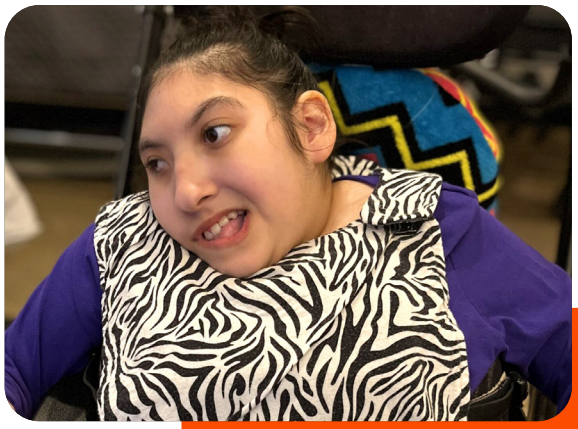

Pediatric Rare Dx

A Parent's Guide to a Rare Disease Diagnosis in Children

Created by the National Organization for Rare Disorders (NORD®) Rare Disease Centers of Excellence

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NORD®
National Organization for Rare Disorders

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The NORD® Rare Disease Centers of Excellence Network is dedicated to diagnosing rare diseases as soon as symptoms appear, while also advancing research to make an accurate diagnosis faster.

Rare Diseases and the Diagnostic Odyssey

In the United States, rare diseases are defined as medical conditions affecting fewer than 200,000 people. Over 30 million Americans, or **close to 1 in 10**, are living with a rare disease. Approximately 80% of these rare diseases either have, or are strongly suspected to have, a genetic cause, and **over half affect children**.

As the parent or guardian of a child with a suspected rare disease, you may find that you and your child are on a **diagnostic odyssey**. This journey often begins when your child experiences unexplained symptoms, continues as the child is referred to specialists, and ends when a final diagnosis is made. The diagnostic odyssey can last months, years, or even decades. Not all rare diseases can be diagnosed at this time, but many can. As medical science advances, more children who have a rare disease will be able to be diagnosed.

The NORD® Rare Disease Centers of Excellence Program aims to diagnose children who have a rare disease as early as possible after symptoms begin and advance science so more rare diseases can be diagnosed.

This guide has been prepared by a working group of doctors, researchers, genetic counselors, laboratory directors, and others at NORD Rare Disease Centers of Excellence to help you better understand the diagnostic pathway and how you and your child can partner with your child's primary care provider (PCP) to work toward finding a diagnosis. This group has also prepared a separate guide to help pediatric PCPs support their patients and patients' caregivers through the diagnostic odyssey (Pediatric Rare Dx: A PCP's Guide to Diagnosing Rare Disease in Children).

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Could my child have a rare disease?

As a parent or guardian, you may *know* or *feel* something is wrong with your child's health, growth, or development but you may not know what to do next. Many people who have a rare disease develop symptoms as an infant or child. Some people who have a rare disease may even have symptoms that are noticed before birth. Others may not have their first symptoms until they are a teenager or adult. Certain symptoms may be specific enough that a rare disease can be diagnosed right away, while the symptoms of other rare diseases can be more challenging to recognize and diagnose.

Signs and symptoms that can raise suspicion of a rare disease include:

Concerns found on an ultrasound during pregnancy, such as slow growth, one or more parts of the body not forming as expected, or low amniotic fluid

Major differences noticed at birth or shortly after (sometimes called birth defects) affecting the way more than one part of the body looks or works, such as a heart defect, large or small head size, cleft palate, or problems urinating

Abnormal newborn screening test results

Feeding issues, such as difficulty swallowing, struggles with breastfeeding, poor appetite, or overeating

Differences in development, such as walking or talking later than average or losing previously developed skills

Seizures, especially if unprovoked, frequent, or difficult to control

Social or behavioral differences, such as not smiling, difficulty interacting with others, constant upset behavior or crying, or a diagnosis of autism

Differences in growth, such as growing too slowly or too quickly, or one part of the body growing out of proportion to the rest

Medical problems affecting more than one part of the body

Health problems that began early in life or that continue to get worse

Illnesses that are excessively frequent, severe, or prolonged, or that do not respond to treatment as expected

Family history of similar unexplained symptoms

If you have these or other concerns about your child, this guide will help you learn ways to work together with your child's primary care provider (PCP) toward finding a diagnosis.

What should I do if I suspect my child has a rare disease?

1. Talk with your child's primary care provider

If you feel like something is wrong with your child's health or development, reach out to your child's primary care provider (PCP) for help. A PCP may be a doctor who practices general medicine, such as a family doctor, pediatrician, or internal medicine doctor. A PCP may also be a nurse practitioner (NP) or physician assistant (PA), both of whom have the ability to perform examinations, make diagnoses, and prescribe medication. Sharing your concerns with your child's PCP is the first step to finding out if there may be a medical problem.

When working toward a diagnosis for your child, it is helpful to have a PCP who listens and understands your concerns and can recognize the possibility of a rare disease. A trusted PCP who knows your child well can recommend testing options, refer to specialists, and provide care tailored to the specific needs of you and your family.

2. Take your child for regular checkups to monitor development

Regularly scheduled checkups with your child's PCP are an opportunity to discuss your child's developmental milestones. Developmental milestones include how your child is growing and when key moments happen, such as smiling, focusing on and reaching for objects, rolling over, walking, talking, and progress in school.

During your child's checkups, review when they learned specific skills and discuss any concerns you have. Your child's PCP can help determine if your child's development is on track, or if they may need extra support. Every child grows and develops on their own timeline, but if your child's development isn't within a certain range, it may alert you and your child's PCP to possible health or developmental concerns.

3. Keep in touch with your child's PCP and care team

Let your child's PCP and care team know if something changes in your child's condition or behavior. The changes you notice may prompt your child's care team to adjust their treatments or care plan. Your child's PCP may be the best person to help coordinate your child's care, especially if they need to be seen by more than one specialist.

4. Work with your child's PCP for referrals to specialists

If the PCP suspects your child's symptoms or developmental concerns may be caused by a rare medical condition that cannot be diagnosed with routine testing and evaluation, referral to a specialist is usually the next step. Discuss referrals to specialists with your PCP. Depending on your child's specific needs, the specialist may be in genetics, neurology, cardiology, nephrology, or another specialty. Each specialist has background and extra training in a specialty area, and there isn't one specialist who can diagnose or treat every condition, so it is important to find specialists or sub-specialists with an understanding of the type of disease your child may have. Some specialists may not think of checking for a rare disease, so it may be helpful to work with your child's PCP to find a specialist who has experience with rare diseases.

5. Find a specialist at a NORD Rare Disease Centers of Excellence

Finding a specialist who knows about rare diseases can be hard, although many community specialists can and do diagnose rare diseases. However, if local specialists are not providing a diagnosis for your child or a referral to medical genetics is needed, talk to your child's PCP about referring to a specialist or sub-specialist at an academic medical center, especially medical centers that are NORD Rare Disease Centers of Excellence. To be a NORD Rare Disease Centers of Excellence, the medical center must show their commitment to diagnosing, managing, and researching many different rare diseases.

What other actions can I take?

1. Make notes of your child's development, symptoms, and illnesses

Having a comprehensive record of your child's health helps you and your child's medical team care for your child better. Keep notes of development, behavior, and diet. When your child is ill, document symptoms and how long illnesses last. It can be helpful to note if other family members or the child's friends or classmates had the same illness.

2. Keep copies of your child's medical records

Many health care systems now use electronic health records that allow you to access or print copies of your child's documents, including visit notes and test results. Although electronic health records and sharing across different hospitals and doctors' offices is improving, all records are often not available to a PCP or different specialists. Save digital or printed copies of your child's health records and keep them accessible, especially if you are moving or going to see a new doctor or specialist. In many cases, your best option is to keep track of your child's records and share them with each doctor you visit.

3. Work with your child's PCP and care team to write a health summary

Before going to a specialist, if your child has many medical records or has a complicated medical history, you may ask your child's PCP or other member of the care team to help you summarize your child's health problems. The summary may include a timeline of symptoms, specialists already seen, current treatments, and important medical test results. Specialist appointments can be time-limited, so having a summary can help the specialist quickly understand your concerns about your child's health, growth, or development.

Health Summary Templates developed by the Undiagnosed Rare Diseases Network International can help your child's PCP write the summary. There is also a template you can use to write a short medical story for your child.

4. Find supportive resources

It is common for parents or caregivers to feel overwhelmed when caring for a child during the journey to a diagnosis. You may also be dealing with financial problems and social pressures, and you may feel isolated and alone. Please take a moment to search for helpful resources in the *'Resources to connect to support'* section (page 13). You may also wish to talk to your child's PCP or your own PCP to find a supportive therapist or medical social worker.

5. Make connections

Many find connecting with other rare disease families who may or may not yet be diagnosed through NORD events very helpful. Learn about NORD events by signing up for NORD's Newsletters or checking the NORD Events page. If there is a specific rare disease that your child's medical care team suspects, consider connecting with a patient advocacy and support group for that disease — most groups are very welcoming of families still on their diagnostic journey. You can search for a rare disease patient group in NORD's Organizational Database.

TAKE ACTION

Getting a diagnosis is important. A diagnosis of a rare or genetic disease may help your child get better access to specialized care such as focused treatment plans, specific health screening, and preventative care.

A diagnosis can also help you connect to other families whose children have the same rare disease. By knowing the diagnosis, you may be able to learn more about your child's rare disease and the changes that may happen over time. With a diagnosis, your child may be able to participate in a clinical trial for a new treatment if that is of interest to your family. A diagnosis may not immediately lead to more information or change the treatment your child is receiving, but with time, as more children are diagnosed with the same disease, more will be known.

Testing for Rare Diseases

Talk to your child's care team about testing options. There are many medical tests that can help with the diagnosis of a rare disease. Ask your child's PCP or specialists about the tests that your child has already had and about the need for further testing. Before you begin testing, confirm that your child had normal newborn screening tests and that no follow-up testing was needed. Further testing may include biochemical testing, imaging such as X-rays, or genetic testing.

Below is a brief introduction to the most common types of testing that may help you and your child's PCP and care team learn about your child's condition.

Biochemical testing

Biochemical testing looks at how our body uses food to make energy and grow, build, and repair itself. These processes are very complex but are well understood. If any of these processes are not working properly, tests of the blood and urine can help uncover the cause. Examples of biochemical testing include comprehensive metabolic panels; ammonia, lactate, or pyruvate levels; plasma amino acid analysis; urine organic acid analysis; and plasma acylcarnitine profile.

Imaging

There are many ways doctors can see inside the body. These types of tests are typically low risk and are important to see if the physical structures of the body, such as the bones, brain, lungs, heart, and kidneys, are formed and working properly. Some common types of imaging include X-ray, ultrasound, and MRI. Each of these imaging methods can be used to visualize the body in different ways.

Genetic testing

Genetic testing is often an important part of searching for a rare disease diagnosis. The genetic code is like an instruction book for the growth and development of our bodies. Our genetic code is made up of 23 pairs of chromosomes. One copy of each chromosome comes from a person's biological mother and the other from the biological father. Each chromosome is like a separate chapter in our instruction book and is made up of a very long strand of DNA bases or letters that spell out instructions. We have about 22,000 genes or paragraphs in our genetic instruction book that tell our body how to make proteins, which our body needs to develop, grow, and function.

Everyone's genetic code is unique, and many genetic differences don't cause health problems. However, some variants (changes or misspellings) in our genetic code can cause health problems or developmental differences. **To the right is a short description of some of the most common types of genetic tests that inspect our genetic code to find important health-related changes.**

NORD EDUCATIONAL RESOURCES: GENETIC TESTING

- NORD Genetic Testing for Rare Diseases [Infographic] | *¿QUÉ son las Pruebas Genéticas? [Infografía]*
- NORD Genetic Testing for Rare and Undiagnosed Diseases [Video] | *Pruebas Genéticas para Enfermedades Poco Comunes y No Diagnosticadas* [Video]
- Why Should I Get Genetic Testing If I Already Have a Diagnosis? [Infographic] | *¿Por qué debería hacerme pruebas genéticas incluso si tengo un diagnóstico? [Infografía]*
- Why Should I Get Genetic Testing If I Already Have a Diagnosis? [Video] | *¿Por qué debería hacerme una prueba genética si ya tengo un diagnóstico? [Video]*

Microarray analysis
A microarray analysis detects if there are sentences, paragraphs, or whole chapters of genetic information that may be extra (duplication) or missing (deletion) in your child's DNA. Sometimes these duplicated or deleted pieces of genetic information can cause medical or developmental differences depending on how much extra or missing genetic information is included in the affected piece and which genes are involved.

Chromosome and fluorescence in situ hybridization (FISH) analysis
Like microarray analysis, chromosome analysis looks to see if there are extra or missing paragraphs or chapters in our genetic instructions. However, chromosome analysis can also see if parts of the genetic code are out of order or rearranged in some way that might be causing health problems. FISH analysis looks at how many copies of one specific paragraph in the genetic code are present and on which chromosome it is found.

Panel, exome, and genome sequencing
Panel, exome, and genome sequencing are more comprehensive genetic tests that look at the spelling of our genetic code. A panel test looks at the sequence or spelling of a few dozen up to a few thousand genes related to a selected category of disease. Exome sequencing looks at the sequence of almost all 22,000 genes looking for misspellings or missing or extra letters. Genome sequencing looks at the sequence of almost all the genes and all the genetic code between genes.

Genetic testing results

Genetic testing results can be complex, and usually a genetic counselor will help you understand your child's genetic testing results. Genetic tests are typically reported as positive, negative, or uncertain.

A positive result means that one or more disease-causing or pathogenic changes called variants in the genetic code were found and are highly likely or likely to be related to your child's current symptoms or symptoms that may develop over time. A positive result generally confirms a genetic diagnosis and can guide next steps for treatment and management. It can also be important to share results with family members if testing indicates that they could be at risk for inherited diseases.

A negative result means that no changes were found that appear to be the cause of your child's symptoms. This doesn't necessarily mean that your child's symptoms are not caused by genetic variants, since our scientific knowledge and technology are not perfect. It just means that based on the testing that was done and what is known at that time, no genetic explanation was found.

An uncertain result means that one or more variants of uncertain significance (VUS) were found. This means that a change was seen in the genetic code, but based on all the information available, the laboratory cannot know for certain if that change is the cause of your child's medical condition. Sometimes additional evaluation or even testing of other family members can help determine if a VUS is related to your child's health problems. In general, making medical decisions based on a VUS is not recommended.

Sometimes genetic testing results indicate that a patient or parent is a carrier of a pathogenic variant in one copy of a specific gene. Typically, someone who is a carrier is not affected with the associated disease, because for the associated disease to cause symptoms, a person would need a pathogenic variant in both copies of the gene. A carrier can pass the genetic change onto their children, but a child would not develop that disease unless the other biological parent passed on a pathogenic variant for the same disease. In these cases, carrier testing for the other biological parent can be considered.

There is no single genetic test that can diagnose all types of rare genetic diseases. Multiple genetic tests might be ordered, so it is important to discuss the benefits and limits of each genetic test with your child's care team. Some genetic testing results may require other relatives and family members to be tested as well to help clarify your child's results. A genetic counselor can help you understand more about what further genetic tests may be needed and what the results mean for your child and your family.

While diagnostic technology has advanced, we are not able to always find the genetic cause. Even with the most comprehensive genetic testing currently available, such as genome sequencing, a genetic cause for a rare disease is only found in 25-50% of patients. There may be differences in someone's genetic code that our technology is not yet able to find, or we may not yet understand how or if a certain difference in a genetic test result can cause symptoms. Other times, the rare disease may not be genetic but may be caused by injuries, infections, exposure to certain pollutants, or other environmental factors.

What Should I Do if My Child Is Diagnosed With a Rare Disease?

Continue your child's routine care

After your child is diagnosed with a rare disease, your child's PCP will remain an important part of the care team and will often co-manage your child's care with their specialists. Your child's PCP will likely take care of common childhood illnesses, coordinate specialists and therapists, and keep an eye on the bigger picture, making sure your child, you, and your family receive the best support available.

If your child has not been seen by a specialist who has treated other children with your child's rare disease or your child requires complex care, their PCP or local specialist may be able to help find a specialist or specialized clinic for your child's rare disease or a complex care program at a NORD Rare Disease Centers of Excellence or other major academic medical center. Your PCP or local specialists can work together with specialists at the major academic center to provide your child with the best care possible.

Learn more about your child's rare disease

Finding information about your child's rare disease can be hard. As a first step, you should talk with your child's PCP and care team to learn more about the rare disease, especially with the specialist who made the diagnosis. If your child has a genetic disorder, a genetic counselor can help you find reliable information. You can also find trusted information in the *'Sources for rare disease information'* section (page 12).

You may decide to search the internet for more information about your child's rare disease but always talk to your child's PCP or medical care team about the information you find. Never try any treatment or care advice provided by patient support groups, social media groups, or other internet sources without checking with your child's PCP and medical care team first.

If you find medical articles about your child's rare disease, always share the articles with your child's medical care team. Please be aware that published case reports often discuss the most serious cases or cases with unusual complications, and therefore the information may not apply to your child.

Build a network of support

In addition to caring for your child's health, you may be caring for other children or family members. You may also be dealing with physical and emotional exhaustion, relationship issues, financial concerns, and social pressures. You may feel isolated and alone. Finding a rare disease support group where others are going through a similar diagnostic journey and keeping in contact with other families can provide much needed support and also offer an opportunity to find and share information and resources. To find patient support groups and other resources that may be helpful, please go to the *'Resources to connect you to support'* section (page 13).

Many times, families and friends want to help but don't know how, so it can be helpful to clearly tell them what you need, such as dropping off groceries or dinner, help with washing or cleaning, taking your other children to activities, or calling or stopping by just to chat. For some, asking for help can be very hard, but letting people help can build a network of support for you and your family.

Manage your expectations

It is important to understand that because a rare disease is rare, there may not be a lot known about the cause of your child's symptoms or how the disease will change over time. There may not be any treatments available to stop your child's symptoms from getting worse. There most likely will not be a cure. But a diagnosis can help connect you and your child with others, gain needed services, try new treatments when they are available, and become involved in research.

Caring for a child with a rare disease can be very time-consuming. Remember to take care of your health, both physical and mental. If you become emotionally overwhelmed, ask your PCP to refer you to a mental health provider for support.

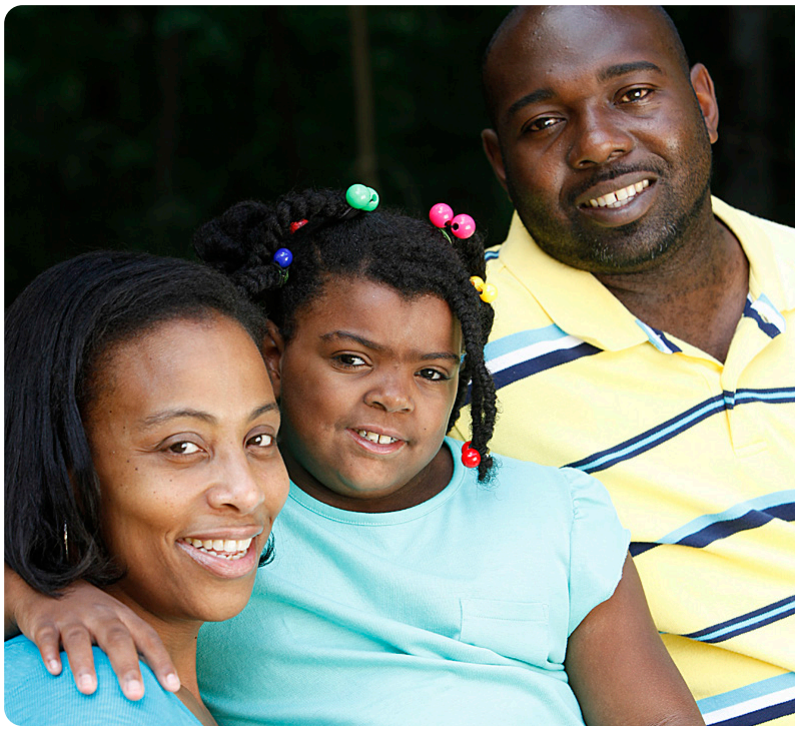
If you have other children, try to make time for them too, or ask family members or friends to help. Do your best to make sure all your children are receiving the attention needed for their proper physical, mental, and social development.

Think about enrolling your child in a rare disease registry

Many rare disease patient advocacy and support (nonprofit) groups have registries. A registry may be as simple as the contact information for families whose children have a specific rare disease. The group may use the contact registry to send out information that may be of interest to you.

Other rare disease registries are designed to learn more about the rare disease from a parent or legal guardian like you. These patient rare disease registries may collect information about how symptoms change over time or differ from child to child. This type of information can be used to learn more about the disease, find new treatments, understand what is most important to families, find gaps in resources and information, and identify families who may be interested in joining future clinical trials.

Registries can be sponsored by a government agency, nonprofit groups, hospital or academic research center, or a private company. It's always good to check first to know who sponsors the registry. Patient registries should be clear about how the information you enter into the registry will be used and what benefits, if any, you or your child may receive. Many patient registries allow you to decide how the information you enter will be shared and when you would like to be contacted. One example might be choosing to be notified of future research opportunities on the consent form.



Learn more about NORD's IAMRARE® Patient Registry

Talk to your child's PCP and specialists about research clinical trials

Research clinical trials may offer testing, monitoring, and new treatments that may not be available outside a research setting. Your child's participation in a clinical trial may also help future children who have the same rare disease. Your child's PCP and/or specialists can talk to you and your child about possible clinical trials. The National Institutes of Health (NIH) Clinical Research Trials and You website provides information that can help you learn more about clinical trials, including The Basics and For Parents and Children.

Many rare diseases will not have a clinical trial, but you can periodically check NORD's Find Clinical Trials and Research Studies or the more comprehensive listing of clinical trials, ClinicalTrials.gov. Research studies and clinical trials are also listed in NORD® Rare Disease Reports found in the NORD® Rare Disease Database. Remember to talk to your child's PCP or specialist before enrolling your child in any clinical trial.

What To Do if My Child Remains Undiagnosed?

Be patient but persistent

Sometimes an answer is not found even when all the best testing is done, and many top specialists have been seen. This does not mean the diagnostic journey is over. Sometimes it takes many months or even many years before your child's doctors can diagnose your child's rare disease. Continue to work with your child's PCP and specialists on a treatment plan as you continue to seek a diagnosis.

Watch for new discoveries

The human body and our genome are complex, and we don't understand everything about them yet. This can mean that information from your child's genetic testing may not find the cause of your child's symptoms now but may in the future as we make new medical advances and gain more knowledge about the genes in our cells. Furthermore, if your child had exome or genome sequencing, the results, such as secondary findings, may be of value throughout your child's life as they may help identify the risk of developing other diseases later in life. Request that your child's genetic specialist order re-analysis of your child's exome or genome testing if new symptoms appear or every few years until a diagnosis is found.

Learn about undiagnosed disease programs

When you and your child's doctors have exhausted all the usual clinically available options, clinical research may be an option for your child. Many of the NORD Rare Disease Centers of Excellence have research-based undiagnosed disease programs designed to discover new causes of disease. Other major academic medical institutions may also have similar programs. Generally, any testing in these research studies is paid for by the study. Ask your child's PCP, specialist, or genetic counselor about research programs that your child may be eligible to join.

Many of the undiagnosed programs at the NORD Rare Disease Centers of Excellence are programs set up by the research hospital system and referrals are made directly. Contact information for these programs can be found using the Directory for NORD® Rare Disease Centers of Excellence. Some of the undiagnosed programs are part of the Undiagnosed Diseases Network (UDN), a research program initially funded by the National Institutes of Health (NIH), and rely on a central application process.

Many NORD® Rare Disease Centers of Excellence have outreach clinics across their home state. Some routinely provide care to rare disease patients in nearby states.

To learn more about your child being seen by a specialist at a NORD Rare Disease Centers of Excellence, please see Patients and Caregivers Frequently Asked Questions. (*en español: Preguntas y respuestas del paciente/cuidador*)

Rare Disease Information and Supportive Resources

Sources for rare disease information

NORD Rare Disease Database

Provides patients and their families with information resources for more than 10,000 rare diseases, including links to GeneReviews, Orphanet, Online Mendelian Inheritance in Man (OMIM), and MedlinePlus Genetics. In addition, the NORD Rare Disease Database contains more than 1,300 NORD Rare Disease Reports in English and more than 500 in Spanish. NORD Rare Disease Reports:

- Tend to be more patient- and family-friendly than other resources but can also be a good place to start for clinicians and allied health professionals.
- Provide an overview of signs and symptoms, causes and inheritance, disorders with similar symptoms, diagnosis, standard therapies, and clinical trials and studies.
- Include a list of relevant patient advocacy, support groups, and other resources for patients.
- List references, and a growing number will include in-text citations.
- Are authored and/or reviewed by medical specialists.

NORD Resource Library

Includes videos, reports, toolkits, infographics, and webinars on everything from genetic testing and drug development to tips on how to advocate for rare diseases. Through the NORD Resource Library, caregivers of children with rare diseases will have access to the information they may find helpful throughout their child's rare disease journey. A growing number of the resources are available in Spanish.

GeneReviews®

Provides information about genetic diseases including clinical description, diagnosis, management, and genetic counseling. Although written for doctors, you may find the information helpful and can share with your child's PCP and care team.

Each chapter in GeneReviews is written by one or more experts on the specific condition or disease, and goes through a rigorous editing and peer review process before being published online. There are more than 800 chapters on specific genetic rare diseases. GeneReviews database is managed by the University of Washington.

MedlinePlus® Genetics

Provides patients and their families with information on more than 1,300 health conditions with a genetic basis, more than 1,400 genes, and basic genetic concepts. The information is written in family-friendly language and includes links to additional information and resources. MedlinePlus is a service of the National Library of Medicine (NLM), which is part of the National Institutes of Health (NIH).

Online Mendelian Inheritance in Man (OMIM®)

Provides a summary of published research about genetic diseases and includes references from the medical literature and links to other genetic information resources. OMIM focuses on the relationship between symptoms of a disease and the genetic changes causing the symptoms. Although written for doctors and researchers, you may find the information helpful and can share with your child's PCP and care team. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Ada Hamosh, MD, MPH.

Orphanet

Provides high quality information on more than 6,000 genetic and rare diseases with the aim of improving the diagnosis, care, and treatment of patients with rare diseases. Some resources are available in languages other than English. Although written for doctors, you may find the information helpful and can share with your child's PCP and care team. Orphanet is based in Europe and funded by the French National Institute for Health and Medical Research and the Health Programme of the European Union.

Resources to connect you to support

NORD's Organizational Database

Offers a disease-searchable listing of non-profit rare disease patient support and advocacy groups to help connect you to needed support. The patient support group may be dedicated to a single rare disease or a group of related or similar rare diseases. Many groups advance research for their rare disease(s). Disease-specific non-profit patient advocacy and support groups may:

- Provide information about their rare disease(s), including diagnosis and management, in family-friendly language.
- Connect families with others living with the same rare disease or facing similar challenges.
- Offer or link to supportive services and assistance programs.
- Share information about rare disease research and clinical study opportunities.
- Include a list of specialists or expert centers for diagnosis or care.

NORD State Resource Center

Contains state-specific organizations that offer free or low-cost programs and services for patients and families impacted by rare disease.

NORD Support Helpline

Please call NORD at **1-800-999-6673** or email us at **informationservices@rarediseases.org**.

*Si deseas hablar con alguien en espanol por favor llame al **1-844-259-7178** para asistencia.*

NORD RareCare® Patient Assistance Programs

Offers help to patients and families who need assistance paying for medical bills, traveling to a treatment center, or participating in clinical trials. NORD also offers support for parents/caregivers through our Caregiver Respite Program and helps cover costs associated with educational programs and conferences.

- Search NORD RareCare Patient Assistance Programs Database: Many of the programs are disease-specific, but new programs are added as funding becomes available.
- Applying for NORD Caregiver Respite: Provides financial assistance to enable parents/caregivers a break to attend a conference or to simply have time away from caregiving.
- Apply for NORD Rare Disease Educational Support Program: Designed to offer rare disease patients, their families, and/or parents/caregivers an opportunity to participate in educational programs and conferences that offer rare disease content.

NORD Events

Connect with others living with a rare diseases at NORD events. You can learn about NORD events by signing up for NORD's Newsletters or checking the NORD events page periodically.

NORD RareLaunch®

Provides resources to help you launch and grow a nonprofit rare disease patient support group if there is not already a group for your child's rare disease. Many social media groups started by parents of a child with a rare disease grow into a non-profit organization.

Facebook or other social medical group

Offers support if there is not a patient support group for your child's rare disease. Facebook or other social media groups started by another parent of a child with the same rare disease may be available. If you can't find one, you can start your own by going to: Create a Facebook group

Insurance Tips

Keep in close contact with your insurance company to know what appointments, tests, and treatments are paid for by insurance and what your out-of-pocket expenses may be. Important things to do:

- Verify coverage for anything ordered by your child's PCP, such as medications, treatments, tests, and referrals.
- Request a "test claim" which will simulate a claim to determine if there will be any problems with coverage and to understand out-of-pocket costs.
- Ask if your child's PCP or certain specialist needs to order referrals or specific tests to be covered by the insurance plan. For example, sometimes referrals need to always come from the PCP, and sometimes only a genetic specialist can order a genetic test.
- If the out-of-pocket cost is too high, talk to your child's PCP or specialist right away. It may be possible to:
  - › Refer your child to a different specialist who is in-network.
  - › Switch to a different:
    - Prescription
    - Medical test
    - Pharmacy
    - Testing center
  - › Help you find financial assistance resources
- If your insurance company refuses to cover an ordered medication, treatment, medical test, or referral because they have decided it is inappropriate for your child's care, talk to the office of the PCP or specialist who wrote the order. Your child's PCP or specialist's staff or office can contact your insurance company to begin a process called "prior authorization" or appeal the decision.
- If you contact your insurance company, it is a good idea to keep a record of the date you called, the name of the person you talked to, and the outcome of the call.
- Never be afraid to ask questions or ask for clarification if you do not understand the information provided to you by a representative of your child's medical insurance plan.
- Keep track of the current status of any of your child's claims. If there are problems, be sure to follow up frequently until the claim is paid.

Members of NORD's Patient and Information Services team

Selected Published Medical Articles

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