

CDKL5 Registry Review Committee Executive Summary

Presented to the ODC on January 30th, 2018

Committee Members*:

Jennifer Farmer:	Committee Chair, Executive Director of Friedreich's Ataxia Research Alliance
Michael Jasulavic:	Parent, Founder of MiaMed
Ana Mingorance, PhD:	Principal at Dracaena Consulting
Alisa Stephens- Shields, PhD:	Assistant Professor of Biostatistics at University of Pennsylvania
Rick Upp:	Parent, Vice President at the International Foundation for CDKL5 Research

**All committee members have no direct affiliation to ODC.*

Charge to the Committee: The CDKL5 Registry Review Committee was convened on August 23, 2017 by the University of Pennsylvania Orphan Disease Center (ODC) to evaluate the needs of the CDKL5 community regarding registries/natural history studies, and to provide recommendations to the ODC.

Process: The committee spent several months interviewing key stakeholders in the CDKL5 community; patient advocates and organizations, clinicians, researchers, industry representatives, the ODC registry team, and their platform technology partner Pulse InfoFrame (Pulse). These interviews explored experiences with registries and natural history studies, how the stakeholders want to leverage or interact with a registry, research priorities, and general concerns and recommendations in developing a CDKL5 registry. A detailed findings and recommendations document was prepared and presented to ODC in January 2018; summary of the findings and key recommendations are provided here.

Findings: It is the opinion of this Committee that at present, there is no CDKL5 registry that facilitates research on longitudinal data (observations over a long period of time) collected from both patient/caregivers and clinicians, and that facilitates international research collaboration. Several other registry efforts exist, but have significant limitations related to growth, sustainability and access. The CDKL5 community shares agreement on the need for a robust registry platform that includes portals for patients/caregivers, clinicians, and researchers. The proposed CDKL5 registry by ODC and Pulse can provide these elements.

Key Recommendations:

- The ODC should proceed with plans for a CDKL5 registry.
- The ODC should make every effort to include existing data sets in the registry, but not at the cost of significant slowing of the effort to launch and establish the registry.
- The ODC registry's patient/caregiver portal should be easy to use, efficient and flexible.
- The ODC registry's steering committee and data access board should have representation from all stakeholders (including patients and caregivers), and should establish a clear mechanism for qualified researchers to request access to the registry data at a low cost and within a short timeframe.
- To ensure sustainability, all stakeholders should collaborate on seeking or participating in financial support of the registry.