

Valuing Rare Disease Treatments in Healthcare: Real Experience, Real Impact

Rare diseases collectively impact over 30 million people in the United States and impose a significant economic burden of \$997 billion per year. However, due to the limited number of patients affected by any individual rare disease, conventional research methods face challenges in effectively studying them. Therefore, novel approaches to measuring outcomes and conducting research are needed. Lack of consensus on assessing the value and effectiveness of treatments for rare diseases hinders the establishment of an evidence-base to inform healthcare decisions and limits patient access to innovative therapies.

The Innovation and Value Initiative (IVI) and the EveryLife Foundation for Rare Diseases collaborated on a project titled, "Valuing Rare Disease Treatments in Healthcare: Real Experience, Real Impact." This project aims to explore common patient-centered outcomes across rare diseases, identify gaps, and achieve a consensus on addressing unique research challenges. The project team convened experts in patient-centered research and those with lived experience to examine the issues and build consensus on patient-centered outcomes.

The project is overseen by a diverse, 15-member Steering Committee comprised of patients, caregivers, clinicians, payers, industry experts, and other stakeholders. Three roundtable discussions were conducted, with the participation of 46 attendees. These discussions aimed to achieve several key objectives, such as identifying expected outcomes significant to rare disease patients and caregivers, promoting communication between payer and patient stakeholders, and reaching a consensus on recommendations for enhancing patient-centered outcomes research. A literature review was also conducted through published literature on rare disease organization reports to obtain informative insights for this research.

From these engagements, key themes emerged on what matters most to patients, caregivers, and other stakeholders in developing approaches in comparative effectiveness research and value assessment, particularly in the context of understanding outcomes important to rare disease patients. The key themes include: patient journey and time, caregiver journey, early and continuing engagement and communication, data and methods, economic impacts, scientific spillover, and identifying common patient-centered outcomes for economic modeling.

In addition to discussing key themes and recommendations, the team examined patient-centered outcomes through a combination of literature reviews and consultations with patients and their families. As part of this analysis, the project team compared outcomes used in research on 11 different rare diseases. Certain outcomes, such as physical functioning (motor, respiratory, and speech), fatigue, social relationships, pain, mental deterioration, mental health, employment/work, economic impacts, and sleep, were common across more than half of these rare diseases. Additionally, outcomes based on the specific type of rare disease were explored, providing valuable insights into the diverse needs and priorities of patients and their families.

This IVI-EveryLife Foundation project has made progress in understanding the challenges of patient-centered outcomes research for rare diseases. The findings will inform the development of more effective research approaches to patient engagement and development of recommendations for patient-centered outcomes in rare disease research studies, leading to more informed selection of outcomes that are important for patients and their families.

SEVEN KEY THEMES



Scientific Spillover

Innovative treatments developed for one rare disease may be re-purposed or modified to treat additional rare diseases. Given the limited resources and data available for the study of rare disease treatments, the value of it could be especially beneficial.



Patient Journey and Time

The patient journey and time, which include the disease's natural history and its evolving impact on patients, is important to consider when identifying and prioritizing outcomes important to rare disease patients.



Caregiver Journey

Throughout a patient's health journey, caregivers provide vital support, assistance, and advocacy for their loved ones. It is essential to understand the health and other impacts on caregivers.



Identifying Common Patient-Centered Outcomes for Economic Modeling

Identifying common patient-centered outcomes across rare diseases can help accelerate cross-cutting research, enhancing our understanding of diseases themselves, patient perspectives, and potentially improve the comparative effectiveness research methods.



Economic Impacts

Understanding the economic impacts on patients and caregivers can help payers and other decision-makers to better design plans and strategies to ultimately improve patients' experiences and outcomes.



Early and Continuing Engagement and Communication

Initiating and continuing patient/caregiver engagement with other stakeholders is critically essential to conducting fully patient-centered research and helpful for decision-making.



Data and Methods

Comprehensive, representative, data plays an important role in value assessment and patient-centered outcomes research, especially when dealing with rare diseases. Collaboration should be encouraged to enhance data collection; and mixed (qualitative and quantitative) methods should also be promoted to incorporate lived experiences as meaningful input.

www.thevalueinitiative.org

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Scan here to learn more about the rare disease patient-centered outcomes project.

