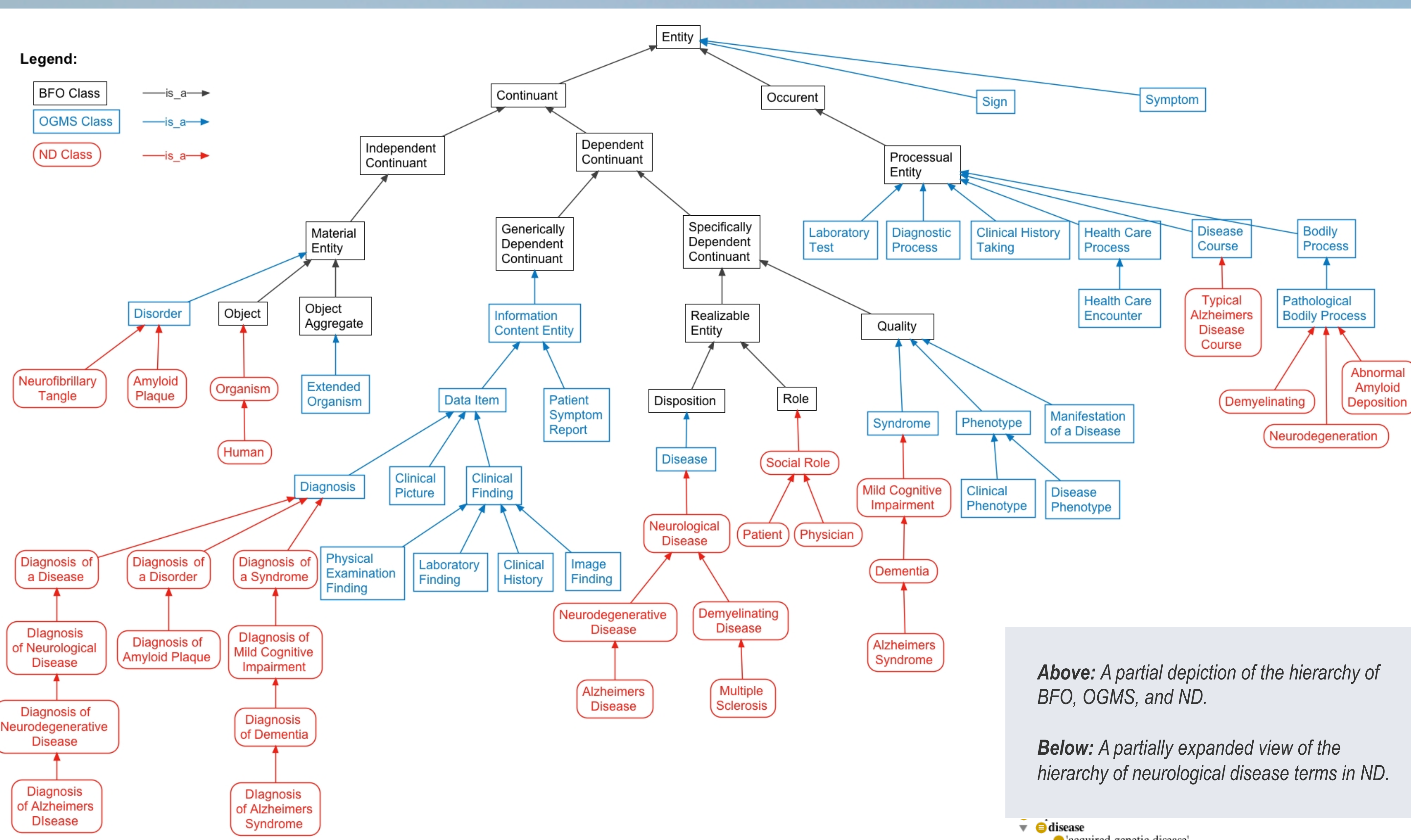# THE NEUROLOGICAL DISEASE ONTOLOGY (ND)

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Above: A partial depiction of the hierarchy of BFO, OGMS, and ND.

Below: A partially expanded view of the hierarchy of neurological disease terms in ND.
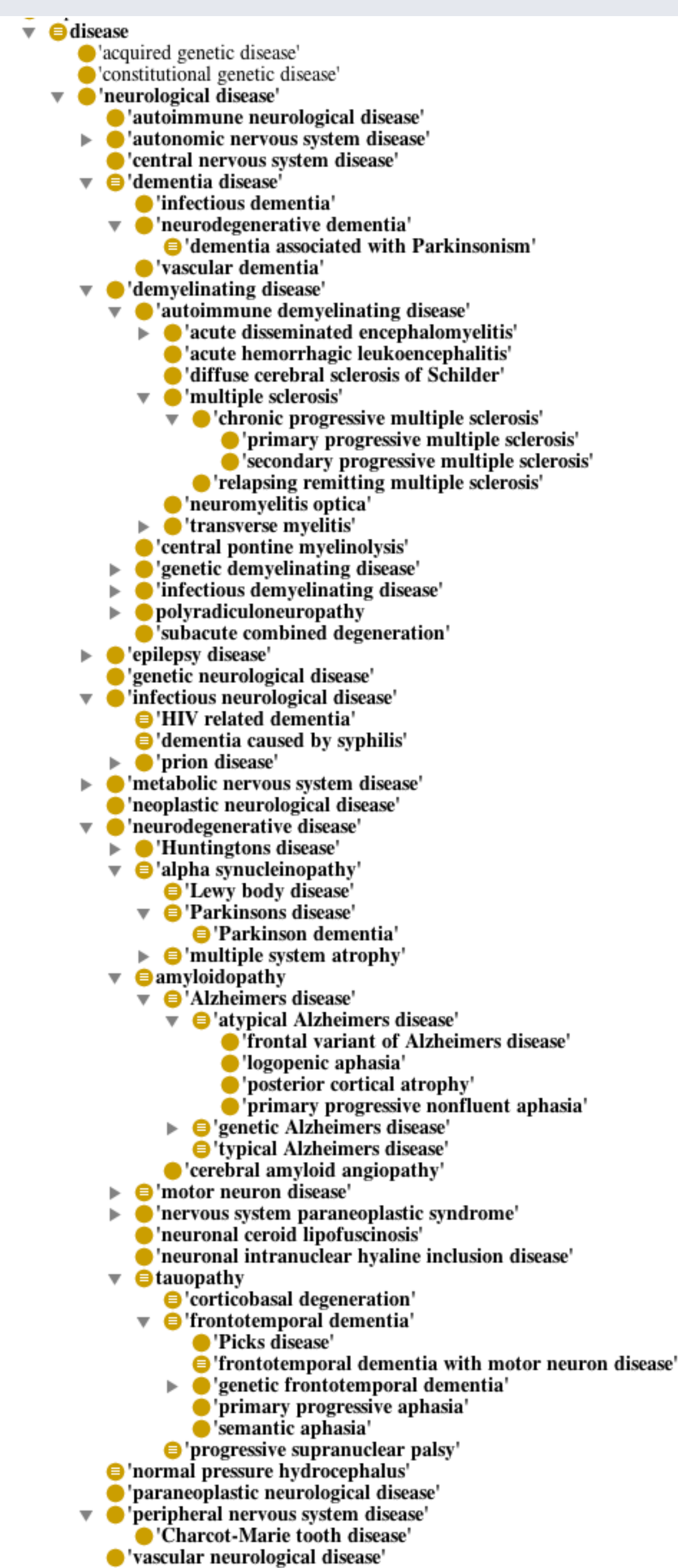
Introduction

We have begun work on the Neurological Disease Ontology (ND) to provide a set of controlled, logically connected terms to describe the range of neurological diseases and their associated signs and symptoms, assessments, diagnoses, and interventions encountered in the course of clinical practice. ND is built as an extension of the Ontology for General Medical Sciences (OGMS) – a high-level candidate OBO Foundry ontology. As seen in the figure above, OGMS provides a set of general classes that can be used to describe general aspects of a disease and its diagnosis, including documenting the role of the physician and other medical professionals in diagnosing and treating patients. ND is being built with carefully axiomatized defined classes that connect and relate to other classes within the ontology itself as well as to classes in reference ontologies such as the Gene Ontology, Cell Ontology, Protein Ontology, Chemical Entities of Biological Interest, and Foundational Model of Anatomy, among others.

Goals

- To provide a comprehensive representation of neurological diseases to support clinicians and researchers in the diagnosis, treatment, and study of these diseases.

- To facilitate querying of medical databases for such purposes as performing quality analysis checks on diagnostic criteria at various stages of a disease's progression.

- To allow physicians and researchers to provide a comprehensive clinical picture of a patient using a standardized language, and to connect and leverage structured descriptions in clinical and translational medicine, in EHRs and published research.

- To develop best practices for the development of other clinically oriented ontologies by identifying a robust set of relations for use with diseases and by providing an applied template for representing temporal entities within a domain.

Referenced Ontologies

The development of ND involves the use of many other ontologies. These include:

Ontology Name:	Use in ND:
Basic Formal Ontology (BFO)	Top-level parent ontology
Ontology for General Medical Science (OGMS)	Mid-level parent ontology
Neuroscience Information Framework (NIF)-Dysfunction	Externally referenced many disease terms
Relations Ontology (RO)	Imported core relations
Protein Ontology (PR)	MIREOTed protein terms
Foundational Model of Anatomy (FMA)	MIREOTed anatomical terms
PATO, ChEBI, GO, CL, and OBI	MIREOTed select terms

Methods

ND is built according to OBO Foundry principles and utilizing Protégé 4.1 and OWL2. It is currently being updated to be compliant with BFO2.0.

Basic Approach:
We identified neurological diseases and outlined the representation of their respective diagnoses. In order to fill in the connections between a disease and its diagnosis, it was necessary to identify the signs and symptoms that characterize each associated syndrome, the material cause (the disorder) and its anatomical location, the processes involved in the development and progression of each disease, and more.

Current Progress

The initial work on ND has focused on Alzheimer's Disease (AD) and other diseases resulting in dementia. Ontology modules to represent muscular sclerosis (MS), and stroke are also under development at the University at Buffalo.

Statistics:
◇ ND currently contains 335 classes
◇ 199 classes have textual definitions
◇ 52 classes have logical definitions
◇ 157 classes have external references
◇ There are 190 children of disease

In addition to disease classes, ND has a heavy focus on diagnosis, syndrome, disorder, and protein classes among others.

ND Spin-Off Project – NPT
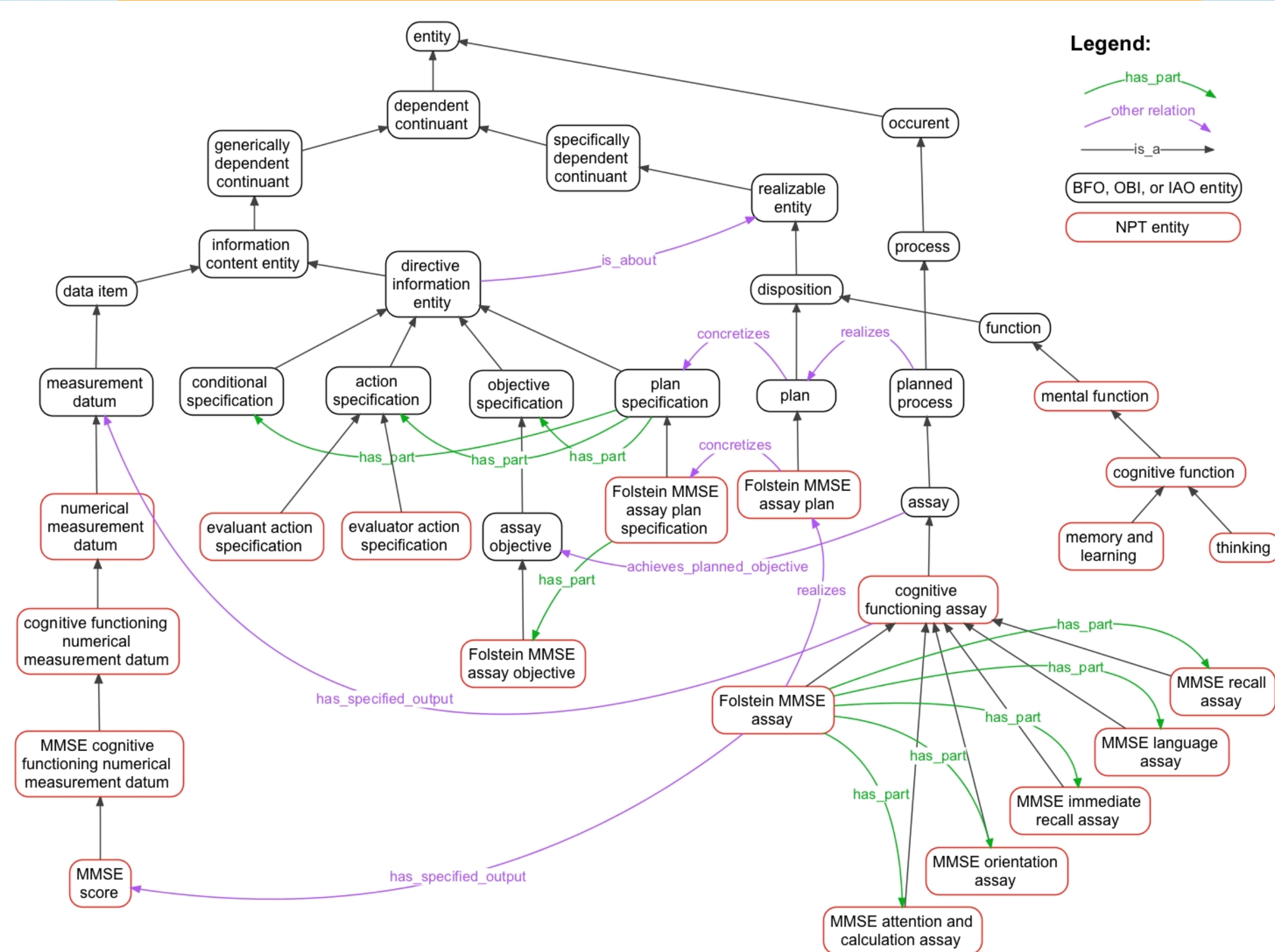
Representation of the Folstein Mini-Mental Examination (MMSE) in NPT.

The Neuropsychological Testing ontology (NPT) represents neuropsychological assessments such as the Folstein Mini-Mental State Examination (MMSE), the Trail Making Test, the Hopkins Verbal Learning Test, and the Wechsler Memory Scale. These standardized assessments are useful for identifying the presence and degree of cognitive impairment in patients. A goal of the NPT project is to test hypotheses about the diagnosis of AD based on the results of neuropsychological assessments.

For More Information:

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We will make a public release of ND by September 2012, at http://code.google.com/p/neurological-disease-ontology/

Background Literature:
- "Toward an ontological treatment of disease and diagnosis," Scheuermann RH, Ceusters W, Smith B., Summit on Translational Bioinformatics. 2009 Mar 1;2009:116-20.0
- "Foundations for a Realist Ontology of Mental Disease" Ceusters and Smith *Journal of Biomedical Semantics* 2010, 1:10