

Course Title: Biomedical Ontology
Course Subject Code: BMI **Course Number:** 508
Type of Instruction: SEM **Class Number:** 24556
Semester: Fall 2020

Version August 23, 2020

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1. Course Information

- Date(s)/Time(s): Wednesdays 10AM – 12.30/1PM
- Location: Anywhere
- Delivery Mode: Remote, real-time: students are considered in attendance when they log-in to the virtual classroom platform on time with their camera on. Webcams should be at eye level and students should refrain from multi-tasking while in class. A link for Zoom or Webex will be sent to students prior to the first class. This link will always be used until stated otherwise by the instructor.
- Number of Credits: 3
- Course director and instructor: Werner Ceusters, MD
contact: wceusters@gmail.com

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).
- As part of the course, students will develop a very small ontology that falls within their domain of interest, ideally useful towards their final MSc or PhD degree requirements. Though small, it must satisfy all quality requirements discussed in the course. It will be gradually developed during the course and presented in a final oral presentation.
- Most classes in the course consists of a theoretical and practical part. The practical parts consist of tests, guided exercises or group work.
- Course prerequisites: BMI 501 unless waived.

3. Student Learning Outcomes (SLO)

Course Learning Outcomes; students will be able to:	BMI PhD Program Outcomes / Competencies	Class	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	• C3, C4, C5	<ul style="list-style-type: none"> • Lit.: R3, R4, R5, R6, R9 • Lecture • Discussion • Guided exercise 	<ul style="list-style-type: none"> • Tests: T1, T2 • Assignments: A2, A3, A4, A6, A10, A11
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	• C3, C4, C5, C6	<ul style="list-style-type: none"> • Lit.: R3, R4, R5, R6, R7, R8, R9 • Lecture • Discussion • Guided exercise 	<ul style="list-style-type: none"> • Tests: T1, T2 • Assignments: A2, A3, A4, A5, A6, A10, A11 • 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	• C8	<ul style="list-style-type: none"> • Lit.: R10, R11 • Lecture • Guided exercise 	<ul style="list-style-type: none"> • Assignment: A7, A10, A11 • Test: T2 • 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	• C1, C2, C6, C10	<ul style="list-style-type: none"> • Lit.: R1, R2, R7, R8, R13 • Lecture • Discussion • Guided exercise 	<ul style="list-style-type: none"> • Assignment: A1, A2, A5, A10, A11 • Test: T1, T2 • Final Exam

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	• C1, C2	• Lit.: R1, R2 • Lecture • Discussion • Guided exercise	• Assignment: A1, A2, A10, A11 • Tests: T1, T2
6. Assess the quality of existing clinical research data repositories and ontologies 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	• C8, C10, C11, C12	• Lit.: R10, R11, R13, R14 • Lecture • Guided exercise • Group exercise	• Assignment: A7, A9, A10, A11 • Test: T2 • Final exam
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	• C6, C7, C13	• Lit.: R7, R8, R9 • Lecture • Guided exercise • Discussion	• Assignment: A5, A6, A10, A11 • Test: T2
8. Understand and apply the principles for life cycle management for biomedical ontologies	O8: The principles for change management and upgrades to biomedical ontologies	• C9	• Lit.: R12 • Lecture • Guided exercise	• Assignment: A8, A10, A11 • Final exam
9. Understand and apply the principles for ontology matching.	O4: Technical approaches to acquiring, modeling, representing and managing healthcare and biomedical research knowledge O8: The principles for change management and upgrades to biomedical ontologies	• C11, C12	• Lit.: R14 • Lecture • Guided exercise • Group exercise	• Assignments A9, A10, A11 • Final exam

4. COURSE REQUIREMENTS

- Students must attend all classes and must participate in class discussions. See attendance policy regulations on page 10 for exceptions,
- There are 14 papers and book chapters listed in the course materials below, indicated as either as required or suggested.
- Some classes will include an in-class test. Students absent for these classes will receive a 0% score for these tests unless the instructor has been informed through email about a valid reason for absence prior to the beginning of the class. When notified of absence in due time, the instructor may propose an alternative test or assignment for that class.
- All assignments need to be completed prior to the deadline specified in the course schedule and send electronically to wceusters@gmail.com. Thus no Google doc links or any other link to a cloud server. The filename should be formatted as this: BMI508-[number of the assignment]-[your UBIT name].
For example, if the course director were a student: "BMI508-A1-ceusters.docx".
- The first line in the document should always be your full name (first and last name).
- The 'sent' date in the metadata header of the message through which the completed assignments will be received will be taken for assessment of in-time delivery.
- When assignments are delivered past the due date, a penalty of 1% of the positive final score will be applied for every 24 hours of delay. When such assignment is not delivered at all, the penalty will be equal to the % for which the assignment counts towards the final grade. Students unable to meet the deadline must inform the instructor by email and provide a valid reason prior to reaching the deadline. An alternative assignment and/or due date can then be agreed upon, be it entirely to the discretion of the instructor.

5. GRADING POLICY

- Grading follows standard graduate policies (<http://grad.buffalo.edu/Academics/Policies-Procedures/Grading-Procedures.html>)
- Counts towards the final score:
 - Two in-class tests (T1, T2): 11%
 - Ten post-class assignments (A1 to A10): 46%
 - Final oral presentation (A11): 13%
 - Final exam: 30%
 - TOTAL: 100%
- 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.

Class	Date	Pre-class required readings	Assessments	Due dates	Final Score weight
C1	02-Sep	R1			
C2	09-Sep	R2	Post-class assignment A1	14-Sep - noon	2%
C3	16-Sep	R3	In-class open book test T1 Post-class assignment A2	21-Sep - noon	4% 4%
C4	23-Sep	R4	Post-class assignment A3	28-Sep - noon	2%
C5	30-Sep	R5, (R6)	Post-class assignment A4	05-Oct - noon	2%
C6	07-Oct	R7, (R8)	Post-class assignment A5	12-Oct - noon	5%
C7	14-Oct	R9	Post-class assignment A6	19-Oct - noon	4%
C8	21-Oct	(R10), (R11)	Post-class assignment A7	26-Oct - noon	6%
C9	28-Oct	(R12)	Post-class assignment A8	09-Nov - noon	1%
C10	04-Nov	R13	In-class open book test T2		7%
C11	11-Nov	R14			
C12	18-Nov		Post-class assignment A9	30-Nov - noon	3%
C13	02-Dec		Post-class assignment A10	07-Dec – noon	17%
C14	09-Dec		In-class presentation A11		13%
EXAM	16-Dec		In-class open-book exam		30%
					100%

Scoring per student learning outcomes and assessments

SLO	Tests		Assignments											Exam	Totals
	T1	T2	A1	A2	A3	A4	A5	A6	A7	A8	A9	A10	A11		
1	1%	1%		1%	1%	1%		1%				2%	2%	2%	12%
2	1%	1%		1%	1%	1%	1%	1%				2%	2%	8%	19%
3		1%							3%			2%	1%	10%	17%
4	1%	1%	1%	1%				2%				2%	2%	1%	11%
5	1%	1%	1%	1%								2%	2%	1%	9%
6		1%							3%		2%	2%	1%	1%	10%
7		1%					2%	2%				2%	1%	1%	9%
8										1%		2%	1%	1%	5%
9											1%	1%	1%	5%	8%
Totals	4%	7%	2%	4%	2%	2%	5%	4%	6%	1%	3%	17%	13%	30%	100%

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. COURSE FEES

Standard UB tuition and fees.

7. Course Organization / Schedule

Reference: <http://registrar.buffalo.edu/calendars/academic/>

C1. Sep 2 / Course introduction – Ontology and representational systems / SLO 4, 5

Pre-class assignments:

a) required reading:

R1 Introduction (pages xix to xxiv) to: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press,: Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

b) After reading **R1**, reflect on the extent to which the issues discussed therein might be applicable to the research and developments you intend to carry out for your future MSc or PhD thesis or project. Be prepared to present and discuss your thoughts informally in class **C1**. No prior submission needed.

Class structure:

- Participant and instructor introduction
- Course introduction, housekeeping rules, expectations, course project work, final exam.
- Traditional lecture on the basics of representational systems
- Guided exercise on on-line available resources
 - SNOMED CT <https://browser.ihtsdotools.org/>
 - Medical Subject Headings <https://meshb.nlm.nih.gov/search>
 - WHO Classification systems <https://www.who.int/classifications/en/>
 - BioPortal 'ontologies' <https://bioportal.bioontology.org/>
- Students will report on pre-class assignment (b) and explain their ideas, each presentation followed by discussion.

Post-class assignment: required reading

R2 What is an ontology? (pages 1-26) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press,: Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

C2. Sep 9 / What is an ontology? / SLO 4, 5

Required reading pre-class:

R2 *What is an ontology?* (pages 1-26) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press, Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

Class structure:

Traditional lecture on what an ontology is, key elements of an ontology, realism-based ontology, what ontologies are useful for.

Post-class assignment:

- a) **A1:** Reading **R1** contains several examples of what went wrong in certain representational systems. Identify them and do for each of these examples the following: (1) explain in 1 sentence what the type of mistake is, (2) find a similar mistake in one of the demonstrated representational systems that, i.e. the example containing a mistake, has not been discussed in class **C1** or in another representational system that you know about that might be useful in your research, (3) use what you learned from **R2** and the **C1** and **C2** lectures to propose a correction.

Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A1-[your name].docx'

Assessment:

All examples identified: 20%

Mistakes correctly explained: 20%

Similar mistakes found in representational system of your choice and explained why: 40%

Soundness of proposed corrections with argumentation and motivation from **R2** and lecture carried out: 20%.

Due date: Sep 14 – noon.

- b) Post-class reading: see **C3**

C3. Sep 16 / Basic Formal Ontology: continuants / SLO 1, 2

Required reading pre-class:

R3 *Introduction to Basic Formal Ontology I: continuants*. (pages 85-120) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press, Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

Class structure:

- a) Traditional lecture
b) **T1:** in-class test (open book) on the application of material discussed thus far in classes **C1** through **C3**
c) Discussion of answers to **T1**

Post-class assignment:

- a) required reading see **C4**

- b) **A2:** prepare a short powerpoint presentation (maximum 5 minutes for presentation during the class) in which you describe in what area and how you intend to develop a small realism-based ontology useful for your final MSc or PhD project/thesis. Required content: precise description of domain, motivation, examples of terms to be represented, challenges you see, intended application with concrete indication of the sort of topic entities it will be applied to. For students in BMI or Biomedical Sciences, the topic entity must be a person. Other students may select another topic type (pieces of equipment, plants, environmental entities, ...). Use previous classes, slides and readings as a guide.

Send your work in a PowerPoint file to wceusters@gmail.com. Name of the file: 'BMI508-A2-[your name].pptx'

Assessment: extent to which the material discussed in classes **C1** to **C3** is appropriately applied.

Due date: Sep 21 – noon

C4. Sep 23 / Basic Formal Ontology: occurrents / SLO 1, 2

Required reading pre-class:

R4 *Introduction to Basic Formal Ontology II: occurrents*. (pages 121-130) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press, Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

Class structure:

Traditional lecture

Post-class assignment:

- a) Required reading see **C5**
- b) **A3**: Create a first version of your ontology consisting of at least 5 continuants and 5 occurrents satisfying the following four requirements: (1) they belong to your selected research domain presented in **A2**, (2) they have not been discussed in class as part of **A1**, although they might have been mentioned, (3) they are *not* terms from BFO, (4) they are *not* all present in one single ontology published on the BioPortal.

For each term, indicate the most specific BFO-term by which it is subsumed.

Send your work in a PowerPoint file to wceusters@gmail.com. Name of the file: 'BMI508-A3-[your name].pptx'

Assessment:

- 1) The ten terms satisfy the three requirements: 40% (1% loss for each requirement not satisfied)
- 2) The ten terms are corrected classified in BFO at the appropriate level according to Fig 5.1 (p. 88) in **R3** for the continuants, and the terms listed in **R4**: 40% (% loss for magnitude of the difference between selected level and correct level)
- 3) Motivation for your decisions: 20%. (2% for each term) A motivation which is plausible or for which reasonable arguments are provided may be positively scored, despite the wrong classification of the term.

Due date: Sep 28 – noon

C5. Sep 30 / Relations in biomedical ontologies / SLO 1, 2**Required reading pre-class:**

R5 *The ontology of relations*. (pages 131-150) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press, Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

Suggested additional reading (for sure for students with concentration in Biomedical Ontology):

R6 Smith, B., et al., *Relations in biomedical ontologies*. *Genome Biol*, 2005. **6**(5): p. R46. [2]

<https://genomebiology.biomedcentral.com/articles/10.1186/gb-2005-6-5-r46>

Class structure:

- a) Traditional lecture
- b) Guided exercise

Post-class assignment:

- a) **A4**: Apply a relational dependency analysis to the terms thus far in your ontology.
Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A4-[your name].docx'

Assessment:

- (1) All dependent entities in version 1 of your ontology must be related to the entities they depend on. If such dependent entity depends only on independent continuants, you are done for now. If this however necessitates the introduction of new dependent entities in your ontology, you need to analyze further until it is for all dependent entities specified upon which independent continuants they directly or indirectly depend: 40%.
- (2) For all types in your ontology thus far you must indicate how instances thereof relate directly or indirectly to an instance of the topic type you selected in A1. If that requires further entities to be introduced, apply step (1) to them as well and continue doing so until for all types in your ontology steps (1) and (2) are done: 40%.
- (3) All relations used must either have been taken from a realism-based source in which the relation is formally defined, OR you create a relation and define it appropriately.

Due date: Oct 5 – noon

- b) required reading see **C6**

C6. Oct 7 / Using Referent Tracking for ontology development / SLO 2, 4, 7**Required reading pre-class:**

R7 Ceusters, W. and B. Smith, *Tracking referents in electronic health records*. *Stud Health Technol Inform*, 2005. **116**: p. 71-6.[3]

PDF downloadable from: <http://ebooks.iospress.nl/publication/10278>

Suggested additional reading (for sure for students with concentration in Biomedical Ontology):

R8 Ceusters, W. and B. Smith, *Strategies for referent tracking in electronic health records*. *J Biomed Inform*, 2006. **39**(3): p. 362-78.[4]

Class structure:

- a) Traditional lecture
- b) Guided exercise

Post-class assignment:

- a) **A5:** Apply a referent tracking analysis to your ontology version 2. This means that you must identify a scenario under which of at least two most specific terms from your ontology an instance of each applies to an instance of your topic type from **A1**. Simulate on paper the population of a Referent Tracking Database with all tuples required to represent the instances of the two selected terms and how they relate directly or indirectly to the instance of the topic type. Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A5-[your name].docx'
Assessment:
 - (1) All required instances, using your ontology as benchmark, are represented: 60%
 - (2) All required tuple types have been correctly used 40%

Due date: Oct 12 – noon

- b) required reading see **C7**

C7. Oct 14 / General principles for realism-based ontology development / SLO 1, 2, 7

Required reading pre-class:

R9 *Principles of best practice I and II*. (pages 43-84) in: Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press, Cambridge, Massachusetts. p. 1 online resource (245 p.) [1]

UB login: <https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo>

Class structure:

- a) interactive lecture
- b) discussion of issues with **A5**
- c) application of **R9** to a few terms from the students' ontologies.

Post-class assignment:

A6: Apply the principles discussed in class **C7** and in **C8** to all the most specific terms in your ontology so as to create version 3. Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A6-[your name].docx'

Due date: Oct 19 – noon.

C8. Oct 21 / Ontology for General Medical Science / SLO 3, 6

Suggested readings:

R10 Scheuermann, R.H., W. Ceusters, and B. Smith, *Toward an ontological treatment of disease and diagnosis*. Summit Transl Bioinform, 2009. **2009**: p. 116-20. [5]

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3041577/>

R11 Smith, B. and W. Ceusters. *Aboutness: Towards foundations for the information artifact ontology*. in *CEUR Workshop Proceedings*. 2015. [6]

<http://ceur-ws.org/Vol-1515/regular10.pdf>

Class structure:

- a) Interactive lecture
- b) Guided exercise: comparing disease related ontologies to OGMS

Post-class assignment:

A7: Add to your ontology 3 terms each one of which (1) is a subtype of OGMS and (2) has thus far not been included in your ontology. Perform the type of analysis as described for **A4** to these terms to ensure that everything that can be related in your ontology, is so related. The result is version 4. Students from other disciplines than biomedical sciences or life sciences may select another relevant ontology than OGMS, but this needs to be discussed with and agreed upon by the instructor in class **C8**. Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A7-[your name].docx'

Due date: Oct 26 – noon.

C9. Oct 28 / Principles for change management in ontologies / SLO 8

Suggested pre-class reading:

R12 Seppälä, S., B. Smith, and W. Ceusters, *Applying the Realism-Based Ontology-Versioning Method for Tracking Changes in the Basic Formal Ontology*, in *Formal Ontology in Information Systems (FOIS 2014)*, P. Garbacz and O. Kutz, Editors. 2014, IOS Press. p. 227-240. [7]

<http://ontology.buffalo.edu/smith/articles/fois2014.pdf>

Class structure:

- a) Traditional lecture.
- b) Guided exercise

Post-class assignments:

- a) **A8:** You have now 4 versions of your ontology, each one the result of **A3**, **A4**, **A6** and **A7** resp. Apply for all the terms in your ontology V4 the change analysis as discussed in **C9**.

Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A8-[your name].docx'

Due date: Nov 9 – noon.

- b) Required reading: see **C10**

C10. Nov 4/ Evaluation of ontologies / SLO 4, 6

Required pre-class reading:

R13: Amith, M., et al., *Assessing the practice of biomedical ontology evaluation: Gaps and opportunities*. J Biomed Inform, 2018. **80**: p. 1-13. [8]

<https://www.sciencedirect.com/science/article/pii/S1532046418300285>

Class structure:

- a) Traditional lecture
- b) **T2:** in-class open book test on material from **C1** to **C8**

Post-lecture assignment: required reading, see **C11**

C11. Nov 11 / Ontology merging and matching I / SLO 6, 9

Required pre-class reading:

R14 Ceusters, W., *Towards a realism-based metric for quality assurance in ontology matching*. Frontiers in Artificial Intelligence and Applications. Vol. 150. 2006. 321-332. [9]

http://ontology.buffalo.edu/bfo/Ontology_Matching.pdf

Class structure:

- a) Interactive lecture
- b) Guided exercise

C12. Nov 18 / Ontology merging and matching II / SLO 6, 9

Class structure:

Group exercise: students will merge the version 4 of their ontologies in one larger ontology. The instructor will guide the exercise, will help where the students together are stuck, but might decide not to provide for certain issues a solution. These issues will be clearly marked, and then need to be resolved by the students individually as part of assignment **A9**.

Post-class assignment:

A9: address the issues left open during class.

Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A9-[your name].docx'

Due date: Nov 30 – noon.

Nov 25 / NO CLASS

C13. Dec 2 / Axioms in ontologies / SLO 7

Class structure:

- a) Traditional lecture about axioms in BFO 2.0
- b) Guided exercise on adding axioms to student ontologies
- c) Instructions for final presentation

Post-class assignment:

A10: prepare a PowerPoint presentation of your ontology. It must contain examples of all elements discussed, including one axiom pertaining to one most specific type in your ontology. It must also include where it fits in the overall merged ontology developed in C. Maximum 10 slides for presentation during **C15** in maximally 15 minutes.

Send your work in a Word doc to wceusters@gmail.com. Name of the file: 'BMI508-A10-[your name].docx'

Due date: Dec 7 – noon.

C14. Dec 9 / Student presentations and discussions / SLO 1 to 9

Each student will present his PP slides in maximally 15 minutes, followed by 10 minutes discussion and questions (**A11**). Presentations will be public, students and faculty of BMI will be invited.

Quality of oral presentation and readiness to answer questions will be assessed independent of quality of the slides which will be assessed as part of **A10**.

C15. FINAL EXAM: Dec 16

The final exam will be held online. It will consist of three phases.

- (1) a group exercise of maximally one hour and a half in which students will work together on solving an ontology evaluation and correction task. At the end of this phase, a document with the obtained results will be emailed to the course instructor. The format in which the results need to be presented will be disclosed at the beginning of the exercise. Students do not have to agree on everything in that document. Evaluation will be based on the completeness of the document with respect to format and content requirements, NOT whether the content is correct. All students will receive the same score for this part, good for 30%.
- (2) During the next 45 minutes, students will individually work further on the document by indicating with which parts they do not agree, thereby indicating how these parts should be corrected. They may not change the original parts of the document, but annotate it with their disagreement and improvement where they see fit. Parts that an individual student does not annotate, will considered to be correct in the eyes of the student and negatively scored when in fact they are wrong. Students must email their individually reworked document at the end of phase 2. No collaboration allowed. Evaluation will this time be based on correctness, i.e. agreement or disagreement with original content, and correctness of improvement. Individual scores for 50% of total.
- (3) During phase 2, the instructor will assess the document submitted at the end of phase 1 and derive from it questions which the students will answer individually during phase 3 for maximally 45 minutes. No collaboration allowed. Evaluation: 20% of total.

8. ATTENDANCE POLICY

Students are expected to attend *all* classes. For religious observances, university sanctioned events, athletic commitments and family/work obligations/emergencies, absences may be granted upon request and subsequent approval by the course director *prior* to the absence. Requests are to be sent by email. Medical absence must be accompanied by a doctor's note. Even if absence is granted, it can have an effect on the finally obtained grade (see grading policy) unless additional coursework in replacement for missed graded activities is performed.

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>)

9. 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>.
- At most two students may develop an ontology together. In that case, all requirements which involve a specified number of elements to be added, need to be doubled (e.g. starting with 20 terms instead of 10 as required for **A3**). This type of collaboration must be agreed upon by the instructor prior to assignment **A3**. All submitted materials must then be clearly labeled as such, with the names of the two 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 which may lead to different scores for these students

on these assignments. Collaboration is not allowed for scored tests and the final exam. For the final presentation, twice the number of slides are allowed, and double the allotted time for presentation. Students must however take care of different aspects of the work, and slides, nor oral presentation to overlap in content.

10. CLASSROOM DECORUM

Students are expected to log on in due time for each class. Video must be active, sound must be muted. Only when the instructor asks questions, students who wish to respond may unmute and talk. Additional rules of conduct, when applicable, will be explained by the instructors prior to the class.

11. 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 course director of this course. The office will provide you with information and review appropriate arrangements for reasonable accommodations. <http://www.student-affairs.buffalo.edu/ods/>
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12. UNIVERSITY SUPPORT SERVICES

Students are often unaware of university support services. For example, the Center for Excellence in Writing provides support for written work, and several tutoring centers on campus provide academic success support and resources.

13. AVAILABLE RESOURCES ON SEXUAL ASSAULT

UB is committed to providing an environment free of all forms of discrimination and sexual harassment, including sexual assault, domestic and dating violence and stalking. If you have experienced gender-based violence (intimate partner violence, attempted or completed sexual assault, harassment, coercion, stalking, etc.), UB has resources to help. This includes academic accommodations, health and counseling services, housing accommodations, helping with legal protective orders, and assistance with reporting the incident to police or other UB officials if you so choose. Please contact UB's Title IX Coordinator at 716-645-2266 for more information. For confidential assistance, you may also contact a Crisis Services Campus Advocate at 716-796-4399.

14. COUNSELING SERVICES:

As a student you may experience a range of issues that can cause barriers to learning or reduce your ability to participate in daily activities. These might include strained relationships, anxiety, high levels of stress, alcohol/drug problems, feeling down, health concerns, or unwanted sexual experiences. Counseling, Health Services, and Health Promotion are here to help with these or other concerns. You learn can more about these programs and services by contacting:

Counseling Services:	120 Richmond Quad (North Campus),	phone 716-645-2720
	202 Michael Hall (South Campus),	phone: 716-829-5800
Health Services:	Michael Hall (South Campus),	phone: 716- 829-3316
Health Promotion:	114 Student Union (North Campus),	phone: 716- 645-2837

15. COURSE MATERIALS

- This course requires the following papers and electronic publications, all of which are available publicly or through the UB Libraries:
 1. Arp, R., B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*. 2015, The MIT Press,: Cambridge, Massachusetts. p. 1 online resource (245 p).
 2. Smith, B., et al., *Relations in biomedical ontologies*. *Genome Biol*, 2005. **6**(5): p. R46.
 3. Ceusters, W. and B. Smith, *Tracking referents in electronic health records*. *Stud Health Technol Inform*, 2005. **116**: p. 71-6.
 4. Ceusters, W. and B. Smith, *Strategies for referent tracking in electronic health records*. *J Biomed Inform*, 2006. **39**(3): p. 362-78.
 5. Scheuermann, R.H., W. Ceusters, and B. Smith, *Toward an ontological treatment of disease and diagnosis*. *Summit Transl Bioinform*, 2009. **2009**: p. 116-20.
 6. Smith, B. and W. Ceusters. *Aboutness: Towards foundations for the information artifact ontology*. in *CEUR Workshop Proceedings*. 2015.
 7. Seppälä, S., B. Smith, and W. Ceusters, *Applying the Realism-Based Ontology-Versioning Method for Tracking Changes in the Basic Formal Ontology*, in *Formal Ontology in Information Systems (FOIS 2014)*, P. Garbacz and O. Kutz, Editors. 2014, IOS Press. p. 227-240.
 8. Amith, M., et al., *Assessing the practice of biomedical ontology evaluation: Gaps and opportunities*. *J Biomed Inform*, 2018. **80**: p. 1-13.
 9. Ceusters, W., *Towards a realism-based metric for quality assurance in ontology matching*. *Frontiers in Artificial Intelligence and Applications*. Vol. 150. 2006. 321-332.