Medical Action Ontology (MAxO)

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Abstract

A standardized, controlled vocabulary allows medical actions to be described in an unambiguous fashion in medical publications and databases. The Medical Action Ontology (MAxO) is being developed to provide a structured vocabulary for medical procedures, interventions, therapies, and treatments for rare diseases. The ontology will capture medical actions used to treat rare diseases from published manuscripts and will be utilized in new computational algorithms that exploit the semantic similarity between related medical actions, phenotypic abnormalities as coded in the Human Phenotype Ontology (HPO), and related genetic mutations using Exomiser to suggest new medical actions for rare diseases. Annotations to publications about rare disease clinical management will be added to the HPO resource. This ontology coupled with computer algorithms does not intend to replace physician's advice, guidelines of medical societies, or textbooks, but rather to complement these sources of information. It provides a convenient method for quickly extracting difficult to find information about treatments for rare diseases. Currently, MAxO contains approximately 300 terms and will be expanded via more data mining from sources such as GeneReviews, PubMed queries, and Rare Disease Network. MAxO is open source and will become freely available. A preliminary version is available at https://github.com/monarch-initiative/MAxO.

Keywords:

Ontology development, medical actions, clinical treatments, Human Phenotype Ontology