RARE DISEASE PATIENT ENGAGEMENT GUIDANCE AND CHECKLIST FOR VALUE RESEARCH

Rare diseases affect relatively small numbers of people, but they place a heavy and often lifelong burden on patients, families and caregivers. Decisions about rare disease treatments are frequently made with limited data, small studies, and outcome measures that do not fully reflect what matters most to people living with these conditions. As a result, comparative clinical effectiveness research (CER) and health technology assessment (HTA) sometimes can overlook important patient and caregiver experiences, and value assessments may not fully support fair and timely access to care.

The **Rare Disease Patient Engagement (RDPE) Guidance and Checklist** was created to address this gap. It is a practical resource that helps value researchers and their partners **plan, implement, and document** efforts to strengthen **patient and caregiver engagement in rare disease value research**. The guidance focuses on the full research lifecycle, from early study planning through dissemination and use of results, and highlights where patient experience data and patient-centered outcomes can and should inform design, analysis, and decision-making. HTA-specific items are highlighted so that users who work closely with payers and health technology assessment bodies can see where their efforts may have the greatest downstream impact.

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Conducted in partnership with the EveryLife Foundation for Rare Diseases and the National Organization for Rare Disorders (NORD), this work is part of the Center for Innovation & Value Research's (Center's) multi-year rare disease project. It is supported by a Patient-Centered Outcomes Research Institute (PCORI) Eugene Washington Engagement Award, with additional support from Alexion, AstraZeneca Rare Disease. An earlier phase of the project identified common patient-centered outcomes across rare diseases and produced recommendations on how to better reflect those outcomes in research and value assessment. Building on those findings, the RDPE Guidance and Checklist translates high-level principles into concrete questions and actions that teams can use in day-to-day work.

What this Guidance Provides

The document has two main components that are meant to be used together:

- **Guidance Text.** This section explains why patient engagement is especially important in rare diseases, outlines key concepts and definitions, and describes core elements of meaningful engagement such as early partnership, budgeting and participation supports, capacity building for all partners, diversity and equity, accessible communication, co-creation, and the use of patient experience data in economic modeling. The guidance also includes short, real-world examples that illustrate how these principles can work in practice.
- **Structured Checklist.** The checklist translates these concepts into specific questions, each linked to a clear objective and response. It is organized into four phases of a research or value assessment project:
    - **Initiation and Planning**
    - **Execution**
    - **Monitoring**
    - **Dissemination and Assessment**

Users can scan the checklist table to see where they are already strong and where gaps may exist. The accompanying explanations give more detail and examples for each item, and HTA-related items are flagged so that users can attend to them explicitly when preparing value assessments or payer-facing materials.

In the **Initiation and Planning** phase, the checklist focuses on engaging patients and caregivers early, budgeting for participation supports and training, and building capacity on both the researcher and patient sides. It prompts teams to consider who is at the table, how representative that group is of the broader rare disease community, and whether transparency, roles, and expectations are clearly defined from the start.

In **Execution**, the checklist addresses representative input and diversity, accessible communication, co-creation through two-way dialogue, and responsible data collection and use. Items ask whether communication is tailored to different languages and literacy levels, whether patients have helped shape study materials and methods, and whether patient experience and preference data, including economic impacts on families, are integrated into value research and modeling where feasible.

The **Monitoring** section centers on documenting what engagement activities were planned and carried out, how patient experience data are incorporated and updated, and how teams acknowledge and respond to evidence gaps and persistent rare disease challenges. It encourages users to describe the strategies they use to mitigate those challenges and to revisit engagement plans as studies evolve.

Finally, **Dissemination and Assessment** focuses on sharing results back to patients, caregivers, and communities in accessible and culturally appropriate ways, recognizing contributors, and maintaining transparent communication about goals, methods, data gaps, and conflicts of interest. The checklist also asks whether patients and caregivers are involved in dissemination activities and whether their feedback is sought to improve clarity and trust.

How this Guidance was Developed

The RDPE Guidance and Checklist reflects an iterative, multi-stakeholder development process. The Center first conducted a rare disease project that combined literature review and stakeholder convenings to identify common patient-centered outcomes and evidence gaps across rare diseases. In 2024, an advisory board of 19 members, including individuals with lived experience, researchers, advocates, and other experts, helped shape the initial framework and checklist.

In 2025, the Center worked with smaller advisory working groups to test and refine the checklist through case studies in three distinct rare disease areas: sickle cell disease, leukodystrophies, and generalized myasthenia gravis. Each case study involved focus group discussions with patients, caregivers, clinical and modeling experts, and advocacy organizations. A public comment period then invited further input from multiple stakeholders, including patient representatives, pharmaceutical companies, HTA organizations, and value researchers. Feedback from these activities led to several important refinements, including clearer language, stronger emphasis on co-creation, better signaling of HTA-relevant items, and expanded examples to support practical use.