PROVIDING BETTER CARE FOR RARE DISEASE PATIENTS: PRIMARY CARE PHYSICIAN GUIDE

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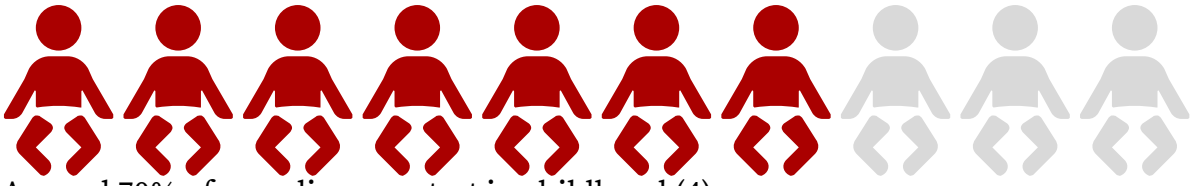
WHAT IS A RARE DISEASE?

The definition of what “rare disease” means varies by country and/or region.

- US: Affects fewer than 200,000 Americans (1)
- Europe: Affects fewer than 1 in 2,000 people (1)
- World Health Organization (WHO): Affects fewer than 65 in 100,000 people (2)

It’s estimated that rare diseases impact 25-30 million people in the United States (3), and over 300 million people worldwide (2)

There have been over 7,000 rare diseases identified to date (2)



Around 70% of rare diseases start in childhood (4)

Genetics are thought to be a factor in many rare diseases (4)

Many rare diseases currently lack effective and/or available treatments

BARRIERS TO CARE

Access to care

Access to care is a significant barrier for rare disease patients. One of the reasons access is an issue is that there is a shortage of medical geneticists, even in high-income countries (5, 6). This insufficient workforce issue is exacerbated by the increased demand for genetic services due to increased awareness and expansion of testing services (7). Both of these things can contribute to long wait times and delays for appointments (8).

Geographical location is another barrier to care (8). Patients may need to travel long distances to see medical geneticists and other specialists. The travel itself may be costly and might require patients or caregivers to take time off work, resulting in lost income (8, 9). Frequent appointments, particularly ones that are far geographically, can also disrupt children's schooling (8, 9). Some people do not have the ability or means to travel, which may lead to appointments being further delayed or not attended at all (7, 8).

Insurance and cost issues

While cost often falls under access to care, we wanted to highlight a few specific issues related to health care costs and insurance separately.

Patients and families with rare disease may face insurance-related barriers, such as being denied coverage for diagnostic testing, specialist visits, or treatments (8, 10). Sometimes insurance will dictate where patients can get specialist care from, and this can further exacerbate travel time and cost concerns. Undiagnosed patients may have an even more difficult time, as a diagnosis can help support medical necessity of a visit, test, or treatment (11). However, even a diagnosis does not prevent all insurance denials.

In the United States, health insurers cannot refuse coverage or charge higher premiums for people because they have a pre-existing condition (12). However, short-term, limited-duration health plans are not required to meet the same quality standards and are sometimes able to discriminate against patients with pre-existing conditions (13). These health plans can also use questionable marketing tactics to lure patients in.

Having health insurance does not guarantee that health care is affordable. A 2025 KFF poll found that 42% of insured US adults found it very or somewhat difficult to afford healthcare costs (14). The poll also found that 36% of respondents said they skipped or postponed care in the last 12 months, and 21% said they have not filled a prescription because of the cost (14).

BARRIERS TO CARE

Poor communication and care coordination

Poor communication and care coordination between clinicians on the care team can be a barrier to optimal care for rare disease patients. Communication issues are among the most frequent causes of harmful medical errors (15). It's estimated that about 67% of communication errors relate to handoffs, which is when patient care responsibility transitions from one clinician to another (15).

Poor communication and a lack of care coordination by clinicians can add unnecessary burden and stress for patients and caregivers (17). One study found that patients (or their caregivers) often took on responsibility for coordinating specialty care but struggled to do so (18). When patients and caregivers take on this extra burden, it can cause fatigue, loss of earnings at work, extra travel costs, and extra time off work and school (17).

Information sharing through electronic medical records (EMRs) can contribute to communication challenges. Data in patient charts may be patchwork (emails, faxes, printed docs, EMR notes, etc.) or not available at all, which can make patient care less effective if clinicians do not have all the relevant health information (6).

Lack of education on rare diseases for primary care physicians (PCPs)

Many surveys have found that physicians worldwide feel they lack sufficient knowledge about rare diseases and genetic testing (5, 19-21). This lack of knowledge can limit the implementation of recommendations and guidelines (16) and can cause issues with diagnostic delay or misdiagnosis (21). Education on rare diseases and genetic testing can vary by institution and country.

Given the large number of rare diseases, physicians may find it more useful to learn about the specific rare diseases that their patients have. But this isn't easy. PCPs often don't have enough time to address all these challenges (5). A 2025 study found that primary care physicians worked an average of 61.8 hours per week (22). Adding in extra work for PCPs without proper tools, resources, or time allotment makes it difficult to put sufficient time into learning about each patient's rare disease and the latest treatment approaches.

BARRIERS TO CARE

Long time to diagnosis

Accurate diagnosis is important to guide treatment, symptom management, and referral to specialists (8). However, many rare disease patients face long delays to diagnosis.

The US Department of Health and Human Services has estimated an average time to diagnosis of five years (8) for rare disease patients. This aligns with findings from a Global Genes study, which reported an average of 4.8 years from symptom onset to correct diagnosis, with some patients experiencing delays up to 20 years (9).

A recent retrospective patient study in Europe found that half of people living with a rare disease waited at least 9 months for a diagnosis after symptom onset, while 25% waited more than 5 years (21). Additionally, a survey conducted in China found a diagnostic delay of an average of 1.4 years for rare disease patients (21).

The diagnostic delay can be taxing, as the typical rare disease patient sees more than seven physicians before getting a diagnosis (9). During this time, patients can experience worsening symptoms or decreased quality of life as they await diagnosis and a more appropriate treatment plan (9).

Lack of guidelines

Guidelines on genetic testing continue to evolve as more information becomes available, but these frequent updates can make it hard for PCPs to stay current on which genetic tests to use and when to use them (7). Even with established guidelines, there are concerns about clarity and feasibility of implementation that may hinder PCPs from scheduling and carrying out recommended tests (16).

WAYS PHYSICIANS CAN IMPROVE CARE FOR RARE DISEASE PATIENTS

Rare disease patients may see their PCPs more often than other patients. One study found that children with a rare disease (through 14 years of follow-up) had an average of 45.4 primary care visits, while children without had an average of 28.2 visits (23). Primary care clinicians can be a great resource for rare disease patients as they manage their condition. Below are a few ways that primary care physicians can help their rare disease patients and families.

(1) Continue to educate yourself on rare diseases

- Continue to learn about rare diseases, especially ones that your patients have (21, 24). This can include more formal educational sessions or independent research with trusted sources (see pg. 11 of this guide for some resources).
- Know where to find up-to-date information on genetic disorders and make these resources known and easily accessible to other clinicians at your clinic (5).
- Have up-to-date resources (educational, support groups, advocacy) to share with rare disease patients and caregivers (6).
- Be aware of common red flags that may indicate a patient has a rare disease (6).

(2) Improve communication and care coordination with other clinicians on a patient's care team

- Work with specialists to clearly define who is taking on which responsibilities for the patients' care (18).
- Know when to communicate through the EMR and when direct communication (such as a phone call) is more effective, such as for complex or timely matters (18).
- Clarify who is responsible for care coordination (primary care physician, specialist, patient, care coordinator, etc.) (18). It may be beneficial to have a care coordination agreement to clearly state roles and responsibilities of each party (18), but only if this is useful and not an additional administrative burden.
- Create a plan to share information with the entire team, such as a short meeting (virtual or in-person), a treatment plan document that everyone signs off on, a portal accessible by both clinicians and patients, or other mode of communication (17).
- Share patient notes with specialists, and take time to review their notes. Patients may be more comfortable asking their PCP questions, so having these notes available (and taking time to review them) can help PCPs better answer patient questions and address patient concerns.

WAYS PHYSICIANS CAN IMPROVE CARE FOR RARE DISEASE PATIENTS

(3) Monitor symptoms related to the patient's rare disease

- In some cases, the PCP may be in charge of surveillance and management of a patient's rare disease, with specialists providing support as needed (7).
- Continue to monitor symptoms and provide health maintenance recommendations, even for patients that see geneticists and other specialists (7, 25).
- Take note of any signs or symptoms that may warrant re-evaluation by a specialist or that may benefit from the expertise of a new specialist that a patient hasn't yet seen (25).
- Engage in problem solving with patients to help ameliorate or treat their rare disease symptoms (24).

(4) Provide referrals to patients for appropriate specialist care

- PCPs can greatly help patients by providing referrals to specialists, as good referral pathways improve patient care (16). PCPs may act as a quarterback or advocate to get their patients to the appropriate specialists and care centers (24).
- Have contact information for local medical genetics and specialty clinics readily available so it can be easily accessed by you and your primary care team (5). As a note, it may be helpful to have contact information for multiple clinics in case there are insurance issues that dictate where a patient can go for covered care.

(5) Small acts of compassion and empathy can make a difference

- Patient and caregiver interviews suggest that supportive PCPs make a positive impact. Supportive behaviors can include asking patients (or caregivers) about the care they received from specialists, showing interest in the patient, learning about their condition, and generally being compassionate (24).

WAYS PHYSICIANS CAN IMPROVE CARE FOR RARE DISEASE PATIENTS

(6) Offer flexibility with telehealth, if available

- If your clinic/practice supports any form of telehealth, offering a blend of in-person and telehealth appointments may help support rare disease patients and caregivers (5, 6).
- Telehealth can help reduce barriers to care, such as
  - Eliminating distance traveled (particularly for those in more remote or geographically isolated areas)
  - Reducing time off of work or school for patients and caregivers
  - Saving money on travel costs
- Telehealth has been found to be a positive tool for chronic disease management (26), including for rare disease patients (27).
- Refer to this Telemedicine Resources for Clinicians link to learn more about telehealth.

(7) Improve knowledge on genetic testing

- Some PCPs may order genetic tests for patients. Individual physicians must assess whether they have adequate training in determining the appropriate test to order and to interpret and counsel the patient or family about the results (7).
- Broader understanding of genetic testing and access to clear testing criteria may help PCPs feel more comfortable ordering genetic tests for patients, which could help speed up the diagnosis process (5, 6).

(8) Provide proper Newborn Screening follow-up, if applicable

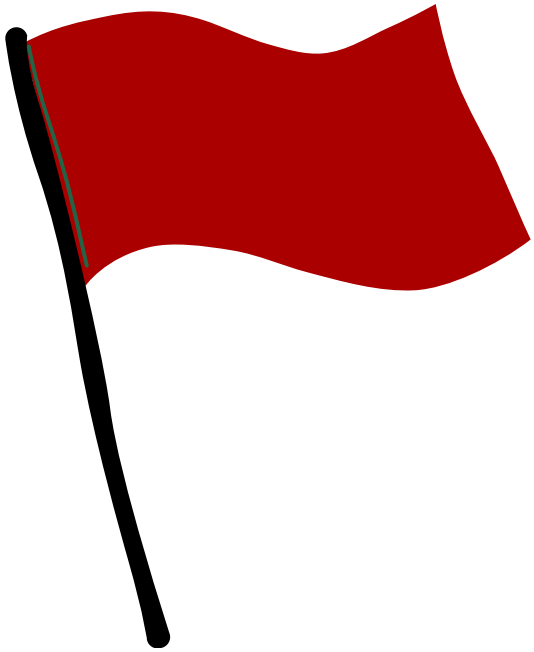
- If newborn screening was not done at birth for whatever reason (ie, home birth), pediatricians or other PCP should get a sample for screening (7). See "Systemic Solutions" section point 3, "Set Clear Guidelines and Standards" for more information on newborn screening in the United States.
- If needed, order repeat screenings or educate the family about the need for additional testing to confirm or refute a diagnosis (7).
- Providing some basic resources to families about the disorder can help relieve some parent or guardian anxiety about the testing (7).

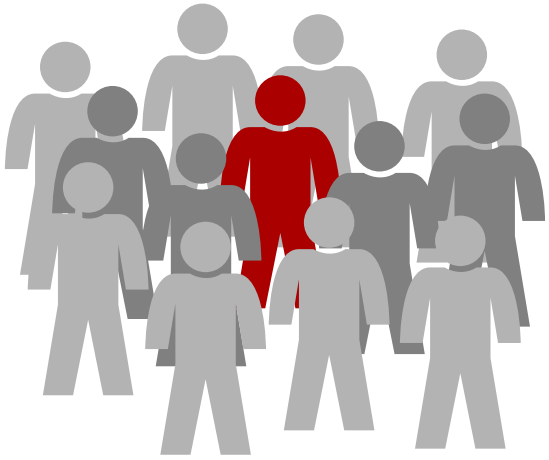
RED FLAGS

Red flags are clinical features that may suggest a patient has a rare disease. Recognizing these red flags can help primary care physicians pursue appropriate testing and potentially reduce time to diagnosis.

Red flags to watch for may include:

- Involvement of three or more systems (21)
- Multiple specialist referrals (21)
- Family history (6, 7, 21)
- Genetic inheritance pattern (21)
- Delayed diagnosis or misdiagnosis (21)
- Prolonged hospital stay (21)
- Failure to standard therapies (21)
- Disease presentation at any age (21)
- Functional impairment (21)
- Clusters of birth defects (21)
- Neurodevelopmental delays or decline (6, 21, 25)
- Severe, extreme, or exceptional pathology (6, 21)
- Congenital anomalies (6, 7)
- Neurological symptoms (7)
- Growth anomalies (7)
- Extreme presentation of common conditions (6, 7, 21, 25)
- Abnormal lab results (7)
- Dysmorphic features (25)
- Physical anomalies (25)





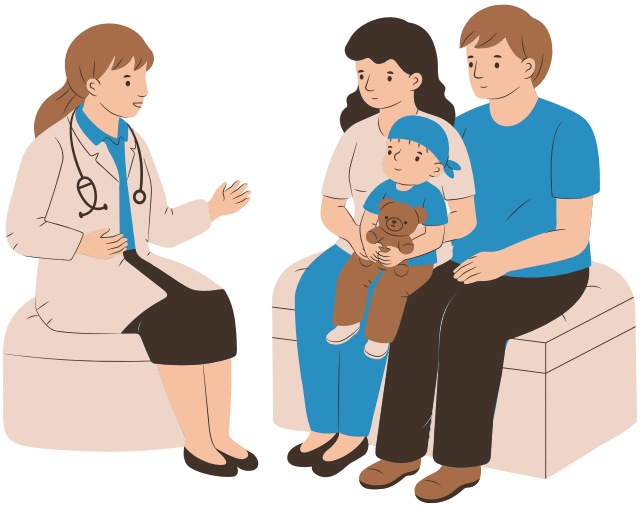
PHYSICIAN RESOURCES

Name and Link	Details
National Organization for Rare Disorders (NORD) CME Videos	These digital courses were created to equip clinicians (and patients) with the information they need to identify the signs and symptoms of rare diseases and make sure they are familiar with available treatment options. *CME Credit Available*
American College of Medical Genetics and Genomics (ACMG) Genetics Academy	ACMG has created educational videos on a variety of genetics topics. *CME Credit Available for Certain Courses*
American Medical Association (AMA) Ed Hub: Genetics and Genomics	Explore the latest in genetics and genomics, including cancer genetics, neurogenetics, and pharmacogenetics through courses and videos. *CME Credit Available for Certain Videos*
GeneReviews	GeneReviews is a resource for clinicians that provides clinically relevant and medically actionable information for inherited conditions that covers diagnosis, management, and genetic counseling for patients and families.
Orphanet	Well-curated resource on rare diseases and orphan drugs.
ACMG ACT Sheets and Algorithms	The ACMG ACT Sheets and accompanying algorithms are resources for clinicians looking for information on genetic conditions to help inform clinical decision making. ACT Sheets and algorithms are excellent refreshers on the conditions, diagnoses, and next steps for patients.
NIH Genetic Testing Registry	The Genetic Testing Registry (GTR®) provides information for clinicians and researchers navigating the landscape of genetic tests.
Heartland Collaborative Resource Center	The Heartland Regional Genetics Network (HRGN) website provides information and resources for professionals, families, and collaborators to facilitate the understanding of genetics and communication.

TALKING TO PATIENTS ABOUT CLINICAL TRIALS

Many rare diseases do not have approved treatments, making clinical trials the best (or only) option for patients. Sometimes patients or caregivers may come to a PCP to ask for guidance about a clinical trial. The following tips may help with navigating these discussions:

- Oftentimes, if patients are interested in a clinical trial and talking to their PCP about it, they have been to specialists, got second opinions, tried other options, etc. Try to understand what these previous conversations covered so you can have better conversations with your patient.
- If a patient sees a medical geneticist or specialist, PCPs may also suggest the patient gets the specialist's opinion about the trial. Specialists may also have connections to those running the trial.
- Focus on getting patients referrals, connections, contact information, etc. to get into the trial if they meet the criteria.
- Be aware of your own knowledge limits, and understand what you do not know. To better answer patient/caregiver questions and concerns, reach out directly to clinical trial coordinators. You can find contact information on clinicaltrials.gov.
- Discuss treatment options outside of the clinical trial and explain how the study differs from current standard treatment (28).



- Encourage patient and relevant caregivers to be involved in the discussion (28).
- Talk clearly, slowly, and avoid jargon. You may need to explain concepts more than once or in different words (29).
- Encourage patients to ask questions. Ask questions in an open-ended manner. For example, ask "what questions do you have?", not, "do you have any questions?" (28).
- Allow ample time to answer all questions that a patient has.
- Check patient and caregiver understanding (29).
- Listen to patients' point of view. Be empathetic and respectful (29).
- Be sure to reiterate that the trial is VOLUNTARY, and it is their choice, and they can leave/withdraw from the trial at any time for any reason (29).
- Don't push the clinical trial on patients, but be honest in answering questions (28).

Where to get info on clinical trials:

- SCGE Gene Therapy Clinical Trial Browser
- clinicaltrials.gov/

SYSTEMIC SOLUTIONS

Systemic changes would be more impactful to improving rare disease care on a large scale. The following pages outline several approaches that, if implemented broadly, could improve care for rare disease patients and their families.

1

Improve Physician Education

- Provide training and informational resources in genomics and rare disease for physicians in practice (5-7).
- Improve genomics education for physicians in training (19).
- Educate clinicians on interpreting guidelines and making informed decisions about genetic testing. This is important, as guidelines and available tests continue to progress as new research and technologies are discovered (25).
- Within clinics or health systems, consider having short sessions (such as a "Lunch and Learn") that specialists host to educate PCPs on the latest news in the field and changes to services and treatments offered by those specialists.

2

Provide Clinicians with Appropriate Tools

- Access to whole genome or exome sequencing could reduce the diagnostic odyssey and potentially improve cost-efficiency compared to single-gene tests (9).
- Providing clinical support and decision-making tools can help PCPs provide better care for rare disease patients (5-7).
- AI and machine learning could be used to analyze health records and develop predictive models to expedite diagnosis of rare diseases and improve care (6).
    - Ask your administrators if you have access to any tools

SYSTEMIC SOLUTIONS

3

Set Clear Guidelines and Standards

- Creating clear, evidence-based guidance on genetic testing can help PCPs with frontline testing, and facilitate timely referral of patients and families to appropriate specialists as needed (5, 6, 25).
- Standardize the approach to getting family history by adopting a universal minimum standard of questions (25). The AMA has a web page called "Collecting a family history" with many good starting points to create these standard questions.
- In the United States, the Recommended Uniform Screening Panel (RUSP) recommends which conditions to test for Newborn Screening, while individual states determine which conditions to include in their screening. As of February 17, 2026, there were 38 core conditions and 26 secondary conditions recommended by RUSP (30). 8 states screen for 64 or more conditions, while 7 states screen for fewer than 38 (31). Standardizing these tests across states may help with better diagnosis (9).

4

Incorporate Genetic Counselors into Primary Care

- Incorporating genetic counselors into primary care clinics could improve patient care (20, 31).
- Genetic counselors could provide primary care support by (31):
  - Selecting genetic tests
  - Interpreting test results
  - Providing pre- and post-test counseling
- Adding genetic counselors to care teams can improve clinical outcomes, increase patient knowledge and engagement, and increase adherence to health plans and healthy behaviors (20, 31).

SYSTEMIC SOLUTIONS

5

Improve Data Sharing and Communication

- Improve capturing and sharing of patient data, both in the EMR and during in-person communication (6).
- Build more time for communication into the PCP daily schedule.
- Improve ways for patients to view their notes and communicate with PCPs.

6

Telehealth and Appointment flexibility

- Offer appropriate telehealth options (6).
- Offer appointment flexibility (17).
  - Can certain routine or frequent tests be done at PCP office or local clinic instead of a specialist office?
  - Can appointments in a similar location be grouped to the same day/times?
- Improve reimbursement for telehealth services.

7

Improve Care Coordination

- Consider adding care coordinators to help patients schedule all necessary appointments in a way that reduces time traveling, time away from work/school, and associated costs.
- Care coordination programs have reported improvements in satisfaction with care, increased interactions with healthcare teams, and reductions in health service use (33).

8

Shorten Time to Diagnosis

- Better genetic testing guidelines may aid PCPs in genetic testing (25).
- Incorporating genetic testing into primary care could streamline testing, reduce time to diagnosis, improve outcomes, and decrease unnecessary specialist visits and travel costs (7, 20, 32).

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ACKNOWLEDGEMENTS

We would like to thank Dr. Donald Basel (Children's Wisconsin, Medical College of Wisconsin), Dr. Michael Muriello (Children's Wisconsin, Medical College of Wisconsin), and Dr. George Leyda, Jr (Hudson Physicians) for their input on this subject matter.