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Improving Clinical Documentation of Rare Neuromuscular Diseases: Development of a Standardised Information Model

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Abstract. Rare neuromuscular diseases (NMDs) encompass various disorders of the nervous system and skeletal muscles, and present intricate challenges in diagnosis, treatment, and research due to their low prevalence and often diverse multisystemic manifestations. Leveraging collected patient data for secondary use and analysis holds promise for advancing medical understanding in this field. However, a certain level of data quality is a prerequisite for the methods that can be used to analyze data. The heterogeneous nature of NMDs poses a significant obstacle to the creation of standardized documentation, as there are still many challenges to accurate diagnosis and many discrepancies in the diagnostic process between different countries. This paper proposes the development of an information model tailored to NMDs, aiming to augment visibility, address deficiencies in documentation, and facilitate comprehensive analysis and research endeavors. By providing a structured framework, this model seeks to propel advancements in understanding and managing NMD, ultimately benefiting patients and healthcare providers worldwide.

Keywords. Rare Diseases, Interoperability, Neuromuscular Diseases, Standards

1. Introduction

Rare diseases (RDs) affect a significant portion of the global population [1], presenting unique challenges to patients, healthcare systems, and researchers alike. Neuromuscular diseases (NMDs) encompass a diverse range of disorders affecting the nervous system and muscles characterized by variability in disease presentation and progression, which requires a comprehensive approach [2]. Standardized documentation is pivotal in enhancing clinical care, facilitating research endeavors, and improving patient outcomes [3,4]. This work attempts to overcome the problem of unstructured collection of NMD patient data through the development of a standardized information model, which goes beyond the collection of routine data and is intended to be used for research purposes.

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2. Methods and Results

The data model developed in collaboration between computer scientists and physicians is based on the European Rare Disease Registry Infrastructure Common Data Set (ERDRI-CDS), the core data set of the German Medical Informatics Initiative (MII-CDS), and the catalog of the ERN NMD registry. Value sets were defined with frequent values for the individual data elements, and in some cases free-text is allowed. Data elements were coded using the ontologies and terminologies proposed by MII-CDS, ERDRI-CDS, the Systematized Nomenclature of Medicine Clinical Terms (SNOMED CT) and the HPO (Human Phenotype Ontology). Documentation of negative findings and exclusions is also permitted, as these can influence analyses. The model implementation in REDCap comprises: personal details, anamnesis of leading motor symptoms, motor impairment, accompanying symptoms, symptom-oriented examination, findings from diagnostic procedures (e.g., muscle MRI) and scores.

3. Discussion and Conclusions

Collected patient data for secondary use can provide many new insights for medical research. Methods like machine learning and AI-based algorithms are becoming increasingly prevalent and require a certain level of data quality including standardization [5]. The development and implementation of information models in different environments are intended to provide coherent access to clinical, genetic, functional, and histological data and imaging findings. Implementation of the same model at different locations facilitates cooperation between the institutions through the comparability of the data, which can be analyzed decentrally, anonymized across locations, and aggregated. To minimize the documentation workload, we use branching logic whenever possible and to integrate the model into hospital information systems. Developing a standardized information model for NMDs represents a crucial endeavor with far-reaching implications for clinical care, research, and patient outcomes.

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