A novel approach at enabling the integration of additional data into the CDKL5 Registry

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INTRODUCTION

CDKL5 Deficiency Disorder (CDD)is a rare developmental epileptic encephalopathy caused by mutations in the CDKL5 gene, and this can manifest in a broad range of clinical symptoms and severity. The hallmarks are early-onset, intractable epilepsy and neurodevelopmental delay impacting cognitive, motor, speech, and visual function. Although rare, it is one of the most common forms of genetic epilepsy.

In 2018 the CDKL5 Registry was launched to the global CDD community. Developed in collaboration with the LouLou Foundation, a UK non-profit dedicated to advancing research into the understanding and development of therapeutics for CDD, and the Orphan Disease Center at the University of Pennsylvania's Perelman School of Medicine, the registry utilises the Pulse Infoframe registry platform. Data is collected for research and pharmaceutical development, as well as helping caregivers track their loved one's symptoms.

OBJECTIVES

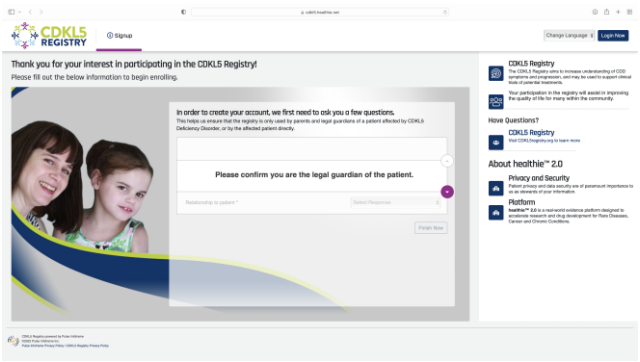
In 2021 an observational endpoint study for CDD called the Clinical Assessment of NeuroDevelopmental measures In CDD (CANDID) was announced. In this study the LouLou Foundation is serving as the study coordinator, and it involves CDD clinical centers worldwide where it is sponsored by an industry consortium of six companies with an interest in CDD.

As part of this study, the CDKL5 registry was further enhanced by the development and integration of a novel global unique identifier (GUID) into the Pulse registry platform. The GUID enables linkage of patient data collected from the three-year CANDID study.

DISCUSSION

By harmonizing the data between the study and registry, researchers can access a broader set of data to answer more nuanced questions around disease progressions and expression, such as those based on geographic location, social determinants of health, and more.

Figure 1. Recruitment Progress



For drug development, linking data through a GUID impacts important regulatory, pricing, and reimbursement-related decisions.

Over time, it becomes valuable for creating synthetic cohorts, which is especially useful in studying rare diseases, where large groups of patients can be difficult to find.

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