

Awareness is key to diagnosing rare diseases

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(StatePoint) Nearly 30 million Americans -- or one in 10 -- are currently affected by a rare disease. Many of these patients are now starting to speak out about the unique challenges they face daily.

"Rare disease patients worldwide face many similar challenges, but because their conditions are so rare, they often feel alone and isolated," said Peter L. Saltonstall, President and CEO of the National Organization for Rare Disorders. "By standing together and sharing our experiences with rare diseases, we can more effectively address these challenges and build a better world for rare disease patients."

In the United States, a disease is considered rare if it affects fewer than 200,000 people. Because of the rarity of their conditions, patients may often be challenged to find information, support, and knowledgeable doctors. Furthermore, it often takes five years or longer to receive an accurate diagnosis of a rare disease. According to the National Institutes of Health, there are about 7000 identified rare diseases, 80 percent of which are genetic.

One such rare disease is Cryopyrin-Associated Periodic Syndromes (CAPS) -- a serious, lifelong autoinflammatory disease affecting about one in one million people in the United States. Because there are so few CAPS patients, online resources play an important role in uniting the community. CAPSConnectsUs.com, a new educational Web site from Novartis Pharmaceuticals Corporation, enables CAPS patients in the United States to connect online to share their stories, encouragement, and advice about managing daily challenges.

Dave Crawford of Dallas knows firsthand the importance of sharing his story with others. He was diagnosed with CAPS in 2010 after suffering from the painful, mysterious symptoms since childhood. He shared his story with his hometown newspaper last year and was contacted by a local man who had read about his experience. He explained that he and several family members had been experiencing the same symptoms that Crawford had described their whole lives, and following a referral to Crawford's physician, the man was also diagnosed with CAPS.

"I take every opportunity to speak out about CAPS, and it's a great feeling to know that my story helped others to get a correct diagnosis," said Crawford. "I remember the frustration of coping with the daily symptoms and not knowing what caused them, and I'm glad I could help end that

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mystery for another family."

For more than 50 years, Dave Crawford experienced daily red bumpy rashes, fevers, conjunctivitis, headaches, joint swelling, and muscle aches. The symptoms often occurred after exposure to cold or damp air, but also flared spontaneously. His mother, grandmother, and great-grandmother had also lived with the same painful symptoms, but never received a diagnosis.

"CAPS is treatable, but often misdiagnosed or undiagnosed because few physicians are familiar with it, and its symptoms can resemble more common conditions," said Dr. Barbara Baxter, an allergist in Dallas. "Rare disease patients are often undiagnosed for years, and accurately diagnosing a patient who has been struggling to get answers is one of the most rewarding experiences for a doctor."

Visit CAPSConnectsUS.com to see a video of Crawford's journey and to learn more about CAPS.