

Ontologies Mapping for the Laboratory Analytics Domain

Key Information

Author Names:

Ian Harrow (Project Manager), Thomas Liener (Paxo author & mapping work) and the Ontologies Mapping Project Team (Funders & Partners below)

Project contact details:

lan.harrow@pistoiaalliance.org

Project website:

https://www.pistoiaalliance.org/projects/ current-projects/ontologies-mapping

Funders (Phase)

BIOVIA 3DS (2,3), GSK (All), Roche (All), Amgen (3), AstraZeneca (3,4), Accenture (3), Bayer (3), Merck & Co (1,2), Novartis (1,2), and Bristol Myers-Squibb (4)

Partners (Phase)

OAEI Organiser, Dr Ernesto Jiménez-Ruiz (2,3,4) EMBL-EBI (3,4), Allotrope Foundation (4), Osthus (All), Eagle Genomics (All), SciBite (All), Linguamatics (All), Novartis (3,4), AbbVie (3,4), Bayer (4) and Elsevier (All)

<u>References</u>

OAEI annual challenge

Ontology Alignment Evaluation Initiative

http://oaei.ontologymatching.org

The Phenotype and disease track has been organised by the Ontology Mapping project over the last four years.

Technical paper from OAEI 2016 authored by the OM project

Ian Harrow, Ernesto Jiménez-Ruiz, Andrea Splendiani, Martin Romacker, Peter Woollard, Scott Markel, Yasmin Alam-Faruque, Martin Koch, James Malone and Arild Waaler (2017)

"Matching disease and phenotype ontologies in the ontology alignment evaluation initiative". *Journal of Biomedical Semantics* 2017 DOI 10.1186/s13326-017-0162-9

Review article authored by the OM project

Ian Harrow, Rama Balakrishnan, Ernesto Jiménez-Ruiz, Simon Jupp, Jane Lomax, Jane Reed, Martin Romacker, Christian Senger, Andrea Splendiani, Jabe Wilson, Peter Woollard

"Ontology mapping for semantically enabled applications" *Drug Discovery Today* 2019 DOI: 10.1016/j.drudis.2019.05.020

Book Recommendation

Jerome Euzenat and Pavel Shvaiko (2013) 2nd ed. Ontology Matching Springer-Verlag DOI: 10.1007/978-3-642-38721-0

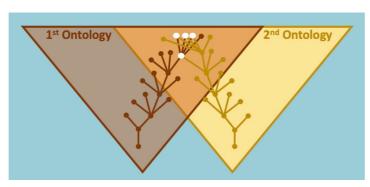
Business Challenge

A growing number of ontologies underpin numerous important applications such as semantic search, data integration, fact extraction and AI/machine learning.

Use of different ontologies within same data domain hampers interoperability and application. This can be solved by mapping between ontologies, as described below.

What are Ontology Mappings?

An Ontology Mapping comprises of pairwise matches between two ontologies. They provide a modular engineering solution to expand coverage at reduced cost of maintenance for building applications.



Summary

- Thomas Liener and Simon Jupp (SPOT group, led by Helen Parkinson at EMBL-EBI) developed the Paxo algorithm for ontology mapping which is openly available on GitHub.
- Mapping "standards" from NCBO BioPortal and the annual OAEI challenge help us to evaluate the quality of the predicted mappings.
- Publication of the review article "Ontology Mapping for Semantic Applications" in Drug Discovery Today.
- Our results demonstrate the successful application of the Paxo mapping algorithm to ontologies in two different domains.
 - o Previously for the phenotype and disease domain
 - $\circ \qquad \text{Here for the laboratory analytics domain (right panel)}$
 - We have demonstrated that the mapping algorithm can be applied to any pair of ontologies hosted by EMBL-EBI.

Plans

- Ideas being explored for 2020 include:
 - Crowd validation of predicted mappings in the OxO repository of ontology mappings, hosted by EMBL-EBI
 - Undertake Ontology Mapping for the clinical domain

Contact Ian Harrow for further info

Selected Ontologies

| Lab analytics domain | Ontology name | Short name |
|----------------------|--|------------|
| Chemistry | Chemical Information Ontology | CHEMINF |
| Chemistry | Physico-Chemical Methods and Properties Ontology | FIX |
| Chemistry | Allotrope Merged Ontology Suite | AFO |
| Chemistry | Chemical Methods Ontology | СНМО |
| Biology | Ontology for Biomedical Investigations | ОВІ |
| Biology | Eagle-I Research Resource Ontology | ERO |
| Biology | Mass Spectrometry Ontology | MS |
| Biology | BioAssay Ontology | BAO |
| Biology | Experimental Factors Ontology | EFO |
| General | National Cancer Institute Thesaurus | NCIT |
| General | Medical Subject Headings | MESH |

• Eleven public ontologies were selected for mapping.

Perceived value of Ontology Mappings

| Ontologies | PVO1 | OBI | 1+2 | ERO | 1+2 | MS | 1+2 | BAO | 1+2 | EFO | 1+2 |
|------------|-------|----------------|-----|------------|-----|------------|-----|-------------|-----|------------|-------------|
| PVO2 | | 23 | | 15 | | 19 | | 26 | | 26 | |
| ERO | 15 | OBI - ERO | 38 | | | | | | | | |
| MS | 19 | OBI - MS | 42 | ERO - MS | 34 | | | | | | $\neg \neg$ |
| BAO | 26 | OBI - BAO | 49 | ERO - BAO | 41 | MS - BAO | 45 | | | | \Box |
| EFO | 26 | OBI - EFO | 49 | ERO - EFO | 41 | MS - EFO | 45 | BAO - EFO | 52 | | \Box |
| MESH | 24 | OBI - MESH | 47 | ERO - MESH | 39 | MS - MESH | 43 | BAO - MESH | 50 | EFO - MESH | 50 |
| NCIT | 25 | OBI - NCIT | 48 | ERO - NCIT | 40 | MS - NCIT | 44 | BAO - NCIT | 51 | EFO - NCIT | 51 |
| Ontologies | PVO1 | CHEMINF | 1+2 | | 1+2 | AFO | 1+2 | СНМО | 1+2 | | |
| PVO2 | 1 401 | 13 | 112 | 11 | 172 | 24 | 112 | 22 | 172 | | |
| | _ | | _ | | + | 24 | _ | 22 | - | | |
| FIX | 11 | CHEMINF - FIX | 24 | | | | | | | | |
| AFO | 24 | CHEMINF - AFO | 37 | FIX - AFO | 35 | | | | | | |
| снмо | 22 | CHEMINF - CHMO | 35 | FIX - CHMO | 33 | AFO - CHMO | 46 | | | | |
| MESH | 24 | CHEMINF - MESH | 37 | FIX - MESH | 35 | AFO - MESH | 48 | CHMO - MESH | 46 | | |
| NCIT | 25 | CHEMINF - NCIT | 38 | FIX - NCIT | 36 | AFO - NCIT | 49 | CHMO - NCIT | 47 | | |
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| Ontologies | PVO1 | OBI | 1+2 | ERO | 1+2 | MS | 1+2 | BAO | 1+2 | EFO | 1+2 |

- The Paxo algorithm generated 54 ontology mappings.
- Each ontology was scored for perceived value by 9 members of the project team, representing different organisations
- Perceived Values (PVs) of each ontology in a mapping pair gave the total score which informed our priorities for evaluation of 13 mappings (below).

Evaluation of selected Ontology Mappings

| Ontology Mapping | Recall % (Silver 3 votes) | Paxo uniques (Silver 3 votes) | Recall % (Silver 2 votes) | Paxo uniques (Silver 2 votes) | Precision % (Uniques, not exact) | |
|---------------------|---------------------------------|--|---------------------------------|--|--|--|
| AFO - CHMO | 99% | 40 | 87% | 24 | 45% | |
| BAO - NCIT | 99% | 206 | 94% | 189 | 62% | |
| EFO - NCIT | 98% | 1,385 | 94% | 1,189 | 95% | |
| OBI - NCIT | 96% | 178 | 90% | 165 | 75% | |
| ERO - NCIT | 96% | 190 | 94% | 181 | 88% | |
| EFO - MESH | 94% | 1,923 | 84% | 712 | 73% | |
| AFO - NCIT | 93% | 203 | 66% | 137 | 68% | |
| MS - NCIT | 92% | 111 | 78% | 93 | 50% | |
| CHMO - NCIT | 89% | 235 | 76% | 221 | 93% | |
| ERO - MESH | 88% | 113 | 68% | 67 | 88% | |
| CHMO - MESH | 86% | 165 | 51% | 116 | 95% | |
| AFO - MESH | 81% | 47 | 52% | 27 | 65% | |
| BAO - MESH | 70% | 100 | 47% | 63 | 70% | |

- Thirteen mappings were selected for evaluation of:
 - o Recall (c.f. silver standard, 3 & 2 vote consensus)
 - Precision (from sampling not exact unique mappings (The evaluation method is detailed in Harrow et al 2017)

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