A Patient-Centered Methodology for Real-World Data Collection in Rare Genetic Diseases: Development and Pilot Implementation

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Purpose

The purpose of this study was to develop a comprehensive methodology for collecting real-world data (RWD) on rare genetic diseases, with a focus on capturing real-world experiences of Spanish-speaking patients and caregivers

This methodology aims to address critical gaps in current data collection practices and provide valuable insights that can inform decision-making, treatment strategies, and orphan drug development.

Background

Rare genetic diseases often lack sufficient real-world data to guide treatment choices and policy decisions.

Recent advancements in therapies, such as gene therapy, present new challenges for patients, caregivers, and healthcare systems, particularly in making informed decisions about treatment options.

There is a need for a patient-centered approach to gather comprehensive data that reflects the complexities of living with these conditions, especially in the Spanish-speaking population that is historically underserved and underrepresented in research and development of orphan drugs

Methods

We used a multi-stakeholder approach, conducting:

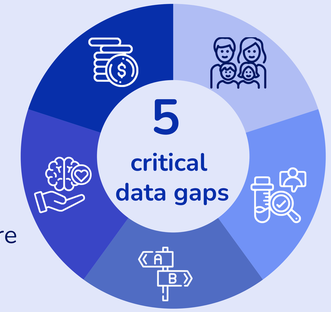
28 **in-depth interviews**

- **15 Patients & Caregivers**
- **3 Healthcare Professionals**
- **10 Industry Representatives** (Medical Affairs, Regulatory experts, Patient Advocacy, Sales & Government Relations professionals, CROs)

In-depth research on national/international patient registries

We identified 5 critical data gaps:

- Economic burdens
- Psychological burdens
- Decision-making processes
- Clinical trial experiences and perceptions
- Family journeys through clinical care



Outcomes:

- Developed a **disease-specific questionnaire using gamification approach** and tested with Spanish-speaking patients and caregivers
- Integrated into **a web app** supporting rare disease-affected families
- Implemented **e-consent & data validation** for ethical collection

Results

Our **Pilot Project is** focused on neuromuscular diseases such as *Duchenne muscular dystrophy*, where treatment options are rapidly evolving. It covers Spanish-speaking countries of the LatAm region



Implementation & Interim results:

- Web app captures real-world data (RWD) on *Patient & caregiver challenges, Decision-making & treatment options, Clinical trial experiences*
- **Gamification** improves engagement & data quality
- **Validated and verified data from patients stored in RWE repository**

Conclusions

Our scalable, patient-centered methodology enables real-world data collection on rare genetic diseases. By integrating validated questionnaires into a user-friendly web app, we captured critical insights to fill data gaps. This approach supports informed decision-making, optimizes treatments, and improves patient outcomes.

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