Reviewer’s report

Title: An ontology-aware integration of clinical models, terminologies and guidelines: an exploratory study of the Scale for the Assessment and Rating of Ataxia (SARA)

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Reviewer: Soon Ae Chun

Reviewer's report:

The paper addresses the computational modeling of rating scales that are used to determine the clinical phenotype observations, and to support the evaluation of clinical phenotype. The scaling system SARA (The Scale of Assessment and Rating of Ataxia) is used to illustrate the implementation on clinical openEHR for ataxia neurological diagnosis. The observation categories/terms are associated with the SARA rating scores, and the evaluation labels/terms are derived through the observation categories. The knowledge that is used in clinical observations and evaluation is modeled with rules in GDL (Guidelines Definition Language) format. The prototype system is either implemented or simulated to illustrate the SARA scoring system and the knowledge to derive the ataxia related severities, and clinical category decisions.

The proposed approach uses the Human Phenotype Ontology (HPO) to annotate the scale related clinical statements. This annotated SARA scales created the Ataxia HPO classes. The scores of scale is mapped to the clinical observations and evaluations through Guidelines (rules). The scores are mapped via GDL rules into one clinical label/term/class of the ataxia observations and evaluations.

The paper is interesting and it will be a useful tool SARA and knowledge system to help the rate the scores and evaluate the symptoms into the right categories. Using Ontology will give a uniform vocabulary (category) across different levels of statements in observations and evaluations.

I recommend the following to be addressed:

- Perhaps a scenario/use case description on how the rating scores are used by clinicians to arrive at the evaluation category can be helpful. This may highlight the problem or challenge that SARA system may support. (Somehow the process of deriving the evaluative category
from a rating score is not there in the intro, so it is difficult to appreciate what problem it is trying to address in the beginning.)

In other words, the motivating scenario description of how it is done will help illustrating the usefulness of SARA/CKM approach.

- Section 2.1: The extraction from textual sources to identify the scale related items using HPO, but please provide some example to understand this part better.

- The binding/annotation with HPO in Fig 3 is also not clear. The figure is unreadable. It is through GDL rules/calculations?

- There are many tools used along the processing, but it looks like there are no integration. So it is unclear how much of prototype system actually is implemented or not. The reasoning part seems to be done in application level rather than using the ontology-based reasoning.

This casts doubts if the system is actually implemented or it is a desired system you plan to implement.

Clarification is needed.

- Please also address if this approach can be generalizable to other domains. What may be an issue?

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

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I am able to assess the statistics

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