Engaging Patients with Rare Disease in Identifying Meaningful Approaches to integrate Patient-Centered Outcomes in Comparative Effectiveness Research and Value Assessment

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BACKGROUND

The need to identify patient-centered outcomes for each rare disease contributes to uncertainty in evidence to inform healthcare decisions and difficulties in assessing the value and effectiveness of treatments for rare diseases, potentially limiting patient access to innovative therapies.

Novel approaches to identifying and measuring patient-centered outcomes are needed.

OBJECTIVE

To build toward consensus on recommendations for engaging patients in patient-centered outcomes research for rare diseases.

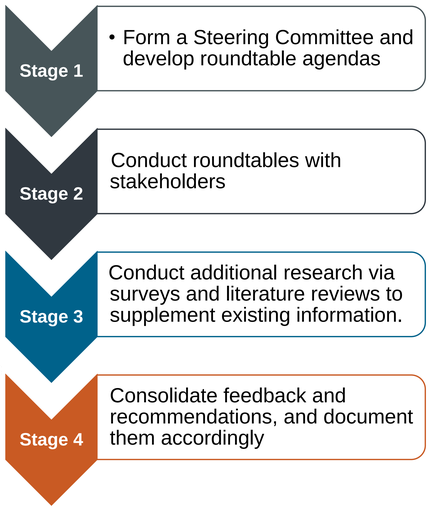
METHODS

A Steering Committee of 15 members was convened in 2023 to guide this research, including patients/caregivers, clinicians, payers, industry experts, and other stakeholders **(Figure 1).** Three multi-stakeholder roundtable discussions were conducted, with participation from 46 attendees, to: discuss processes for engaging rare disease patients to identify outcomes important to them and their caregivers, promote communication between payer and patient stakeholders, and develop recommendations for enhancing patient-centered outcomes research in rare diseases.

Discussions identified several themes important to consider when identifying patient-centered rare disease outcomes. With multiple recommendations per theme, a ranking survey was conducted to derive a list of prioritized recommendations within each theme, based on urgency and feasibility.

A literature review was also conducted in published literature and rare disease organization websites to obtain informative insights for this research.

Figure 1. Study Approach



RESULTS

Seven key themes and several recommendations per theme 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.

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 (Figure 2).**

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RESULTS (continued)

Prioritized recommendations include **earlier engagement and collaboration with patients/caregivers throughout the drug development timeline, continuous advocacy for research on outcomes significant to patients, and consistent data collection on these outcomes over time**. Additionally, gaining insights into the caregiver's journey and systematically collecting data on various aspects of their experiences is crucial. Prioritized recommendations ranked high on bother urgency and feasibility are shown in **Table 1**. More details can be read in the **Valuing Rare Disease Treatments in Healthcare: Real Experience, Real Impact** report.

Figure 2. Seven Key Themes

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.

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 (quantitative and qualitative) methods should also be promoted to incorporate lived experiences as meaningful input.

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.

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.

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 comparativeness effectiveness research methods.

Table 1. Prioritized Recommendations

Domain	Prioritized Recommendations ranked high on both Urgency and on Feasibility
Patient Journey/Time	• Researchers should collaborate with patients, advocacy groups, and rare disease organizations to include meaningful outcomes in clinical trials and studies. • Attributes like time to diagnosis, age at diagnosis, disease severity, treatment delays, time on treatment, and caregiver time require further exploration.
Caregiver Journey	• Researchers should understand the caregiver journey in relation to the individual's condition and note the conditions diagnosed in both over time.
Engagement and Communication	• Pharma companies and patient groups should collaborate earlier in drug development to ease time and resource pressures (early engagement). • Research and value assessment should use virtual strategies to involve patients and caregivers facing geographical or mobility challenges.
Data	• Promote research on outcomes considered important to patients and collect data on those outcomes over time. • Data collection should include caregiver perspectives, particularly in pediatric diseases with communication and cognitive challenges.
Methods	• Studies should be designed to consider the natural history of rare diseases, including the broad spectrum of symptoms, changes in symptom severity over time, and the time of onset.
Economic Impacts	• Value assessors and payers should consider condition variations and personal circumstances to lessen economic impacts on rare disease patients and families. • Researchers should design studies to capture broad economic impacts important to patients, including medical costs, caregiver impacts, and ability to work.
Scientific Spillover	• Research should evaluate the potential scientific spillover effects of rare disease research and treatments, including advancements that could impact common outcomes and apply to more common diseases.
Identifying Common Patient-Centered Outcomes	• Researchers should survey and interview patients, caregivers, researchers, and clinicians for a patient-centered view on outcome priorities. • Value assessors should partner with rare disease advocacy groups for insights (e.g., patient/caregiver advisory boards).

CONCLUSIONS

Strategic convenings with rare disease patients and other stakeholders led to key themes and a set of prioritized recommendation for identifying patient-centered outcomes. The key themes identified here offer a strategic framework for addressing research challenges in rare disease therapy evaluation.

Implementing these recommendations would represent a pivotal step towards advancing patient-centered outcomes research for rare diseases.