

Epilepsy disease classification: a community effort to enhance the Mondo Disease Ontology

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Abstract

Motivation: Epilepsy is a diverse group of neurological disorders affecting over 50 million people worldwide. While common epilepsy types are well studied, rare epilepsies—often severe and genetically complex—pose significant challenges in diagnosis, research, and treatment. Accurate and interoperable etiology and disease classifications are critical for improving data sharing, supporting clinical decision-making, and advancing rare disease research. **Results:** To enhance the accuracy of epilepsy-related disease concept representation within the Mondo Disease Ontology (Mondo), we conducted a series of expert-driven workshops in collaboration with the team from the Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP). Specialists in epileptology, genetics, neurodevelopment, biomedical ontology, and patient community advocates systematically reviewed and revised the epilepsy hierarchy in Mondo, aligning it with the International League Against Epilepsy (ILAE) classification system. These updates include reclassification of epilepsy subtypes, including syndromes, age-related epilepsies, and developmental epileptic encephalopathies, resulting in a more granular, standardized, and clinically relevant structure. Mondo now offers an enhanced framework for integrating epilepsy data across resources, enabling improved interoperability and facilitating rare disease research and data curation, with continued efforts underway to further refine and expand this integration.

Read this publication in its original format on *The Journal of Biological Databases and Curation*, [here](#).