SNOMED CT Expo 2015
Presentation or Poster Abstract

Extending the coverage of phenotypes in SNOMED CT through post-coordination

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Audience

Developers and users of SNOMED CT interested in the representation of phenotypes.

Objectives

1. To learn about the coverage of phenotypes in SNOMED CT (pre-coordinated concepts);
2. To understand how logical definitions can serve as templates for new post-coordinated concepts;
3. To learn how such templates can be automatically populated for phenotypes not currently represented in SNOMED CT.

Abstract

In previous work, we demonstrated that only 30% of the 10,000 phenotypes from the Human Phenotype Ontology (HPO) were represented in SNOMED CT as pre-coordinated concepts, which is insufficient for supporting the deep phenotyping required for detailed genetic studies.

Our objective is to extend the coverage of phenotypes in SNOMED CT through post-coordination. The specific contribution of this work is to identify templates in SNOMED CT, based on the logical definitions of existing concepts, for the creation of post-coordinated expressions for phenotype concepts from HPO.

For example, based on the logical definition of “Congenital hypoplasia of kidney”, we can create a post-coordinated expression for “Congenital hypoplasia of macula” by substituting “Kidney structure” with “Macula lutea structure”. This post-coordinated expression provides a mapping for the HPO concept “Macular hypoplasia” (interpreted as being congenital in the context of HPO). Moreover, the post-coordinated expression created for “Congenital hypoplasia of macula” can be generalized to a template for the congenital hypoplasia of any anatomical structure.

We created 12 templates (e.g., "abnormality of <ANATOMICAL_STRUCTURE>") from which we were able to derive post-coordinated expressions for 1,617 HPO concepts for which there was no equivalent pre-coordinated concept in SNOMED CT. Through this novel approach, we were able to increase the current number of mappings between HPO and SNOMED CT from 3,081 to 4,698 (+52%). Post-coordination is an efficient way of extending the coverage of phenotypes in SNOMED CT.

References

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