

Common Primary Care Issues That Arise When Treating Patients With Rare Diseases

Hegarty J, Smith PK, Rappa E, Giffen-Castro M, Osetto L, Cini J; Rare Opportunities in Primary Care, Health and Wellness Education Partners



Abstract

Objective: To characterize clinician perspectives on unmet needs in the management of primary care issues for patients with rare diseases. Methods: An electronic survey was distributed in August 2015 to assess clinician perspectives on providing primary care to patients with rare diseases. Questions assessed practice protocols, information sources, and clinician confidence in providing routine primary care for patients with rare diseases.¹ Results: Multiple specialties and provider types were represented among the 536 respondents who identified themselves as clinicians who manage primary care issues for patients with rare diseases. Respondents included primary care physicians/internists, pediatricians, nurse practitioners, and physician assistants; specialists represented among the respondents included oncologist/hematologists, pulmonologists, rheumatologists, gastroenterologists, and dermatologists. The majority of respondents (71%) indicated that their practices did not have any protocols for managing primary care issues for their patients with rare diseases. When respondents were asked about their main source of current information for managing their patients with rare diseases, they most frequently cited communication with specialists (34%). Online medical publications (29%) and websites (23%) were also frequently mentioned. With regard to confidence across clinical skills needed for optimal management of patients with rare diseases, nearly half of all respondents (42% to 50%), regardless of specialty, indicated low confidence levels (ratings of 1, 2, or 3 on a scale of 1 to 5, with 1 representing "not at all confident" and 5 representing "highly confident"). Areas where these clinicians lacked confidence included interpreting laboratory results, identifying patients for referral, providing routine vaccinations, and providing care for other unrelated health issues. Similarly, survey respondents indicated low confidence levels in prescribing medications (lifestyle medications, medications for preexisting comorbidities and/or comorbidities associated with the rare disease, and treatments to mitigate side effects) for patients with rare diseases. There were high levels of interest across training topics, including education on best practices for providing primary care services for patients with rare diseases, interpreting laboratory results, and advances in therapeutics. Conclusion: Survey results indicate that there is an immediate and imperative need for education, guidance, and support for clinicians providing primary care for patients with rare diseases. It is important to note that almost half of the respondents were specialists, demonstrating that the need for education in this area does not apply solely to primary care providers. Although specialists may have expertise in treating certain rare diseases, it is out of the purview of their everyday work to manage primary care issues such as giving patients routine vaccinations and providing care for other unrelated health issues. On the other hand, primary care providers may typically be better equipped to manage these routine issues, but their confidence levels drop when treating patients with rare diseases. Given that there are 7000 known rare diseases and an estimated 30 million affected people in the United States, it has become more imperative than ever to address these gaps in everyday patient care.²

Background:

Patients living with a rare disease commonly visit their primary care physician for routine illnesses and preventive medicine.³ Many primary care clinicians routinely see patients with rare diseases and are faced with challenges of meeting their health care needs.³ Despite primary care clinicians' role in routine management of the rare disease patient, primary care clinicians do not receive special training on how to administer or adapt care for their patients with rare diseases. Given that there are an estimated 30 million rare disease patients in the United States, 7000 different known rare diseases, and new treatments for rare diseases in the drug development pipeline, it is imperative to address these gaps in everyday patient care.²

Objective:

To characterize clinician perspectives on unmet needs in the management of primary care issues for patients with rare disease.

Methods:

An electronic survey was developed to better understand the clinical perspectives on factors influencing care of patients with rare diseases in a primary care setting. In August 2015, the survey was electronically distributed through SurveyMonkey to a broad sample of clinicians largely comprising primary care physicians/internists, pediatricians, and nurse practitioners. Clinicians who did not have patients receiving specialist care for a rare disease were ineligible to participate in the survey. Participants were asked about the presence of standardized protocols in their practice setting and about their main source of current information related to management of primary care issues for patients with rare diseases. Additionally, survey participants were asked to self-assess their level of confidence in several areas associated

Multiple specialties were represented

71% lack standard protocols for

managing primary care issues in patients

with rare diseases (n = 444)

In your practice setting, are there standardized

protocols for managing primary care issues for

patients with rare diseases?

No

71%

Nurse Practitioner

10%

with routine management of patients with rare diseases. Participants were then asked to rate their level of confidence in prescribing routine treatments (lifestyle medications, medications for comorbidities, or treatments to mitigate side effects) for their patients with rare diseases. Lastly, participants were asked about their level of interest in training on skills associated with rare disease management as well as specific medical education tactics. The number of respondents varied per question; aggregated survey responses are summarized using descriptive statistics.

Results:

Multiple specialties and types of providers were represented among the 536 respondents who identified themselves as clinicians who manage primary care issues for patients with rare diseases. Respondents included primary care physicians/internists (28%), pediatricians (12%), nurse practitioners (10%), and physician assistants (6%) as well as several types of specialists including dermatologists (8%), rheumatologists (7%), pulmonologists (5%), gastroenterologists (5%), oncologist/ hematologists (2%); and other unspecified specialty type (17%) (Figure 1).

The majority of respondents (71%, n = 444) indicated that their practices did not have any protocols for managing primary care issues for their patients with rare diseases (Figure 2).

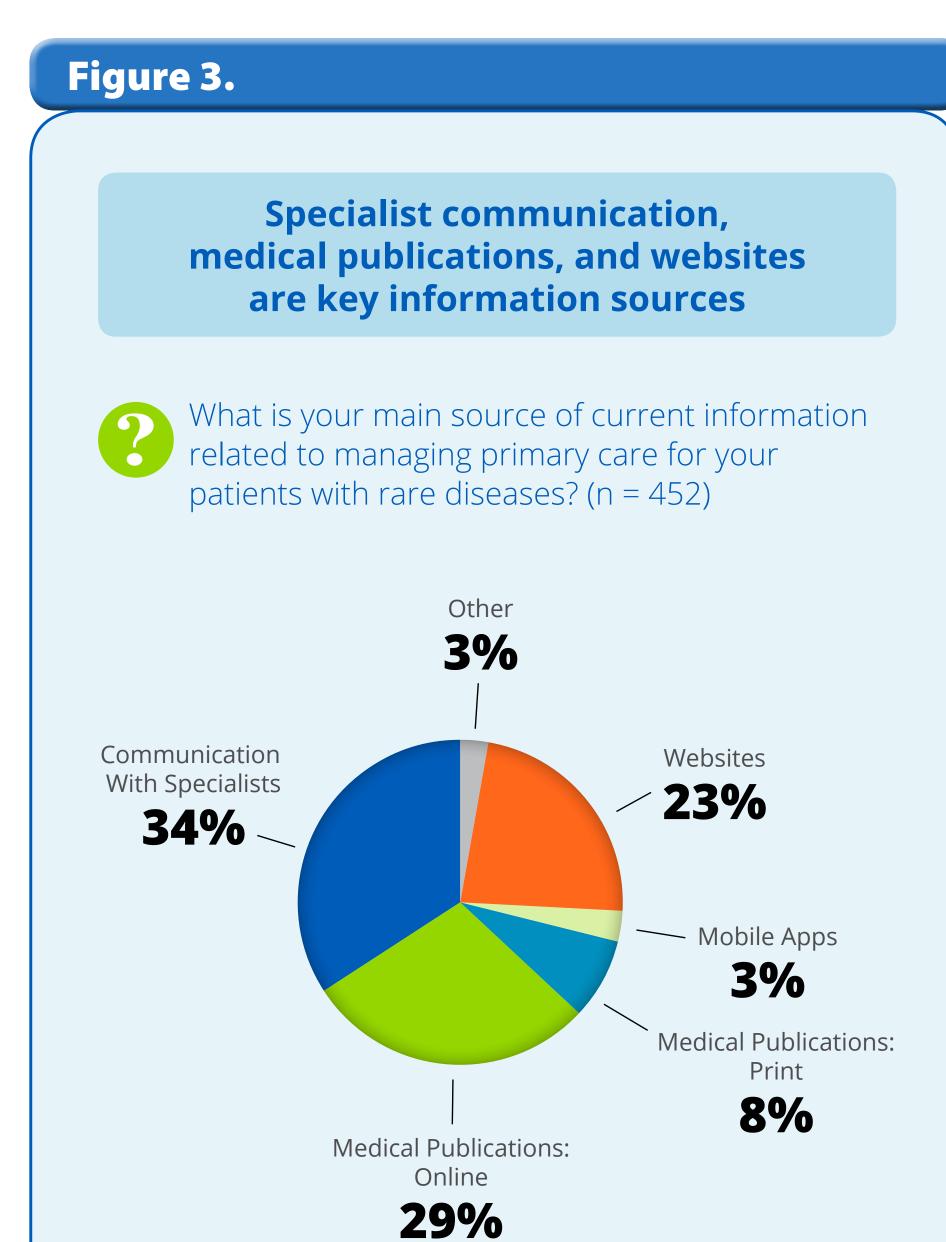
Respondents were asked about their main source of current information related to managing their patients with rare diseases. Among the 452 respondents, communication with specialists (34%) was the most frequently cited source.

Online medical publications (29%) and websites (23%) were also frequently mentioned. Print medical publications (8%), mobile apps (3%), and other sources (3%) were less frequently mentioned (Figure 3).

With regard to confidence across clinical skills needed for optimal management of patients with rare diseases, nearly half of all respondents (42% to 50%), regardless of specialty, indicated low confidence levels (ratings of 1, 2, or 3 on a scale of 1 to 5, with 1 representing "not at all confident" and 5 representing "highly confident") (Figure 4). A significant proportion of clinicians selfreported low levels of confidence in identifying patients for referral (42%, n = 452); providing routine vaccinations (49%, n = 450); providing care for other unrelated health issues (44%, n = 449); interpreting clinical lab test data (50%, n = 449); and refilling prescriptions prescribed by a specialist (47%, n = 448).

Similarly, survey respondents indicated low confidence levels in prescribing medications for patients with rare diseases (Figure 5). Specifically, respondents lacked confidence in prescribing lifestyle medications (54%, n = 450), medications for preexisting comorbidities (45%, n = 449) and/or comorbidities associated with the rare disease (51%, n = 445), and treatments to mitigate side effects (61%, n = 447).

When asked to indicate level of interest in obtaining additional training on topics related to providing comprehensive care for patients with rare diseases, respondents expressed high levels of interest (ratings of 4 or 5 on a scale of 1 to 5, with 1 representing "not interested" and 5 representing "highly interested") across training topics (Figure 6). Specifically, respondents showed high interest in training on identifying patients for referral (78%, n = 436), advances in therapeutics (76%, n = 441), best practices for providing primary care services for patients with rare diseases (80%, n = 445), attending to the whole patient (76%, n = 446), and interpreting clinical laboratory test data (79%, n = 442).



Approximately half of providers are

for patients with rare diseases

Indicate your level of confidence in the following:

■ Identifying patients for referral (n = 452)

■ Providing routine vaccinations (n = 450)

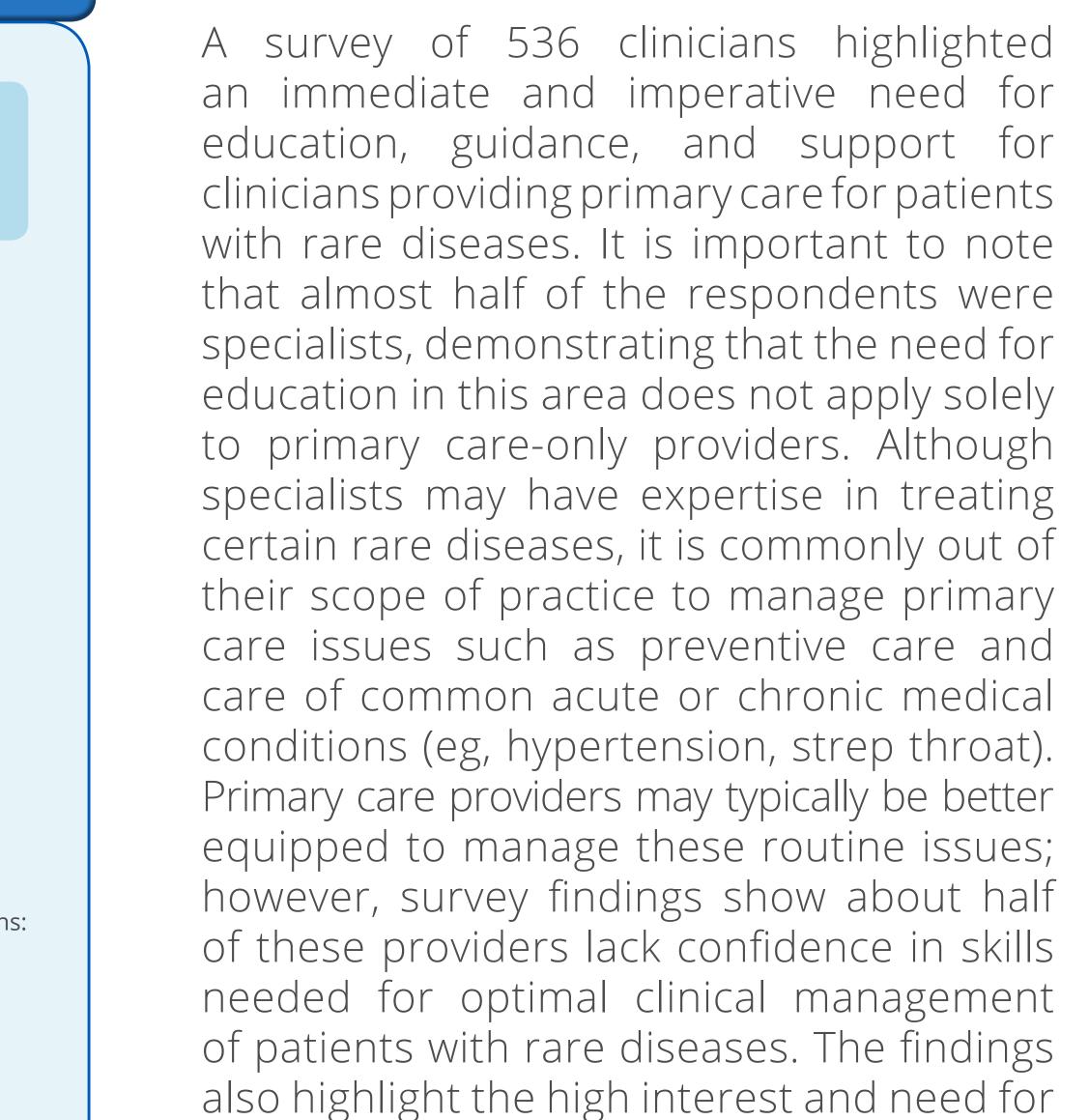
■ Interpreting clinical lab test data (n = 449)

Note: Percentages rounded to the nearest whole number.

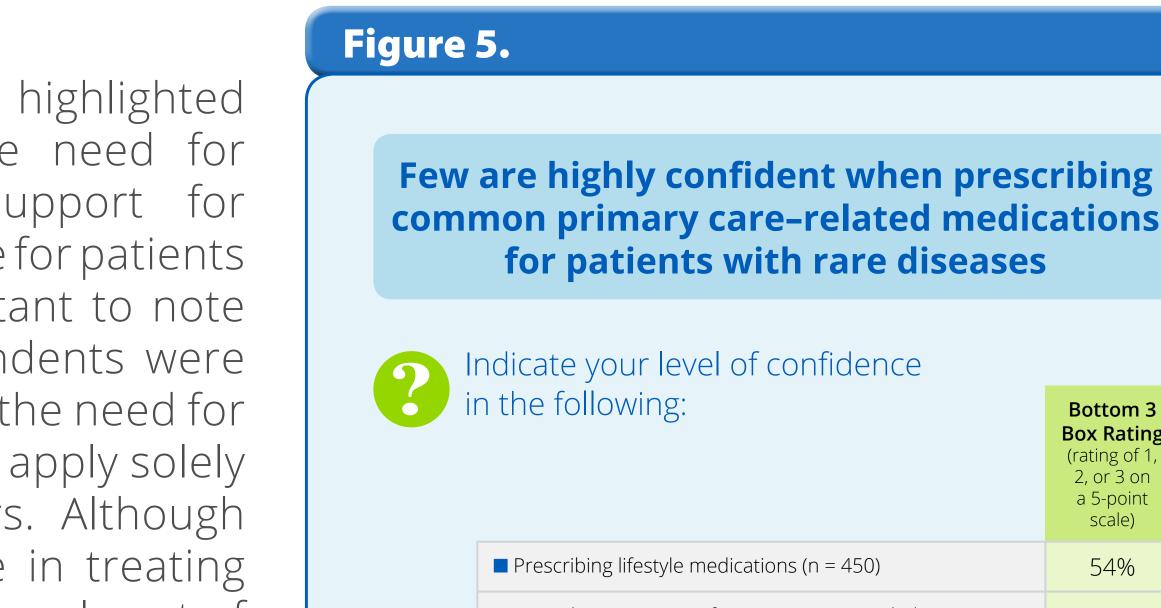
confident in providing routine primary care

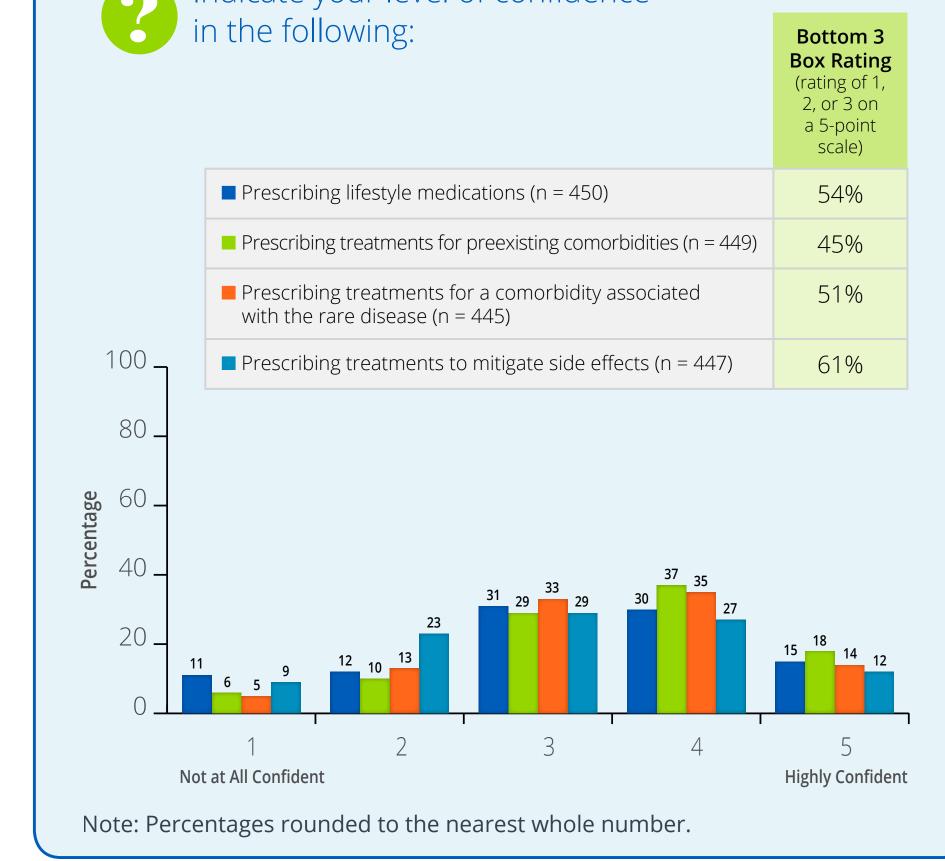
■ Providing care for other unrelated health issues (n = 449) 44%

■ Refilling prescriptions prescribed by specialist (n = 448) 47%



Conclusion:







Rare Opportunities in Primary Care is a that focuses specifically on improving the

education on topics related to optimizing

care for patients with rare diseases.

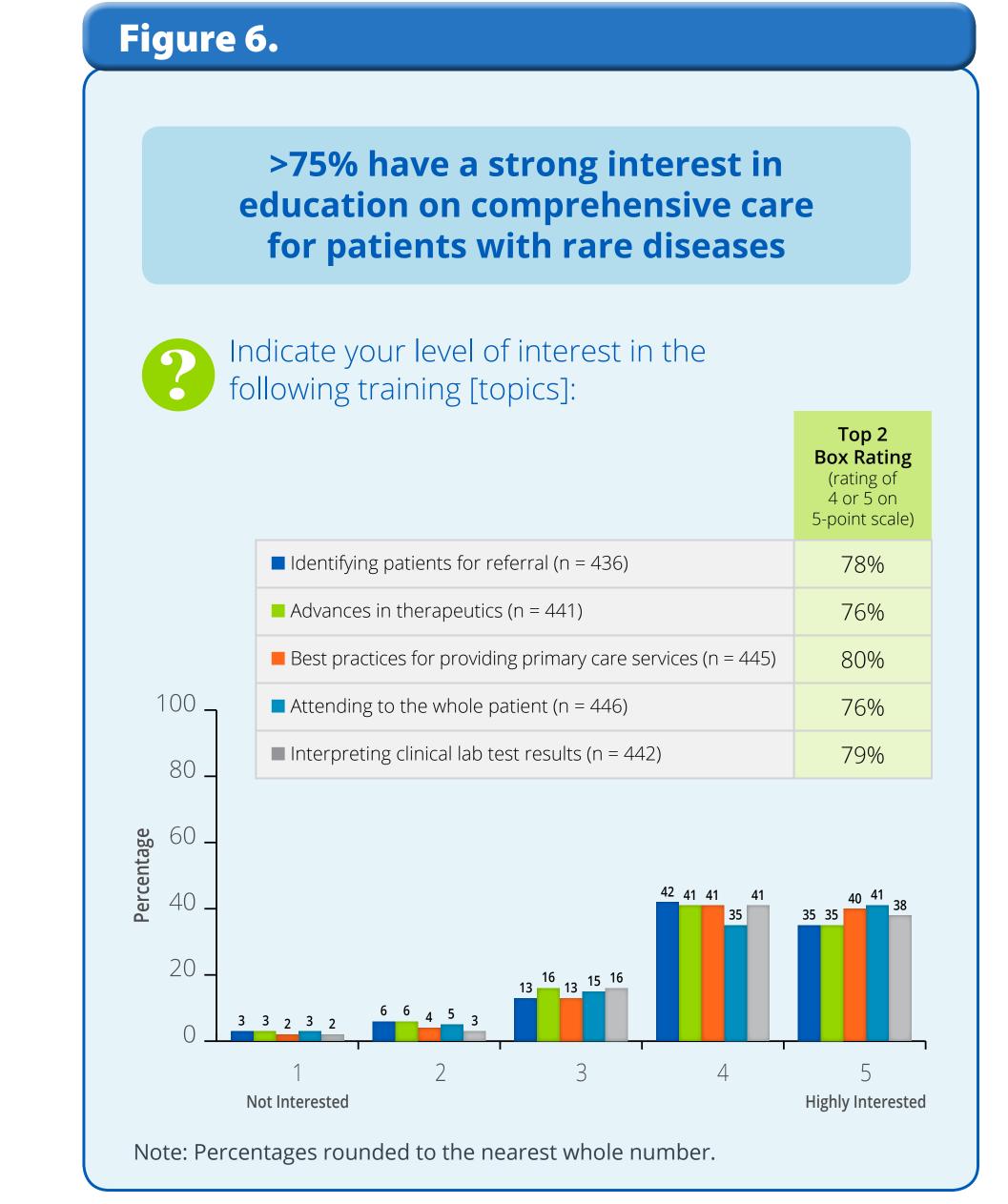
best practices for managing these conditions

 Equipping providers with tools and resources to address patients' unmet

 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

is an inititive sponsored by Health and Wellness Education Partners.



new ally in the rare disease community quality of primary care for people living with rare diseases, by: Championing clinical and administrative

Rare Opportunities in Primary Care

1. ACCORD.* Rare disease management survey. Ramsey, NJ; 2015. 2. National Organization for Rare Disorders. Neurological rare disease special report. Neurology Reviews. 2015;23(suppl 1):S1-S52. **3.** Phillips WR. Zebras on the commons: rare conditions in family practice. *J Am Board Fam Pract*. 2004;17(4):283-286.

Health and Wellness Education Partners, LLC | 70 Hilltop Road, Suite 2300-B, Ramsey, NJ 07446 | Phone: 201.661.5480 | Email: info@hwepnj.com * We would like to acknowledge Frontline Medical Communications for distribution of the survey