University at Buffalo
The State University of New York

JACOBS SCHOOL OF MEDICINE AND BIOMEDICAL SCIENCES
Department of Biomedical Informatics

Course Title: Biomedical Ontology
Course Subject Code: BMI **Course Number:** 508
Type of Instruction: SEM **Class Number:** 18006
Semester: Fall 2025 **Delivery mode**: Remote (exam in person)

Version Aug 25, 2025

1. *Course Information*

• Date(s)/Time(s):	Wednesdays 10AM – 12.50/1PM
• 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. Students must register for the Zoom sessions through the following link: https://buffalo.zoom.us/meeting/register/2J7RXTPCS06NGdIniXRCSg Upon registration, students will receive the link to connect to all sessions. Final exam is in person unless all attendants are US citizens or Green Card holders!
• Number of Credits:	3
• Course director and instructor: contact:	Werner Ceusters, MD wceusters@gmail.com
• Office hours:	There are no fixed office hours. Students may however request a meeting by sending an email to the instructor thereby indicating clearly dates and times they are available. When sent between 8am and 5pm, a same-day response will be provided.

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: none. This course is itself a prerequisite for BMI708.

3. *Student Learning Outcomes (SLO)*

Institutional Learning Outcomes (UB ILO) mapped to individual classes and assessments

1. Critical Reasoning: C1 – C13; A1, T1, A3, A4, A5, A6, A7, A8, T2, A9, final exam
2. Literacy Skills: C14; A1, A2, A10, A11
3. Ethics and Responsibility: applied attendance, participation and assignment policies for this course
4. Local and Global Diversity: not applicable
5. Collaborate Positively: C11, C12, C13
6. Personal Skills: C1 – C14; A2, A11
7. Service Engagement: not applicable

Course and program learning outcomes

Course Learning Outcomes; students will be able to:	BMI 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	• Lit.: R3, R4, R5, R6, R9 • Lecture • Discussion • Guided exercise	• Tests: T1, T2 • Assignments: A2, A3, A4, A5, 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	• Lit.: R3, R4, R5, R6, R7, R8, R9 • Lecture • Discussion • Guided exercise	• 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	• Lit.: R10, R11 • Lecture • Guided exercise	• 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	• Lit.: R1, R2, R7, R8, R13 • Lecture • Discussion • Guided exercise	• Assignment: A1, A2, A6, 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: A6, A4, 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 11 for exceptions. Classes are online through Zoom, while the final exam is in person.
- There are 12 papers and book chapters listed in the course materials below, indicated either as required or suggested. Reading materials will not exhaustively be covered in class, but students can at the beginning of the class ask for clarifications where needed. The content of required readings may be the topic of final exam questions while that is not the case for suggested

readings unless the material is explicitly covered in class. Students with an orientation in Ontology are strongly suggested to also read the suggested papers.

- Some classes will include an in-class test, i.e. a test during the scheduled class time. 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 for that student an alternative test or assignment, or transfer the allocation of earnable points to a later assignment or the final exam.
- All assignments need to be completed prior to the deadline specified in the course schedule and uploaded as documents in the format specified for the assignment to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. No Google doc links or any other links to a cloud server are allowed. The filename before the file extension should be formatted as this: BMI508-[number of the assignment]-[your UBIT name].
  For example, if the course director were a student and the dedicated format for the assignment would be MS Word, the name would be: "BMI508-A1-ceusters.docx".
- The first line in the document should always be your full name (first and last name) unless otherwise specified.
- The 'upload' date and time in the metadata header of the uploaded file 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.
- You may email the instructor at any time. Mails related to the class should in the subject line be prefixed with 'BMI508', otherwise they will be ignored and not answered.
- To send email to the instructor, only use wceusters@gmail.com. Mails to ceusters@buffalo.edu will be ignored. The subject line of each email must start with 'BMI508:' (without quotes, but with colon). Mails without this prefix in the subject line will be ignored.

5. ***Grading Policy***

- Grading follows standard graduate policies (https://www.buffalo.edu/grad/succeed/current-students/policy-library.html?q=grades)
- Counts towards the final score:
  - Two in-class tests (T1, T2): 11%
  - Ten post-class assignments (A1 toA10): 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. When applied, a student's grade will never be lower than the original grade.

Class	Date	Pre-class readings	Assessments	Due dates	Final Score weight
C1	27-Aug	R1			
C2	03-Sep	R2 (R2b, R2c)	Post-class assignment A1	9-Sep - noon	2%
C3	10-Sep	R3	In-class open book test T1 Post-class assignment A2	 16-Sep - noon	4% 4%
C4	17-Sep	R4	Post-class assignment A3	23-Sep - noon	2%
C5	24-Sep	R9	Post-class assignment A4	30-Sep - noon	4%
C6	01-Oct	R5, (R6)	Post-class assignment A5	07-Oct - noon	2%
C7	08-Oct	R7, (R8)	Post-class assignment A6	14-Oct - noon	5%
C8	15-Oct	(R10), (R11)	Post-class assignment A7	21-Oct - noon	6%
C9	22-Oct	(R12)			
C10	29-Oct	R13	In-class open book test T2 Post-class assignment A8	 04-Nov - noon	7% 1%
C11	05-Nov	R14 (R15)			
C12	12-Nov		Post-class assignment A9	18-Nov - noon	3%
C13	19-Nov		Post-class assignment A10	02-Dec – noon	17%
No class	26-Nov				
C14	03-Dec		In-class presentation A11		13%
EXAM	10-Dec		In-class open-book exam		30%
					100%

Scoring per student learning outcomes and assessments

	Tests		Assignments											Exam	Totals
SLO	**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%**	**4%**	**2%**	**5%**	**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

The following F-grades might be assigned:

- F1 (for a student who participated beyond the 60% point of the class)
- F2 (for a student who started participating, but stopped prior to the 60% point of the class)
- F3 (for a student who did not participate in the class)

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 any of the standard grades from the table above, including F1 and F2. The default grade shall become the permanent course grade of record if the 'I/x' grade is not changed through formal notice by the instructor upon the student's completion of the course.
An incomplete grade can be assigned only if successful completion of unfulfilled course requirements can result in a final grade better than the default grade.

6. Course Fees

Standard UB tuition and fees.

7. Course Organization / Schedule

C1. Aug 27 / 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:

a) Participant and instructor introduction
b) Course introduction, housekeeping rules, expectations, course project work, final exam.
c) Traditional lecture on the basics of representational systems
d) Guided exercise on on-line available resources
   a. SNOMED CT https://browser.ihtsdotools.org/?
   b. Medical Subject Headings https://meshb.nlm.nih.gov/search
   c. WHO Classification systems https://www.who.int/classifications/en/
   d. BioPortal 'ontologies' https://bioportal.bioontology.org/
e) 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]
UB login: https://ebookcentral-proquest-com.gate.lib.buffalo.edu/lib/buffalo/detail.action?docID=3433795&pq-origsite=primo
This paper describes ontology from a realism-based perspective, the paradigm that students have to follow for their assignments.

Suggested readings:

R2b: Guarino, N., D. Oberle, and S. Staab, ***What Is an Ontology?,*** in Handbook on Ontologies, S. Staab and R. Studer, Editors. 2009, Springer Berlin Heidelberg: Berlin, Heidelberg. p. 1-17. [2]
https://link.springer.com/chapter/10.1007/978-3-540-92673-3_0
This paper focuses on the logical aspects of ontologies. Although the authors take a conceptualist stance, the logical principles they describe are equally applicable to realism-based ontologies.

R2c: Rector, A., et al., ***On beyond Gruber: "Ontologies" in today's biomedical information systems and the limits of OWL***. J Biomed Inform, 2019. 100s: p. 100002. [3]
https://pubmed.ncbi.nlm.nih.gov/34384571/
This paper clarifies the many confusions about the various artifacts that are nowadays called 'ontologies', most of them unwarranted.

C2. Sep 3 / 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.
   Upload your work in a Word doc to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A1-[your ubitname].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 9 – noon.

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

C3. Sep 10 / 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**

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.
Upload your work in a PowerPoint file to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A2-[your ubitname].pptx'
Assessment: extent to which the material discussed in classes **C1** to **C3** is appropriately applied.
Due date: Sep 15 – noon

C4. Sep 17 / 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:

a) Discussion of answers to **T1**
b) Student presentations of assignments **A2**
c) 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 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 or anywhere else, (5) no two entity types you selected may be directly subsumed by the same BFO universal, (6) no selected entity type may be subsumed by another selected entity type.
For each term, indicate the most specific BFO-term by which it is subsumed.
Upload your work in an Excel file to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A3-[your ubitname].xlsx'
c) Assessment:
    1) The ten terms satisfy the six 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 22 – noon

C5. Sep 24 / 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 **A3**
c) application of **R9** to a few terms from the students' ontologies.

Post-class assignment:
A4: Apply the principles for term selection, replacement, formatting and definitions, discussed in class and in **R9** to all the terms in your ontology so as to create version 2. Do this by amending your file for **A3**.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A4-[your ubitname].XXX', where XXX is the extension of the selected format (Excel or Word). Note: Keep the numbering of the assignment as indicated here in the syllabus.
Due date: Sept 29 – noon.

C6. Oct 1 / 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. [4]
https://genomebiology.biomedcentral.com/articles/10.1186/gb-2005-6-5-r46

Class structure:
a) Traditional lecture
b) Guided exercise

Post-class assignment:
a) **A5**: Apply a relational dependency analysis to the definitions (to be corrected from assignment A4) indicated by the instructor so as to create one large relational PoR as exemplified in slide 23. This may require further entities to be defined to tie the entities referenced in the definitions together by means of relations. 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.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A5-[your ubitname].XXX'
Assessment:
(1) Definitions appropriately corrected in Aristotelian form and jointly complete: 20%
(2) BFO relations correctly applied or new ones correctly defined: 40%
(3) Adequate motivations for decisions made in (1) and (2): 40%
Due date: Oct 6 – noon
b) required reading see **C7**

C7. Oct 8 / 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.[5]
PDF downloadable from: http://ebooks.iospress.nl/publication/10278

Suggested additional reading (for sure for students with concentration in Biomedical Ontology as well as PhD students):
R8 Ceusters, W., The Place of Referent Tracking in Biomedical Informatics, in Terminology, Ontology and their Implementations, P.L. Elkin, Editor. 2023, Springer International Publishing: Cham. p. 171-218. [6]
https://osf.io/q8hts

Class structure:
a) Traditional lecture
b) Guided exercise

Post-class assignment:

a) **A6**: Apply a referent tracking analysis to your ontology version 3 so as to create version 4 by amending your **A5** file. 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.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A6-[your ubitname].XXX'
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 13 – noon

b) required reading see **C8**

C8. Oct 15 / 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. [7]
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. [8]
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, by amending your **A6**-file, 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 **A5** to these terms to ensure that everything that can be related in your ontology, is so related. The result is version 5. 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**.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A7-[your ubitname].XXX'
Due date: Oct 20 – noon.

C9. Oct 22 / 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. [9]
http://ontology.buffalo.edu/smith/articles/fois2014.pdf

Class structure:

a) Traditional lecture.
b) Guided exercise

Post-class assignments:

Required reading: see **C10**

C10. Oct 29 / 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. [10]
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**

a) **A8**: You have now 5 versions of your ontology, each one the result of **A3**, **A4**, **A5**, **A6** and **A7** resp. Apply for five terms in your ontology V5 the change analysis as discussed in **C9** and **C10**.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A8-[your ubitname].XXX'
Due date: Nov 3 – noon.
b) Required reading: see **C11**

C11. Nov 5 / 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. [11]
http://ontology.buffalo.edu/bfo/Ontology_Matching.pdf

Suggested reading (for sure for students with concentration in Biomedical Ontology as well as PhD students):

R15 Pengput, A. and W. Ceusters, *Setting the Scene to Link SNOMED CT to Realism-Based Ontologies* in *19th World Congress on Medical and Health Informatics (MEDINFO)*. Sydney, Australia. [12]
http://www.referent-tracking.com/RTU/files/PengputCeustersMEDINFO2023final.pdf

Class structure:

a) Interactive lecture
b) Guided exercise

C12. Nov 12 / Ontology merging and matching II / SLO 6, 9

Class structure:

Group exercise: students will merge the version 5 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.
Upload your work to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A9-[your ubitname].XXX'
Due date: Nov 17 – noon.

C13. Nov 19 / Axioms in ontologies / SLO 7

Class structure:

a) Traditional lecture about axioms in BFO2020
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 number of slides for presentation during **C15** and maximal time for presentation will be determined by the number of students able to present.
Upload your work in a PowerPoint to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. Name of the file: 'BMI508-A10-[your ubitname].pptx'
Due date: Dec 1 – noon.

Nov 26 / NO CLASS

C14. **Dec 3 / Student presentations and discussions / SLO 1 to 9**

Each student will present his PP slides in the time for presentation allotted, followed by 10 minutes discussion and questions (**A11**).

Presentations might be public.

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 11**

The final exam will be held in person. 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 uploaded to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1. 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 upload their individually reworked document to https://buffalo.box.com/s/j45edsmldrecnj3k53tawt406mua32i1 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 orally and competitively in cross-examination style during phase 3 for maximally 45 minutes. Evaluation: 20% of total.

8. Attendance Policy

Students are expected to attend ***all*** lectures and exercises. 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 (https://emergency.buffalo.edu/) for cancellations/delays due to weather or other unforeseen events.

9. Academic Integrity

Academic integrity is a fundamental university value and critical to the learning process. 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. It is your responsibility as a student to complete your work in an honest fashion, upholding the expectations your individual instructors have for you in this regard. The ultimate goal is to ensure that you learn the content in your courses in accordance with UB's academic integrity principles, regardless of whether instruction is in-person or remote. Thank you for upholding your own personal integrity and ensuring UB's tradition of academic excellence. See Graduate Academic Integrity Policy and Graduate Academic Integrity Procedures.

Specific for this course: If the class is attended by more than 2 students, 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 in 60 Capen Hall, 716-645-2608 and also the instructor of this course during the first week of class. The office will provide you with information and review appropriate arrangements for reasonable accommodations, which can be found on the web at: http://www.buffalo.edu/studentlife/who-we-are/departments/accessibility.html.

12. Protection of course material

All materials prepared and/or assigned by me for this course are for the students' educational benefit. Other than for permitted collaborative work, students may not photograph, record, reproduce, transmit, distribute, upload, sell or exchange course materials, without my prior written permission. "Course materials" include, but are not limited to, all instructor-prepared and assigned materials, such as lectures; lecture notes; discussion prompts; study aids; tests and assignments; and presentation materials such as PowerPoint slides, or transparencies; and course packets or handouts. Public distribution of such materials may also constitute copyright infringement in violation of federal or state law. Students who violate this policy will be required to complete an educational sanction about the value of intellectual property. More serious and/or repeat violations of this policy may be treated as acts of "academic dishonesty" and/or subject a student to disciplinary charges under the "Student Code of Conduct." (https://www.buffalo.edu/studentlife/life-on-campus/community/rules.html)

13. University Support Services

Students are often unaware of university support services available to them. 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.
Other vital support for graduate students include counseling services and sexual violence resources. A support service section of your syllabus might include information about those. Feel free to add the following text in your syllabus as you see fit.

counseling service
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 issues you may experience. You learn can more about these programs and services by contacting:

Counseling Services
120 Richmond Quad (North Campus), 716-645-2720
202 Michael Hall (South Campus), 716-829-5800
https://www.buffalo.edu/studentlife/who-we-are/departments/counseling.html

Health Services
Michael Hall (South Campus), 716-829-3316
https://www.buffalo.edu/studentlife/who-we-are/departments/health.html

Office of Health Promotion
114 Student Union (North Campus), 716-645-2837
https://www.buffalo.edu/studentlife/who-we-are/departments/health-promotion.html .

sexual violence
UB is committed to providing a safe learning 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.
Please be aware UB faculty are mandated to report violence or harassment on the basis of sex or gender. This means that if you tell me about a situation, I will need to report it to the Office of Equity, Diversity and Inclusion. You will still have options about how the situation will be handled, including whether or not you wish to pursue a formal complaint. Please know that if you do not wish to have UB proceed with an investigation, your request will be honored unless UB's failure to act does not adequately mitigate the risk of harm to you or other members of the university community. You also have the option of speaking with trained counselors who can maintain

complete confidentiality. UB's Options for Confidentially Disclosing Sexual Violence provides a full explanation of the resources available, as well as contact information. You may call UB's Office of Equity, Diversity and Inclusion at 716-645-2266 for more information, and you have the option of calling that office anonymously if you would prefer not to disclose your identity.

14. Technology Recommendations

To effectively participate in this course, regardless of mode of instruction, the university recommends you have access to a Windows or Mac computer with webcam and broadband. Your best opportunity for success in the blended UB course delivery environment (in-person, hybrid and remote) will require these minimum capabilities.

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. Guarino, N., D. Oberle, and S. Staab, *What Is an Ontology?*, in *Handbook on Ontologies*, S. Staab and R. Studer, Editors. 2009, Springer Berlin Heidelberg: Berlin, Heidelberg. p. 1-17.
3. Rector, A., et al., *On beyond Gruber: "Ontologies" in today's biomedical information systems and the limits of OWL.* J Biomed Inform, 2019. **100s**: p. 100002.
4. Smith, B., et al., *Relations in biomedical ontologies.* Genome Biol, 2005. **6**(5): p. R46.
5. Ceusters, W. and B. Smith, *Tracking referents in electronic health records.* Stud Health Technol Inform, 2005. **116**: p. 71-6.
6. Ceusters, W., *The Place of Referent Tracking in Biomedical Informatics*, in *Terminology, Ontology and their Implementations*, P.L. Elkin, Editor. 2023, Springer International Publishing: Cham. p. 171-218.
7. Scheuermann, R.H., W. Ceusters, and B. Smith, *Toward an ontological treatment of disease and diagnosis.* Summit Transl Bioinform, 2009. **2009**: p. 116-20.
8. Smith, B. and W. Ceusters. *Aboutness: Towards foundations for the information artifact ontology*. in *CEUR Workshop Proceedings*. 2015.
9. 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.
10. Amith, M., et al., *Assessing the practice of biomedical ontology evaluation: Gaps and opportunities.* J Biomed Inform, 2018. **80**: p. 1-13.
11. Ceusters, W., *Towards a realism-based metric for quality assurance in ontology matching*. Frontiers in Artificial Intelligence and Applications. Vol. 150. 2006. 321-332.
12. Pengput, A. and W. Ceusters, *Setting the Scene to Link SNOMED CT to Realism-Based Ontologies* in *19th World Congress on Medical and Health Informatics (MEDINFO)*. Sydney, Australia.