

Empowering patients with rare diseases: Strategies for overcoming challenges and promoting self-advocacy



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Abstract

Patients with rare diseases often face significant challenges, including delayed diagnosis, limited treatment options and inadequate support systems. Empowering these patients involves a multifaceted approach that enhances their advocacy, education and access to care. We can significantly improve the quality of life for rare disease patients by collaboration among stakeholders, patients, healthcare providers, advocacy groups and focusing on the key areas offering them hope and the potential for better health outcomes.

1. Introduction

The medical conditions that affect a relatively small number of people compared to other, more common illnesses are termed as Orphan and rare diseases. These Orphan diseases generally receives less attention may be due to limited market potential for developing treatments. Many patient advocacy groups work globally to support research, raise awareness and improve the lives of patients with rare diseases. Patient advocacy plays a vital role in the Orphan and rare disease community and improving outcomes for patients and their families. Raising awareness in rare disease patient empowerment is a multifaceted effort aimed at increasing knowledge and understanding of rare disease among general public, healthcare providers and researchers.

2. Education and outreach programs

Various educational programs and outreach initiatives were develop and implement by Patient advocacy organizations to raise awareness about specific rare diseases. These programs may include webinars, workshops, seminars and providing educational materials targeting patients, their families and healthcare providers. Patient empowering starts by providing them useful information about their condition, including its causes, symptoms, prognosis, treatments option and various lifestyle adjustments or management strategies. Along with this, various media outlets that highlights the challenges and needs associated with rare diseases are also comprehensively informed. Education and outreach programs focused on rare diseases serve an important role in bridging gaps in knowledge, support and resources. It helps in empowering patients and families better equipped to manage health conditions. This knowledge enables patients to make informed decisions about their care and advocate for themselves effectively (1).

3. Public awareness campaigns

Advocacy groups may set up public awareness campaigns to increase visibility, understanding and support for rare diseases. It helps in increasing public awareness about existence and challenges of rare diseases or for the conditions that are often overlooked. It generates interest and funding resources for the development related to treatment and potential cures. Awareness campaigns require careful planning, robust engagement strategies as because rare diseases affects small number of people that makes it challenging to capture public interest. These campaigns often utilize traditional and digital media channels, social media platforms, and community events to reach a wide audience and share information about rare diseases, their symptoms, diagnosis and available resources (2).

4. Advocacy events and awareness days

One of the advocacy event like Rare Disease Day celebrated on 28th February of every year to provide opportunities for individuals and organizations to come together to raise awareness, share information and advocate for policy change. Advocacy events are designed to engage with policy makers, healthcare providers and broader public to drive changes that benefit the rare disease community. The purpose of this event is to educate the stakeholders about importance of supporting research funding, healthcare reforms and policies. These advocacy events often include rallies, legislative conferences, testimony from patients and experts, panel discussions on specific needs, fundraisers and social media campaigns to amplify the voices of the rare disease community (3).

Capitol Hill days that directly engages elected officials to discuss rare disease community needs, are kind of advocacy events. In this event, group of individuals gather in the capital city, often in the United States, to meet with lawmakers and advocate for specific policies or causes. It plays an important role in pushing for changes that significantly impact healthcare policy and research funding and also in improvement of drug development incentives. Awareness days are vital for educating the public and rallying support around the globe. Each year, on awareness day a thematic campaign is organized that highlights a particular aspect of rare diseases. Such events and awareness days are awareness tools in fight to bring attention and resources to rare disease community (4).

5. Storytelling and patient narratives

Storytelling and patient narratives are one of the powerful tools for raising awareness, fostering empathy and driving change within rare disease community. These narratives provide a human face to the statistics and medical jargon, helping to connect with audiences on a personal level. These are the personal stories and experiences of individuals affected by rare diseases. Patient advocacy organizations highlight patient narratives through websites, social media and written publications to humanize the impact of rare diseases and inspire action (5).

6. Collaboration and partnerships

Collaboration and partnerships are essential in addressing the complex challenges associated with rare diseases. No single entity can effectively tackle such issues alone having limited resources, expertise and patient population. Different groups or collaborations like research includes pharmaceutical companies that can accelerate drug discovery and clinical trials, academic institutions that conduct basic research, government agencies i.e National Institute of Health (NIH) or European medicines agency that provides funding and regulatory support. Collaboration with biotech firms helps in developing diagnostic tests, biomarkers and personalized therapies for rare diseases and also patient-reported data or exchange of genomic data for research purpose by data sharing companies (6). Partnership leads to enhanced patient care, improving outcomes and quality of life. It strengthens advocacy by increasing influence in policy discussions and decision making process. Partnerships can accelerate research among academia, industries and government. Besides having enormous benefits, collaboration and partnership faces challenges like sustainability, effective communication among stakeholders, data protection, regulation ensurance as well as clear agreements regarding Intellectual Property Rights (7).

7. Legislative and self-advocacy

Advocacy groups advocate for legislative initiatives and policies that benefit individuals affected by rare diseases. This may include advocating for increases funding for research, incentives for orphan drug development, improved access to healthcare services and protections for patient's rights. Empowering patients involves encouraging them to advocate for their needs and rights within the healthcare system and society at large. Also increases access to treatments, participating in research initiatives, raising awareness about their condition and promoting policy changes that benefit the rare disease community (8).

Legislative advocacy and self- advocacy both are essential strategies for addressing the unique challenges faced by rare disease patients. Legislative advocacy helps in identifying priorities of research funding through various government agencies, push for regulatory reforms that streamline the orphan drug development process and incentives. Also plays an important role in building relationships with elected officials, advocacy groups, healthcare providers for amplify the collective voice of rare disease community. It provides policy education by briefing information, data and personal stories that highlight the importance of addressing rare diseases and participation in advisory panels before experts. Self- advocacy empowers individuals affected by rare diseases to assert their rights, access necessary resources and advocate for their own needs. It builds up knowledge and awareness in understanding rights as a patient, assessment of healthcare and privacy protection including option for treatment and creating a network of support and solidarity (9).

8. Shared decision-making

Encouraging shared decision- making between patients and healthcare providers allows patients to actively participate in their treatment plans. By involving patients in discussions about treatment options, risks and benefits. Healthcare providers can ensure that care plans align with patient's preferences, values and goals. Providing patients with clear, unbiased information about their condition, treatment options, risks, benefits and uncertainties helps in patient autonomy. The key components behind shared decision- making are Information exchange about patient's condition, deliberation, improved communication. It also incorporates with many challenges and barriers i.e. time constraints, cultural and linguistic differences, health literacy and power imbalance (10).

9. Health literacy and communication building

Empowering patients involves promoting health literacy and communication skills to effectively navigate the complexities of the healthcare system, communicate with healthcare providers, ask questions and express concerns. This helps patients in managing their health. Communication building in the context of rare diseases involves creating networks of support, advocacy and collaboration among patients, caregivers, healthcare providers, researchers and stakeholders. Patient and family support by online forums and support group, peer mentoring programs, various campaigns and events or collaborative advocacy efforts builds up good communication. Research collaboration involves patients and caregivers in all stages of research process, from study design and recruitment to data analysis and dissemination of results. Adopts Community- based Participatory research (CBPR) approaches that prioritize rare disease community (11).

10. Self-management skills

Patient empowerment involves equipping individuals with the skills and resources they need to actively manage their condition on a day- to- day basis. This may include teaching patients techniques for symptom management, monitoring their health status, adhering to treatment regimens and recognizing signs of complications. Self-management skills include health literacy by understanding the condition and familiarize to medical terminologies. Symptom monitoring by

using symptom tracking tools such as diaries, apps or wearable devices, to document symptoms, triggers and patterns over time. Medication management by adhering to prescribed medications, including dosage, schedules and potential side effects. Healthy lifestyle practices by adopting a balanced diet incorporate regular physical exercise and healthy sleep habits. Stress management by relaxation techniques, mindfulness and stress reduction activities. By empowering patients with the knowledge, skills and resources they need to take an active role in their own care, we can improve quality of life and foster resilience in the face of rare diseases (12).

11. Conclusion

Empowering patients with rare diseases is a multifaceted endeavour that requires collaboration among patients, caregivers, healthcare providers, researchers and policymakers. By empowering patients to become active participants in their own care, we can foster resilience, promote well-being and create a more inclusive and supportive healthcare system for individuals affected by rare diseases.

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