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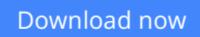
Peter N. Robinson, Sebastian Bauer

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Introduction to Bio-Ontologies explores the computational background of ontologies. Emphasizing computational and algorithmic issues surrounding bio-ontologies, this self-contained text helps readers understand ontological algorithms and their applications.

The first part of the book defines ontology and bio-ontologies. It also explains the importance of mathematical logic for understanding concepts of inference in bio-ontologies, discusses the probability and statistics topics necessary for understanding ontology algorithms, and describes ontology languages, including OBO (the preeminent language for bio-ontologies), RDF, RDFS, and OWL.

The second part covers significant bio-ontologies and their applications. The book presents the Gene Ontology; upper-level ontologies, such as the Basic Formal Ontology and the Relation Ontology; and current bio-ontologies, including several anatomy ontologies, Chemical Entities of Biological Interest, Sequence Ontology, Mammalian Phenotype Ontology, and Human Phenotype Ontology.

The third part of the text introduces the major graph-based algorithms for bio-ontologies. The authors discuss how these algorithms are used in overrepresentation analysis, model-based procedures, semantic similarity analysis, and Bayesian networks for molecular biology and biomedical applications.

With a focus on computational reasoning topics, the final part describes the ontology languages of the Semantic Web and their applications for inference. It covers the formal semantics of RDF and RDFS, OWL inference rules, a key inference algorithm, the SPARQL query language, and the state of the art for querying OWL ontologies.

Web Resource

Software and data designed to complement material in the text are available on the book's website: http://bio-ontologies-book.org The site provides the R Robo package developed for the book, along with a compressed archive of data and ontology files used in some of the exercises. It also offers teaching/presentation slides and links to other relevant websites.

This book provides readers with the foundation to use ontologies as a starting point for new bioinformatics research projects or to support current molecular genetics research projects. By supplying a self-contained introduction to OBO ontologies and the Semantic Web, it bridges the gap between both fields and helps readers see what each can contribute to the analysis and understanding of biomedical data.

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