Engaging Patients with Rare Disease in Identifying Meaningful Approaches to Comparative Effectiveness Research and Value Assessment

Project Overview

Background

Rare diseases represent a broad spectrum of illness. While each rare disease affects a relatively small patient population (defined as fewer than 200,000 individuals in the U.S.[i]), in aggregate more than 30 million Americans live with rare diseases and disorders[ii]. One study has estimated the total economic burden of rare diseases in the U.S. as $997 billion in 2019[iii].

With such a small number of individuals impacted by any given rare disease, researchers often have difficulty meeting traditional standards for comparative effectiveness research[1] and other research studies. The growing ability to identify, diagnose, and develop new therapies demands new approaches in outcomes and comparative effectiveness research.

Researchers and decision-makers across the U.S. healthcare system are seeking direction on the full range of outcomes that should be considered when evaluating treatment options in the rare disease space. With a lack of consensus on how to assess comparative effectiveness and value in this arena[iv], the challenges to building the evidence base on effectiveness and value of emerging therapies may pose a significant barrier to patient access. The current approach includes a focus on one disease at a time, given the lack of consensus on common clinical and patient-reported outcomes across diseases. But with an estimated 10,000+ rare diseases and disorders, this approach will not keep pace with the number of therapies being developed.

Project Objectives

Using IVI's open-source "learning laboratory" approach to value assessment, IVI, in partnership with the EveryLife Foundation for Rare Diseases, seeks to convene experts from across the patient and family, research, and healthcare communities to engage in an iterative process to plan for the development of new approaches in outcomes and comparative effectiveness research that advance our understanding of patient-centered outcomes in rare disease.

IVI is uniquely qualified to convene patients and a range of stakeholders to contribute to the design of new approaches for understanding the value of interventions for rare diseases, including cell and gene therapies. IVI has experience in stakeholder engagement, patient preference research, and developing novel methods and practices to inform comparative effectiveness research and value assessment. The specific objectives of this project include:

- Explore existing gaps in data, challenges for conducting research and treatment evaluations, and resources or guidance to assist stakeholders in evaluating evidence for therapies for rare diseases.

[1] Definition: The generation and synthesis of evidence that compares the benefits and harms of alternative methods to prevent, diagnose, treat, and monitor a clinical condition or to improve the delivery of care. The purpose of comparative effectiveness research is to assist consumers, clinicians, purchasers, and policymakers to make informed decisions that will improve health care at both the individual and population levels.

This project is partially funded through a Patient-Centered Outcomes Research Institute® (PCORI) Eugene Washington PCORI Engagement Award (EASCS-26726) and Alexion Pharmaceuticals

- Explore the full range of outcomes for patients living with rare disease that are important to patients and caregivers, and prioritize common outcomes across multiple rare conditions that inform decision-making for therapies for rare diseases.
- Develop recommendations for evaluating evidence, including patient and caregiver outcomes, as part of comparative effectiveness research.
- Build consensus on approaches and methods for engaging patients, caregivers, and other stakeholders to address research challenges in evaluating therapies for rare diseases.

Methods/Activities

1. Convene a Steering Committee that includes patient leaders, researchers, and other experts in the field to assist and advise IVI on project implementation.

2. Conduct a literature review of outcomes and comparative effectiveness research in rare diseases and of existing economic models, and produce a summary of the literature review findings to inform project activities. This review will assess value assessment frameworks to evaluate novel therapies for rare diseases, and guide selection of up to three disease areas (based on input from the Steering Committee and other advisors) to further evaluate outcomes and comparative effectiveness research and economic models.

3. Host three Roundtable Meetings of patient community representatives and other stakeholders to explore key concepts, including gaps in outcomes and comparative effectiveness research, patient preference research, and data inputs.

4. Produce a stakeholder convening proceedings report based on the Roundtable Meetings. Consensus and recommendations included in the report can inform future outcomes and comparative effectiveness research studies, including on the types of economic burdens that should be incorporated in research, data gaps that are important to bridge in order to improve patient-centered decision making, and potential value assessment model inputs.

[i] *Rare Diseases Act of 2002.* 107 P.L. 280. 42 USC 201. 2002 Enacted H.R. 4013.

[ii] *Rare Diseases Act of 2002.* 107 P.L. 280. 42 USC 201. 2002 Enacted H.R. 4013.

[iii] Yang G, Cintina I, Pariser A, Oehrlein E, Sullivan J, Kennedy A. The national economic burden of rare disease in the United States in 2019. Orphanet Journal of Rare Diseases (2022) 17:163. https://doi.org/10.1186/s13023-022-02299-5

[iv] Morel T, Arickx F, Befrits G, Siviero P, van der Meijden C, Xoxi E, et al. Reconciling uncertainty of costs and outcomes with the need for access to orphan medicinal products: a comparative study of managed entry agreements across seven European countries. Orphanet J Rare Dis. 2013;8:198.

This project is partially funded through a Patient-Centered Outcomes Research Institute® (PCORI) Eugene Washington PCORI Engagement Award (EASCS-26726) and Alexion Pharmaceuticals