LANDSCAPE ANALYSIS OF INITIATIVES TO IMPROVE ACCESS TO ORPHAN DRUGS LED BY INDUSTRY PATIENT AND ACADEMIC STAKEHOLDERS

Richard Macaulay[1], Anika Mavinkurve[1], Joel Iff[2*], Susan J. Ward[3]

[1] Precision AQ, London, UK; [2]Sarepta, USA; [3]Collaborative Trajectory Analysis Project, USA; *For further information, contact richard.macaulay@precisionaq.com*

INTRODUCTION

- 95% of rare diseases have no FDA-approved therapy, and there are even fewer EMA-approved rare disease therapies
- Further, orphan medicines that receive EMA-approval often fail the reimbursement/health technology assessment (HTA) hurdle
- Reasons are complex and include difficulties in meeting payer evidence standards in the face of challenging data collection[1]
- Multiple groups have undertaken initiatives to examine and potentially address the underlying challenges to secure approval and HTA/payer requirements. This research critically evaluates these groups and their activities

METHODS

- A targeted literature review was conducted in May 2025 using a structured search strategy to identify academic, industry, and patient organizations with publications, reports, and advocacy materials related to access challenges in rare diseases
- The approach combined both top-down and bottom-up searches via PubMed and Google to ensure comprehensive coverage and identification of rare disease access-focused groups
- The aims and activities of each group were extracted into a spreadsheet in the context of 4 pre-specified pillars, derived from ISPOR's Top 10 HEOR trends 2024-2025 and the ISPOR 2030 Strategic Plan: 1) Payer evidence standards, 2) Acceptability of real-world evidence (RWE), 3) Patient centredness/voice in HTA, 4) Affordability of new healthcare innovations
- Each group was rated on recency of deliverables, quality / relevance to impact across the relevant pillar for rare diseases, and likelihood of future activities planned

RESULTS

- The total number of identified groups was 21, with objectives distributed across the four pillars as follows: 5, 6, 11, and 8 groups
- The number of groups reporting recent (2024/2025) results/activities by pillar was only: 1/5, 2/6, 3/11, & 1/8, respectively *(Figure 1)*
- Majority (17/21) of these groups' activities focus on awareness-building initiatives such as webinars, white papers, and reports on challenges within pillars, rather than undertakings to specifically address/action identified challenges
- While some organizations have developed specific tools including frameworks/models to assess value of new therapies, educational decision-making support resources, and RWE/data-collection tools, these all tended to be country and/or disease-specific, limiting widespread usability *(Figure 2)*
- Current efforts are valuable but fragmented with regard to actionable solutions, leaving significant opportunity for novel and scalable approaches to address key challenges

Figure 1: Identified group policy activities and gaps by pillar

Pillar	Groups identified: Total	Groups identified: Rec. Active	Gap/Opportunity for Policy Change Initiatives
Addressing **payer evidence standards** in rare disease	5	1	• HTAi groups (and ISPOR, though less active recently) largely focus on this pillar; they advocate for rare disease and drug development, aiming to deliver actionable outputs that drive change
Acceptability of **Real World Evidence** in payer/HTA assessments	6	2	• Various groups are working to increase RWE acceptance in payer/HTA assessments, but rarely with a rare disease focus • GAP: It is unclear whether current efforts fully address RWE use in rare disease HTA assessments
Patient centeredness / voice in payer / HTA assessments	11	3	• There is growing momentum for patient engagement in HTA with major organizations, many closely linked to HTA bodies, launching targeted initiatives likely to yield actionable outcomes • GAP: Most efforts to date have focused on presentations and webinars rather than deeper engagement
Affordability of new healthcare innovations	8	1	• Despite growing emphasis on affordability, few organizations make it a core focus; most efforts center on advocating for value of rare disease innovations—e.g., contextualizing costs, integrating broader data, and amplifying patient voices in HTA decisions • GAP: A gap remains for initiatives addressing affordability directly rather than justifying cost and value

Figure 2: Resources developed to support HTA

In addition to policy-focused activities, several organizations have developed tools/resources to support HTA of rare disease therapies, though these tend to be tailored to specific countries or diseases

Models	Decision-Making Support	Data Collection
Organizations are developing frameworks and tools to assess the value of new therapies and ensure that healthcare advancements are accessible	Organizations are producing educational resources to improve understanding, support informed decision-making, and ensure healthcare advancements are effective & accessible	Organizations are developing frameworks & tools to collect and analyze real-world data, ensuring patient experiences & outcomes are central to healthcare policy and decision-making
Example: Duchenne UK developed a **natural history model & a core economic model** to help evaluate the clinical and economic impact of new treatments in DMD	Example: The Association for British Pharmaceutical Industry published a **UK-focused paper**, outlining **strategies & new payment models** to help lower costs of high-cost therapies for patients	Example: EURORDIS launched the Rare Barometer surveys globally to **assess impact of rare diseases on patient lives**, **sharing findings with decision makers to inform policy recommendations**

CONCLUSIONS

- Current gaps center on RWE acceptability and affordability, with patient voice in HTA still evolving despite ongoing initiatives
- Additionally, rare disease representation remains limited across all pillars, highlighting the need for targeted efforts to empower the global rare disease community to navigate complex HTA processes and improve access to innovative therapies
- These gaps could potentially be addressed by a rare disease consortium focused on prioritizing challenges and developing innovative solutions and policy recommendations to improve RWE acceptability, demonstrate value, and enhance access to therapies

PRECISION AQ™

cTAP 10 YEARS COLLABORATING TO ACCELERATE PROGRESS

SAREPTA THERAPEUTICS

[1]Nestler-Parr, S. et al., 2018. Challenges in research and health technology assessment of rare disease technologies: Report of the ISPOR Rare Disease Special Interest Group. *Value in Health*, 21(5), pp.493–500. Available at: https://doi.org/10.1016/j.jval.2018.03.004

* JI was an employee of Sarepta at the time of abstract submission