The Ocular Disease Ontology

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1 INTRODUCTION

The Ocular Disease Ontology (ODO) is an ontology designed to represent ocular diseases for both clinical and research purposes. This constitutes an effort to build an ontology that represents, to the fullest possible extent, ocular diseases. ODO makes use of the following: Cell Ontology (CL), Ontology for General Medical Science (OGMS), Neuronal CL, Gene Ontology (GO), Uberanatomy Ontology (UBERON), and the Neurological Disease Ontology (ND). Much of the work on ODO focuses efforts on integrating recent work representing retinal cell types from CL and diseases from OGMS. Ocular diseases are generally classified in medical literature depending on the anatomical regions in which they originate (or, more accurately, where the disease has a material basis). The impetus behind the work in ODO rests on the hypothesis that ocular diseases are limited in physical location and physiological effect. Given the somewhat simple anatomical structures that comprise the eye and immediate surrounding area, it is possible to define most ocular diseases using current resources. As there have been no prior attempts to represent ocular disease comprehensively in a formal ontology, we believe that ODO represents a useful and straightforward extension of the OGMS based on these considerations.

Long-term goals: Integration with ND, OGMS, and other existing ontologies under the BFO umbrella, complete interoperability, i.e., the ability of terms generated in ODO to function within other BFO ontologies, and coverage of all identified ocular diseases.

The work so far: Under the umbrella class 'ocular disease' there are eight subclasses that correspond to eight major anatomical regions of the eye (lacrimal apparatus, cornea, lens, pupil, retina, sclera, uvea, vitreous) as well as two subclasses for diseases that escape characterization based on anatomical region (ocular albinism, optic neuropathy). There are currently over 100 terms in ODO (not including synonyms and relations).

The motivation behind such methodology is to allow for classification of ocular diseases whose particular physiological mechanisms are as of yet unknown. Employment of current methods allows for classification of ocular diseases where current research has yet to provide a detailed description of disease course but general locational information is available. In this way, ODO terms can be used in the same way that researchers and practitioners currently use them, not merely in the ideal sense where one knows the particular disease course of an ocular disease. In addition, ODO leverages existing work in other ontologies such that ocular diseases are treated as one among many different diseases of the body. This guarantees that ODO will grow and change inasmuch and insofar as the ontologies related to ocular structures and diseases change in general. This interoperability is essential for functioning ontologies.

Ideally, one would appeal merely to the anatomical region where the material basis of disease in located as much as possible for ease of representation. However, this approach will not work to differentiate all of the ocular diseases. For this reason, multiple differentia are employed to capture ocular diseases. For example, the two subclasses of 'ocular albinism' and 'optic neuropathy' are subclasses of 'ocular disease' that do not have a specific and predictable associated anatomical region.

Future work: Continued development of ODO in conjunction with existing ontologies is slated for the near future. Ultimately, ODO will capture all known existing ocular diseases and remain flexible enough to accurately capture future ocular diseases. One immediate extension is classification of ocular diseases related to neurological diseases.

Examination of specific use cases has shown promising results. For example, the class of ocular diseases known as retinal degenerative diseases (retinal degeneration) has provided valuable insight into the structuring of ODO.

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