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New York State Center of Excellence in Bioinformatics & Life Sciences

MSPD: An ontological representation and analysis of patient-reported and clinical outcomes for multiple sclerosis

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Introduction

We have developed the Multiple Sclerosis Patient Data Ontology (MSPD) to represent clinical measures and patient reported outcomes obtained from enrollment forms used by centers participating in the New York State Multiple Sclerosis Consortium (NYSMSC). In order to better understand the relationship between treatments for multiple sclerosis and both patient and physician reported outcomes, it is important to see how practitioners' assessments track with patients' perception of their well-being. In this poster, we detail our representational structure and our use of both logical and textual definitions for classes. Of particular interest to our project is understanding the relationship between current treatments, clinical outcomes, and patients' perceptions of their own outcomes, known as patient reported outcomes (PRO), to see how practitioners' assessed clinical efficacy and PRO track with each other.

Goals

- Annotate a subset of data from the NYSMSC Registry using MSPD
- Create an inferred hierarchy between classes through reasoning
- Develop unique queries of the data looking for term enrichment
- Compare patient reported outcomes to clinical outcomes
- Track quality of life assessments with clinical measures

Multiple Sclerosis

Multiple sclerosis (MS) is an autoimmune demyelinating disease of the central nervous system affecting over 2 million people worldwide. MS presents clinically through varied neurological symptoms such as loss of motor control and balance, visual and cognitive impairment, and sensory disturbances. Both genetic and environmental factors play a role in both the underlying etiology and in influencing the heterogeneous MS disease course as it presents in individual patients. 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). Disability in MS is assessed using the Kurtzke Expanded Disability Status Scale (EDSS). In recent years a variety of new treatments have improved outcomes for many MS patients, yet the disease is considered incurable and progressive in its course.

New York State Multiple Sclerosis Consortium

The NYSMSC is an alliance of treatment centers organized to prospectively assess clinical attributes of MS patients and allow for population-based research collaboration. The NYSMSC database includes more than 15 MS centers across New York State and is the largest clinical-based cohort of MS patients in the United States with over 10,000 registrants and 17,200 plus follow-up visits. NYSMSC uses the LIFEware system as part of its data collection activities to record patients' perceptions of their physical and psychosocial impairment as a way of representing their quality of life and well-being. During the enrollment process, in addition to certain demographic information, patients are asked to rate their perception of their own functional abilities and affective states.

Example of Forms Used by NYSMSC

Clinician Reported Form

33. Functional Scores and EDSS

Pyramidal	Cerebellar	Brain Stem	Sensory	Bowel & Bladder	Visual	Cerebral	Other functions	Kurtzke EDSS at registration
○	○	○	○	○	○	○	○	○ 0.0 ○ 5.5
○	○	○	○	○	○	○		○ 1.0 ○ 6.0
○	○	○	○	○	○	○		○ 1.5 ○ 6.5
○	○	○	○	○	○	○	○	○ 2.0 ○ 7.0
○	○	○	○	○	○	○		○ 2.5 ○ 7.5
○	○	○	○	○	○	○		○ 3.0 ○ 8.0
○	○	○	○	○	○	○		○ 3.5 ○ 8.5
○	○	○	○	○	○	○		○ 4.0 ○ 9.0
	○				○		○	○ 4.5 ○ 9.5
○	○	○	○	○	○	○		○ 5.0

34. Timed Ambulation for 25 feet in seconds

0 ○○○ ○
1 ○○○ ○
2 ○○○ ○
3 ○○○ ○
4 ○○○ ○
5 ○○○ ○
6 ○○○ ○
7 ○○○ ○
8 ○○○ ○

Level of Assistance
○ Unassisted ○ Assist of one ○ Assist of two ○ Unable

Is the patient wearing an AFO?
○ Yes ○ No

Patient Reported Form

19. Are you having any pain? (fill in only one): ○ No ○ Yes

If YES, what was the extent of your pain during the past 3 days including today (fill in only one)?
○ Mild pain ○ Discomforting pain ○ Distressing pain ○ Horrible pain ○ Excruciating pain

20. How satisifed are you with life in general (fill in only one)?
○ Very well satisfied ○ Fairly well satisifed ○ More satisfied than not satisfied ○ Not satisfied

21. How much are you limited in each of the following areas:

	No limitation / None to mild limitation / Mild limitation / Mild to moderate limitation / Moderate limitation / Moderate to severe limitation / Severe limitation
Right upper limb	○ ○ ○ ○ ○ ○ ○
Left upper limb	○ ○ ○ ○ ○ ○ ○
Right lower limb	○ ○ ○ ○ ○ ○ ○
Left lower limb	○ ○ ○ ○ ○ ○ ○
Bowel continence	○ ○ ○ ○ ○ ○ ○
Bladder continence	○ ○ ○ ○ ○ ○ ○
Fatiguability	○ ○ ○ ○ ○ ○ ○
Vision	○ ○ ○ ○ ○ ○ ○

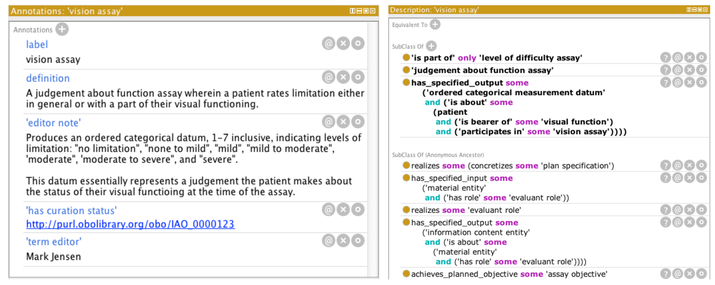
22. For each of the following feelings or moods, please fill in one response indicating how much you have been bothered or worried during the last 7 days:

Are you feeling...	Not bothered or worried / Mildly / Moderately / Quite a bit / Extremely
Lonesome or isolated	○ ○ ○ ○ ○
Pessimistic about future	○ ○ ○ ○ ○
Uptight, tense or stressed	○ ○ ○ ○ ○
Panic attacks	○ ○ ○ ○ ○
Are you feeling...	Not bothered or worried / Mildly / Moderately / Quite a bit / Extremely
Easily irritated or annoyed	○ ○ ○ ○ ○
Morbid or gloomy thoughts	○ ○ ○ ○ ○
Blaming yourself or guilt	○ ○ ○ ○ ○

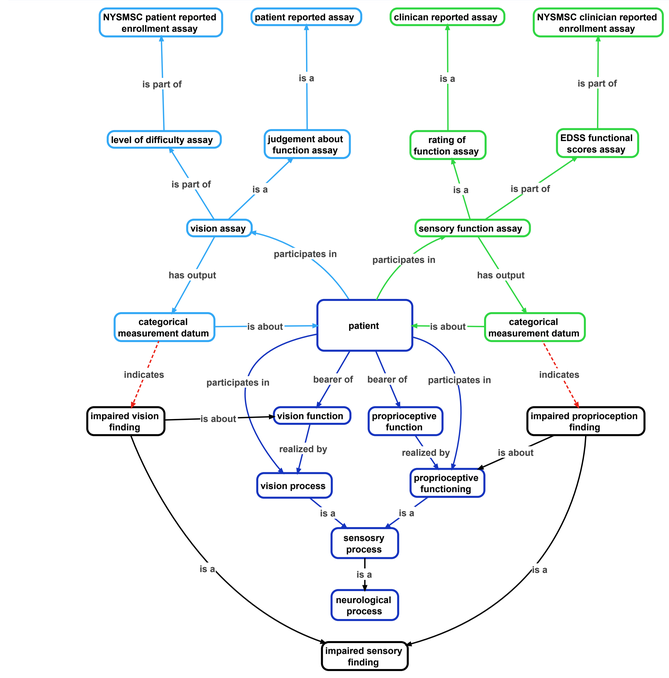
MSPD 'assay' Hierarchy

- assay
  - 'clinician reported assay'
    - '9-hole peg test assay'
    - 'currently exacerbating assay'
    - 'disease status assay'
    - 'dominate hand assay'
    - 'functional scores and EDSS assay'
    - 'level of assistance in timed ambulation assay'
    - 'MS diagnosis according to McDonald criteria assay'
    - 'MS diagnosis according to Poser criteria assay'
    - 'MS type assay'
    - 'number of relapses previous year assay'
    - 'NYSMSC clinician reported enrollment assay'
    - 'patient using an ankle foot orthotic assay'
    - 'rating of function assay'
      - 'bowel and bladder function assay'
      - 'brain stem function assay'
      - 'cerebellar function assay'
      - 'cerebral function assay'
      - 'Kurtzke EDSS at registration assay'
      - 'other function assay'
      - 'pyramidal function assay'
      - 'sensory function assay'
      - 'visual function assay'
    - 'time for left hand in 9-hole peg test assay'
    - 'time for right hand in 9-hole peg test assay'
    - 'time to walk 25 feet assay'
    - 'timed ambulation assay'
  - 'NYSMSC enrollment form assay'
  - 'patient reported assay'
    - 'fact-based assay'
    - 'NYSMSC patient reported enrollment assay'
    - 'self-appraisal assay'
      - 'fatigability assay'
      - 'feelings and moods assay'
      - 'judgement about affect assay'
        - 'blame guilt assay'
        - 'experiencing pain assay'
        - 'irritated annoyed assay'
        - 'life satisfaction assay'
        - 'lonesome isolated assay'
        - 'morbid gloomy thoughts assay'
        - 'pain rating assay'
        - 'panic attack assay'
        - 'pessimistic about future assay'
        - 'uptight tense stressed assay'
      - 'judgement about function assay'
        - 'bladder continence assay'
        - 'bowel continence assay'
        - 'difficulty of climbing stairs assay'
        - 'difficulty of driving assay'
        - 'difficulty of getting up assay'
        - 'difficulty of prolonged standing assay'
        - 'left lower limb assay'
        - 'left upper limb assay'
        - 'right lower limb assay'
        - 'right upper limb assay'
        - 'vision assay'
  - 'level of difficulty assay'
  - 'limitations assay'
  - 'pain judgement assay'

MSPD Class: 'vision assay'



Relations Between Classes



Methods and Current Progress

MSPD is an OWL ontology built using Protégé 4.3. It is compliant with OBO Foundry principles and based on the methodological framework of the Ontology for Biomedical Investigations (OBI). In an effort to be interoperable and collaborative, MSPD utilizes classes from other ontologies, such as the Ontology for General Medical Science, Neuroscience Information Framework Standard Ontology, Gene Ontology, Emotion Ontology, and Human Phenotype Ontology. We share our design standards with our sibling ontology, the Neuropsychological Testing Ontology, and use a template for creating textual definitions adopted from BFO. MSPD currently has over 100 unique classes and is developed in conjunction with the Neurological Disease Ontology.

Results

- Developed a hierarchy of classes representing assays, neurological functions and processes, clinical findings and measurement data that distinguishes between patient's perception of their functional abilities vs. a clinician's assessment of particular realizations of patient functions.
- Created logical definitions that incorporate classes from external ontologies to formally define classes in MSPD.
- Performed a preliminary analysis of a subset of NYSMSC data to aid in annotations of clinical finings.

Acknowledgments

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Future Work

- Build an RDF triple-store annotating NYSMSC data sets with classes from the ontology.
- Employ reasoners to check the ontology for consistency to ensure robust queries.
- Begin clinician supported analysis of data utilizing term enrichment to spot patterns in data between patient perceived outcomes and clinical measures.
- Import classes from ND to establish relationships between disease classes and patient and clinician reported data.

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