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Standardized Data Structures in Rare Diseases: CDISC User Guides for Duchenne Muscular Dystrophy and Huntington's Disease

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Abstract

Interest in drug development for rare diseases has expanded dramatically since the Orphan Drug Act was passed in 1983, with 40% of new drug approvals in 2019 targeting orphan indications. However, limited quantitative understanding of natural history and disease progression hinders progress and increases the risks associated with rare disease drug development. Use of international data standards can assist in data harmonization and enable data exchange, integration into larger datasets, and a quantitative understanding of disease natural history. The US Food and Drug Administration (FDA) requires the use of Clinical Data Interchange Consortium (CDISC) Standards in new drug submissions to help the agency efficiently and effectively receive, process, review, and archive submissions, as well as to help integrate data to answer research questions. Such databases have been at the core of biomarker qualification efforts and fit-for-purpose models endorsed by the regulators. We describe the development of CDISC therapeutic area user guides for Duchenne muscular dystrophy and Huntington's disease through Critical Path Institute consortia. These guides describe formalized data structures and controlled terminology to map and integrate data from different sources. This will result in increased standardization of data collection and allow integration and comparison of data from multiple studies. Integration of multiple data sets enables a quantitative understanding of disease progression, which can help overcome common challenges in clinical trial design in these and other rare diseases. Ultimately, clinical data standardization will lead to a faster path to regulatory approval of urgently needed new therapies for patients.

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