

BECOMING AN EMPOWERED PATIENT: A TOOLKIT FOR THE UNDIAGNOSED



Introduction

Have you been searching for answers from many physicians and caregivers? And have you been feeling frustrated and confused with the lack of answers? Then this toolkit is for you.

The goal of this toolkit is to move you from feeling afraid and anxious to feeling prepared, confident and educated on how to manage the next steps in your healthcare journey.

What Does it Mean to be an “Undiagnosed” Patient?

“In the beginning of my daughter’s undiagnosed journey, I felt extremely sad, alone, and frustrated. I expected answers about my daughter, and we never got any. This is a journey I never expected to be on, and I didn’t know anyone that had any experience or advice either. Where do I go? What do we do? What is my daughter’s future? I am her mother and I can’t fix it. It was extremely scary!”

—April Alvillar, parent of an undiagnosed child

Alvillar, a parent of an undiagnosed child, expresses the pressing concerns felt when caring for someone with a rare disease. While the path to a diagnosis may vary from patient to patient, everyone searching for a diagnosis shares this period of frustration. But the diagnostic process can become an odyssey if the symptoms continue to elude diagnosis, and the person remains undiagnosed long after the first concern arises.

For many people with a rare disease, obtaining a correct diagnosis takes time.

The **Shire Rare Disease Impact Report** (<http://rarediseaseimpact.com/>) found:

- The average patient spends five to seven years searching for a proper diagnosis.
- They typically consult with up to eight physicians, receiving two to three misdiagnoses.
- Physicians (both primary care and specialists) often don’t have the time, resources, or information to properly diagnose/manage patients with rare diseases, compared to more common diseases.

And that time to a diagnosis is often extended by the inadequate preparation primary care physicians receive on rare disease identification and care. The **Journal of Rare Disorders’** article *Physician and Patient Perceptions Regarding Physician Training in Rare Diseases: The Need for Stronger Educational Initiatives for Physicians* found:

- Fifty-seven percent of primary care physicians rated their training in rare diseases as neutral, ineffective, or very ineffective.
- Forty percent of primary care physicians and twenty-four percent of specialists indicated that they lack sufficient time to do a workup for a rare disease even when they suspect the patient may have one.
- Only approximately twenty-three percent of primary care physicians and twenty-five percent of specialists were likely to consult someone they considered a “disease expert” to help them make the diagnosis.

INTRODUCTION

Becoming Empowered as an Undiagnosed Patient

Being an **empowered patient** means taking an equal and active role in the care of yourself or a loved one, managing healthcare information, proactively researching and learning the most about the disease or condition, and seeking needed answers. Being equipped in this way can improve the chances of doctors listening and providing better care.

Having a rare or undiagnosed condition can be difficult. It is common to feel isolated, alone, afraid, anxious, and misunderstood. But when you seek help for the symptoms, and instead, leave the physician's office feeling dismissed and patronized, those feelings can turn to hopelessness. The best protection against developing these feelings are well-kept medical records, patience, being organized, and the confidence to seek another opinion when you feel it's required.

This toolkit outlines many of the steps of becoming an empowered patient. But there are other resources on this important subject, including:

- **Center for Advancing Health** (<http://www.cfah.org/>): This nonprofit organization works to increase people's engagement in their healthcare. They listen to patient perspectives and provide resources that encourage direct participation in one's healthcare.
- **Dr.Greene.com** (<http://www.drgreene.com/mission-statement/>): This community aims to improve children's health by informing and inspiring those who care for them. It provides information and perspective to prepare parents to become knowledgeable partners who can work with their children's physicians in a new and rich way.

- **The Empowered Patient Coalition** (www.empoweredpatientcoalition.org): The coalition strongly feels that the first crucial steps in patient empowerment and patient safety efforts are information and education. They provide needed tools, strategies and support to assist the public in becoming informed and engaged medical consumers.
- **E-Patient.net** (<http://e-patients.net/>): This blog publishes the latest news and information on healthcare. Its founder, Tom Ferguson, coined the term "e-patients" to describe individuals who are equipped, enabled, empowered and engaged in their health and healthcare decisions. He envisioned healthcare as an equal partnership between e-patients and health professionals and systems that support them.
- **The Journal of Participatory Medicine** (<http://www.jopm.org/about/>): This peer-reviewed, open access journal's mission is to advance the understanding and practice of participatory medicine among healthcare professionals and patients.
- **The Society for Participatory Medicine** (<http://participatorymedicine.org/>): This nonprofit organization is devoted to promoting the concept of participatory medicine, a movement in which networked patients become responsible drivers of their health. It provides resources, reference tools, and best practice standards that support participatory medicine.

One of the best and most important resources is support from other patients and their families. Whether you are experiencing a specific sign or symptom, feel lost or confused with the journey of finding a diagnosis, or want additional advice on best practices, other patient advocates provide a myriad of benefits.

"The best advice I ever receive is from other parents. They live this life 24/7, and they know what works and where to go. Our children are all dealing with very different syndromes/diseases, but a lot of the symptoms are similar between children," says April Alvillar, parent of an undiagnosed child. "This is where you need to ask parents questions. Who is your doctor and what has been your experience with them, how is their child treated for the symptom, how do you manage the symptom at home, what is their diagnosis, and what have they been tested for?"

Alvillar says being a parent of an undiagnosed child can be isolating. Even though her daughter was without a diagnosis, she suffers from seizures. So her family got involved in the **Epilepsy Foundation of Los Angeles**. Even if epilepsy is not her daughter's only issue, any advancements in research on epilepsy she felt would benefit her daughter. The group's website (<http://www.end-epilepsy.org/>) offers a vast array of resources and events for people suffering from epilepsy. "Getting involved makes me feel like we are doing something about it," says Alvillar. "It brings us closer as a family, and we feel we are making a difference."

See the **Appendix** to learn more about the importance of patient support.

SECTION 1: PREPARING FOR APPOINTMENTS



Proper preparation for a doctor's appointment can assure you get the most from your visit. Compiling the information needed before a doctor's appointment can ensure that the experience goes smoothly, better information is provided, and a good doctor-patient relationship is strengthened. See the **Appendix** for a checklist of what to bring to appointments.

How to Speak to Doctors

It is crucial for you to have your care materials organized and available at each appointment in order to receive better care—but so is knowing what you want to ask and say before walking into an appointment. How well you and your doctor communicate with each other is one of the most key parts to finding a diagnosis for a rare disease.

Here are some dos and don'ts when speaking to your doctor:

- **Do** think ahead about what to say, especially if this is an important appointment about a serious condition.
- **Do** know what questions you want to ask *before* the appointment. Being prepared can make sure the time with your doctor is used most effectively.
- **Do** tell the doctor about your most pressing concerns as early in the visit as possible. Don't wait until the doctor is on the way out to say, "By the way, I'm having chest pain," or "I think my son suffers from epilepsy."
- **Do** try to be specific and honest when describing symptoms to a doctor. Where are

you feeling pain, for how long? What other symptoms are you having?

- **Do** ask your doctor to help explain what's been learned elsewhere. Show respect for the doctor's expertise by asking, "*How does this information apply to me?*"
- **Do** ask questions. If you don't understand something, ask for clarification. Studies show that doctors and patients often have very different ideas of what the patient is going to do after leaving the doctor's office.
- **Do** take notes or ask to use a tape recorder. Most doctors won't mind if you explain that you want to remember the conversation or share it with loved ones.
- **Do** review the conversation and check to see if all of your questions have been answered.
- **Do** consider bringing a friend or loved one as a second listener, but set ground rules. For example, you might ask your spouse to jump into the conversation only if signaled to do so.
- **Do** also consider bringing all of your medicines, or a list of them, for the doctor to go over. More than one-third of adults struggle to understand how to take their medicines. But reviewing medicines can help you and your doctor discover mistakes, such as prescriptions for two drugs that shouldn't be taken together.
- **Don't** give up if all questions aren't answered during a single appointment. Instead, ask the doctor for another appointment, follow up by phone or e-mail, or seek help from another member of the medical team. Also reach out to your doctor when waiting for test results or especially if symptoms worsen.
- **Do** remember to say "thank you" from time to time.

For more information on how to speak to doctors, especially when they seem unwilling to listen, see the **Appendix**.

SECTION 1: PREPARING FOR APPOINTMENTS

Getting Multiple Opinions

Sometimes it is valuable to gain a second, third, or even fourth opinion when seeking a diagnosis. No one provider has all the knowledge of all the diseases and how they vary. Exploring other specialties can prove useful because not all specialists understand the body systems or diseases outside their own area of specialization. However, it can be difficult to know what specialist to see for what symptom. Reviewing this list of medical specialist definitions may be helpful in identify what type of doctors to seek: <http://intermountainhealthcare.org/providers/specialties.html>.

Getting additional opinions not only provides an opportunity for you to learn more about your condition, it also offers peace of mind that you are approaching your new diagnosis with the best chances for a favorable outcome. If you feel the treatment prescribed requires long-term or chronic care, it may be best to seek another opinion. There may be other treatment options available.

Also consider consulting medical professionals active in research. Professionals who work in academic medical centers may be good sources for identifying difficult-to-diagnose diseases because of their interests. If you are at a loss as to how to find additional doctors and specialists that can review your case, review the diagnostic programs and centers listed within the **Resource Guide** of this toolkit.

Understanding Testing Options

There are many different types of tests that can aid in the diagnostic process. Most people are aware of imaging tests (X-Ray, Ultrasound, CT scan, and MRI), or blood tests. These types of tests can provide clues for what direction a physician may need to take when providing care and treatment. But it is important for you to know more about these tests, what

information they may be able to provide, and their limitations.

A revolution in sequencing has made **genetic testing** fast and affordable. These tests can identify changes in chromosomes, genes, or proteins. The results of this test can confirm suspected genetic conditions. It may be necessary to get genetic testing done if your doctor hasn't been able to diagnose the condition based on its signs and symptoms.

If the undiagnosed condition is believed to be genetic, it is likely that these types of tests have been performed or considered:

- **Karyotype:** This test gives a picture of all of a person's chromosomes from the largest to the smallest. This type of testing can identify changes in chromosome number and large changes in DNA structure. A karyotype would, for instance, identify Down syndrome.
- **FISH:** This test identifies certain regions on chromosomes using fluorescent DNA probes. FISH analysis can find small pieces of chromosomes that are missing or have extra copies. These small changes can be missed by the overall karyotype test.
- **Microarray:** This test looks at specific DNA sequences. They can be useful in detecting much smaller changes than routine karyotypes. They can identify deletions, duplications, and unbalanced translocations.
- **Exome Sequencing:** This test selects out and sequences only about 20,000 genes in the human genome that are responsible for making proteins. These regions of the human genome contain 85 percent of all known disease-causing mutations and can readily be interpreted.
- **Genome Sequencing:** This test will sequence all 3 billion base pairs in the human genome.

To learn more about genetics and testing, explore the **Understanding Rare Inherited Genetic Diseases Toolkit**.

SECTION 2: OBTAINING YOUR OWN MEDICAL RECORDS



There are multiple steps to becoming an empowered patient. One of these is becoming knowledgeable about your own health information by requesting, reviewing, and organizing your medical records.

Having copies of your medical records will establish your standing as a strong and active member of the care team. Families are often the ones that manage the coordination of care, so maintaining the most up-to-date information is essential to becoming an empowered partner in the care of you or a family member.

“When patients and/or caregivers are faced with managing a complex, chronic medical condition, it is vital that they make a habit of obtaining copies of their medical records for their personal files in order to maintain a complete, congruent medical history,” says Heather Long, Co-founder and Director of Advocacy for U.R. Our Hope. “This is especially important because a patient’s medical records are often scattered amongst numerous healthcare providers and plans utilizing numerous independent health information management tools.”

Understanding Your Rights as a Patient

We’ve all done it—sneaked a peak at our patient folder, while waiting for the doctor to enter the room. It’s usually not until we hear footsteps coming towards the exam room that we wonder, while shutting our file closed, was I allowed to read that?

The federal Health Information Portability and Accountability Act (**HIPAA**, pronounced hip-a) gives patients the right to obtain a copy of their

medical records from any medical provider, with a few exceptions. Patients may inspect, review, and receive a copy of their medical and billing records.

“The HIPPA law created a new system of protecting privacy and making one’s health information more secure,” says Joel White, President of Horizon Government Affairs. “It was amended in 2009 with additional protections that reflect the growing use of health information technology and electronic use and exchange of personal health information.”

Who may get records? According to **HIPAA**, you may request your own records, someone else’s records if you are their legal guardian or their designated representative (if you obtained permission, in writing, to act as their representative).

A parent may not get a child’s records if:

- The child has consented to medical care and parental consent is not required under state law.
- The child gets medical care at the direction of a court.
- The parent agrees that the minor and the medical provider have a confidential relationship. And therefore, the physician believes the parent’s access to the child’s medical records would have a detrimental effect on the physician’s professional relationship with the patient or the patient’s physical safety or psychological well-being.

If a provider denies a request for medical records, it must provide a denial letter. But in some cases, denials can be appealed. If you need help with the appeal process, the **Patient Advocate Foundation** (<http://www.patientadvocate.org/resources.php?p=36>) has a step-by-step guide on how to appeal a denial and sample appeal letters.

SECTION 2: OBTAINING YOUR OWN MEDICAL RECORDS

For a more detailed explanation of your rights to medical records and some difficulties associated with obtaining them, see the **Appendix**.

Requesting Medical Records

All hospitals and most doctors’ offices have **release forms** that can be used to request your medical records. In most cases, you can request the medical information directly from the doctor’s office or medical records department at a hospital. Keep in mind that offices may only keep records for a certain amount of time as required by each state, so call the office to be sure the records are still available.

If the doctor’s office does not have a release form, you can compose a letter to request the medical records. This letter should include:

- The patient’s birth date, full name (including any name changes), address, and phone number
- The time range when the patient was seen (for example, July 2012 to September 2013—Be specific by including the approximate months and years)
- The specific types of information wanted (such as reports from a brain scan, your cholesterol levels, blood tests, etc.)

Often there is no charge for medical records if they are being sent directly to another health professional. However, if you request that the paper-based medical records be sent to you, or you want to pick them up in person, there may be a fee that reflects the reasonable cost of copying and mailing. In New York, the maximum fee a facility can charge is 75 cents per page; in California it is 25 cents per page. For diagnostic film copies, such as an X-ray, MRI, CT and PET scans, providers can charge the actual cost of copying the films.

In the case where a provider has implemented an electronic health record system, the law provides individuals with a right to obtain their health information in an electronic format.

An individual can also designate that a third party be the recipient of the information. The law limits any charges to the labor cost for an electronic request.

“One way to avoid or limit the costs associated with obtaining medical records is to request the copies at each appointment, visit, hospitalization, etc. instead of waiting and requesting copies of records that span over a lengthy period of time and involve numerous pages of records,” says Heather Long, Co-founder and Director of Advocacy for U.R. Our Hope. “Another way to reduce the cost and time associated with obtaining medical records is to use a third party company that specializes in obtaining and storing medical records for individuals for a fee.”

In most cases, **HIPAA** requires medical providers to supply copies of medical records within 30 days of a request. If it will take longer than 30 days, the medical provider must give a reason for the delay.

State laws may also regulate how quickly those records must be supplied to a patient as well. In some states, you’ll be given access to review records in a doctor’s office but may have to wait from 10-60 days to obtain your own copies. Some states require a quicker turnaround. For example, in California, providers must allow patients to see their records within five days of the request and provide copies within 15 days.

For a list of state laws governing access to medical records, visit the **Center on Medical Records Rights and Privacy at Georgetown University’s Health Policy Institute’s** website at <http://hpi.georgetown.edu/privacy/records.html>.



SECTION 3: BECOMING YOUR OWN RESEARCH ADVOCATE

What do you do when you have already gotten a second or third opinion, but there is still no diagnosis? Chances are your doctors may have categorized the condition in some manner or there is a defining sign or symptom. You may even have a condition that looks similar to a specific disease but doesn't quite fit the criteria. By conducting your own research, you may learn how to better manage symptoms.

Jill Wood, a CFO and Co-Founder of Jonah's Just Begun-Foundation to Cure Sanfilippo, began by downloading research papers from **PubMed** and asked her genetic counselor to get the papers she couldn't.

"First we hit PubMed, downloaded all the papers we could get, and asked our genetic counselor to get us the papers we couldn't. We purchased college text books and taught ourselves the basics. Once you have read about 20 papers, it starts to sink in. Highlight the portions you don't understand and make an appointment to discuss on the phone with the author," she says. "Our advisers were great about helping us understand the science. Find an adviser, a genetic counselor or a scientist. Anyone can go to a conference and track down their scientist of choice and ask to have a private talk. That's really the best advice: meet in person and build a rapport."

Interpreting the Records

Once you have obtained your medical records, the next important step is to review and understand what is in them. If you don't understand something within them, highlight it, consult with professionals, or use resources to decipher the meanings of terms.

And researching key words at webpages such as **OMIM** (<http://www.ncbi.nlm.nih.gov/omim/>), **PubMed** (<http://www.ncbi.nlm.nih.gov/pubmed/>), or even **Google** (<http://www.google.com>) is one way to start. This will provide articles written about research that has been done on particular conditions. Access to the full article may not be available, but abstracts can be accessed. Viewing the abstract will provide an indication if the article will be useful to you.

Articles can be requested from a local medical library directly from **PubMed** through a program called **Loansome Doc** (http://www.nlm.nih.gov/pubs/factsheets/loansome_doc.html).

How to use **Loansome Doc**:

1. You will need to establish an agreement with a medical library that uses **DOCLINE**. If you are not affiliated with a medical library, you will need to find one that serves the general public. U.S. users can call 1-800-338-RMLS (7657). International users need to call medical libraries in their country to see if they are on **NLM DOCLINE system**.
2. Sign up for **Loansome Doc** (<https://docline.gov/loansome/login.cfm>) and choose the medical library to use for ordering.

SECTION 3: BECOMING YOUR OWN RESEARCH ADVOCATE

3. Find the article in **PubMed**, and in the upper right hand area of the page. Often there will be a box for the full text of the article. To the left of that is a "send to" drop down. Some articles can be obtained for free by clicking on the full text version, so there will be no need to order it. If it is not free, click on the "send to" drop down and choose order. Log in and follow the ordering instructions.
4. Some medical libraries may have fees for this service, while others will not.

Additional valuable information can be found within the abstract, including the names of the researchers who participated in the study. It may be possible to contact them or have one of your doctors contact them. They may be doing a current study, or they may know colleagues that can be consulted.

You can be a partner in researching your own health by running a search of the signs and symptoms to see which conditions share a similar pattern. There are two tools today open to the public to conduct these types of searches, although both of them can be difficult to maneuver initially.

- **Orphanet** (http://www.orpha.net/consor/cgi-bin/Disease_DiagnosisAssistance.php?lng=EN): It is best to use the Thesaurus button to find the signs related to the condition. Doing so will bring up a list of categories to help locate specific signs. For example, Microcephaly will be under skull/cranial. Clicking on it will enter it into the search criteria. You may also want to pick "mandatory" for the clinical sign to avoid getting too many results. Once you have entered all of the appropriate search criteria, click on "ok."

- **Phenomizer** (<http://compbio.charite.de/phenomizer/>) has a manual that can be downloaded to help guide you through the process. To find the manual, click on "help."

Recording Ongoing Health History

A personal **care notebook** serves as an organizing tool to help patients keep track of important information about their healthcare history. A care notebook can help you maintain an ongoing record of care, services, providers, and notes, empowering you to become an expert of your own or a loved one's care. Be sure to update the records included within the notebook regularly, especially after medical procedures, vaccinations, major illnesses or diagnoses, health test results, or other relevant events. The care notebook can also be brought to medical appointments, conferences, and travel, and can be an invaluable resource for patients that change physicians.

To view sample documents or sheets that can be printed and inserted within the notebook, please refer to the **online version** of this toolkit (<http://globalgenes.org/toolkits>). And additional care notebook downloadable samples can be found in the **Resource Guide**.

Another option for tracking and maintaining a patient's medical history is the use of **personal electronic health records**, or applications that allow patients to manage and access their health information in an environment that is confidential, private, secure, and organized.

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In general, these records are controlled and maintained by the individual and can be shared with others, such as caregivers, family members, or providers. This electronic database offers a complete summary of your medical history based on information you enter. Using such a tool can provide doctors with a more complete picture so they can provide higher quality care from the outset. This will allow them to better aggregate your information and learn, for example, if you are taking a particular drug that is causing adverse effects.

Some advantages to using such electronic applications include:

- Health history tracking in an easy, controlled manner
- Quick access to vital information during an emergency
- The ability to manage and improve your health and track conditions/symptoms
- A means to organize appointments, screenings, tests, medications, etc.
- A lower chance of medical errors and duplicative tests
- Fewer communication problems between you and your doctor, caregiver, or family members

There are many benefits for patients having their medical history centralized online, says Courtney Larned, Vice President of Marketing and Communications for **CareSync**.

“Having access to medical information, no matter where you are, is not just a luxury; it’s a requirement when you are managing illness or seeking a diagnosis,” she says. “Being able to quickly share information in the case of an emergency can be the difference between life and death. For many chronic patients, baselines are not the same as they are for someone not managing chronic conditions, and being able to convey important information in an emergency is incredibly important to receive effective care.”

For people who are undiagnosed, she says, this is even more important. One reason is such information can often play a greater role in a diagnosis than tests and procedure. Being able to provide a complete medical story with providers will help the entire team work towards a diagnosis, and ultimately make the best choices for care, says Larned.

There are several applications available that can help you in maintaining your own medical information. These include **Medefile** (<http://www.medefile.com/>), **Zweena** (<http://zweena-health.com/>), **CareZone** (<https://carezone.com/home>), and **CareSync** (<http://www.caresync.com/>).



APPENDIX: THE IMPORTANCE OF PATIENT SUPPORT

Amy Clugston, SWAN USA

There are many benefits to finding support from others when you are faced with something that is unfamiliar. These situations can include being diagnosed with a specific condition, experiencing a specific sign or symptoms, or even being undiagnosed for an extended period of time.

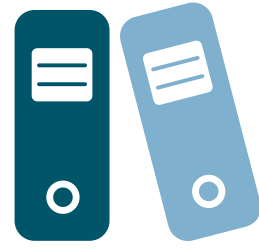
You might find support in different ways and places. For some, support from family and friends will be enough, while others will seek support elsewhere. You might find local support groups where you can meet face-to-face with others, which can be useful when gathering local resources and connections. But nevertheless, you may not encounter people who experience the same challenges you face when not having a specific diagnosis.

There are also online support groups that may provide a helpful way to connect with others who are also undiagnosed. The downside to online support is the lack of local resources and face-to-face contact. You may want to become involved with both types of support to realize the benefits each offers.

Some Online Patient Communities to Explore:

- **Facebook Groups** (<http://www.facebook.com/help/>): Facebook reached over a billion monthly active users in September 2012, making it easy for users to find and connect to others through supportive community groups.

- **Google Groups** (<https://groups.google.com/forum/?fromgroups#!overview>): Google Groups lets users participate in online discussions. Connected to Google, this social media site allows users to stay organized.
- **Inspire** (<http://www.inspire.com>): Inspire connects patients, families, friends, caregivers and health professionals, providing health and wellness support.
- **PatientsLikeMe** (<http://www.patientslikeme.com/>): Users can join this registry to connect with others in similar situations. Access this site to read through other patients’ stories, learn from their experiences, and share your own.
- **Rare Connect** (<https://www.rareconnect.org/en>): Hosted by trusted patient advocates, this is a place where rare disease patients can connect with others globally. Joining or forming a group through Rare Connect can help users provide support to others, while learning from their experiences.
- **Rare Share** (<http://www.rareshare.org/>): RareShare is a social hub that helps build communities for patients, families, and healthcare professionals affected by rare conditions.
- **Yahoo Groups** (<http://groups.yahoo.com/>): With millions of groups at your fingertips, it’s easy to find a group through this site. Establishing a group through Yahoo allows users to connect with a world of rare disease patients and parent advocates, inspiring and empowering others.



APPENDIX: COMPLICATIONS ASSOCIATED WITH OBTAINING RECORDS

Heather Long, U.R. Our Hope

Although the Health Insurance Portability and Accountability Act (**HIPAA**) Privacy Rule (<http://www.hhs.gov/ocr/privacy/hipaa/understanding/consumers/index.html>) provides patients or their authorized representatives with the right to inspect, review, and receive copies of their medical and billing records that are held by healthcare providers and health plans, there are some cases where patients or their representatives will not have the ability to obtain copies of their medical records.

Often patients are asked to sign a release when participating in a research study or clinical trial. Care should be taken to review the terms and conditions of the release carefully because the language in the release could waive any rights a patient may have to obtain copies of the medical records derived during the study or trial.

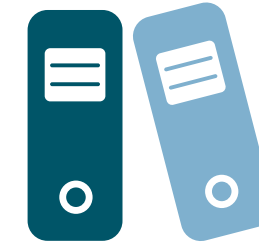
If patients have laboratory testing done, they may or may not have direct access to lab test results. There are two federal regulations that govern a patient's direct access to lab test results. These regulations are **HIPAA** and the **Clinical Laboratory Improvement Amendments of 1988** (CLIA) <http://www.cms.gov/>

Regulations-and-Guidance/Legislation/CLIA/index.html. The HIPAA Privacy Rule exempts CLIA certified laboratories from providing lab test results directly to patients unless such access is granted at the state level.

While under CLIA, a lab can only disclose patient test results to a referring lab, an individual responsible for using the test results for treatment, or an "authorized person." Individual states can define an authorized person as a healthcare provider and/or a patient. However, if a state does not define who an authorized person is, it may rely upon HIPAA's definition.

Recommendations:

1. Ask your healthcare providers for access to your medical records.
2. Learn the laws regarding patient access to medical records at both the federal and state levels.
3. Research which agencies govern your healthcare providers, so you know who to contact if denied access to your health information.
4. Make a habit of obtaining copies of your health records after every office visit, hospitalization, etc.
5. Find a way to organize and maintain your health information in a manner that best suits you and your needs.



APPENDIX: WHAT TO DO WHEN THEY SAY, "IT'S ALL INSIDE YOUR HEAD"

Iiana Jacqueline, Let's Feel Better

"You look perfectly healthy to me. So you must be faking it."

Fists up, rare readers. Because if you haven't heard a phrase like this before—chances are, you will. Whether your symptoms are bizarre and frequent, or obnoxious and chronic, at some point you're going to get some flak for seeking necessary medical care.

For those with undiagnosed illnesses, preparing to go to a new doctor can feel like preparing for battle. Having been knocked down by prejudice in the past, they're walking into each waiting room wearing the layers of judgments from past appointments. And many lose confidence in their ability to recognize their body's own signs and symptoms.

There is no roadmap and little support for undiagnosed patients. To not have a name for your disease and to not know what kind of doctor should be managing it can be frustrating and isolating.

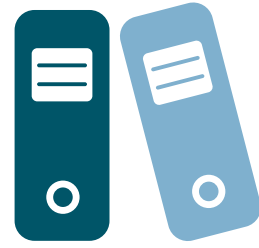
Doctors also have their own set of challenges. The demands of today's healthcare sector means they see more patients than they have time for—and as a result, they see those patients repeatedly with the same issues unresolved. They are inherently overworked, frustrated, and blamed for everyone's problems. This explains why your doctor may seem like a "total jerk." But just because doctors may feel overwhelmed, doesn't mean they aren't necessarily good at their job. If, on the other hand, they ignore vital information, refuse to

check for what you feel are necessary tests, fail to explain their methods or provide information on prescribed medications—then jerk or no jerk—they are bad doctors.

Recommendations:

1. **Be patient, but firm**
Patients should give a full history of their condition, use their medical records, and ask their doctor about their diagnosis. If patients feel there's something about their diagnosis the doctor is missing, they should be firm in asking for a more extensive exam.
2. **Don't be afraid to date around**
I once asked my cardiologist's office if they could transfer my files for a second opinion. It didn't go over well. So I had to remind them that we weren't exclusive and I had every right to get a second, third or eighth opinion if that's what made me feel most comfortable.
3. **Ask yourself "Am I dating Dr. Wrong?"**
Like many bad first dates, you may be able to predict right away just how quickly your new relationship with a doctor is going to go downhill. Other times you may feel trapped into continuing with a doctor who isn't serving your needs. So ask yourself: Do I feel like I'm getting answers? Am I being treated with respect?

The goal is always to feel better and you shouldn't feel ashamed or embarrassed just because you strive to have enough strength to go about your normal day's activities. You deserve to feel well enough to enjoy your life and don't let a close-minded physician convince you of anything different.



APPENDIX: DOCTOR'S APPOINTMENT CHECKLIST

- ❑ **List of questions:** Come prepared with a list of questions to ask your doctor. Brainstorm the list well before the appointment and have a concise list of questions, starting with the ones that are most urgent and must be answered. Don't leave the doctor's office without asking these questions!
- ❑ **List of symptoms:** Keep a journal that documents all symptoms and how they fluctuate throughout the days and weeks. Bring this journal to medical appointments to help the doctor better understand what's going on and how the symptoms affect your daily life.
- ❑ **Blank paper and a pen for notes:** This may seem obvious, but your doctor may not always have writing equipment readily accessible, and it's important to have a paper and a pen to take notes. Write down things that don't make sense to ask for clarification later. If there are words mentioned that you've never heard of, ask your doctor to spell them.
- ❑ **Medical history:** Although it would be ideal for every doctor to have a full list of each patient's medical history, there is no nationally accessible medical record system in place. Be sure to provide the doctor with key information, including medical problems, past surgeries, doctors' names, health insurance, and medical allergies.
- ❑ **Any changes to your medical record:** Bring any test results since your last visit.
- ❑ **Your List of medications** (or a list with doses and frequency): Be as specific as possible about drug and dose. The easiest way to do this is by bringing the actual medication bottles with the labels on them. It's important that patients be honest about their medications and let their doctors know if they've stopped using a drug or have not used it as prescribed. Otherwise, your doctor may wrongly conclude a drug is not working and prescribe more.
- ❑ **Forms:** Make sure to complete and bring any requested forms.
- ❑ **Previous doctor information:** If referred by another physician, bring the name, address, and telephone number of the doctor.
- ❑ **A family member or friend:** Patients need support. And having someone will not only provide company and support, but also may remind you of important questions and concerns.
- ❑ **Additional doctor appointment information:** If you have been to another doctor for the same complaint, bring all of the information from that doctor's visit such as test results, x-rays, medication, or treatments prescribed.



RESOURCE GUIDE

Creating a Care Notebook

Maryland's Department of Health and Mental Hygiene (http://phpa.dhmh.maryland.gov/genetics/SitePages/create_care_notebook.aspx): These steps easily lay out how to create a care notebook.

Medical Home Portal (<http://www.medicalhomeportal.org/living-with-child/after-a-diagnosis-or-problem-is-identified/caring-for-children-with-special-health-care-needs/managing-and-coordinating-care/care-notebook>): This resource provides a completed, downloadable notebook that patients can compile into a binder or download its individual forms separately.

National Center for Medical Home Implementation (http://www.medicalhomeinfo.org/for_families/care_notebook/): This site describes what a care notebook is, its importance, and how to build it out, while also providing free examples.

Parent to Parent of NYS Care Notebook Sample (http://parenttoparentnys.org/images/uploads/pdfs/health_care_notebook_10_13_with_eval_included.pdf): This PDF booklet includes quick tips you should review before getting started on developing a care notebook and important sample forms and links.

Getting Multiple Opinions, Diagnostic Programs

Blank Children's Hospital Diagnostic Pediatrics (<http://www.blankchildrens.org/diagnostic-clinic.aspx>): Diagnostic pediatricians assess challenging or unusual pediatric patients. As background knowledge and medical history is essential to diagnosis, the diagnostic team works closely with the primary care physician to determine a clinical diagnosis and formulate a treatment plan.

Duke University's Task Force for Neonatal Genomics (<https://www.dukegenes.org/>): The Duke Task Force for Neonatal Genomics was formed to bring patients, researchers, and clinicians together to use genomic technologies in order to assist the diagnosis of newborns and young children with challenging clinical cases. By sequencing children early in life, clinicians can make use of personalized genomic information that, in combination with all of the other tools doctors have at their disposal, can improve patient management and outcomes.

Genomic Medicine Clinic at Children's Hospital of Wisconsin (<http://www.chw.org/medical-care/genetics-and-genomics-program/programs-and-services/genomic-medicine-program/>): This program provides services to individuals with unidentified complex health problems thought to have genetic causes. Some of their services include medical evaluations, pre-test education, genome sequencing, result education, and treatment planning.

RESOURCE GUIDE



Mayo Clinic's Division of Consultative and Diagnostic Medicine (<http://www.mayoclinic.org/gim-jax/>): The Division of Consultative and Diagnostic Medicine provides comprehensive evaluations to adults with complex medical problems. General internists combine the characteristics of a diagnostician and a consultant in disease prevention and health promotion.

Mayo Clinic's Individualized Medicine Clinic (<http://mayoresearch.mayo.edu/mayo/research/center-for-individualized-medicine/individualized-medicine-clinic.asp>): The Individualized Medicine Clinic (IM Clinic) offers genomic testing to patients in order to further personalize care. The IM Clinic treats patients with advanced cancers and those with rare or undiagnosed diseases believed to have genetic causes.

Mayo Clinic's Pediatric Diagnostic and Referral Clinic (<http://www.mayoclinic.org/pediatrics-rst/pdrc.html>): Mayo Clinic Children's Center includes providers from more than 40 medical and surgical specialties, all focused on children's health care needs. Each year, its teams provide expert care to children and teens, from common problems to the most complex situations.

NIH's Undiagnosed Diseases Program (<http://rarediseases.info.nih.gov/research/pages/27/undiagnosed-diseases-program>): The two key goals of the NIH's Undiagnosed Diseases Program are to provide answers to patients with mysterious conditions that have long eluded diagnosis and to advance medical knowledge about rare and common diseases.

Patrick S. Pasquariello Jr. Diagnostic and Complex Care Center at The Children's Hospital of Philadelphia (<http://www.chop.edu/service/diagnostic-and-complex-care-center/>): This center is designed to meet the needs of patients with complex or difficult-to-diagnose illnesses. Their physicians and staff work closely with referring physicians in managing your child's overall medical care.

Paul C. Gaffney Diagnostic Service (<http://www.chp.edu/CHP/gaffney+diagnostic+service>): This service helps families and supports primary care physicians by identifying a child's specialized medical needs while coordinating the best possible advanced treatment. They have years of experience in diagnosing and managing a wide variety of the most complex pediatric medical conditions.

Pediatric Diagnostic Referral Service at North Carolina Children's Hospital (<http://www.nc-childrenshospital.org/ourservices/raleighclinic/diagnosticdilemmas>): Specialists evaluate complex diagnostic dilemmas, including cases of complex symptoms of unknown cause and patients that do not fit into a particular specialty category.

Riley Hospital for Children's Diagnostic Clinic (<http://iuhealth.org/riley/community-pediatrics/diagnostic-clinic/>): The Diagnostic Clinic at Riley Hospital for Children at Indiana University Health assists physicians who wish to refer infants, children, and teens for second opinions or consultations. They help physicians in managing complex problems or difficult-to-diagnose illnesses.

RESOURCE GUIDE



TGen's Center for Rare Childhood Disorders (<https://www.tgen.org/research/center-for-rare-childhood-disorders.aspx>): This center applies the latest tools of genomic medicine to provide answers for parents wanting to identify the disease or disorder affecting their child. Its mission is to better diagnose, treat, and ultimately cure rare childhood genetic disorders.

Undiagnosed Disease Center at Rady Children's Hospital (www.rchsd.org/programsservices/a-z/u-z/undiagnoseddiseasecenter/index.htm): At the Undiagnosed Disease Center, a multidisciplinary team of specialists from Rady Children's and the University of California, San Diego School of Medicine seeks to provide a diagnosis and improve treatment for children. To do this, they collaborate with leading basic science researchers and use cutting-edge technologies, such as genome sequencing.

University of Alabama, Birmingham's Undiagnosed Diseases Program (<http://www.uab.edu/medicine/genetics/undiagnosed-diseases>): This program seeks to meet the needs of patients with severe chronic medical conditions in which a diagnosis has not been made despite extensive efforts by the referring physician. Patients who could benefit from this program include those with rare diseases, those with conditions not previously known to exist, and those who have an atypical presentation of a common disease or disorder.

Joining Patient Support Groups

Facebook Groups (<http://www.facebook.com/help/>): Facebook reached over a billion monthly active users in September 2012, making it easy for users to find and connect to others through supportive community groups.

Google Groups (<https://groups.google.com/forum/?fromgroups#!overview>): Google Groups lets users participate in online discussions. Connected to Google, this social media site allows users to stay organized.

Inspire (<http://www.inspire.com>): Inspire connects patients, families, friends, caregivers and health professionals, providing health and wellness support.

PatientsLikeMe (<http://www.patientslikeme.com/>): Users can join this registry to connect with others in similar situations. Access this site to read through other patients' stories, learn from their experiences, and share your own.

Rare Connect (<https://www.rareconnect.org/en>): Hosted by trusted patient advocates, this is a place where rare disease patients can connect with others globally. Joining or forming a group through Rare Connect can help users provide support to others, while learning from their experiences.

Rare Share (<http://www.rareshare.org/>): RareShare is a social hub that helps build communities for patients, families, and healthcare professionals affected by rare conditions.

Yahoo Groups (<http://groups.yahoo.com/>): With millions of groups at your fingertips, it's easy to find a group through this site. Establishing a group through Yahoo allows users to connect with a world of rare disease patients and parent advocates, inspiring and empowering others.

RESOURCE GUIDE



Learning to Be Empowered

Center for Advancing Health (<http://www.cfah.org/>): This nonprofit organization works to increase people's engagement in their healthcare. They listen to patient perspectives and provide resources that encourage direct participation in one's healthcare.

Dr.Greene.com (<http://www.drgreene.com/mission-statement/>): This community aims to improve children's health by informing and inspiring those who care for them. It provides information and perspective to prepare parents to become knowledgeable partners who can work with their children's physicians in a new and rich way.

The Empowered Patient Coalition (www.empoweredpatientcoalition.org): The coalition strongly feels that the first crucial steps in patient empowerment and patient safety efforts are information and education. They provide needed tools, strategies and support to assist the public in becoming informed and engaged medical consumers.

E-Patient.net (<http://e-patients.net>): This blog publishes the latest news and information on healthcare. Its founder, Tom Ferguson, coined the term "e-patients" to describe individuals who are equipped, enabled, empowered and engaged in their health and healthcare decisions. He envisioned healthcare as an equal partnership between e-patients and health professionals and systems that support them.

The Journal of Participatory Medicine (<http://www.jopm.org/about/>): This peer-reviewed, open access journal's mission is to advance the understanding and practice of participatory medicine among healthcare professionals and patients.

The Society for Participatory Medicine (<http://participatorymedicine.org/>): This nonprofit organization is devoted to promoting the concept of participatory medicine, a movement in which networked patients become responsible drivers of their health. It provides resources, reference tools, and best practice standards that support participatory medicine.

Obtaining Medical Records

Center on Medical Records Rights & Privacy at Georgetown University's Health Policy Institute (<http://hpi.georgetown.edu/privacy/records.html>): When requesting you or a loved one's medical records, you are subject to federal and state laws. You can view your rights according your state through this site.

Memorial Hospital Medical Release Form Sample (<http://www.memorialhospitalnh.org/docs/AuthorizationToReleaseInformation-2.pdf>): This is a sample of a medical release form. When requesting information, it is often through filling out and submitting a form similar to this one.

Patient Advocate Foundation (<http://www.patientadvocate.org/resources.php?p=36>): A complete guide on navigating through the appeals process for obtaining medical records can be found through this foundation's website.

RESOURCE GUIDE



Preparing for Medical Appointments

Agency for Healthcare Research and Quality Question Builder (<http://www.ahrq.gov/apps/qb/>): This tool can be used before visiting your doctor (or other medical professional) to know what questions to ask and make sure you are prepared and ready.

Personal Electronic Medical Records

CareSync (<http://www.caresync.com/>): Recognizing that managing illness can be a full-time job, CareSync's tools and services help patients coordinate care before, during, and after the doctor's appointment.

CareZone (<https://carezone.com/>): CareZone simplifies the lives of those caring for children, partners, and aging parents, by providing a simple and private environment where family and helpers can stay organized and coordinated.

Medefile (<http://www.medefile.com/>): Medefile helps you get the best possible treatment, making your vital information available whenever it is needed.

Zweena (<http://zweenahealth.com/>): Zweena is changing the way its clients know and utilize their own healthcare information. It offers numerous applications, such as personalized health dashboards, that transform personal health information.

Researching Medical Terms and Information

Intermountain Healthcare Medical Specialist Definitions (<http://intermountainhealthcare.org/providers/specialties.html>): If you feel unsure about which medical professional or specialty you should consult, review this list of definitions. It provides definitions of specialties and subspecialties, guiding its readers through multiple medical professionals available.

Loansome Doc (http://www.nlm.nih.gov/pubs/factsheets/loansome_doc.html): Loansome Doc connects individuals to medical libraries. A user can order articles from a list of citations retrieved from PubMed by sending requests to a library for the full-text documents.

Medline Medical Dictionary (<http://www.nlm.nih.gov/medlineplus/mplusdictionary.html>): This search engine can help you research the medical terms often used by professionals, within diagnoses or described within symptoms.

OMIM (<http://www.ncbi.nlm.nih.gov/omim/>): OMIM is a continuously updated catalog of human genes and genetic disorders and traits, with particular focus on the molecular relationship between genetic variation and phenotypic expression. This resource can be used to search and learn more about medical conditions and symptoms.

PubMed (<http://www.ncbi.nlm.nih.gov/pubmed>): PubMed is an information hub for articles. Search PubMed to read and learn more about certain conditions, issues, and complications.



LET OTHERS BENEFIT FROM YOUR KNOWLEDGE BY SHARING YOUR TIPS AND TRICKS!

If you would like to contribute your experience or have a comment/suggestion, please enter it online at <http://globalgenes.org/toolkits>.

Contributors:

Amy Clugston

Founder and President, SWAN USA
<http://www.undiagnosed-usa.org>

Amy Gleason

Chief Operating Officer, CareSync
<http://www.caresync.com>

April Alvillar

Parent of an Undiagnosed Child

Courtney Larned

Vice President of Marketing and Communications, CareSync
<http://www.caresync.com>

Heather Long

Co-Founder and Director of Advocacy, U.R. Our Hope
<http://www.urourhope.org>

Ilana Jacqueline

Patient Advocate and Blogger, Let's Feel Better
<http://www.letsfeelbetter.com>

Jill Wood

CFO and Co-founder, Jonah's Just Begun-Foundation to Cure Sanfilippo Inc.
<http://jonahsjustbegun.org>

Joel White

President, Horizon Government Affairs
<http://horizondc.com>



Global Genes
SWAN (Syndromes Without A Name)

<http://globalgenes.org/toolkits>