Hiremath G, Dellon ES. Commentary: Individuals affected by Eosinophilic Gastrointestinal Disorders Have Complex Unmet Needs and Experience Barriers to Care. J Rare Dis Res Treat. (2018) 3(2): 34-36

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Journal of Rare Diseases Research & Treatment

Commentary Open Access

Commentary: Individuals affected by Eosinophilic Gastrointestinal Disorders Have Complex Unmet Needs and Experience Barriers to Care

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Article Info

Article Notes
Received: June 16, 2018
Accepted: July 17, 2018

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By definition, rare diseases have low prevalence but they represent a large and diverse group of life-threatening or chronically debilitating conditions some of which may have no cure, existing pathways of care, or appropriate medical interventions. Collectively rare diseases pose significant public health burden and social issues as they affect approximately 6-10% of the world's population[1]. Individuals affected by rare diseases are often dispersed and face a variety of challenges including lack of or barriers to specialized care, delays in diagnosis, negative social consequences and other psychosocial burden[2].

In recent years, the medical, scientific and political communities have demonstrated renewed interest in rare diseases and rare diseases research. This in part is due to increased recognition of these conditions by the healthcare community, improved understanding of the underlying pathophysiology, advances to bring therapies to the market, and greater public awareness[3]. The Patient Advocacy Groups (PAGs) that promote the needs and priorities of patients play a crucial role in propagating the public awareness. Over the years, PAGs have evolved as one of the major stakeholders in advancing the care and research in rare diseases by forging collaborative partnerships with other stakeholders. They also have an important role in adoption of public policies, advocating for research funding, and fostering conducive environment for rare diseases research[4]. Despite all these advancements made in the realm of rare diseases, our understanding of the impact of these diseases on the lives of individuals affected is incomplete. This could be true for many of the common diseases as well. For instance, we do not fully comprehend the extent of their problems nor do we sufficiently understand the barriers to care experienced and/or perceived by the individuals affected by rare diseases.

Eosinophilic gastrointestinal disorders (EGIDs) are a group of rare diseases and are being increasingly recognized as an emerging healthcare problem. They represent heterogeneous conditions characterized by eosinophilic inflammation affecting various segments of the gastrointestinal tract, such as esophagus, stomach, small intestine and colon, referred to as eosinophilic esophagitis (EoE), eosinophilic gastritis (EG), eosinophilic gastroenteritis (EGE), and eosinophilic colitis (EC), respectively[5]. Of these, EoE is the most well studied entity and is estimated to affect 56.7 per 100,000 persons or 152,152 individuals in the United States (US).

While the exact burden of EG, EGE and EC is unclear, the estimated prevalence of EG is 6.3, EGE is 8.4, and EC is 3.3 per 100,000 persons in the US[6-8]. Recently, an NIH-funded multi-center Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR) have been studying these conditions[9]. Even though significant progress being made towards understanding the epidemiologic and genetic underpinnings of the EGIDs and EoE in particular, the diagnosis and monitoring of these conditions remains expensive and burdensome, and the commonly used treatment options are largely empiric or experimental[10]. As a result, individuals affected by EGIDs, like many of those affected by other rare diseases, have unmet needs and experience barriers to care. The article by Hiremath et al is first to systematically investigate the unmet needs and barriers perceived by the EGID community[11]. Their results indicate that the individuals affected by EGIDs have a constellation of unmet needs and experience barriers to care across multiple domains meaningful to them.

In this study, the authors used a community-based participatory approach wherein the PAGs, as representatives of the EGID community, were actively involved in conceptualizing study design, development of study protocol and survey questionnaire, and disseminating the survey through their web-based platforms (including online portals and social media outlets). The domains and questions most relevant to the EGID community were identified through focus group interviews conducted by the PAGs. The final questionnaire consisted of 58 questions spanning across medical, healthcare, social, and emotional impact domains. The responses were collected in a 5 point Likert scale (1=Totally agree; 5=Totally disagree). By leveraging partnership between PAGs and medical researchers and using the web-based approach this study was able to achieve participation from a large and diverse group of individuals affected by EGIDs. In all, 414 responses from adults diagnosed with EGIDs and adult caregivers of children (less than 18 years of age) with EGIDs were received. Of these, 361 were analyzed with 280 (78%) being complete in all aspects. The majority of respondents were adult caregivers of children with EGIDs, and the participants were from 9 countries.

In medical domain, over half of the participants indicated that they had to consult multiple providers prior to being diagnosed with EGID indicating that it is challenging to readily make this diagnosis in the field. Only a small proportion of participants indicated that current approach to diagnose EGID requiring repeated endoscopy was convenient (19%), and about one in three participants indicated that repeated endoscopy was affordable (33%). A large majority of respondents indicated that the current treatment for their EGID (i.e. dietary elimination, topical steroids, immunomodulators, elemental diet) was neither easy to adhere nor convenient. The responses in the healthcare domain emphasized the lack of insurance coverage for most common treatment modalities for EGIDs such as topical corticosteroids and elemental formulas. A notable proportion (64%) of respondents indicated that they did not have easy access to dieticians or nutritionists who they felt understood the challenges faced by EGID patients. In the social domain, the large majority (79%) of respondents indicated that they used social media to connect with peers and get social support. Approximately 62% had experienced food-related discrimination due to the dietary restrictions they need to adhere to as part of their treatment. The respondents also emphasized lack of knowledge about EGIDs in the school and at workplaces . With regards to the emotional impact, the large proportion (70%) of respondents indicated that they had support from their family members and friends. A large proportion of participants indicated that the EGID had placed significant emotional burden, and stress as a result of out-of-pocket costs related to EGIDs. When asked to prioritize the domains for improvement, the participants emphasized on improving the medical domain followed by healthcare and emotional impact domains. They felt that least amount of improvement was needed in the social challenges domain. Overall, the perceptions of unmet needs and barriers were similar between adult EGID patients and the adult caregivers of children with EGIDs.

The methodology used in this study has multiple limitations. Most importantly, the responses could be subject to significant bias and the authors were unable to estimate the response rate. Nonetheless, the real strength of this study is that it has initiated the process of appreciating unmet needs and barriers across multiple domains identified by the EGID community to be most relevant to them. Understanding the unmet needs and perceived barriers will serve as an important step towards inspiring future patient-centered research and informing the policies to bridge gaps and to ultimately advance the field of EGIDs and rare diseases research.

In conclusion, this study underscores the importance of PAGs in advocating for individuals with rare diseases, and is an example of advancing the field through partnership between medical research community and the PAGs. Understanding unmet needs and barriers to care experienced by the affected individuals will provide direction to the medical and health care research in rare diseases.

Acknowledgement

G.H and E.S.D are supported by Consortium of Gastrointestinal Eosinophilic Disease Researchers (CEGIR; U54 AI117804). CEGIR is part of the Rare Disease Clinical Research Network (RDCRN), an initiative of the Office of Rare Diseases Research (ORDR), National Center for

Advancing Translational Sciences (NCATS), and is cofunded by the National Institute of Allergy and Infectious Diseases (NIAID), National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), and NCATS. CEGIR is also supported by patient advocacy groups including the American Partnership for Eosinophilic Disorders (APFED), Campaign Urging Research for Eosinophilic Disease (CURED), and Eosinophilic Family Coalition (EFC).

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