Rare Diseases: Common Medical Needs

People with rare diseases have common medical needs.
In fact, rare diseases are often discovered in the primary care setting, where patients continue to be seen for routine medical care throughout their lives. Despite primary care clinicians assuming responsibility for the management and care of these patients' common needs, the majority of primary care clinicians do not receive any special training on how to administer or adjust routine care for their patients with rare diseases.

Therefore, it may not be surprising that the hundreds of health care professionals (HCPs) Rare Opportunities in Primary Care spoke with over the last year highlighted this distressing trend: HCPs lack confidence in their ability to provide primary care for patients with rare diseases.*

What is Rare Opportunities in Primary Care?
Rare Opportunities in Primary Care is a new ally in the rare disease community that focuses specifically on improving the quality of primary care for people living with rare diseases, by:

- Championing clinical and administrative best practices for managing these conditions
- Equipping providers with tools and resources to address patients' unmet needs
- Facilitating dialogue between primary care and specialty care providers

Rare Opportunities in Primary Care is committed to enhancing primary care for patients with rare diseases. Learn more at: www.RareOpportunities.com.

*The research presented in this paper was conducted under the organization's former name, the Association for Comprehensive Care in Rare Diseases (ACCORD). The survey conducted was distributed with the help of Frontline Medical Communications.

Faculty Discussants

The following faculty members participated in a December 2015 call to discuss the results of Rare Opportunities in Primary Care's August 2015 survey on the topic of unmet needs in patients with rare diseases.

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Having a rare disease does not make a person "rare." "Cumulatively, rare diseases affect the lives of 30 million Americans," noted Robert Saul, MD, FACMG, FAAP, of the Children's Hospital of the Greenville Health System in Greenville, South Carolina, and the American Academy of Pediatrics Genetics in Primary Care Institute in Elk Grove Village, Illinois. "Clearly, primary care providers must be better prepared to help identify and care for these patients."

"It's not surprising that HCPs are uncomfortable dealing with rare diseases," commented Christie M. Ballantyne, MD, of Baylor College of Medicine and Houston Methodist DeBakey Heart & Vascular Center in Houston, Texas. Although 1 in every 10 Americans has a rare disease,[1] "each disease itself is rare. Therefore, HCPs do not have much practical experience with individual conditions."

Indeed, these thoughts reflect those of the 536 clinicians (Figure 1) who participated in an August 2015 survey designed to identify educational gaps associated with providing primary care for patients with rare diseases and gauge a level of interest in the creation of an association for HCPs who provide such care.[2]

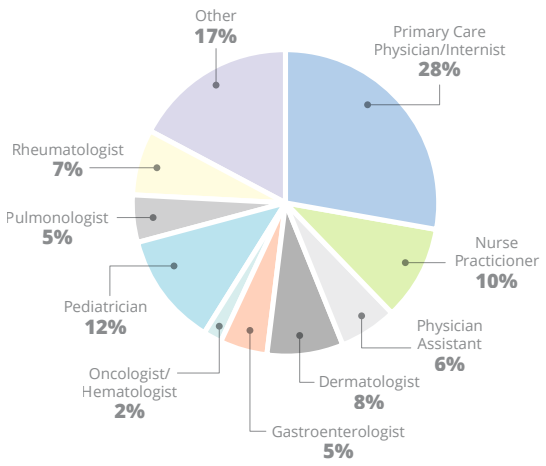
"It's becoming more and more common for primary care providers (PCPs) to see patients with rare diseases," said Jason C. Gallagher, PharmD, FCCP, FIDSA, BCPS, of Temple University in Philadelphia, Pennsylvania. "However, they simultaneously know less about them."

Appropriate Treatment Must Begin with an Accurate Diagnosis

One of the most common difficulties clinicians encounter when dealing with rare diseases is pinpointing an accurate diagnosis.[3] In fact, 60% of the 452 survey participants who responded to a question about diagnosis highlighted this fact, noting their lack of confidence when it came to identifying patients presenting with symptoms that might necessitate referral.[2]

"Referring appropriately is essential," noted Kristene C. Diggins, DNP, of Liberty University in Lynchburg, Virginia, Kaplan University in Indianapolis, Indiana, and MinuteClinic in Woonsocket, Rhode Island. "Often, initial assessments are based on health history alone. Clinicians must be able to identify anything that seems unusual. For instance, when diagnosing a suspected sore throat, they should look for anything that deviates from the virus' typical etiology. If they find any deviation—even if they don't understand exactly what it is—they should refer the patient to a specialist who might be able to provide more answers."

Figure 1. Participant Specialties (N = 536)[2]

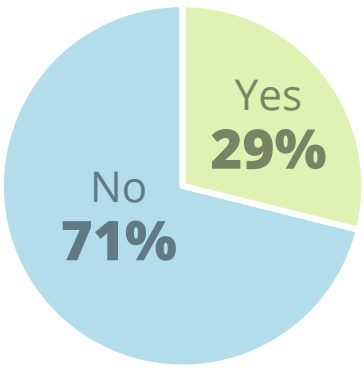


HCPs Need Standard Protocols for Providing Primary Care for Patients with Rare Diseases

One of the most important revelations uncovered by the August 2015 survey was that 71% of the 444 participants who responded to a question about protocols said that their practices did not have standardized protocols for managing patients with rare diseases (Figure 2).[2]

Such protocols might help prevent or delay disease progression by prompting earlier testing, noted Elizabeth A. Nguyen, RN, of the Worldwide Syringomyelia & Chiari Task Force Inc.

Figure 2. Respondents with Standardized Protocols for Managing Primary Care Issues in Patients with Rare Diseases (n = 444)[2]



in Lawrenceville, Georgia, citing the protocols emergency departments typically have for specific patient populations. "When I worked as an emergency room nurse, we had a certain protocol we followed when a patient came in having a stroke or in cardiac arrest. When we had codes, we used a pediatric crash cart or an adult crash cart—we followed a protocol that made the situation less stressful and more efficient for better outcomes," she explained. "I think it would be phenomenal if a similar type of protocol could be implemented for patients with rare diseases—it could save lives."

Given the scarcity of standardized protocols for treating patients with rare diseases, it may not be surprising that many of the HCPs surveyed noted that they lacked confidence in their abilities to provide routine primary care for patients with rare diseases (Table).[2]

"HCPs are still going to be treating issues such as sinus infections, strep throat, and influenza in these patients," noted Dr Diggins. "Being aware of the special considerations associated with treating these patients is very important."

Table. Sample Areas in Which HCPs Lack Confidence When Providing Routine Primary Care for Patients with Rare Diseases[2]

- Interpreting clinical lab test data
- Identifying which patients need to be referred
- Providing care for unrelated health issues
- Writing/refilling prescriptions
- Providing routine vaccinations

Note: n's varied.

In addition, patients who need multiple medications may experience drug interactions, added Todd I. Stone, PA-C, of Highland Medical Associates in Hendersonville, North Carolina, possibly explaining the lack of confidence revealed when survey respondents were asked about how comfortable they felt writing prescriptions for patients with rare diseases (Table).[2]

"Most people feel inadequately prepared when dealing with these types of issues," explained Dr Saul. "There is a great need for education in this space."

The Importance of Comprehensive Primary Care for Patients with Rare Diseases

"Even if primary care physicians can't offer a treatment or cure, patients just want to know that someone is taking care of them and monitoring them as they navigate the rare disease," Ms Nguyen explained. "The rare disease patient wants to have continuity of care when they leave the specialist, ongoing medical management, and collaborative care. Collaboration is required when complications arise that are outside the primary care physician's scope of practice. Rare disease patients are tired of "falling through the cracks." Poor medical management results in higher morbidity and mortality rates that could have been prevented."

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– **Robert Saul, MD, FACMG, FAAP**
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Moreover, as Kevin B. Gebke, MD, of Indiana University School of Medicine and Indiana University Health Physicians in Indianapolis, Indiana noted, ongoing primary care is so essential for patients with rare diseases because "rare diseases affect the body as a whole. Their impact is not necessarily limited to one section."

Comprehensive primary care includes not only screening for and managing physical symptoms but also monitoring patients for mental health issues and providing care when appropriate. "These conditions need to be managed," noted Daniel P. Duffy, MD, of the Medical College of Wisconsin and Columbia St. Mary's Community Physicians in Milwaukee, Wisconsin. "Many patients have anxiety and depression. We need to make sure they obtain appropriate treatment."

Certain Patient Populations Need Special Considerations

HCPs should be aware that special considerations may need to be addressed in women and children.[4-6]

Females with Rare Diseases

"I think that addressing the various aspects of being a female with a rare disease—from birth to postmenopause—is something that has been

severely lacking in the past," commented Dr Saul. "It's vital that we start to focus on these issues."

Children with Rare Diseases

Given that at least half of patients with rare diseases are younger than 18 years,[1] it also may be prudent for HCPs to pay special mind to issues that specifically affect children with rare diseases, including many physical, social, and developmental complications that may need to be addressed.[6]

In particular, HCPs may need to address concerns related to diet, nutrition, and exercise, said Kathi Kinnett, MSN, CNP, of Parent Project Muscular Dystrophy in Hackensack, New Jersey. Additionally, she noted, "Adolescents need age-appropriate care, and this group is often missed."

The Importance of Collaborative Care

"Often, best outcomes for patients stem from interprofessional collaboration," commented Dr Diggins.

"The ability to collaborate with specialists is so important," added Dr Duffy, noting that although he can easily find basic information on specific rare diseases with a few strokes of the computer, having a network of experts to call upon would make him feel more confident when working with patients with rare diseases.

Although all of the HCPs who participated in a December 2015 call on this topic agreed that successful interprofessional collaboration is vital when treating patients with rare diseases, the survey itself revealed that only about one-third of respondents rely on communication with specialists as their main source of information when caring for these patients (Figure 3).[2]

This, however, does not mean that there isn't a way forward. Both survey respondents[2] and the faculty discussants noted that there was a great interest in and need for education in the area of providing primary care for patients with rare diseases.

"Best outcomes for patients stem from interprofessional collaboration."

– **Kristene C. Diggins, DNP**

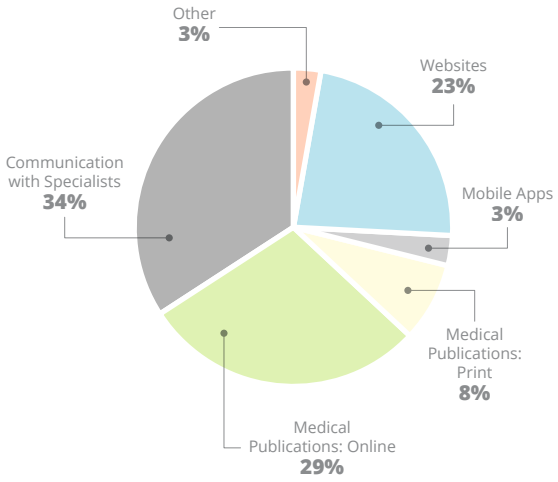
Liberty University Online, Lynchburg, VA
Kaplan University Online, Indianapolis, IN
MinuteClinic, Woonsocket, RI

"My experience definitely aligns with these survey results," noted Mr Stone. "The greatest unmet need in this area centers on access to information."

A New Path Forward

As noted in the box on page 1, Rare Opportunities in Primary Care was specifically founded with this in mind—with a mission of improving the quality of primary care for people living with rare diseases.

Figure 3. Clinicians' Main Source of Current Information on Managing Primary Care Issues for Patients with Rare Diseases (n = 452)[2]



Rare Opportunities in Primary Care hopes to bring these issues to light, as well as raise the bar for the quality of primary care provided to patients with rare diseases through its Web-based CME initiative, *Optimizing Primary Care for Patients with Rare Diseases*. The first program of its kind, this CME initiative will endeavor to help clinicians:

- Identify clinical clues that expedite diagnosis of rare diseases in different clinical settings
- Describe the role of genetic testing in diagnosing patients with suspected rare diseases
- Develop successful referral networks
- Optimize care for pediatric patients
- Utilize tools and resources to manage the whole patient
- Locate resources for clinical practice

"I'm very delighted to be a part of this," commented Dr Saul, who is serving as program chair. "We need to start taking action."

Accreditation

University of Cincinnati

This activity has been planned and implemented in accordance with the accreditation requirements and policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint providership of the University of Cincinnati and Health and Wellness Education Partners (HWeP).

The University of Cincinnati is accredited by the ACCME to provide continuing medical education for physicians.

Physicians | The University of Cincinnati designates this activity for a maximum of 5 *AMA PRA Category 1 Credits*™. Physicians should claim only the credits commensurate with the extent of their participation in the activity.

University of Cincinnati College of Nursing

Nurses | 5.25 continuing education contact hours for nurses approved by the Ohio Board of Nursing through the OBN Approver Unit at the University of Cincinnati College of Nursing (OBN-011- 93).

American Academy of Family Physicians

This activity, **Optimizing Primary Care for Patients with Rare Diseases**, with a beginning date of 07/01/2016, has been reviewed and is acceptable for up to 5 Prescribed credit(s) by the American Academy of Family Physicians. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

References

1. Global Genes. Rare diseases: facts and statistics. https://globalgenes.org/rare-diseases-facts-statistics/. Published 2015. Accessed June 8, 2016.
2. ACCORD. Rare disease management survey. Ramsey, NJ; 2015.
3. Engel PA, Bagal S, Broback M, Boice N. Physician and patient perceptions regarding physician training in rare diseases: the need for stronger educational initiatives for physicians. *J Rare Disord*. 2013;1(2):1-15.
4. Lara B, Fornet I, Goya M, et al. Contraception, pregnancy and rare respiratory diseases. *Arch Bronconeumol*. 2012;48(10):372-378.
5. Zimran A, Morris E, Mengel E, et al. The female Gaucher patient: the impact of enzyme replacement therapy around key reproductive events (menstruation, pregnancy and menopause). *Blood Cells Mol Dis*. 2009;43(3):264-288.
6. Poisson A, Nicolas A, Cochat P, et al. Behavioral disturbance and treatment strategies in Smith-Magenis syndrome. *Orphanet J Rare Dis*. 2015;10:111.