

# Operationalizing Patient Engagement in Rare Disease Value Research: Lessons from Disease-Specific Use Cases

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## OBJECTIVE

**Problem:** While patient engagement is widely endorsed and crucial for rare diseases, researchers often lack practical, standardized "how-to" frameworks for partnering with patients and caregivers to improve real-world evidence generation and support timely access decisions.

**Objective:** Synthesize lessons from multiple disease-specific engagement activities to develop a [Rare Disease Patient Engagement \(RDPE\) Guidance and Checklist \(Figure 1\)](#) that can improve patient-centered CER and value research and support decision-making for access and coverage.

Figure 1. RDPE Guidance & Checklist At A Glance

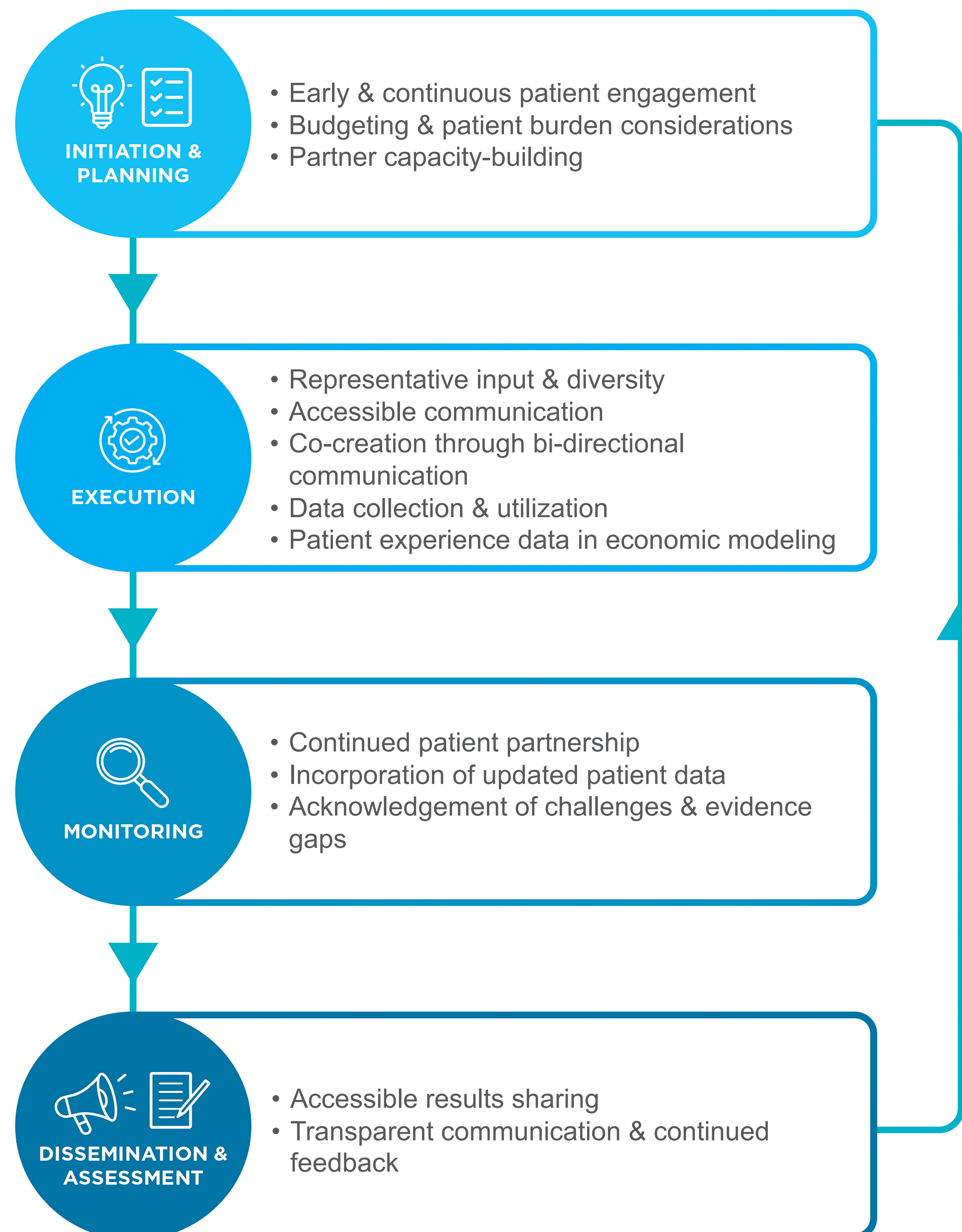


Table 1. Cross-Cutting Themes from Disease-Specific Use Cases

Cross-Cutting Themes	Details
<b>Early &amp; Continuous Engagement</b>	<ul style="list-style-type: none"> <li>• Start engagement at early planning phase</li> <li>• Maintain involvement throughout lifecycle</li> <li>• Define clear roles &amp; shared decision-making</li> </ul>
<b>Accessibility of Materials</b>	<ul style="list-style-type: none"> <li>• Use plain language</li> <li>• Provide practical examples</li> <li>• Ensure materials are usable for non-expert stakeholders</li> </ul>
<b>Inclusive &amp; Feasible Participation</b>	<ul style="list-style-type: none"> <li>• Improve representativeness – checklist question example: Are caregivers or proxies engaged for patients who cannot themselves be engaged (e.g., too young, severely cognitively impaired, other health reasons)?</li> <li>• Reduce participation burden</li> <li>• Use flexible engagement formats</li> </ul>
<b>Meaningful Outcomes</b>	<ul style="list-style-type: none"> <li>• Go beyond clinical endpoints to include broader patient-centered outcomes, such as caregiver impact</li> <li>• Align outcomes with real-world value discussions</li> </ul>
<b>Need for “How-To” Guidance</b>	<ul style="list-style-type: none"> <li>• Stakeholders want clear implementation steps, and standardized documentation practices</li> <li>• Transparency is critical for credibility</li> </ul>

## METHODS

The RDPE Guidance and Checklist was developed using an **iterative, multi-stakeholder** development process. The initial phase combined a 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, a smaller **advisory working group** with 10 members tested and refined 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 generated 11 submissions from diverse stakeholders, including patient representatives, pharmaceutical companies, HTA organizations, and value researchers, which informed further revisions to the checklist and guidance.

## RESULTS

Across disease-specific use cases, stakeholders consistently highlighted several key themes, presented in **Table 1**.

Based on these inputs, the **RDPE Guidance and Checklist (Figure 1)** was refined to offer high-level yet actionable guidance, accompanied by brief explanations, relevant examples, and links to supporting tools and resources where available.

- **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

## CONCLUSION

Patient engagement insights from disease-specific use cases can be converted into concrete guidance that helps teams operationalize patient partnership in rare disease CER and value research. The RDPE Guidance and Checklist aims to improve consistency, transparency, and relevance of evidence to inform access and coverage decisions, while supporting more meaningful participation by rare disease communities.

**View RDPE Guidance and Checklist**



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