Aligning stakeholder terminologies and priorities in clinical trials to enhance outcomes for Lysosomal Storage Disorder (LSD) patients.

Authors

Daniel Lewi1, Kathleen Coolidge1, Femida Gwadry-Sridhar1

Affiliations

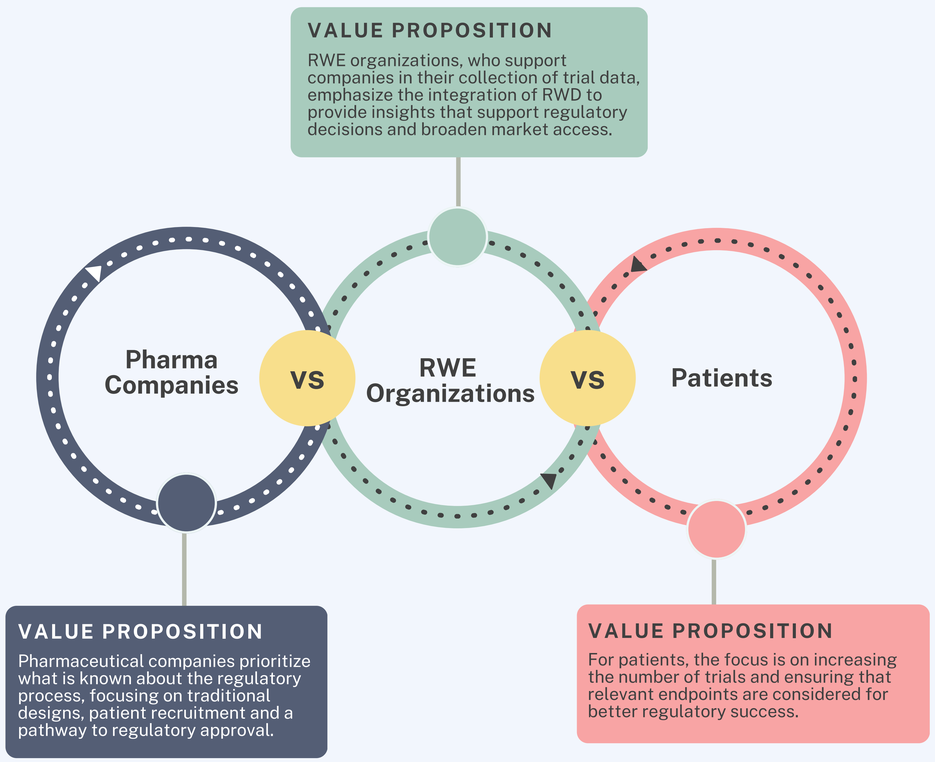
1. Pulse Infoframe, Inc., London, ON, Canada;

Introduction

The complexities of designing rare disease clinical trials are often exacerbated by the differing priorities and terminologies of stakeholders, including pharmaceutical companies, RWE (Real World Evidence) companies, and advocacy groups. In Lysosomal Storage Disorders (LSDs), these diverse perspectives can challenge innovation in trial design and hinder patient access to much-needed therapies.

Bridging these gaps requires collaboration and a shared framework to align goals and communication. Real World Data (RWD) serves as a crucial conduit for better understanding how to design studies that reflect real-world patient experiences, yet it remains underutilized due to barriers like data fragmentation and limited cross-stakeholder engagement.

Background



Analysis

The pharmaceutical industry, real-world evidence (RWE) companies, and patient advocacy groups all share a common goal: advancing treatment options for lysosomal storage disorders (LSDs) through increased clinical trial activity.

However, their approaches and priorities often differ, and discussions surrounding optimal study design remain insufficient.

Path for Improvement

	PHARMA	RWE COMPANIES	PATIENT COMMUNITIES
FAVORED APPROACH	Traditional randomized controlled trials (RCTs)	Leverage real-world data (RWD) to complement or substitute for trial components	Timely access to effective therapies
PROS OF APPROACH	Gold standard for regulatory approval	Reduce timelines, capture diverse patient populations, insights into long-term treatment outcomes	Advocacy drives interest in adaptive trial designs. Patients' real-world experiences help improve trial design and endpoints.
CONS OF APPROACH	Time-consuming and resource-intensive	Inconsistencies in data quality, standardization, and validation slow RWE's widespread acceptance	Urgent need for treatment conflicts with time needed to collect long-term safety data

Despite these shared interests, the potential of RWE as a pathway to accelerate treatment development through innovative trial designs—such as hybrid or pragmatic trials—remains underexplored.

Enhanced collaboration and dialogue among these stakeholders could help bridge these gaps, ensuring that study designs meet scientific, regulatory, and patient-centered goals.

Conclusion

The outcome of this review underscores the need for LSD patients to collect RWD. RWD studies should be designed to bridge the gap between industry requirements and patient needs ensuring that RWE effectively enhances trial recruitment, streamlines regulatory approvals and facilitates access to treatments.

This approach recognizes that small sample sizes, when accompanied by a well-defined and relevant data set that includes appropriate endpoints, can provide greater value than overly complicated study designs. By leveraging these insights, we can align research efforts with the needs of those most affected.

Acknowledgements

Thank you to those who agreed to be interviewed for this poster.

Mark Dant
Volunteer Executive Director
Ryan Foundation

Cyndi Frank
Co-President, Co-Founder
Gaucher Community Alliance

Patricia Durao
Co-Founder, Executive Director
CATS Foundation