

Semantic Characterization of Hypertrophic Cardiomyopathy Disease

Abstract:

The application of a translational medicine approach to the study of diseases enables personalized clinical diagnosis and prognosis.

Hypertrophic cardiomyopathy (HCM) is a disease that can benefit from such an approach, since it combines a variable clinical presentation with a genetic heterogeneity denoted by 640 known mutations, in more than 20 genes. This is a relatively common genetic myocardial disorder and the most frequent cause of sudden cardiac death in young people and athletes.

This article presents a novel semantic model representing the integration of phenotype and genotype data, mandatory for the characterization of HCM.

The model, developed in OWL Lite, comprises three connected modules: *HCM Clinical Evaluation*, *Genotype Analysis* and *Medical Classifications*. The RDF/XML representation of each module is available at

<https://sites.google.com/site/hcmsemanticmodel/home-1>.

The lexicon of the model was based on controlled vocabularies, namely SNOMED CT, NCI Thesaurus and OCRE, with a total of 78% linked concepts.

The model will provide the basic framework for a biomedical system that will improve the diagnosis and prognosis of HCM. This improvement will be accomplished through the utilization of data mining techniques that will identify associations between the presence of certain mutations and the resulting physical traits.

Keywords: Translational Medicine; Semantic Web; Domain knowledge representation; Semantic Modeling; Hypertrophic Cardiomyopathy