Rare Disease Case Studies

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Case study:
End-to-End Evidence Generation in Dilated Cardiomyopathy (DCM)

Enabling progression from feasibility assessment to natural history study to external control arm (ECA) within a single integrated platform

Project Context

- Rare and heterogeneous disease (DCM) requires longitudinal real-world data to capture variability in disease progression, patient outcomes, and clinical treatment patterns
- In rare disease, conventional randomized trials alone are insufficient to support study design including definition of clinically meaningful endpoints in support of regulatory submissions
- Need for a scalable platform that supports feasibility analysis, natural history data collection, external control arm development, and long-term safety follow-up.
- Importance of regulatory alignment, patient inclusion, and site engagement across global geographies

Pulse implemented a staged, platform-enabled approach to accelerate evidence generation that scales with evolving sponsor requirements

Phase I: Discovery & Feasibility Analysis:

Multi-stakeholder interviews and site landscape review

Identified relevant endpoints, data availability and regulatory/scientific requirements

Assessed potential for decentralized patient recruitment

Phase II: Platform Configuration

Configured DCM, specific data structure (clinical, genomics, PROs)

Designed to support protocol and informed consent development , regulatory submissions and site onboarding

Phase III: Natural History Study Execution & ECA development

Launching across 8 global sites with real-time analytics dashboards

Collecting longitudinal, standardized data over 18 months

Developing a regulatory-ready External Control-Arm dataset

Case study:
Decentralized recruitment methodology

Prospective Observational Study to Assess the Long-term Safety of Olipudase Alfa in Pediatric Patients Less Than 2 Years of Age with ASMD: Study Design

Pablo Bianculli, Sefika Uslu Cil, Daniel Lewi, Kathleen Coolidge, Femida Gwadry-Sridhar, Judy Hull, Antonio Oliveira-dos-Santos

[Click here to read the poster]

An agile an efficient, decentralized study design by Pulse and accepted by FDA for an ultra rare disease

The project was to support a study to assess long-term safety and immune response of a drug in pediatric patients treated before age 2 and in patients with no age restriction.

Evaluate the relationship between treatment-related antibodies and safety outcomes Data Collection Timing: At every infusion (every 2 weeks) during dose escalation, every 3 months up to 24 months, and at 36 months..

A decentralized approach was implemented to reduce the burden of clinical visits, using digital technology for remote clinical data and lab test collection.

Decentralized design provided a variety of advantages including a more efficient approach than opening multiple sites, faster enrollment, no care transfer needed, local care convenience, reduced geographic barriers, and real-world data collection.

Case study:
Feasibility process for GM2 gangliosidosis

Pulse presented its feasibility assessment approach at the WORLD*Symposium* 2024 in relation to a project for GM2 gangliosidosis

- Real-world data (RWD) collection for GM2 gangliosidosis has long been fragmented and incomplete.
- Pulse Infoframe led a shift toward a more collaborative and standardized approach, aligning with FDA guidance and addressing key gaps such as missing clinical, genomic, and patient-reported data.
- By engaging clinicians, researchers, and advocates, Pulse developed a unified framework that supports transparent, high-quality data collection across drug development.
- This model not only improves research for GM2 but also offers a blueprint for advancing rare disease understanding more broadly.

Click here to read the poster

CDKL5 Global Registry

Key statistics

2 PARTNERS

The CDLK5 Registry was created in collaboration with the LouLou Foundation and Orphan Disease Center (UPenn)

LAUNCH

The CDLK5 Registry was launched in 2018 and was in development for 2 years. Now, 7 pharma have collaborated on an endpoint-enabling study with data linked to the CDKL5 registry through our GUID service.

PUBLICATIONS

Data from the CDLK5 Registry has been used in publications to increase the understanding of the disease

PATIENTS ENROLLED

Since its launch, the CDLK5 registry has enrolled over 170 patients and the number of anticipated patients is ~300

Global Access

The CDLK5 Registry has enrolled patients around the world, with the largest dataset collected on the CDLK5 community coming from the US and data shared with Australian researchers through our GUID.

Case study:
GUID-Enabled Data Linkage

CDKL5 REGISTRY

Enabling multi-sponsor collaboration through the CDKL5 Registry to link patient data across independent trials and real-world registries without compromising privacy

Project Context

- **Multi-sponsor registry initiative supporting multiple studies in a rare disease population**
- **Need to link patient data across independent trials and real-world registries without compromising privacy**
- **Key challenges: duplicate enrollment, inconsistent patient identifiers, and fragmented data sources**
- **Registry required a secure, standardized solution to harmonize data while complying with global data protection laws**
- **Ensuring data integrity and longitudinal tracking across studies was essential for regulatory-grade evidence**

Pulse implemented a Global Unique Identifier (GUID) system to assign each patient a consistent, anonymized ID across data sources.

GUID creation enabled secure data linkage without exposing personally identifiable information (PII).

Developed a privacy-preserving workflow that allows clinical sites to generate GUIDs independently which also allowed patients participating in multiple studies to be tracked and linked within the registry

Enabled a multi-sponsor registry infrastructure with clean, linkable patient data across studies which enhanced data quality and analytic depth by capturing full patient journeys across research contexts

Supported regulatory-grade evidence generation through consistent and traceable patient records which demonstrated Pulse's ability to implement privacy-first, interoperable data systems to support real-world research in rare diseases

Timeline

2016

Collaboration formed with the LouLou Foundation and Orphan Disease Center (UPenn)

2018

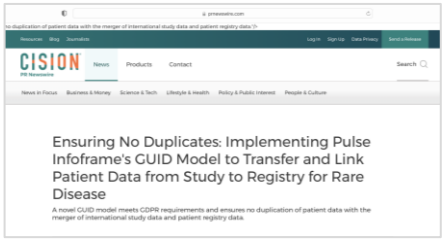
The CDLK5 Registry is launched, powered by Pulse's platform

2021

Pre-competitive pharma consortium established to initiate an endpoint-enabling study: CANDID

2022

Pulse GUID implemented to link CDKL5 Registry and CANDID Study data



2024-

Registry infrastructure and GUID leveraged to support additional observational studies

CDKL5 Global Registry

Goals, Impact, and Vision

WHAT?

A significant gap in the assessment of patient treatment and outcomes because of the lack of systematically captured, reported or analyzed Natural History of Disease.

WHY?

Physicians lacked insights on:

- Survivorship
- Quality of life (patient and caregiver)
- Lifetime cost of treatment
- Social impact of treatment
- Disease progression

HOW?

The CDKL5 Registry collects information from family/caregivers that links to clinical data.

Families are fully consented to learn about opportunities for CDKL5 clinical studies.

Pulse Infoframe provides a secure, data privacy-compliant, centralized evidence generation platform which meets regulatory requirements for clinical data.

RESULTS

Capturing Natural History of Disease data has led to a deeper understanding of:

- Implications and impact of genotyping
- Quality of life
- Impact of reduced time-to-diagnosis
- Guidance for symptom management
- Patient and caregiver preferences
- Implications for large patient cohorts and population health

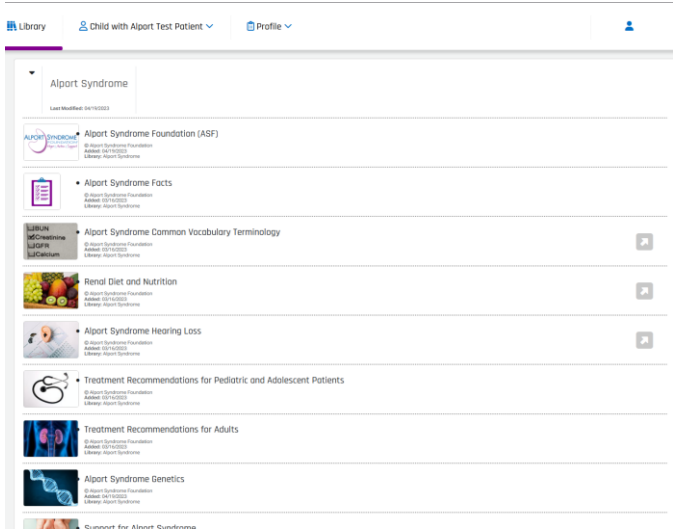
Incredible value for industry, physicians, payers, and most importantly patients has been unlocked, including::

- Greater number of companies being involved in drug development
- Endpoint-enabling study
- An MOU was established by Pulse to link data between the CDKL5 ODC registry and the IFCR funded Australian work
- What's next ? Additional studies and forming a central repository for CDKL5 data

Global Collaboration and Voice

Case study:
Patient engagement

Co-designed with direct feedback through ASF's in-person and virtual patient meetings, surveys, and private online support group played a vital role in identifying data elements.



Launched in August 2023, this IRB-approved, decentralized, ambispective, longitudinal natural history study has over 450 fully consented adult and minor participants

Collaborative approach with patient engagement group played a vital role in identifying data elements that position the registry at the forefront of patient-centered drug development in Alport syndrome

The platform offers interactivity and ease of use for participants, ongoing record collection every six months to monitor disease progression/symptoms/medication changes, and a library of educational resources for patients.

The registry has been designed to capture data to provide insight into the unmet need within a rapidly evolving treatment landscape: genetics, treatment history, hearing loss included.

CATNAP Narcolepsy Registry

Key statistics

2 PARTNERS

Children, Adolescents, and Their providers: the Narcolepsy Assessment Partnership (CATNAP™) is a retrospective and prospective longitudinal, multicenter web-based pediatric narcolepsy registry

PATIENTS ENROLLED

Since its launch, the **CDLK5 registry** has enrolled over 170 patients and the number of anticipated patients is ~300.

GROWTH

GUID leveraged to support additional observational studies, including an endpoint-enabling study launched in 2021 with a pre-competitive pharma consortium.

PUBLICATIONS

Data from the **CDLK5 Registry** has been used in publications to increase the understanding of the disease.

Global Access

The **CDLK5 Registry** has enrolled patients around the world, with the largest dataset collected on the CDLK5 community coming from the US and data shared with Australian researchers through our GUID.

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Case study:
A Patient-Centered Approach to Primary Biliary Cholangitis (PBC) Research

Challenge

- Rare autoimmune liver disease with severely impactful pruritus
- Small, dispersed patient population with cultural, language, and digital literacy barriers
- Difficulty linking symptom burden (pruritus, sleep disruption) to clinical and patient-reported outcomes

Solution

- Feasibility-driven, patient-first hybrid study design
- Multilingual, inclusive platform for decentralized participation
- Stakeholder collaboration and patient empowerment tools
- Integrated clinical and patient-reported data capture

Outcome

- Multilingual platform ready for observational data collection
- Clinical site activation in US and Germany
- Tools enhancing patient retention and experience
- Scalable model for future rare disease research registries

Pulse Infoframe's approach bridges stakeholder input, platform flexibility, and patient empowerment to ensure compliance, technical readiness, and a patient-centric model grounded in real-world patient voice

Thank You!

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