#### **ABSTRACT**

It is increasingly challenging to analyze the data produced in biomedicine, even more so when relying on manual analysis methods. My hypothesis is that using a common representation of knowledge, implemented via standard tools, and logically formalized can make those datasets computationally amenable, help with data integration from multiple sources and allow answering complex queries.

The first part of this dissertation demonstrates that ontologies can be used as common knowledge models, and details several use cases where they have been applied to existing information in the domain of biomedical investigations, clinical data and vaccine representation.

In a second part, I address current issues in developing and implementing ontologies, and proposes solutions to make ontologies and the datasets they are applied to available on the Semantic Web, increasing their visibility and reuse.

The last part of my thesis then builds upon the first two, and applies their results to pharmacovigilance, and specifically to analysis of reports of adverse events following immunization. I encoded existing standard clinical guidelines from the Brighton Collaboration in Web Ontology Language (OWL) in the Adverse Events Reporting Ontology (AERO) I developed within the framework of the Open Biological and Biomedical Ontologies Foundry. I show that it is possible to automate the classification of adverse events using the AERO with very high specificity (97%). I also demonstrate that AERO can be used with other types of guidelines. Finally, my pipeline relies on open and widely used data standards (Resource Description Framework (RDF), OWL, SPARQL) for implementation, making the system easily transposable to other domains.

This thesis validates the usefulness of ontologies as semantic models in biomedicine enabling automated, computational processing of large datasets. It also fulfills the goal of raising awareness of semantic technologies in the clinical community of users. Following my results the Brighton Collaboration is moving towards providing a logical representation of their guidelines.

#### **BIOGRAPHICAL NOTES**

Place of Birth: Saint Louis, France

Academic Studies: B. Sc. Biochemistry, Université Louis Pasteur, 2000

M. Sc. Computer Science, Université Louis Pasteur, 2002

Current Position: Post-doctoral fellow, MBB Department, SFU

**GRADUATE STUDIES** 

Field of Study: Bioinformatics

#### **SELECTED AWARDS**

[2012] Research Grant: Co-investigator for the PHAC/CIHR Influenza Research Network

[2012] PHAC/CIHR Influenza Research Network scholarship (2nd award)

[2011] PHAC/CIHR Influenza Research Network scholarship

[2010] Michael Smith Foundation for Health Research Trainee award

[2009] Research Grant: Co-investigator for the PHAC/CIHR Influenza Research Network

#### **SELECTED PUBLICATIONS**

**Courtot M.,** Brinkman RR and Ruttenberg A. "The logic of surveillance guidelines: An analysis of vaccine adverse event reports from an ontological perspective" PLoS ONE **Courtot M.,** Juty N., Knupfer C., Waltemath D., Zhukova A., Drger A., Dumontier M., Finney A., Golebiewski M., Hastings J., Hoops S., Keating S, Kell DB., Kerrien S., Lawson J., Lister A., Lu J., Machne R., Mendes P., Pocock M., Rodriguez N., Villeger A., Wilkinson DJ., Wimalaratne S., Laibe C., Hucka M., Le Novere N. "Controlled vocabularies and semantics in Systems Biology" Molecular Systems Biology 7(1) (2011)

**Courtot M.,** Gibson F., Lister AL., Malone J., Schober D., Brinkman RR and Ruttenberg A. "MIREOT: the Minimum Information to Reference an External Ontology Term" Applied Ontology. 6 (1): 23-33 (2011)

## **SELECTED PRESENTATIONS**

[2014] Effective automated classification using ontology-based annotation: experience with analysis of adverse event reports, Seventh International Biocuration Conference (ISB2014), April 2014, Toronto, Canada

[2013] OBO Foundry 101, Tutorial faculty, July 7th 2013, Montreal, Canada

[2013] Diagnostic criteria and clinical guidelines standardization to automate case classification, International Conference on Biomedical Ontologies (ICBO) 2013, July 2013, Montreal, Canada

**[2012]** Adverse events following immunization: standardization, automatic case classification and signal detection, Conference on Semantics in Healthcare and Life Sciences (CSHALS) 2012, February 24th 2012, Boston, USA

## SUPERVISORY COMMITTEE

Dr. Ryan R. Brinkman (Research supervisor)

Dr. Raymond Ng (since Dec 2011)

Dr. Paul Pavlidis

Dr. Margaret-Anne Storey

Dr. Mark Wilkinson (Sep 2009 - Dec 2011)



# **Graduate and Postdoctoral Studies**

### **PROGRAMME**

The Final Oral Examination For the Degree of

DOCTOR OF PHILOSOPHY (Bioinformatics)

## **MELANIE COURTOT**

B. Sc. Biochemistry, Université Louis Pasteur, 2000 M. Sc. Computer Science, Université Louis Pasteur, 2002

Monday, May 12, 2014, 12:30pm Room 203, Graduate Student Centre Latecomers will not be admitted

"Semantic Models in Biomedicine: Building Interoperating Ontologies for Biomedical Data Representation and Processing in Pharmacovigilance"

## **EXAMINING COMMITTEE**

#### Chair:

Dr. Haydn Pritchard (Pathology and Laboratory Medicine)

## Supervisory Committee:

Dr. Ryan R. Brinkman, Research Supervisor (Bioinformatics)

Dr. Paul Pavlidis (Bioinformatics)

## University Examiners:

Dr. Julie Bettinger (Population and Public Health)

Dr. Rachel Pottinger (Computer Science)

#### External Examiner:

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