Rare Diseases and Educational Tools for Advanced Practitioners

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A rare disease is defined as a condition that affects fewer than 200,000 people. Yet cumulatively, rare diseases affect over 25 million individuals in the United States. Treatments for the over 7,000 rare diseases have historically lacked pharmaceutical development due to the high cost of research and potential loss of profits (Roberts & Wadhwa, 2025; Haendel et al., 2020).

The Orphan Drug Act, passed by the United States Congress in 1983, was intended to foster innovation and incentivize pharmaceutical companies to develop drugs to treat rare diseases. This law has increased access to treatment in many countries, including the United States. Despite more drugs available to treat these diseases, prompt and accurate diagnoses are not being made. In fact, many patients with rare diseases see an average of five to seven providers across multiple specialties before receiving an accurate diagnosis (Kittleson et al., 2023; Jennings et al., 2018).

Rare Mendelian diseases that hematology and oncology advanced practitioners (APs) may encounter include Huntington's disease, cystic fibrosis, and sickle cell anemia. Other types of rare diseases that can mimic other conditions include neurologic disorders such as amyotrophic lateral sclerosis (ALS), blood disorders such as Gaucher's or Castleman's disease, or uncommon cancers such as myeloproliferative neoplasms or amyloidosis. Each of these conditions has unique characteristics and complexities, yet all rare diseases share similar clinical challenges. Many clinicians are unlikely to have experience recognizing or managing these rare conditions in busy practices, where more common conditions are routinely encountered.

In 2023, Rohani-Montez and colleagues published a report highlighting gaps in knowledge on rare diseases and assessed learning preferences among clinicians in the United States and European Union. Survey respondents mentioned that case-based education (64%) and short-format (< 15 minutes) lectures (59%) were the preferred learning methods. Articles or publications were also deemed valuable for gaining rare disease education (57%).

In this issue, Kurtin and colleagues report the findings of a study that assessed patient and AP perspectives on symptom burden and symptom management in a rare condition: indolent systemic mastocytosis. Based on this research, a comprehensive Toolkit for Systemic Mastocytosis was developed by the Advanced Practitioner Society for

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Hematology and Oncology (APSHO). This provides an AP-focused resource for overcoming some of the barriers and gaps identified in the study. You can access this toolkit at https://www.apsho.org/apsho-aptoolkit-sm.

Highly educated APs are well suited to play a crucial role as experts in diagnosing and managing rare diseases and providing patient care throughout the care continuum. Given the complexity of rare diseases, timely diagnosis is difficult, which causes mental, physical, and financial burdens for patients and their families. The AP can provide appropriate patient education on how to manage the disease.

What is your experience with rare disorders, and are there any best practices you would like to share? I would love to hear from you, and I am sure your colleagues would as well. You can tag me and #JADPRO or #APSHO on LinkedIn: https://www.linkedin.com/in/bethfaiman/ or X: @Bethfaiman.

In This Issue

In addition to learning about indolent systemic mastocytosis and systemic mastocytosis, read about an APRN-led intervention for supporting patients as they transition from completing a phase I clinical trial. Using data from a Texas cancer screening program, the article by Chen and colleagues examined the impact of geographical factors, specifically rurality, persistent poverty, and racial residential segregation, on colorectal cancer screening adherence. In this issue's review article, get pharmacist perspectives and strategies for talquetamab for relapsed/refractory multiple myeloma. A case report in this issue highlights a patient who developed progressive multifocal leukoencephalopathy just 2 months after CAR T-cell therapy and underscores the need for better prevention and treatment strategies in immunocompromised patients. Another case describes a 10-year discordance of imaging that proved to be high-grade ductal carcinoma in situ, and therefore emphasizes the importance of understanding imaging protocols, recognizing imaging discrepancies, and guiding follow-up imaging procedures. A pilot program researched a walking plan and nurse education program to address cancer-related fatigue in patients undergoing cancer treatment.

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