Course Title: Biomedical Ontology
Version Jun 22, 2018
Partially cross-listed with PHI598

	Department of Biomedical Informatics Jacobs School of Medicine and Biomedical Sciences	Department of Philosophy School of Arts and Sciences
Course Subject Code:	BMI	PHI
Course Number:	508	598
Type of Instruction:	SEM	SEM
Class Number:	24643	24696
Semester:	Fall 2018	Fall 2018

1. *Course Information*

- Date(s)/Time(s): Aug 27 – Oct 15 and Dec 3, 1-4 PM: 200G Baldy, North Campus
  Oct 22 – Dec 10, except Dec 3, 4-7 PM: Room 506, 77 Goodell street, 5th floor (BMI Dept)
- Delivery Mode: Traditional
- Number of Credits: 3

- Instructors
  Course directors: Biomedical Informatics: Werner Ceusters, MD (contact: 77 Goodell Street, 5th floor, by appointment only through wceusters@gmail.com)
  Philosophy: Barry Smith, PhD (contact: 126 Park Hall, N Campus, by appointment only through phismith@buffalo.edu)
  Lecturers: Werner Ceusters (WC), Barry Smith (BS), Brian Donohue (BD)

2. *Course Description*

- This course provides an introduction to biomedical ontology. It will review how data and information are generated through biological and biomedical experiments and through patient care, and show how ontologies are used in accessing, maintaining and exploiting the results. We will describe how biomedical ontologies are developed and evaluated and provide a comparative critical analysis of the principal current ontology resources. We will also review the major theories, methods and tools for the development of ontologies, and illustrate how these are being used in different areas of biomedical research and healthcare. The methods and tools for applied ontology as well as the management and maintenance of biomedical ontologies will be discussed in detail including the principles of ontological realism and the implementation thereof in the Basic Formal Ontology (BFO). Students will gain some experience with the Web Ontology Language (OWL) and Protégé.

- Course prerequisites:
  For BMI students: BMI 501
  For other students: Any 400-level course in either philosophy or computer science
  Prerequisites can be waived under special circumstances to be discussed with the course directors.

3. *Student Learning Outcomes (SLO)*

Course Learning Outcomes; students will be able to:	BMI PhD Program Outcomes / Competencies	Instructional Method(s)	Assessment Method(s)
1. Understand and apply the principals of ontological realism	O1: Ontological Realism, the Basic Formal Ontology (BFO), and the Ontology of General Medical Science	• Lecture • Discussion	• Final Exam
2. Understand and apply the Basic Formal Ontology in ontology design	O1: Ontological Realism, the Basic Formal Ontology (BFO), and the Ontology of General Medical Science	• Lecture • Discussion	• Final Exam
3. Understand and apply the Ontology of General Medical Science in biomedical ontology design	O1: Ontological Realism, the Basic Formal Ontology (BFO), and the Ontology of General Medical Science	• Lecture • Discussion	• Assignment W3 • Final exam

4. Understand the strengths and weaknesses of prevailing biomedical data, information and knowledge management paradigms	O2:methods of data representation, manipulation, storage, analysis and mining in healthcare and biomedical research databases O3: Information retrieval and critical analysis skills	• Lectures • In-class exercises	• Final exam • Assignments (W1, W5)
5. Identify opportunities, risks and challenges to current biomedical data, information and knowledge management paradigms	O2: Methods of data representation, manipulation, storage, analysis and mining in healthcare and biomedical research databases	• Lecture	• Final exam
6. Assess the quality of existing clinical research data repositories using ontological principles	O3: Information retrieval and critical analysis skills O4: Technical approaches to acquiring, modeling, representing and managing healthcare and biomedical research knowledge O5: The evaluation of biomedical ontologies and the published biomedical ontology research literature	• Lecture	• Final exam • Student presentation
7. Develop ontology-based solutions for quality improvement of biomedical and clinical research data	O4: Technical approaches to acquiring, modeling, representing and managing healthcare and biomedical research knowledge O6: Advanced methods and tools for managing biomedical ontologies (including the Web Ontology Language, OWL) O7: Use of ontology editors and add-on tools (e.g., Protégé) to build a realism-based biomedical ontology	• Lecture	• Final exam • Assignment W11 • Student presentation
8. Understand and apply the principles for life cycle management for biomedical ontologies	O8: The principles for change management and upgrades to biomedical ontologies	• Lecture	• Assignment W12 • Final exam

4. Course Requirements

- Students are required to read 3 book chapters and 10 papers and electronic publications as listed in the course materials below.
- All assignments need to be completed prior to the deadline specified in the course schedule and send by email to wceusters@gmail.com.

5. Grading Policy

- Grading follows standard graduate policies (http://grad.buffalo.edu/Academics/Policies-Procedures/Grading-Procedures.html.
- Learning assessments will be graded based on rubric criteria and weighted according to the following detailed break-down. If the final results for all students are outside the expected range, curve grading might be used at the discretion of the course director.
- Assignments submitted after the deadline will be subject to a 1% penalty for the total final score. Assignments which are more than one week late will not be graded and will be subject to a penalty equal to the score weight unless the student requested during that week an extension which was approved by the course director.
- Some classes will start with a pre-test based on the required readings prior to the lecture, this to assess the student's preparedness for the class. Students absent for these classes will receive a negative score of 2% for these tests unless the instructor and the course director have been informed through email about the reason for absence prior to the beginning of the class. When notified of absence in due time, the instructor will propose an alternative test or assignment for that class. The maximum negative score for non-participation in pre-class tests is 10%.

Week	2018	Assignment post-lecture	Due Date	Score weight
W1	Aug 27	Write a 2-page essay summarizing the key elements of an ontology as identified in the lecture and comparing them to the view of an ontology offered in (Hoehndorf, Schofield & Gkoutos, 2015). Evaluation criteria: % of issues covered, soundness of argumentation.	Sept 6, noon	**5%**
W3	Sep 10	Summarize in a 2 page essay the issues discussed in pages 16-21 of (Haendel et. al., 2018) and describe how the framework offered by (Scheuermann, Ceusters & Smith 2009) might resolve them. Evaluation criteria: % of issues covered, soundness of argumentation.	Sept 24, noon	**5%**
W5	Sep 24	Summarize in a 2 page essay the problems discussed in (Smith & Koppel, 2014) and describe how Basic Formal Ontology can assist in dealing solving them. Evaluation criteria: % of issues covered, soundness of argumentation.	Oct 18, noon	**5%**
W7	Oct 8	Create a simple ontology using Protégé. Evaluation criteria: adherence to relevant criteria covered in W1, W3 and W4.	Oct 11, noon	**5%**
W11	Nov 5	Apply Referent Tracking to some problematic issue in the student's research/interest topic. Evaluation criteria: % of issues covered, adherence to reference tracking and BFO principles.	Nov 12, noon	**5%**
W12	Nov 12	Correct and improve the W11 assignment adhering to the principles of change management. Evaluation criteria: mistakes corrected, all principles thus far discussed applied.	Nov 19, noon	**5%**
W15	Dec 3	Student presentation. Evaluation criteria: consistent story line, application of principles discussed, assertions backed up by literature or sound arguments, clear take home message, areas for further research.		**35%**
W16	Dec 10	FINAL EXAM		**35%**

Final Grades:

Grade	Quality Points	Percentage
A	4.0	93.0% -100.00%
A-	3.67	90.0% - 92.9%
B+	3.33	87.0% - 89.9%
B	3.00	83.0% - 86.9%
B-	2.67	80.0% - 82.9%
C+	2.33	77.0% - 79.9%
C	2.00	73.0% - 76.9%
C-	1.67	70.0% - 72.9%
D+	1.33	67.0% - 69.9%
D	1.00	60.0% - 66.9%
F	0	59.9% or below

An interim grade of Incomplete (I) may be assigned if the student has not completed all requirements for the course. An interim grade of 'I' shall not be assigned to a student who did not attend the course. The default grade accompanying an interim grade of 'I' shall be 'U' and will be displayed on the UB record as 'IU.' The default Unsatisfactory (U) grade shall become the permanent course grade of record if the 'IU' is not changed through formal notice by the instructor upon the student's completion of the course.
Assignment of an interim 'IU' is at the discretion of the instructor. A grade of 'IU' can be assigned only if successful completion of unfulfilled course requirements can result in a final grade better than the default 'U' grade. The student should have a passing average in the requirements already completed. The instructor shall provide the student specification, in writing, of the requirements to be fulfilled.

6. ACADEMIC INTEGRITY

Academic integrity is a fundamental university value. Through the honest completion of academic work, students sustain the integrity of the university while facilitating the university's imperative for the transmission of knowledge and culture based upon the generation of new and innovative ideas. See http://grad.buffalo.edu/Academics/Policies-Procedures/Academic-Integrity.html.
Students may collaborate for the assignments in which case the submitted materials should be clearly labeled as such, with the names of all collaborating students. In case students who collaborate cannot come to a consensus for certain parts of the work, alternate solutions proposed by individual students should be clearly marked as such. Grading of individual students will take into account such alternatives.

7. ACCESSIBILITY RESOURCES

If you have any disability which requires reasonable accommodations to enable you to participate in this course, please contact the Office of Accessibility Resources, 25 Capen Hall, 645-2608, and also the instructor of this course .. The office will provide you with information and review appropriate arrangements for reasonable accommodations. http://www.student-affairs.buffalo.edu/ods/

8. COURSE FEES

Standard UB tuition and fees. No extra costs.

9. Course Organization / Schedule

Reference: http://registrar.buffalo.edu/calendars/academic/
The first part of this course (W1-W8) overlaps with PHI 598 and will be taught at the North Campus. The content is primarily theoretic.
Classes W9-W14 will be taught at the Department of Biomedical Informatics and will focus on specific problems or issues that students are or have been confronted with in their personal research projects or mentored research rotations and for which the application of the right sort of ontology might be beneficial. In class W10, students will discuss such issues and select one as the topic for their presentation during W15. Classes W11-W14 will in part be theoretical, in part used to apply the theories seen thus far to develop the foundations for a domain or application ontology that addresses the selected issue.

Week	2018	SLO	Instr.	Topic	Required readings prior to lecture	Assignment post-lecture	Due Date
W1	Aug 27	4	BS, WC	• Course overview • What is an ontology? • Key elements of an ontology • What are ontologies useful for?	(Hoehndorf, Schofield & Gkoutos, 2015).	Write a 2-page essay summarizing the key elements of an ontology as identified in the lecture and comparing them to the view of an ontology offered in (Hoehndorf, Schofield & Gkoutos, 2015).	Sept 6, noon
W2	Sep 3			NO CLASS			
W3	Sep 10	3	WC	Ontology of Disease: • disease from the clinician's perspective, • ontological approaches to disease, • the Ontology for General Medical Science	(Scheuermann, Ceusters & Smith 2009) (Haendel *et. al*., 2018)	Summarize in a 2 page essay the issues discussed in pages 16-21 of (Haendel et. al., 2018) and describe how the framework offered by (Scheuermann, Ceusters & Smith 2009) might resolve them.	Sept 24, noon
W4	Sep 17	2	BS	The Basic Formal Ontology	(Arp, Smith & Spear, 2015) chapters 5-6.		
W5	Sep 24	4	WC	Ontology and Information Engineering (IE) in the Healthcare Domain: • Ontology look-alikes, • Use of ontology and look-alikes in biomedical applications, • BFO as benchmark for biomedical IE	(Smith & Koppel, 2014)	Summarize in a 2 page essay the problems discussed in (Smith & Koppel, 2014) and describe how Basic Formal Ontology can assist in dealing solving them.	Oct 18, noon
W6	Oct 1	5	BS	Big Data and How to Overcome the Problems It Causes: • Definition of 'Big Data' • Overview of machine learning and other approaches to the exploitation of Big Data • Role of ontology in Data Science	(Merelli et. al, 2014)		
W7	Oct 8	7	BD	Introduction to ontology editors (e.g. Protégé) and add-on tools		Create a simple ontology using Protégé.	Oct 11, noon.

W8	Oct 15	1	BS	Ontological Realism: • Conceptual vs. realist approaches to ontology • Universals and defined classes • Realism about universals	(Smith & Ceusters, 2010) (Maojo et al., 2011) (Brochhausen et. al., 2011)		
W9	Oct 22	1, 2, 4, 6	WC	Using BFO's relations in biomedical applications	(Arp, Smith & Spear, 2015) chapter 7.		
W10	Oct 29	5, 6, 7	WC	Application of ontology in students' research			
W11	Nov 5	7	WC	Using Referent Tracking for ontology development and use	(Ceusters et. al., 2011)	Apply Referent Tracking to some problematic issue in the student's research/interest topic	Nov 12, noon
W12	Nov 12	8	WC	Principles for change management in ontologies	(Ceusters 2009)	Correct and improve the W11 assignment adhering to the principles of change management.	Nov 19, noon
W13	Nov 19	4,5,7	WC	Principles for upgrading to new (versions of) ontologies in biomedical information systems	(Ceusters 2011)		
W14	Nov 26	4,6,8	WC	Evaluation of ontologies	(Obrst et. al., 2007)		
W15	Dec 3	6, 7	WC+ BS	Student presentations			
W16	Dec 10		WC	Final Exam			

10. Course Materials

This course requires the following books, papers and electronic publications, all of which are available publicly or through the UB Libraries:

- (Arp, Smith & Spear, 2015) Arp R, Smith B, Spear AD. Building ontologies with Basic Formal Ontology. MIT Press, 2015.
- (Brochhausen *et. al*., 2011) Brochhausen M, Burgun A, Ceusters W, Hasman A, Leong TY, Musen M, Oliveira JL, Peleg M, Rector A, Schulz S. Discussion of "biomedical ontologies: toward scientific debate". Methods Inf Med. 2011;50(3):217-36.
- (Ceusters 2009) Ceusters W. Applying Evolutionary Terminology Auditing to the Gene Ontology. Journal of Biomedical Informatics 2009;42:518–529.
- (Ceusters 2011) Ceusters W. SNOMED CT Revisions and Coded Data Repositories: When to Upgrade? In American Medical Informatics Association 2011 Annual Symposium Proceedings, Washington DC, October 22-26, 2011:197-206
- (Ceusters *et. al.*, 2011) Ceusters W, Capolupo M, De Moor G, Devlies J, Smith B. An Evolutionary Approach to Realism-Based Adverse Event Representations. Methods of Information in Medicine, 2011;50(1):62-73.
- (Haendel *et. al.*, 2018) Haendel MA, McMurry JA, Relevo R, Mungall CJ, Robinson PN, and Chute CG. A Census of Disease Ontologies. Annu. Rev. Biomed. Data Sci. 2018. 1:305–31.
- (Hoehndorf, Schofield & Gkoutos, 2015) Robert Hoehndorf, Paul N. Schofield and Georgios V. Gkoutos. The role of ontologies in biological and biomedical research: a functional perspective. Briefings in Bioinformatics, 2015, 1–12
- (Merelli *et. al.*, 2014) Merelli I, Pérez-Sánchez H, Gesing S, and D'Agostino D. Managing, Analysing, and Integrating Big Data in Medical Bioinformatics: Open Problems and Future Perspectives. BioMed Research International Volume 2014, Article ID 134023, 13 pages. http://dx.doi.org/10.1155/2014/134023
- (Obrst *et. al.*, 2007) Obrst L, Ceusters W, Mani I, Ray S, Smith B. The Evaluation of Ontologies: toward Improved Semantic Interoperability. In: Baker, Christopher J.O.; Cheung, Kei-Hoi (Eds.) Semantic Web: Revolutionizing Knowledge Discovery in the Life Sciences. Springer, Heidelberg, 2007;:139-58.
- (Scheuermann, Ceusters & Smith, 2009) Scheuermann RH, Ceusters W, Smith B. Toward an ontological treatment of disease and diagnosis. Summit Transl Bioinform. 2009 Mar 1;2009:116-20.
- (Smith & Ceusters, 2010) Smith B, Ceusters W. Ontological Realism as a Methodology for Coordinated Evolution of Scientific Ontologies. Applied Ontology, 2010;5(3-4):139-188.
- (Smith & Koppel, 2014) Smith SW & Koppel R, Healthcare information technology's relativity problems: a typology of how patients' physical reality, clinicians' mental models, and healthcare information technology differ. J Am Med Inform Assoc 2014;21:117–131. doi:10.1136/amiajnl-2012-001419.

11. Attendance Policy

Students are expected to attend ***all*** lectures. For religious observances, university sanctioned events, athletic commitments and family/work obligations/emergencies, absences may be granted upon request but can have an effect on the finally obtained grade (see grading policy)
For course cancellation/emergency planning, see the university website for cancellations/delays due to weather or other unforeseen events (http://emergency.buffalo.edu/campus-weather-alerts.html)

12. Classroom Decorum

Students are expected to arrive in due time for each class. Some lectures will start with a pre-lecture test to assess the student's level of preparation for the class. This test contributes to the final grading. Use of cell phones and laptops is allowed for the purposes of the class, but not for private reasons. Phone calls are not allowed.