Representing Disease Courses: An Application of the Neurological Disease Ontology to Multiple Sclerosis Typology

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1 INTRODUCTION

The Neurological Disease Ontology (ND) is being developed to provide a comprehensive framework for the representation of neurological diseases (Diehl et al., 2013). ND utilizes the model established by the Ontology for General Medical Science (OGMS) for the representation of entities in medicine and disease (Scheuermann et al., 2009). The goal of ND is to include information for each disease concerning its molecular, genetic, and environmental origins, the processes involved in its etiology and realization, as well as its clinical presentation including signs and symptoms. ND builds upon the Neuroscience Information Framework Standard ontology module, NIF_Dysfunction, which represents diseases affecting the nervous system (Bug et al., 2008).

Multiple sclerosis (MS) is an autoimmune demyelinating disease of the central nervous system. MS presents clinically through varied neurological symptoms including loss of motor control and balance, as well as visual and cognitive impairments (NMSS, 2013). A hallmark of MS is its manifestation through one or more patterns of neurological impairment: relapsing remitting (RRMS), secondary progressive (SPMS), or primary progressive (PPMS). Diagnoses of these clinical variants are based primarily on the frequency and intensity of episodes of neurological worsening. Disability in MS is assessed using the Kurtzke Expanded Disability Status Scale (EDSS).

2 RESULTS

ND and OGMS recognize that representing the complexity of disease manifestation requires multiple interrelated ontological classes. At the core of the OGMS extended picture of disease are the disease itself, its material basis (disorder), and the pathological processes that realize the disease (disease course). Existing MS disease hierarchies use terms that are often ambiguous or vague with respect to what aspect of the complete disease picture terms refer to, such as the material basis of a disease versus the processes that realize it. Diseases are not identical with their symptoms or stages of progression. Symptoms result from pathological processes realized in the patient the disease inheres in. This type of ambiguity must be addressed in order to provide a precise representation of the domain, which is essential for reliable storage, use, and reuse of medical data.

In this poster, we illustrate our solution to representing MS and MS disease courses. A unique aspect of these MS disease courses is the pattern of neurological worsening that occurs as the disease progresses. EDSS provides a way of quantifying the qualitative nature of disease progression. We contend that MS is best represented as a disease realized via multiple disease courses, or clinical variants, which present with one of three typical patterns of progression of disability. We formalized RRMS, SPMS, and PPMS as variants of disease courses differentiated by their unique patterns of disability, assessed using EDSS scores. In our approach, the distinction between the MS variants is not between unique disease types; rather, it is between which pattern of progression, characterized by neurological disability, the disease course has at a particular time. These patterns describe the general clinical variants of MS – not necessarily distinct diseases.

ND's use of the development versions of OGMS and BFO 2 has proved to be an excellent framework to accomplish this task. We extend OGMS:'disease course' to create the needed subtypes for each variant and add logical relations to connect associated classes, such as ND:'diagnosis of relapsing remitting multiple sclerosis' and ND:'demyelinating disorder'. We believe this disease model provides the most realistic ontological representation of MS thus far.

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