involvement could look like. Could you:

- Help decide how data should be cleaned, organized, or analyzed?
- Develop or refine an analysis plan with your mentor?
- Create a visualization that helps the research team understand or communicate its findings?
- Compare different approaches to answering the same question?
- Help interpret results, not just generate them?
- Present the work at a lab meeting, student research day, or scientific conference?
- Contribute to an abstract, poster, or manuscript?

You don't need to do all of these things. The goal is to **begin moving from “I know how to use this tool” toward “I know how to use this tool to help answer a research question.”**

Learn how to ask good questions. Technical skills matter, but knowing *why* you are running an experiment or analysis matters just as much as knowing how to use the tools. As you gain experience, ask how the work you're doing connects to the research question, what the results mean, and what question should come next.

Connect your existing experience to rare disease. Your current research doesn't have to focus on rare disease to prepare you for rare disease research. If you're already working in computational genomics, statistics, machine learning, epidemiology, or another data-focused field, ask yourself: ***How could the skills I'm developing be applied to questions that matter in rare disease?*** You may be able to bring the expertise you're already developing into a new scientific context.

Clinical & Translational Research

Are you interested in how scientific discoveries ultimately help people?

Clinical research studies health and disease in people. Translational research helps move discoveries to practical applications like diagnostic tools or treatments and brings observations and questions from patients back into the research process.

Clinical and translational researchers design clinical studies; study how a disease changes over time (natural history); test different approaches to treatment including small-molecule drugs, antibody-based therapies, RNA-based therapies, and gene therapies; develop better ways to measure whether a treatment is working (such as finding molecular signs in blood or tissue or tracking genetic changes over time), or work with patients and caregivers to understand what outcomes matter most to them.

You might enjoy this type of research if you:

- Are interested in connecting science with patient care
- Like asking questions about how diseases affect people differently
- Want to understand how potential treatments are studied and evaluated
- Enjoy collaborating with people from different scientific and clinical backgrounds
- Are interested in working with patients, caregivers, or health information

Explore It

“I’m interested, but I’m not sure what clinical research experience actually looks like.” That’s okay! There are many ways to get involved. You might help a research team collect or organize study data, work with a patient registry or natural history study, assist with participant recruitment, review medical records for a research study, or contribute to a clinical trial. Scientists, physicians, nurses, genetic counselors, psychologists, statisticians, data scientists, research coordinators, and many others contribute to this work.

How do I get exposure to this?

Clinical and translational research may not be happening at your school, and that’s okay. In addition to the general strategies at the beginning of this section, here are two ways to specifically look for clinical and translational research:

- **Use clinical research terms in your search.** Try searches such as “cystic fibrosis clinical research Baltimore,” “rare disease clinical trials Boston,” or “genetics research Chicago.” You can also search for a particular disease plus terms like “natural history study,” “registry,” or “clinical research.”
- **Look up clinical studies themselves.** [ClinicalTrials.gov](https://clinicaltrials.gov) lets you search for studies by disease, location, researcher, or institution. **You can use it to discover who is conducting research that interests you and where that research is happening.**

Here’s an example of what this could look like:

- A student is interested in leukodystrophies.
- They search ClinicalTrials.gov.
- They see which institutions are conducting studies.

- They discover a children's hospital 30 miles away.
- They click around that institution's research program and identify investigators.
- They contact someone and find out there's a summer research program they can apply to.

Your research experience doesn't have to happen at your school. If your college or university doesn't have a medical school, teaching hospital, or clinical research program, look outward. Hospitals, academic medical centers, research institutes, and other universities may offer volunteer, summer, internship, or paid research opportunities to students from other institutions.

Some terms you may come across:

- **Clinical research:** Research involving people, health information, or human samples that helps people better understand health, disease, diagnosis, treatment, or care
- **Clinical research methods:** Structured, systematic rules and procedures used to design, conduct, and analyze studies involving human participants
- **Clinical trials:** Research studies that test a medical, surgical, or behavioral intervention in people. These are the primary ways that researchers determine if a new form of treatment or prevention (a new drug, medical device, or diet) is safe and effective in people.
- **Natural history study:** A study that collects information on how a disease would develop, change, or progress over time
- **Observational study:** A study in which researchers observe and collect information without assigning participants to a particular intervention or treatment
- **Outcomes research:** Research that is concerned with the effectiveness of public health interventions and health services, including real-world effectiveness, costs, and quality of life rather than just looking at lab results or controlled clinical settings⁴
- **Patient registry:** An organized collection of information about people who share a disease or condition
- **Regulatory science:** Developing new tools, standards, and approaches to assess the safety, efficacy, quality, and performance of FDA-regulated products⁵

⁴ <https://pubmed.ncbi.nlm.nih.gov/12603584/>

⁵ <https://www.fda.gov/science-research/focus-areas-regulatory-science-report/focus-areas-regulatory-science-introduction>

- **Translational research:** Research that helps turn scientific discoveries into practical applications for human health and can also bring observations and questions from patients back into scientific investigation

Build on It

Already have some clinical or research experience? Look for opportunities to contribute to **study design, natural history studies, patient registries, clinical trials, outcomes research, or regulatory science.**

Consider where you could gain experience helping to shape a research question, develop a study protocol, select or develop outcome measures, analyze or interpret data, or understand how findings inform the next stage of research or patient care.

You don't need experience in all of these areas. Instead, ask yourself: ***What part of the clinical or translational research process would I like more experience with, and where could I gain that experience?*** Then talk with your principal investigator (PI), research mentor, clinical mentor, or another member of the research team about opportunities to take on more responsibility, contribute to another stage of a study, or collaborate with people whose expertise differs from your own.

Good to Know: Translation doesn't travel in only one direction. Translational research is sometimes described as moving discoveries from the "bench to the bedside," but research doesn't always follow a straight line. Questions that come up in patient care can lead researchers back to the lab, and results from clinical trials can generate new scientific questions. Progress often involves communication back and forth from lab research and clinical research and patient care.

See Rare Disease Research in Action

Rare disease research can take many forms – from investigating what is happening inside cells and developing disease models to improving diagnosis, studying treatments, and figuring out how advances can reach patients. The publications below offer a few examples from researchers featured in Rare Research Launchpad.

You do not need to read every paper from beginning to end. Start by looking at the research question, abstract, figures, or discussion. Notice what the researchers wanted to learn, how they approached the question, who they collaborated with, and what questions remain.

Vici syndrome: Using a disease model to understand what drives progression

Thornton BT, Hardinger AG, Pence L, et al. [Progressive neuroinflammation and deficits in motor function in a mouse model with an Epg5 pathogenic variant of Vici syndrome](#). *Experimental & Molecular Medicine*. 2026;58(1):284-295. doi: 10.1038/s12276-026-01644-z

Researchers used a mouse model of Vici syndrome to investigate biological changes associated with progressive neurological disease and create a platform for studying potential therapeutic approaches. **Rare Research Launchpad faculty contributor Jay L. Vivian, PhD, is a coauthor of the study.**

Undiagnosed diseases: Using model organisms to help solve human genetic mysteries

Baldrige D, Wangler MF, Bowman AN, et al. [Model organisms contribute to diagnosis and discovery in the Undiagnosed Diseases Network: current state and a future vision](#). *Orphanet Journal of Rare Diseases*. 2021;16(1). doi: 10.1186/s13023-021-01839-9

This paper shows how researchers can use model organisms (such as worms, flies, zebrafish, or others) to investigate candidate genes and genetic variants identified in people with undiagnosed conditions. **Rare Research Launchpad faculty contributor Stephen C. Pak, PhD is a coauthor.**

Krabbe disease: Turning newborn screening into timely clinical action

Thompson Stone R, White AL, Rubin JP, et al. [Expert Recommendations for Rapid Response to Positive Newborn Screen for Infantile Krabbe Disease](#). *Neurology Genetics*. 2026; 12(3). doi:10.1212/NXG.0000000000200383

Experts outline how clinical teams can rapidly evaluate and refer infants after a positive Krabbe newborn screen, where treatment must occur very early to provide the greatest potential benefit. **Rare Research Launchpad faculty contributor Joanne Kurtzberg, MD is a coauthor.**

Cystic fibrosis: Asking what research is needed after a major breakthrough

Fajac I, Jain R, Mall MA, Rubin BK, Flume PA. [The clinical, psychosocial, and economic burden of cystic fibrosis lung disease in the era of CFTR modulator therapy](#). *Annals of the American Thoracic Society*. 2026;23(1):17-29. doi:10.1513/annalsats.202408-800fr

The authors examine the disease and treatment burdens that remain in CF despite transformative advances in CFTR modulator therapy. **Rare Research Launchpad faculty contributor Raksha Jain, MD, MSCI is a coauthor.**

Rare neurodevelopmental disorders: Connecting genetic diagnosis with precision treatment

Assadourian AA, Martinez-Agosto JA. [Precision diagnostic and therapeutic interventions in rare genetic neurodevelopmental disorders](#). *Pediatric Research*. 2025;98(7):2491-2502. doi: 10.1038/s41390-025-04611-y

This review explores how genetic diagnosis can guide more individualized care and how approaches such as small-molecule drugs, gene therapy, and RNA-based treatments are creating new possibilities and new translational challenges for rare neurodevelopmental disorders. **Rare Research Launchpad faculty contributor Julian A. Martinez, MD, PhD, FACMG is a coauthor.**

Try this with an area that interests you! Find a few recent publications from a lab, researcher, disease area, or scientific field you're curious about. You don't need to understand every detail. Look for the questions researchers are asking, the approaches they use, the people they collaborate with, and what they say still needs to be learned. Those clues can help you identify active areas of research and think about where your own interests or skills might fit.

PREPARE

Step 2: Build the Right Foundation

Don't worry about building the "perfect" transcript.

The researchers who contributed to this toolkit recommended courses such as genetics, cell biology, molecular biology, neuroscience, statistics, bioinformatics, epidemiology, and pharmacology as a starting point.

This doesn't mean you need to take all of them. Your options will depend on your major, your school, and where you are in your education. Use this list to explore rather than as a checklist of requirements.

If you have room in your schedule for an elective...

These courses can help build skills in reading scientific literature, critical thinking, and experimental design (a scientific framework to find cause-and-effect relationships).

Interested in lab research?

Consider genetics, cell biology, molecular biology, biochemistry, or neuroscience.

Interested in data?

Consider statistics, data science, or a research-related programming course (such as Programming for Scientists, Scientific Computing with Python, etc.).

Interested in clinical research?

Consider statistics, epidemiology, public health, pharmacology, or research methods.

Don't have room for electives, or your school doesn't offer them?

That's okay. Coursework is only one way to build your foundation. Look for workshops, seminars, online courses, journal clubs, or research experiences where you can begin developing the same skills. Ask a professor or researcher, *"If I can't take a course in this area right now, is there a resource you'd recommend for learning the basics?"*

Step 3: Start Getting Experience

Your first research experience does not have to be in rare disease. When you're getting started, experience in a real research environment can help you learn how research works, build skills, and discover what you enjoy (and sometimes discover what you don't!). A lab studying cancer biology, a neuroscience project, a public health dataset, or a clinical study may teach you skills you can then bring to rare disease research.

And your first experience doesn't have to become your permanent path. It's okay to learn from one research experience and then take those skills somewhere new. Each experience can help you make a more informed decision about what you want to try next.

If you've never done research before, try one of these first experiences:

- **Attend a research talk.** Universities often host free seminars where scientists present their work. Look at the events pages for your medical school, or biology, genetics, neuroscience, or other science departments. You don't need to understand everything. Go to hear what researchers actually talk about.
- **Read about a lab at your school.** Choose one faculty member whose work sounds interesting. Read the lab website. What questions are they trying to answer?
- **Ask a professor about research.** You don't have to ask them for a job. Try: *"I'm interested in getting some research experience, but I'm not sure where to start. Do you have any advice?"*
- (If you are an undergrad) **Look for undergraduate research programs.** Search your school's website for terms like "undergraduate research," "summer research," "research fellowship," or "student research opportunities."

- **Explore rare disease patient organizations' websites.** Patient organizations often support research, fund grants, maintain registries, host scientific meetings, or partner with researchers. It's a great way to see how science connects with the people it is intended to help.
- **Attend a national research conference.** Many research organizations offer students registration, and many sponsor their own undergraduate student research conferences. You do not need to present your research to participate, and if one is available close to where you live, it can be an opportunity to learn more and meet others that share your interests.

Ready to move from exploring to doing?

Look for a manageable first role where someone can teach you. That might be volunteering for a few hours in a lab, helping with an existing faculty project, completing a research-for-credit experience, applying for a summer program, working as a student research assistant, or contributing to a clinical or data-focused project.

You don't need to lead your own project. Your first goal is to **join a research environment where you can learn how research is done.**

Before you can start, you may need some training.

Finding a research opportunity doesn't always mean you can begin research immediately. Depending on the type of work you'll be doing, your institution or research team may require training before you can participate. For example:

- **Working with people or their health information?** You may need training in human subjects research, research ethics, privacy, or your institution's Institutional Review Board (IRB) requirements. Some institutions use the CITI (Collaborative Institutional Training Initiative) Program for this training. It covers topics such as human research ethics, regulations, and protections that research team members may be required before participating in human subjects research.
- **Working in a lab?** You may need laboratory safety training before you can work independently with equipment, chemicals, biological materials, or other potential hazards.
- **Working with animals?** Additional institution-specific training and approval requirements may apply.

Don't worry if you haven't completed these trainings before applying for your first research role. Your research mentor or institution should tell you what is required and how to complete it. **If you're offered an opportunity, ask what training or onboarding you'll need before you can begin.**

CONNECT

Step 4: Find Opportunities

Don't wait for opportunities to come to you. Researchers across NORD's Rare Disease Centers of Excellence Network highly recommend that you:

- Reach out directly to faculty and researchers with personalized emails about your interest in working in their lab and gaining experience.
- Ask your mentors to introduce you to researchers they know.
- Apply broadly and early to summer research programs.

Want to see where rare disease research is happening? Explore the [NORD Rare Disease Centers of Excellence directory](#) to find academic medical and research institutions around the country with expertise in rare disease care and research. Once you identify a Center, don't stop at the Center's main webpage. Explore its affiliated departments, faculty profiles, laboratories, research programs, and training opportunities.

- Look beyond your own institution.

Remember that researchers were students once too. If someone's work interests you, reach out to them. Be specific about what caught your attention and what you're hoping to learn. Not everyone will respond or have an opening, but **one thoughtful email can lead to a conversation, a connection, or an opportunity you wouldn't have found otherwise.**

- Build relationships early.

How to contact a researcher you don't know

You don't need a perfect email. Personalize it and keep it short. Here is an example:

Dear Dr. [Last Name],

I am a second-year biology student at [University] interested in learning more about rare disease research. I recently came across your work on [specific topic], and I was especially interested in [one genuine detail].

I'm hoping to gain research experience and would be grateful to learn whether there might be opportunities for students to contribute to your lab, now or in the future. Can you please let me know if there might be a current or upcoming opportunity for me to explore with your lab, and if so, when we might be able to meet in person or virtually to talk about that opportunity?

Thank you very much for your time.

Sincerely,

[Your First & Last Name]

Do not be discouraged if you don't hear back. Researchers receive a lot of email, and a lack of response usually doesn't tell you anything about your potential. Contact several researchers whose work genuinely interests you. If someone says their lab doesn't have space, you can also ask whether they know another researcher you might contact.

If you don't hear back: Wait one to two weeks and send one short, polite follow-up. Here is an example:

Dear Dr. [Last Name],

I'm following up on my message below about possible opportunities to gain research experience with your lab. I remain very interested in your work on [topic] and would be grateful for the opportunity to speak with you if you anticipate any openings for students. Thank you again for your time.

Including your resume can let others know your school performance, any awards you have received, and activities that demonstrate your ability to work well with others.

If you still haven't received a response, move on and contact someone else whose work interests you. Researchers are busy, and a nonresponse isn't necessarily a rejection.

Use These Tools:

As you explore labs, research programs, summer experiences, internships, jobs, and other opportunities, use the [Research Opportunity Search Planner](#) at the end of this toolkit to keep track of what you find and decide which opportunities are worth pursuing.

Found a few opportunities? Now compare them to find which most strongly aligns with what you need next by using the [Research Opportunity Comparison Tool](#) at the end of this toolkit.

Step 5: Find a Mentor

Good mentors challenge you, answer questions, introduce you to others, and help you grow. **However, a mentor doesn't have to be one person who guides your entire career.** One professor might help you find your first lab. A graduate student may teach you lab techniques. Another researcher might help you think about graduate school. Over time, you'll likely build a network of people who help you in different ways.

Look for good mentorship, not just an impressive name. A well-known lab or institution can be exciting, but the people you work with will shape how much you learn. Look for opportunities where someone has the time and willingness to teach you, answer questions, give feedback, and help you take on more responsibility as you're ready.

Who could be a mentor to you right now?

- A professor whose class you enjoyed or a teaching assistant (TA)

- Your academic advisor
- A researcher in a lab where you volunteer or work
- A graduate student, postdoctoral fellow, or research coordinator
- Someone you meet through a summer research program or at a scientific conference

Good To Know: You do not need to find one mentor who does everything. Different people may help you build skills, make connections, understand career options, or navigate opportunities.

Use This Tool: Use the [Mentor Map: Building Your Network tool](#) at the end of this guide to think about what you need help with, reflect on who you already have in your network, and plan to find the kind of mentor connection you may be missing.

You don't have to ask, "Will you be my mentor?"

Mentoring relationships often develop naturally. Start with a smaller ask such as a brief conversation, ask for advice about a decision you're making, or follow up with someone you met through a class or at a seminar or conference. If their advice is helpful, stay in touch and let them know how things go. For example:

"Your advice about looking for clinical research positions was really helpful. I applied to three programs and have an interview next week."

Step 6: Participate in Conferences and Communities

Students often think that medical or scientific conferences are for famous professors. They're not. They're where you discover research, meet mentors, hear new ideas, and find opportunities to get involved.

You don't need to be presenting research to attend many scientific meetings. Look for student registration rates, travel awards, trainee programs, virtual attendance options, and opportunities to volunteer. If a full national conference isn't realistic for you yet, start with a local research symposium or scientific meeting at your own university.

If you attend, give yourself three goals:

- ☐ Attend one session on something you know little about.
- ☐ Introduce yourself to one researcher or trainee.
- ☐ Write down one question or topic you want to explore afterward.

Not sure what to say when introducing yourself to a researcher? Keep it simple.

After a session, try something like: *“Hi, I’m [Your Name], and I’m a [student/trainee] at [institution]. I’m interested in [research area/topic], and I really liked what you said about [specific point they made]. I’m still exploring the field. Do you have any advice for someone hoping to get experience in this area?”*

If you have a good conversation, ask whether it’s okay to connect by email or LinkedIn afterward. Within a few days, send a short note reminding them where you met and thanking them for their advice.

TAKE YOUR NEXT STEP

Step 7: Find Programs and Funding That Match Your Stage

Research opportunities look different depending on where you are in your training. An undergraduate may be looking for a first lab experience or summer research program, while a medical or graduate student may be looking for an internship, protected research time, specialized training, a fellowship, or funding to pursue an independent project.

Before you apply, look beyond the program title. Read the program description carefully and find out:

- What would you actually be doing?
- Who would mentor you, and how is that mentorship structured?
- What skills or experiences could you gain?
- Is the experience paid or funded?
- What is the time commitment?
- Are there opportunities to present, publish, or continue the work?
- What do former participants go on to do?

Then, use the [Research Opportunity Comparison Tool](#) at the end of this toolkit to compare your strongest options.

You don't need to pursue every opportunity below. Start with the section that best describes where you are now – or where you're headed next.

Undergraduate Students

What you might be looking for:

Your first substantial research experience, exposure to a field you're considering, or the chance to spend a summer working full-time in research.

Look for:

- **Research opportunities at your own college or university:** Faculty labs may hire undergraduate research assistants, offer research-for-credit opportunities, or welcome students through structured undergraduate research programs. Your undergraduate research office, academic department, or professors can help you find opportunities that may not be widely advertised.
- **Summer undergraduate research programs:** Universities, academic medical centers, research institutes, and other organizations offer structured summer experiences where students work with a research mentor. Many programs provide a stipend or other financial support.
- **Research Experiences for Undergraduates (REUs):** These structured programs, supported by the National Science Foundation and other institutions, give undergraduate students the opportunity to spend a summer conducting research, often at an institution other than their own.
- **[National Institutes of Health \(NIH\) Summer Internship Program \(SIP\)](#):** College students can spend the summer conducting biomedical, behavioral, or social science research in an NIH research group. These are paid, full-time research experiences.
- **Paid research positions:** Search your university's student employment system and the career pages of nearby universities, hospitals, and research institutes for positions such as undergraduate research assistant, lab assistant, or student research assistant.
- **Research opportunities during the academic year:** Don't limit your search to summer. Some labs hire students part-time during the semester, and programs such as work-study, research-for-credit, or institutionally funded undergraduate research may make it possible to gain experience while you're taking classes.

Good to Know: Research experience does not have to mean unpaid work.

Volunteering can be one way to gain experience, but it isn't financially realistic for everyone. Look for paid research assistant positions, paid summer programs such as REUs and NIH internships, work-study positions that can be used for research, undergraduate research fellowships, and other funded opportunities. If you find a lab or researcher whose work interests you, it's okay to ask whether paid positions, funding or other forms of support are available.

Where to start:

There are several ways to begin finding opportunities that fit your interests and stage of training. Here are some practical places to start:

1. **Find out whether your school has an undergraduate research office.** Search “[school name] undergraduate research,” “student research,” or “summer research.”
2. **Look at faculty research pages.** Start with departments related to your interests and identify 3 or more faculty members whose work sounds interesting.
3. **Ask professors and advisors,** especially professors whose classes you've enjoyed. Ask whether they know of labs looking for undergraduate help.
4. **Search beyond your institution.** Explore hospitals, research institutions, universities, NIH programs, and Research Experiences for Undergraduates (REUs). Try terms such as “summer biomedical research undergraduate,” “rare disease summer research,” “genetics undergraduate research,” or “REU molecular biology.” Many programs accept students from other universities.
5. **Start early for summer opportunities.** Many summer programs begin accepting applications **months before summer**, so start looking in the fall and early winter.
6. **Don't rule yourself out too quickly.** Read eligibility requirements carefully, but don't assume you need prior research experience unless the program actually says it is mandatory.
7. **Ask about funding you may not know exists.** If a faculty member is interested in having you join their research team, but funding is a concern, ask whether there are institutional undergraduate research funds, work-study opportunities, scholarships, or grant-supported options that could support your participation. Some NIH-funded researchers may also be able to request supplemental funding to support eligible students through programs designed to broaden participation in biomedical research. Your mentor or institution's research office can help determine whether an option like this applies to you.

Medical Students

What you might be looking for:

A way to explore research despite a packed medical school curriculum, a summer research experience, a scholarly concentration/project, dedicated research time, or mentorship in a specialty you're considering.

Look for:

- **Summer research programs for medical students:** Many medical schools, academic medical centers, NIH programs, foundations, and other professional organizations offer structured summer research experiences. These can be especially useful between the first and second years of medical school, when students may have a block of time available for research.
- **Research or scholarly concentrations:** Your med school may offer a pathway, track, concentration, or required scholarly project that allows you to pursue a research question with a faculty mentor over an extended period. A project doesn't have to carry a "rare disease" label to help you build relevant expertise.
- **Funded research fellowships:** Look for fellowships that provide financial support to conduct a mentored research project. Some support summer projects, while others fund longer periods of dedicated research.
- **Research years or other dedicated research periods:** Some medical students take several months or a full year away from the standard curriculum to focus primarily on research. This can provide time to take greater ownership of a project and further develop your research skills.
- **Clinical and translational research opportunities:** Academic medical centers may offer opportunities to contribute to clinical trials, natural history studies, patient registries, chart review studies, outcomes research, or other research involving patients and health data.
- **Opportunities through specialty medical societies:** If you're considering a field such as pediatrics, neurology, genetics, hematology, immunology, or another specialty, explore its professional societies. Some offer student research programs, mentorship, fellowships, travel awards, or other opportunities for trainees.

- **NIH-supported research and training opportunities:** NIH offers and funds research experiences at different stages of medical training. Some take place at NIH, while others are offered through universities and academic medical centers.

Medical school doesn't always leave much room for electives. You don't need to add a long list of courses to become involved in rare disease research. A mentored project, summer experience, longitudinal scholarly project, or dedicated research period may be a much more realistic way to build experience.

Where to start:

1. Find out what's already available at your medical school.

Start with your school's research office, student affairs office, scholarly concentration program, or the equivalent. Search the website for terms like "medical student research," "summer research," "scholarly project," "research fellowship," and "research year." You may already have access to funding, mentors or structured programs that aren't widely advertised outside of your institution.

2. Look beyond the medical school itself.

Don't assume that all research opportunities will be listed through your medical school's student research office. Explore the websites of affiliated hospitals, children's hospitals, genetics clinics, rare disease programs and disease-specific specialty centers, and other research centers within your health system. A faculty member who teaches at your school may conduct their research at an affiliated institution.

3. Search by the specialty or science that interests you (not just "rare disease").

A student interested in rare neurological diseases might find an excellent training opportunity in neuroscience, neurogenetics, gene therapy, or neuroimmunology that never uses the words "rare disease." The same applies to immunology, metabolism, genomics, bioinformatics, cell biology, and other fields.

4. Identify a few faculty members whose work genuinely interests you.

Read their faculty profiles and lab or research program pages. You don't need to understand every publication. Learn enough to identify *why* their work interests you, then reach out with a short, specific message asking whether they mentor medical students or know of opportunities to get involved.

5. Ask students ahead of you.

Upper-year medical students, residents, fellows, and MD/PhD students can be excellent sources of practical information. Ask who mentors students well, which programs were worthwhile, and how they fit research into medical training.

6. Be realistic about the time you can commit.

Different opportunities require different levels of commitment. If you have a defined block of time available (like eight weeks available this summer), look for a defined project or structured summer program. If you want to pursue research alongside your regular coursework and clinical training, look for a longitudinal project designed for medical student participation. If you know you want more intensive research training, explore whether a dedicated research period or research year fits your goals.

You don't have to choose between medicine and research.

Physicians contribute to rare disease research in many ways. They identify questions that emerge in patient care, lead clinical studies, collaborate with lab scientists, analyze data, and develop and test new treatments. Your research involvement can grow and change throughout your medical training and career.

Graduate Students

What you might be looking for:

Opportunities to expand your research expertise, gain skills outside your primary lab or discipline, build collaborations, secure funding for your own research or training, and begin developing an independent research identity.

Look for:

- **NIH-funded institutional training programs (T32 programs):** Training programs based at universities and academic medical centers that support predoctoral and/or postdoctoral researchers in specific areas of biomedical research
 - Start here: Ask whether your institution has T32 programs related to genetics, neuroscience, translational science, clinical research, data science, or another area relevant to your interests.
- **Individual predoctoral fellowships (such as the NIH F31):** Fellowships that support your own research training
 - These generally require you to develop a research and training plan with a faculty sponsor and can be an important early opportunity to gain experience developing a funding proposal.

- **Rare disease foundation and patient advocacy organization funding:** Many disease-specific organizations offer fellowships, research grants, trainee awards, pilot funding, or other support for early-career researchers. *See more in the section below on [Opportunities that Cut Across Stages](#).*
- **Specialized research training:** Look for short courses, workshops, summer institutes, or intensive training programs in areas such as genomics, bioinformatics, clinical and translational research, drug development, gene and cell therapy, statistics, or other methods relevant to your work.
- **Opportunities to learn a new technique or approach:** If your research project would benefit from a method your lab doesn't use, talk with your advisor or a mentor about spending time with another lab or collaborator to learn it. This could mean gaining experience with a lab technique, computational method, animal model, clinical dataset, or another research method.
- **Conference travel and trainee awards:** Scientific societies, universities, foundations and patient organizations may offer travel grants, poster awards, trainee scholarships, or reduced registration for graduate students. These can make it possible to attend meetings that would otherwise be too expensive.

Good to Know: You may already be building skills that rare disease research needs. **Your current project or dissertation does not necessarily have to be labeled “rare disease research”** to prepare you for this field. Skills and discoveries in genetics, protein biology, immunology, neuroscience, cell biology, computational genomics, bioinformatics, drug development, clinical research methods, and many other areas can translate directly into rare disease research.

Where to start:

1. Start with your advisor or program director.

Ask: “Are there training grants, fellowships, or funding opportunities I should be considering at this stage of my training?” They may know about institutional programs that aren't obvious from a general web search.

2. Find out which NIH training grants exist at your institution.

Search your university or academic medical center website for terms such as “T32,” “predoctoral training,” “NIH training grant,” and your research area. Your graduate program or Office of Graduate Education may also maintain a list.

3. Explore individual fellowship opportunities early.

If you're interested in an NIH F31 or another fellowship, talk with your advisor or mentor and your institution's research or grants office well before the deadline. Developing a competitive application takes time.

4. Search by the science (not only by "rare disease").

See more in the section above for [Medical Students](#).

5. Look at organizations connected to the diseases you study.

See more in the section below on [Opportunities that Cut Across Stages](#).

6. Ask researchers one step ahead of you.

A senior graduate student, postdoc, or recently funded fellow can often tell you about opportunities you wouldn't know to search for and what was actually useful to them.

Gap Year and Post-Baccalaureate Trainees

What you might be looking for:

A year or two of focused research experience before medical school, graduate school, or another next step; an opportunity to strengthen your research skills, more time to explore what kind of research interests you; or experience that will help you decide whether a research-focused career is right for you.

Look for:

- **NIH Postbaccalaureate Research Program:** [An opportunity for recent college graduates to conduct full-time biomedical research alongside NIH scientists](#). These can provide intensive research experience, mentorship, and exposure to a large scientific community before graduate or professional school.
- **Research assistant or research technician positions:** Paid, full-time jobs in academic or research labs. Depending on the lab, you might help conduct experiments, maintain cell cultures, collect and analyze data, manage lab materials, or contribute to ongoing research projects. These positions can be an excellent way to build substantial hands-on experience.
- **Clinical research coordinator positions:** Paid roles that support research involving patients. Depending on the position, you might help recruit study participants, coordinate study visits, collect research data, maintain study records, or support clinical trials and natural history studies. These can be especially useful if you're interested in medicine or clinical and translational research.
- **Structured post-baccalaureate research programs:** Universities, academic medical centers, and research institutions may offer one- or two-year programs specifically for recent graduates. Unlike other jobs in a lab, these programs may combine research with mentorship, professional

development, seminars, and preparation for graduate or medical school.

- **Data or computational research positions:** If your interests are in bioinformatics, genomics, statistics, or data science, look for entry-level research positions involving biological, genetic, or clinical data. Not every research opportunity happens at a lab bench.
- **Research positions at hospitals and academic medical centers:** Rare disease research often happens within departments such as genetics, neurology, pediatrics, immunology, hematology, or other specialties. Searching the employment pages of major academic medical centers can uncover opportunities that aren't advertised as "rare disease research."

Where to start:

1. Decide what you want most from the year.

You don't need a detailed career plan, but ask yourself: ***What do I want? Hands-on lab experience? Experience working with patients? Stronger data skills? A chance to explore whether I enjoy research?*** That will help narrow your search.

2. Don't search only for "post-baccalaureate programs."

Many great gap year opportunities are jobs, not programs. Search job boards using titles such as "research assistant," "research technician," "laboratory (or lab) technician," "clinical research assistant," "clinical research coordinator," and "research associate" along with a scientific area that interests you.

3. Search by scientific interest, not just "rare disease."

For example, if you're interested in rare neurological diseases, try searches involving neuroscience, neurogenetics, leukodystrophy, gene therapy, or neuromuscular disease. **A lab may conduct rare disease research without describing itself that way.**

4. Look directly at research institutions.

Explore the career and research pages of universities, medical schools, children's hospitals, academic medical centers, NIH, and independent research institutes. If you discover a researcher whose work interests you, look at both the lab's website **and** the institution's employment listings.

5. Ask professors and researchers you already know.

Ask: "I'm considering spending a year doing research before my next step. Do you know of labs or investigators who might be looking for a research assistant?" Faculty often hear about openings through their professional networks before students do.

6. Start looking well before you want to begin.

Formal programs may have application deadlines months in advance, while research jobs are often posted throughout the year and filled closer to the desired start date. Start researching formal programs early and continue checking for job openings as your planned gap year gets

closer.

Good to Know: A job can be a research opportunity. You don't necessarily need to find a program with "fellowship" in the title. Entry-level research assistant, laboratory technician, and clinical research coordinator positions can provide substantial hands-on experience **while you're being paid.**

Opportunities that Cut Across Stages

- **NIH Opportunities:** The National Institutes of Health (NIH) supports research training **at many career stages**, from summer experiences and post-baccalaureate programs to graduate and postdoctoral fellowships. Some opportunities take place at NIH itself, while others are NIH-funded programs offered through universities at research institutions.
Start here:
 - o Explore NIH-funded research training opportunities by career stage: [NIH Research Training & Career Development](#)
 - o Interested in conducting research at NIH? Explore internships, post-baccalaureate, graduate, and postdoctoral research opportunities through the [NIH Office of Intramural Training and Education](#).
- **Rare Disease Foundations and Patient Advocacy Organizations:** Disease-specific foundations and patient advocacy organizations don't only support patients and families. Many also fund research, offer fellowships or travel awards, host scientific meetings, connect researchers, and support early-career investigators.

Start here: If there's a rare disease or disease area you're interested in, find the organizations

Good to Know: [NORD's Member List](#) could be a great place to start researching patient advocacy organizations for rare conditions.

serving that community and look for sections of their websites labeled "Researchers," "For Researchers," "Grants," "Grant Program," "Young Investigators," or "Scientific Programs."

- **Industry:** Pharmaceutical, biotechnology, diagnostics, and other life science companies employ scientists and clinicians working on rare diseases at nearly every stage of therapy development. Opportunities may include internships, fellowships, co-ops, entry-level research positions, clinical development, data science, regulatory science, medical affairs, and other roles.

Start here: When you hear about a therapy or technology that interests you, find out which company developed it. Visit its Careers page on its website and look at the backgrounds and skills required for internships and early-career positions, even if you aren't ready to apply yet.

Advice from Rare Disease Researchers

We asked researchers and clinician-researchers in [NORD's Rare Disease Centers of Excellence Network](#) what they wish students and trainees knew as they explore the field. Here are some of the messages that came through most clearly.

- **Get involved and start where you can.** Look for opportunities to join a lab or clinical research team, participate in a summer research program, shadow researchers or clinicians, attend research seminars and conferences, or connect with patient advocacy organizations. You don't need extensive experience or a clear career plan before you begin. **Start gaining exposure early and use those experiences to find out what interests you.**
- **Don't wait for an opportunity to be posted.** Many research opportunities are **not** advertised. Rare disease researchers cautioned against relying only on posted positions and encouraged students to identify researchers whose work interests you and reach out directly. Before contacting someone, **learn about their work and explain specifically what interests you (about their current project or recent publication of theirs) and why you're reaching out.** Avoid sending the same generic email to dozens of researchers.
- **Be curious, reliable, and willing to take initiative.** Knowing every technique isn't what makes someone a strong beginning researcher. Rare disease researchers emphasized qualities such as **curiosity, self-directed learning, initiative, and reliability.** If you join a research team, show up prepared, follow through on what you agreed to do, ask thoughtful questions, and look for opportunities to learn rather than waiting for someone to map out the next steps for you.
- **Get close to the people and problems behind the research.** Rare disease researchers recommended asking for opportunities to shadow clinicians and researchers, learn about diagnosis and care, and engage with patient advocacy organizations and rare disease

communities. They emphasized that understanding disease manifestations, diagnosis, care, and treatment gives researchers important context for **why the research matters**.

Good to Know: Engage with rare disease communities with respect and humility.

Patient organizations, people living with rare diseases, and families can offer perspectives that researchers cannot gain from scientific literature alone. If you have opportunities to learn from or engage with a rare disease community, **approach those relationships as opportunities to listen and learn** (not simply to recruit participants, collect information, or advance your own research).

Respect people's time, privacy, and lived experience. Don't assume that one person's experience represents everyone with a condition and be thoughtful about how you ask questions or talk about disease. When possible, follow the lead of established patient organizations and community partners, and remember that meaningful engagement is built on trust and mutual respect.

- **Give yourself enough time to actually learn.** Research takes time. Rare disease researchers cautioned against underestimating the commitment involved or taking on so much that you can't meaningfully contribute. **When considering an opportunity, be realistic about what you can commit** and give yourself enough time to learn, contribute, make mistakes, and grow from the experience.

Your Rare Research Launchpad: The Next 30 Days

You've explored different types of rare disease research, ways to build experience, people who can help you, and programs and opportunities available at different stages of training. **Now, choose a few actions you can realistically take in the next 30 days.**

If you're just starting to explore rare disease research...

Choose 3:

- ☐ **Explore one type of research.** Read about a lab, watch a research lecture, try one of the resources in this toolkit, or learn about a project that sounds interesting.
- ☐ **Identify three researchers or research groups** whose work interests you. Use faculty webpages, research institute or hospital websites, ClinicalTrials.gov, or other strategies from this toolkit.
- ☐ **Talk to one person about research.** Ask a professor, advisor, grad student, or researcher about how they got involved and what they recommend as a first step.

- ☐ **Try something.** Complete a beginner bioinformatics tutorial, attend a research seminar, visit a lab open house, or attend a patient advocacy organization research event.
- ☐ **Look for one real opportunity.** Find a summer program, internship, paid or volunteer research position, research-for-credit option, lab opportunity, or other experience you could realistically pursue.
- ☐ **Reach out.** Contact one researcher whose work genuinely interests you and ask about opportunities or advice for getting started.

If you already have some research experience...

Choose 3:

- ☐ **Identify what you want next.** Is there a skill you want to build, a type of research you want to try, a disease area you want to explore, or a part of the research process you haven't experienced yet?
- ☐ **Take greater ownership of something you're already doing.** Ask whether you can help plan an experiment, work with a different dataset, contribute to an analysis, attend a study design meeting, present findings, or take responsibility for a small piece of a project.
- ☐ **Expand your network.** Identify and contact one researcher, clinician, data scientist, patient organization, or other person outside your current research group whose work connects with your interests.
- ☐ **Explore how your current expertise could apply to rare disease.** Find two or three rare disease researchers or teams using the skills, methods, or scientific area you're already developing.
- ☐ **Find one next-stage opportunity.** Identify a fellowship, research year, training grant, conference, travel award, internship, research position, or other opportunity appropriate to your career stage.
- ☐ **Have a next step conversation with a mentor.** Tell them what you're interested in developing and ask for one concrete suggestion, connection, or opportunity that could help you get there.

Rare disease research needs people who are willing to ask questions, learn from others, and keep looking for answers. You don't need to know today exactly where you'll fit or where your path will lead. Start with the next opportunity to learn, contribute, or connect, and see where it takes you!

Research Opportunity Search Planner

Use this tool to keep track of research opportunities you find and identify the next step you can take to explore each one.

Research Area / Research Question I'm Interested In	Search Terms I Used	Opportunity (Lab / Program)	Researcher or Contact	What I Need to Find Out / My Next Step	Follow- Up/Deadline

Research Opportunity Comparison Tool

Use this tool to compare your opportunities and determine which are the best fit for what you need next.

Opportunity	What Could I Learn?	What Would I Actually Do?	Location / Remote	Time Commitment	Paid / Funded?	Mentorship / Training Provided?	Chance to Present / Publish?	Deadline / Next Step

Mentor Map: Building Your Network

Use this tool to identify the people who can support different parts of your research journey and plan where you may want to build new connections.

What I Need Help With	Who Could Help?	Person I Already Know	Person I could Meet	My Next Step
Learning a research skill	Graduate student, postdoc, lab manager			
Finding a research opportunity	Professor, advisor, PI			
Understanding a career path	Physician-scientist, PhD researcher, clinician-researcher			
Applying for funding or programs	Advisor, program director, grants office			
Meeting people in the field	Conference contact, medical society member, patient advocacy organization			
Figuring out my next step	Someone 1-2 stages ahead of me			