Ontology-driven formal conceptual data modelling for biological data analysis

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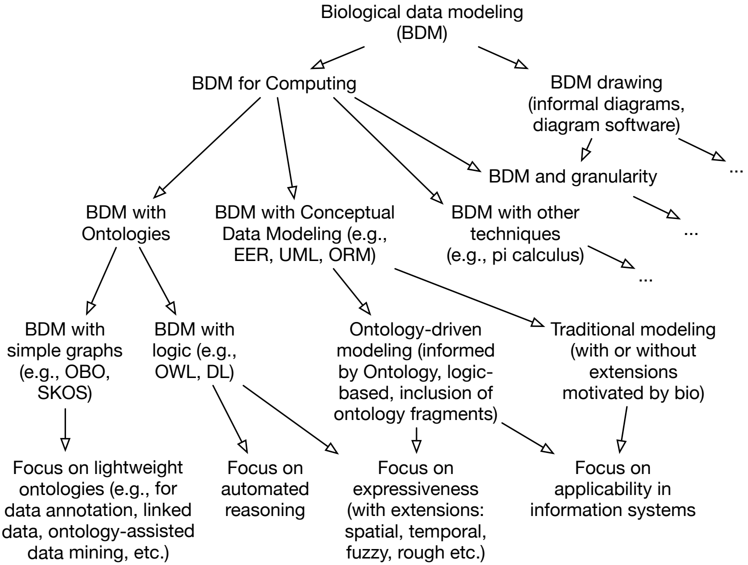
Masters Artificial Intelligence spring School 2011 (MAIS'11)
University of KwaZulu-Natal, 26-30 September 2011

Talk based on the forthcoming book chapter in *Biological Knowledge Discovery Handbook: Preprocessing, Mining and Postprocessing of Biological Data*, Mourad Elloumi and Albert Y. Zomaya (Eds.). Wiley.


Biological data modeling (BDM)
BDM for Computing
BDM drawing (informal diagrams, diagram software)
BDM and granularity
...
BDM with Ontologies
BDM with Conceptual Data Modeling (e.g., EER, UML, ORM)
BDM with other techniques (e.g., pi calculus)
...
...
BDM with simple graphs (e.g., OBO, SKOS)
BDM with logic (e.g., OWL, DL)
Ontology-driven modeling (informed by Ontology, logic-based, inclusion of ontology fragments)
Traditional modeling (with or without extensions motivated by bio)
Focus on lightweight ontologies (e.g., for data annotation, linked data, ontology-assisted data mining, etc.)
Focus on automated reasoning
Focus on expressiveness (with extensions: spatial, temporal, fuzzy, rough etc.)
Focus on applicability in information systems

Motivation

- Scope: ontology-driven formal conceptual data modelling
- Audience (here at MAIS): I assume you know the motivations for conceptual data modelling and ontologies
  - Conceptual model for an *implementation*-independent view
  - Ontology for an *application*-independent view
- What we will look at:
  - Formal conceptual data modelling
  - Ontology-driven conceptual data modelling
- Both take a scientific approach to improving the quality of conceptual data models (hence, also the resulting applications), and facilitate use and reuse (interoperability)

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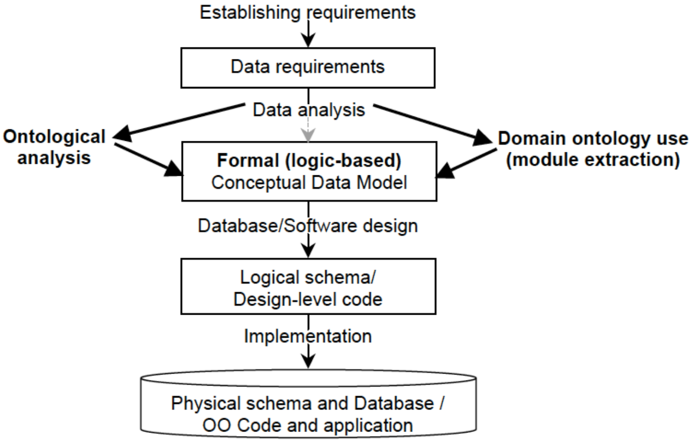
Establishing requirements
Data requirements
Data analysis
Ontological analysis
Domain ontology use (module extraction)
Formal (logic-based) Conceptual Data Model
Database/Software design
Logical schema/ Design-level code
Implementation
Physical schema and Database / OO Code and application

CDM for biological data analysis

- Many one-off bioinformatics tools (perl scripts etc.) and boutique databases
- Use of conceptual data modeling in bioinformatics limited; e.g., [BBP02, CKN$^+$10, EJF07, Kee03, PLC$^+$10, SZ05]
- Mainly EER and various type of UML diagrams
- Neither a link with ontologies (except for [EGOMA06]) nor a formal approach

Ontology-driven CDM for biological data analysis

- More precise and correct representations for correct (automated) inferences and biological knowledge discovery, surpassing human capacity (e.g., [WSH07, KRM07])
- Better data management, hence, way to make (better) use of the 'write-only' databases and 'data silos' (e.g., [CKN$^+$10])
- Reduce redundancy in scientific experiments (e.g., [MBSJ08])
- Ontological guidance for recurring modelling issues (e.g., [Kee03, AGK08, KA08, EJF07])
- Avoid adding to the pile of one-off tools
- Reusability of the information represented at the conceptual layer

1. DLs for CDM
   - $\mathcal{DLR}_{ifd}$ syntax and semantics
   - $\mathcal{CM}_{com}$
2. Extended CDM
   - Ontology-driven modeling
   - Very expressive languages
3. Automated reasoning
4. Conclusions

Introduction

Q: Which language features are 'essential' for biological data modeling?

Q: Does it make any difference which conceptual data modeling language we use for biological data modeling?

⇒ Some claim so, and different languages are used (EER, UML Class diagrams, UML Activity Diagrams, and UML Sequence Diagrams, ORM)

Q: What is the greatest common denominator (or core) of the industry-grade conceptual data modeling languages?

⇒ Compare ER, EER, UML class diagrams, ORM, and ORM2 and identify greatest common denominator. [Kee08]

- (Extends and refines [CDGL+98, CLN98, CLN99, ACK+07, Kee09])
- $\mathcal{DLR}_{ifd}$ used to formally define the generic common conceptual data modeling language $\mathcal{CM}_{com}$, i.e., with syntax and (model-theoretic) semantics, and a mapping between the two

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The base language: $\mathcal{DLR}$

Take atomic relations (**P**) and atomic concepts A as the basic elements of $\mathcal{DLR}$, which allows us to construct arbitrary relations (arity ≥ 2) and arbitrary concepts according to the syntax:

$\mathbf{R} \longrightarrow \top_n | \; \mathbf{P} \; | \; (\$i/n : C) \; | \; \neg \mathbf{R} \; | \; \mathbf{R}_1 \sqcap \mathbf{R}_2$

$C \longrightarrow \top_1 | \; A \; | \; \neg C \; | \; C_1 \sqcap C_2 \; | \; \exists[\$i]\mathbf{R} \; | \; \leq k[\$i]\mathbf{R}$

i denotes a component of a relation; if components are not named, then integer numbers between 1 and n_{max} are used, where n is the arity of the relation. Only relations of the same arity can be combined to form expressions of type $\mathbf{R}_1 \sqcap \mathbf{R}_2$, and $i \leq n$

The base language: $\mathcal{DLR}$

The model-theoretic semantics of $\mathcal{DLR}$ is specified through the usual notion of interpretation, where $\mathcal{I}= (\Delta^{\mathcal{I}}, \cdot^{\mathcal{I}})$, and the interpretation function $\cdot^{\mathcal{I}}$ assigns to each concept C a subset $C^{\mathcal{I}}$ of $\Delta^{\mathcal{I}}$ and to each n-ary $\mathbf{R}$ a subset $\mathbf{R}^{\mathcal{I}}$ of $(\Delta^{\mathcal{I}})^n$, such that the conditions are satisfied following:

$$\begin{array}{ll}
\top_n^{\mathcal{I}} \subseteq (\Delta^{\mathcal{I}})^n & (\mathbf{R}_1 \sqcap \mathbf{R}_2)^{\mathcal{I}} = \mathbf{R}_1^{\mathcal{I}} \cap \mathbf{R}_2^{\mathcal{I}} \\
\mathbf{P}^{\mathcal{I}} \subseteq \top_n^{\mathcal{I}} & (\neg C)^{\mathcal{I}} = \Delta^{\mathcal{I}} \setminus C^{\mathcal{I}} \\
(\neg \mathbf{R})^{\mathcal{I}} = \top_n^{\mathcal{I}} \setminus \mathbf{R}^{\mathcal{I}} & (C_1 \sqcap C_2)^{\mathcal{I}} = C_1^{\mathcal{I}} \cap C_2^{\mathcal{I}} \\
A^{\mathcal{I}} \subseteq \Delta^{\mathcal{I}} & (\$i/n : C)^{\mathcal{I}} = \{(d_1, ..., d_n) \in \top_n^{\mathcal{I}} | d_i \in C^{\mathcal{I}}\} \\
\top_1^{\mathcal{I}} = \Delta^{\mathcal{I}} & (\exists[\$i]\mathbf{R})^{\mathcal{I}} = \{d \in \Delta^{\mathcal{I}} | \exists (d_1, ..., d_n) \in \mathbf{R}^{\mathcal{I}}.d_i = d\} \\
 & (\leq k[\$i]\mathbf{R})^{\mathcal{I}} = \{d \in \Delta^{\mathcal{I}} || \{(d_1, ..., d_n) \in \mathbf{R}_1^{\mathcal{I}} | d_i = d|\} \leq k\}
\end{array}$$

The base language: $\mathcal{DLR}$

A knowledge base is a finite set $\mathcal{KB}$ of $\mathcal{DLR}$ (or $\mathcal{DLR}_{ifd}$) axioms of the form $C_1 \sqsubseteq C_2$ and $R_1 \sqsubseteq R_2$.

An interpretation $\mathcal{I}$ satisfies $C_1 \sqsubseteq C_2$ ($R_1 \sqsubseteq R_2$) if and only if the interpretation of C_1 (R_1) is included in the interpretation of C_2 (R_2), i.e. $C_1^{\mathcal{I}(t)} \subseteq C_2^{\mathcal{I}(t)}$ ($R_1^{\mathcal{I}(t)} \subseteq R_2^{\mathcal{I}(t)}$).

$\top_1$ denotes the interpretation domain, $\top_n$ for $n \geq 1$ denotes a subset of the n-cartesian product of the domain, which covers all introduced n-ary relations.

($\$i/n : C$) denotes all tuples in $\top_n$ that have an instance of C as their i-th component.

$\mathcal{DLR}_{ifd}$

- $\mathcal{DLR}_{ifd}$ has two additional constructs compared to $\mathcal{DLR}$:
  - identification assertions on a concept C, which has the form $(\textbf{id}\ C[i_1]R_1, ..., [i_h]R_h)$, where each R_j is a relation and each i_j denotes one component of R_j.
  - Non-unary functional dependency assertions on a relation R, which has the form $(\textbf{fd}\ R\ i_1, ..., i_h \rightarrow j)$, where $h \geq 2$, and $i_1, ..., i_h, j$ denote components of R
- Syntax and semantics as for $\mathcal{DLR}$

$\mathcal{CM}_{com}$ syntax

Definition 1 (Conceptual Data Model $\mathcal{CM}_{com}$ syntax)

A $\mathcal{CM}_{com}$ conceptual data model is a tuple $\Sigma = (\mathcal{L}, \text{REL}, \text{ATT}, \text{CARD}_R, \text{CARD}_A, \text{ISA}_C, \text{ISA}_R, \text{ISA}_U, \text{DISJ}_C, \text{COVER}_C, \text{DISJ}_R, \text{KEY}, \text{EXTK}, \text{FD}, \text{OBJ}, \text{REX}, \text{RDM})$ such that:

- $\mathcal{L}$ is a finite alphabet partitioned into the sets: $\mathcal{C}$ (*class* symbols), $\mathcal{A}$ (*attribute* symbols), $\mathcal{R}$ (*relationship* symbols), $\mathcal{U}$ (*role* symbols), and $\mathcal{D}$ (*domain* symbols); the tuple $(\mathcal{C}, \mathcal{A}, \mathcal{R}, \mathcal{U}, \mathcal{D})$ is the *signature* of the conceptual data model Σ.
- REL is a function that maps a relationship symbol in $\mathcal{R}$ to an $\mathcal{U}$-labeled tuple over $\mathcal{C}$, $\text{REL}(R) = \langle U_1 : C_1, \ldots, U_k : C_k \rangle$, and k is the *arity* of R.
- ...

$\mathcal{CM}_{com}$ semantics

Definition 2 ($\mathcal{CM}_{com}$ Semantics)

Let Σ be a $\mathcal{CM}_{com}$ conceptual data model. An *interpretation* for the conceptual model Σ is a tuple $\mathcal{B} = (\Delta^{\mathcal{B}} \cup \Delta^{\mathcal{B}}_D, \cdot^{\mathcal{B}})$, such that:

- $\Delta^{\mathcal{B}}$ is a nonempty set of abstract objects disjoint from $\Delta^{\mathcal{B}}_D$;
- $\Delta^{\mathcal{B}}_D = \bigcup_{D_i \in \mathcal{D}} \Delta^{\mathcal{B}}_{D_i}$ is the set of basic domain values used in Σ; and
- $\cdot^{\mathcal{B}}$ is a function that maps:
  - Every basic domain symbol $D \in \mathcal{D}$ into a set $D^{\mathcal{B}} = \Delta^{\mathcal{B}}_{D_i}$.
  - ...
  - Every attribute $A \in \mathcal{A}$ to a set $A^{\mathcal{B}} \subseteq \Delta^{\mathcal{B}} \times \Delta^{\mathcal{B}}_D$, such that, for each $C \in \mathcal{C}$, if $\text{ATT}(C) = \langle A_1 : D_1, \ldots, A_h : D_h \rangle$, then, $o \in C^{\mathcal{B}} \rightarrow (\forall i \in \{1, \ldots, h\}, \exists a_i.$ $\langle o, a_i \rangle \in A_i^{\mathcal{B}} \wedge \forall a_i . \langle o, a_i \rangle \in A_i^{\mathcal{B}} \rightarrow a_i \in \Delta^{\mathcal{B}}_{D_i})$.
- ...

$\mathcal{CM}_{com}$ semantics

Definition 3 (Mapping $\mathcal{CM}_{com}$ into $\mathcal{DLR}_{ifd}$)

Let $\Sigma = (\mathcal{L}, \mathrm{REL}, \mathrm{ATT}, \mathrm{CARD}_R, \mathrm{CARD}_A, \mathrm{ISA}_C, \mathrm{ISA}_R, \mathrm{ISA}_U, \mathrm{DISJ}_C, \mathrm{COVER}_C, \mathrm{DISJ}_R, \mathrm{KEY}, \mathrm{EXTK}, \mathrm{FD}, \mathrm{OBJ}, \mathrm{REX}, \mathrm{RDM})$ be a $\mathcal{CM}_{com}$ conceptual data model. The $\mathcal{DLR}_{ifd}$ knowledge base, $\mathcal{K}$, mapping Σ is as follows.

- For each $A \in \mathcal{A}$, then, $A \sqsubseteq \mathtt{From}:\top \sqcap \mathtt{To}:\top \in \mathcal{K}$;
- If $C_1 \ \mathrm{ISA}_C \ C_2 \in \Sigma$, then, $C_1 \sqsubseteq C_2 \in \mathcal{K}$;
- If $R_1 \ \mathrm{ISA}_R \ R_2 \in \Sigma$, then, $R_1 \sqsubseteq R_2 \in \mathcal{K}$;
- If $U_1 \ \mathrm{ISA}_U \ U_2 \in \Sigma$, then $\mathcal{K}$ contains: $[U_1]R_1 \sqsubseteq [U_2]R_2$; $R_1 \sqsubseteq \neg R_2$;
- If $\mathrm{REL}(R) = \{U_1 : C_1, \ldots, U_k : C_k\} \in \Sigma$, then $R \sqsubseteq U_1 : C_1 \sqcap \ldots \sqcap U_k : C_k \in \mathcal{K}$;
- ...

Example: mapping to icons

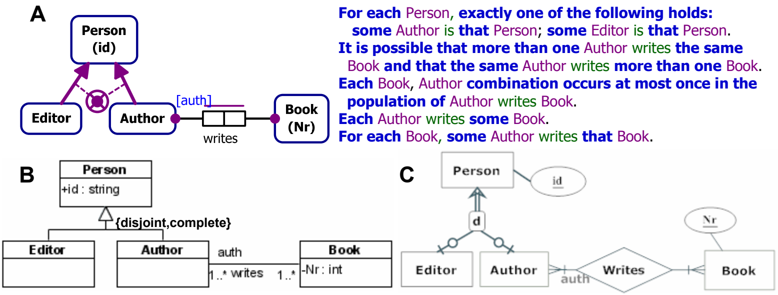

Figure: Examples of graphical syntax for $\mathcal{CM}_{com}$with ORM2 drawn in NORMA (A), UML class diagram drawn in VP-UML (B), and EER drawn with SmartDraw (C).

Example: mapping to icons

- Author ISA Person *(directed arrow in UML, EER, ORM2)*
  CARD(Author, Writes, auth) = $(1, n)$
  (1.. in UML, craw's feet and line in EER, blob and line in ORM2)*
  KEY(Person) = id *(underlined* id *in EER,* (id) *in ORM2)*
  {Author, Editor} DISJ Person
  ({disjoint} *in UML, encircled* d *in EER, encircled* X *in ORM2)*
  {Author, Editor} COVER Person
  ({complete} *in UML, open shaft arrow in EER, encircled blob in ORM2)*
- Equivalent representation in $\mathcal{DLR}_{ifd}$ as: Author $\sqsubseteq$ Person (subsumption), Author $\sqsubseteq$ $\exists$[auth]writes (at least one), Author $\sqsubseteq$ $\neg$Editor (disjoint), Person $\sqsubseteq$ Author $\sqcup$ Editor (covering), and Person $\sqsubseteq \exists^{=1}$[From]id, $\top \sqsubseteq \exists^{\leq 1}$[To](id $\sqcap$ [From] : Person) (key)

Example: mapping to icons

- $\texttt{Author}\ \textsc{isa}\ \texttt{Person}$ *(directed arrow in UML, EER, ORM2)*
  $\textsc{card}(\texttt{Author}, \texttt{Writes}, \texttt{auth}) = (1, n)$
  (1.. in UML, craw's feet and line in EER, blob and line in ORM2)*
  $\textsc{key}(\texttt{Person}) = \texttt{id}$ *(underlined* id *in EER,* (id) *in ORM2)*
  $\{\texttt{Author}, \texttt{Editor}\}\ \textsc{disj}\ \texttt{Person}$
  ({disjoint} in UML, encircled d *in EER, encircled* X *in ORM2)*
  $\{\texttt{Author}, \texttt{Editor}\}\ \textsc{cover}\ \texttt{Person}$
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EER, UML, and ORM in terms of $\mathcal{CM}_{com}$

Definition 4

($\mathcal{CM}_{EER}$) A $\mathcal{CM}_{EER}$ conceptual data model is a tuple $\Sigma = (\mathcal{L}, \text{REL}, \text{ATT}, \text{CARD}_R, \text{ISA}_C, \text{DISJ}_C, \text{COVER}_C, \text{KEY}, \text{EXTK})$ adhering to $\mathcal{CM}_{com}$ syntax and semantics.

Definition 5

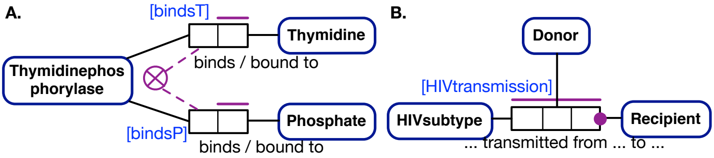
($\mathcal{CM}_{UML}$) A $\mathcal{CM}_{UML}$ conceptual data model is a tuple $\Sigma = (\mathcal{L}, \text{REL}, \text{ATT}, \text{CARD}_R, \text{ISA}_C, \text{ISA}_R, \text{DISJ}_C, \text{COVER}_C, \text{KEY}, \text{EXTK}, \text{FD}, \text{OBJ}, \text{PW})$
adhering to $\mathcal{CM}_{com}$ syntax and semantics, except for the aggregation association PW, with syntax $\text{PW} = \{U_1 : C_1, U_2 : C_2\}$, that has no defined semantics.

EER, UML, and ORM in terms of $\mathcal{CM}_{com}$

Definition 6

A $\mathcal{CM}_{ORM2^-}$ conceptual data model is a tuple
$\Sigma = (\mathcal{L}, \text{REL}, \text{ATT}, \text{CARD}_R, \text{CARD}_A, \text{ISA}_C, \text{ISA}_R, \text{ISA}_U, \text{DISJ}_C, \text{COVER}_C, \text{KEY}, \text{EXTK}, \text{FD}, \text{OBJ}, \text{DISJ}_R, \text{REX}, \text{RDM})$
adhering to $\mathcal{CM}_{com}$ syntax and semantics.

Example: constraints among relations



in $\mathcal{CM}_{com}$:

$\{\texttt{bindsT}, \texttt{bindsP}\}$ REX $\texttt{binds}$

In $\mathcal{DLR}_{ifd}$:

$\texttt{ThymidinePhosphorylase} \sqsubseteq (\exists^{\leq 1}[\texttt{bindsT}]\texttt{binds}_1 \sqcup \exists^{\leq 1}[\texttt{bindsP}]\texttt{binds}_2)$,

$[\texttt{bindsT}]\texttt{binds}_1 \sqsubseteq \neg[\texttt{bindsP}]\texttt{binds}_2$

Three main scenarios

- Provide a solution to a recurring modeling problem, informed by ontology and foundational ontologies (e.g., part-whole relations [KA08, AGK08])
- Use an ontology to generate several conceptual data models (e.g., [EGOMA06, JDM03, SS06])
- Integrate (a section of) an ontology into the conceptual data model that subsequently is converted into data in the database (e.g., KEGG, GO for annotation)

Solution to a recurring modeling problem

- (Re-)Usable components of foundational ontologies (e.g., BFO, DOLCE, GFO, ...)
  - High-level categories
  - Generic relationships (parthood, participation, dependency, constitution, etc.)
- Modeling guidance; e.g., OntoClean, ONTOPARTS
- Informs and refines language features; e.g., relational properties, role/relation-components (positionalism), keys/identification

⇒ Tells how, has justification why

Example: catalysis

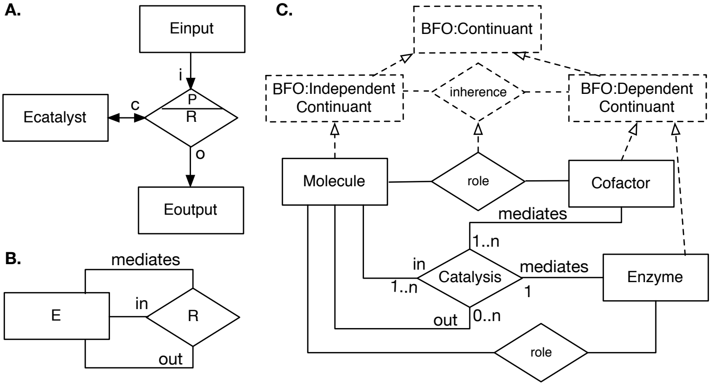
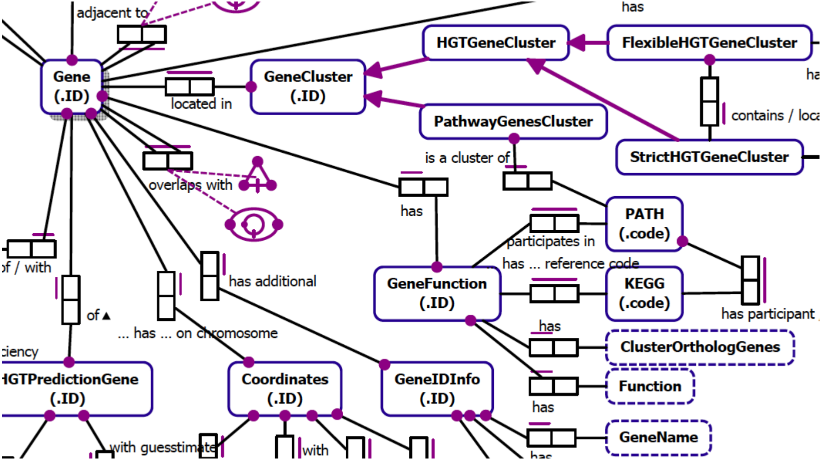
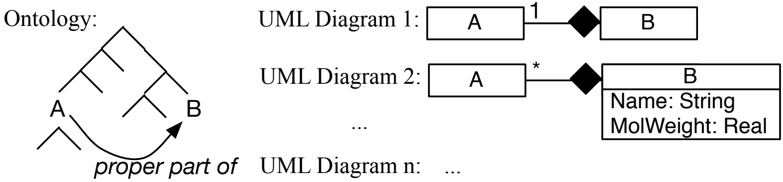


Figure: Static aspects of modeling single processes (catalytic reactions) in EER. A: Elmasri *et al.*'s [EJF07] proposal, with input, output and catalyst molecules; B: The essential roles played; C: An example of a more refined representation of catalysis,

Example: use of scenario 1 and 3



Generation of CDMs from one ontology



Need for language extensions

- Metabolic pathways (temporal)
- Central Dogma, viral infection (temporal)
- Development and transformations (temporal)
- `SmallMolecule` etc (fuzzy)
- 'typical' and default cases (probabilistic)

Temporal: examples

- Use, e.g., $\mathcal{DLR}_{\mathcal{US}}$ and $\mathcal{ER}_{\mathcal{VT}}$, with refinements on relation migration [APS07, KA10]
- $\texttt{hasRole}$ $\textsc{RDex}$ $\texttt{autocatalysis}$

  $\texttt{R}$ $\textsc{RDex}$ $\texttt{R}'$ if and only if $\langle o_1, o_2\rangle \in \texttt{R}^{\mathcal{I}(t)} \rightarrow \exists t' > t.\langle o_1, o_2\rangle \in \texttt{R}'^{\mathcal{I}(t')}$

  $\textsc{rel}(\texttt{hasRole}) = \{\texttt{bearer} : \texttt{RNAmolecule}, \texttt{role} : \texttt{Ribozyme}\}$

  $\textsc{rel}(\texttt{autocatalysis}) = \{\texttt{substrate} : \texttt{RNAmolecule}, \texttt{catalyst} : \texttt{Ribozyme}\}$
- $\texttt{Monocyte}$ $\textsc{dev}$ $\texttt{Macrophage}$

  $\texttt{Monocyte} \sqsubseteq \Diamond^{+}(\texttt{Macrophage} \sqcap \neg\texttt{Monocyte})$
- Viral entry: binding, membrane fusion or detach, viral entry (details in chapter)

Standard reasoning services

- CDM consistency
- Class consistency
- Class subsumption
- Refinement of multiplicities
- (instance classification and retrieval)
- Examples: discovery of new protein phosphatase, quickly finding suitable rubber or pharma molecules

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Querying and other reasoning services

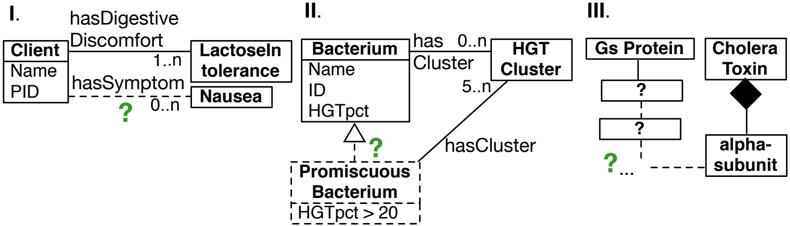

Figure: Graphical depictions of the three query patterns to find 'new' classes or relationships supported by the data; (i): correlation; (ii): hypothesis about existence of subclass PromiscuousBacterium; (iii): path query to check whether the Gs protein somehow relates to the alpha-subunit of the CholeraToxin.

Conclusions and future directions

- Substantiated advantages of ontology-driven formal conceptual data modeling:
  - Formal foundation for UML, EER, ORM, with $\mathcal{CM}_{com}$, which has an equi-satisfiable $\mathcal{DLR}_{ifd}$ knowledge base
  - Ontological guidance to motivate better modeling choices, illustrated with a refinement for representing catalytic reactions
  - Claimed to be 'non-representable' biological knowledge can be represented in $\mathcal{CM}_{com}$ (n-aries, constraints among relationships)
- Language extensions for, a.o., temporal knowledge; demonstrated with related processes in a cascade of reactions
- Automated reasoning services were illustrated for taxonomic classification
- Three different query patterns to find new type-level information

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Outlook

- Ontology-driven formal conceptual data modeling is still a relatively young field
- How to handle incomplete information in hypothesis testing [Kee10]?
- how OntoClean [GW04] ideas can be incorporated in conceptual data modeling methodologies
- Formal link between ontologies and conceptual data models
- Development of CASE tools with both a unifying formalism and an integrated automated reasoner, and multiple language interfaces
- Temporal reasoning beyond $\mathcal{ER}_{\mathcal{VT}}$ and its $\mathcal{DLR}_{\mathcal{US}}$ foundation, principally either as extension to UML Class Diagrams or as formalization of Sequence and Activity Diagrams

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Thank you for your attention