UZH BIO390

Semantic web, RDF, Ontologies and Knowledge Graphs in biomedical sciences

Ahmad Aghaebrahimian

Zurich University of Applied Sciences
agha@zhaw.ch

Introduction

- Ahmad Aghaebrahimian

- Research Associate at ZHAW

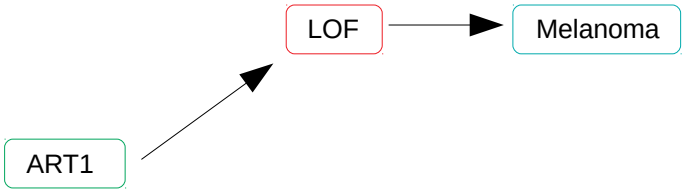
- Ph.D. Computer Sciences focusing on Computational Linguistics

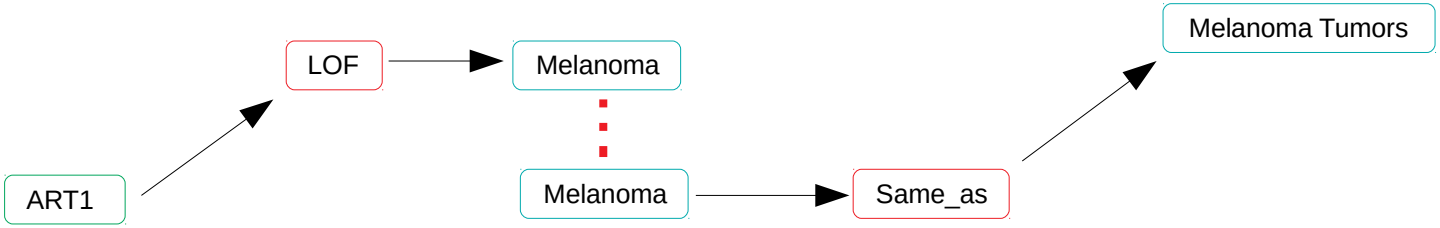
- Area of interests:
  - Machine Learning
  - Deep Neural Networks
  - Biomedical text analytics
  - Natural Language Processing
  - Semantic Web

Email: agha@zhaw.ch

Session Content

- Introduction
- Stack of standards (URI, XML, RDF, SPARQL, OWL, …)
- RDF: Entities and Relationships
- Ontology
- Knowledge graphs


LOF
Melanoma
ART1


Melanoma Tumors
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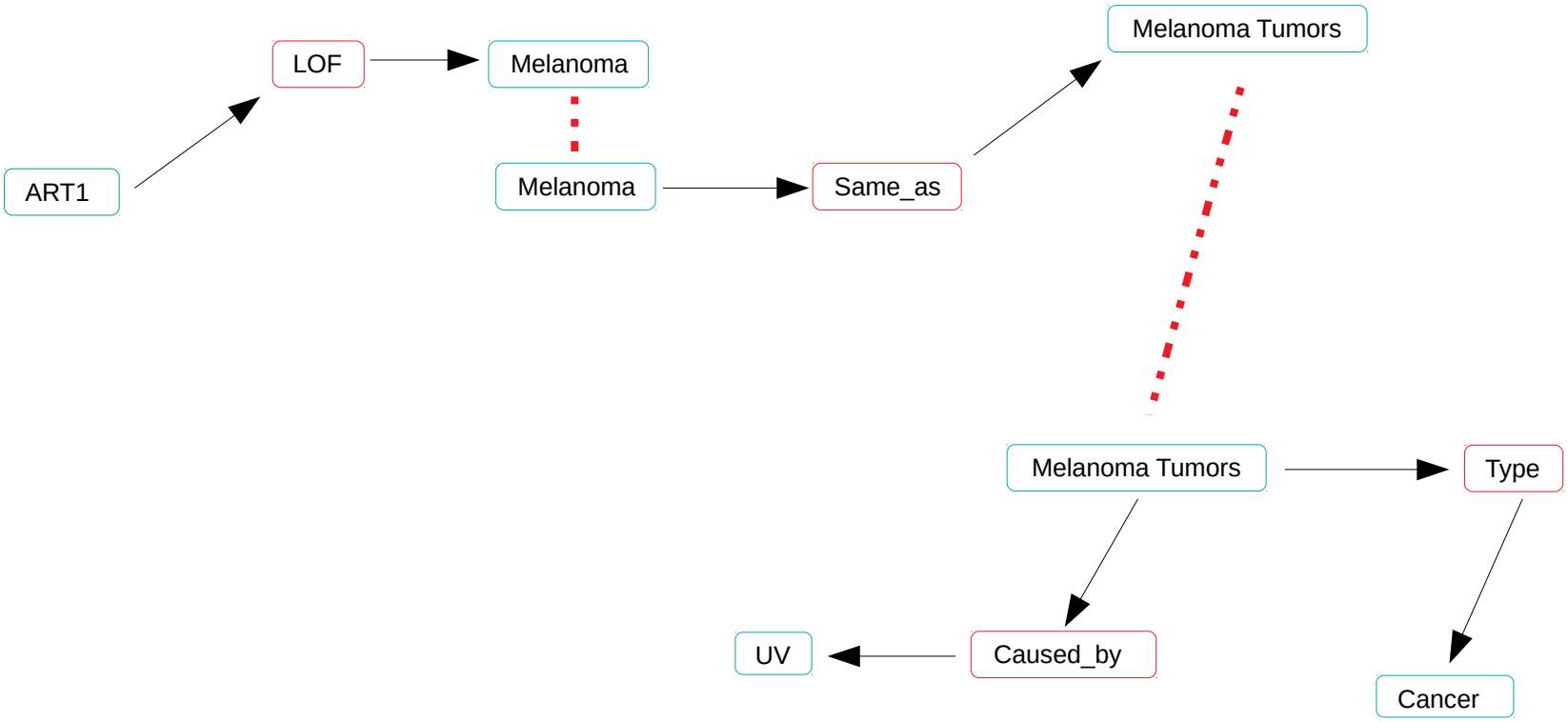
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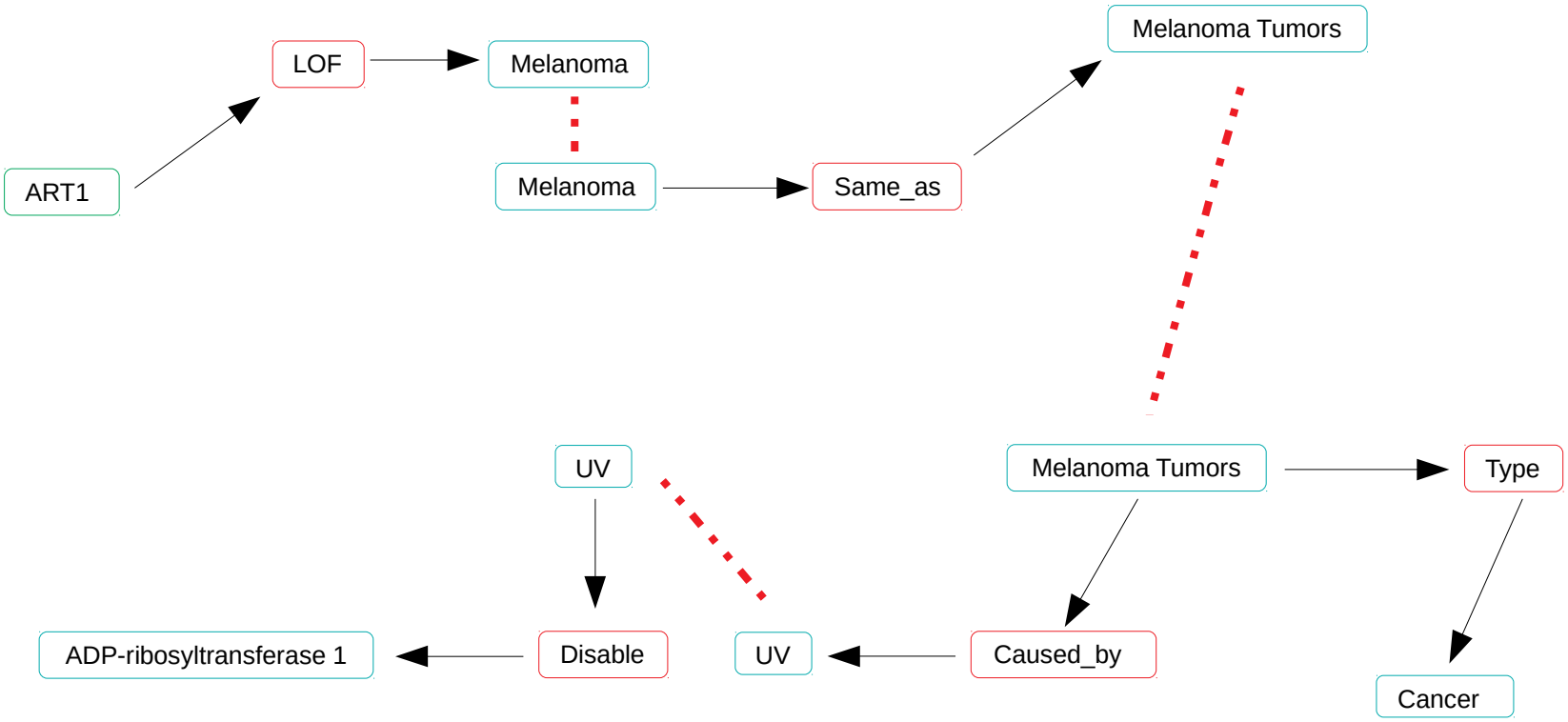
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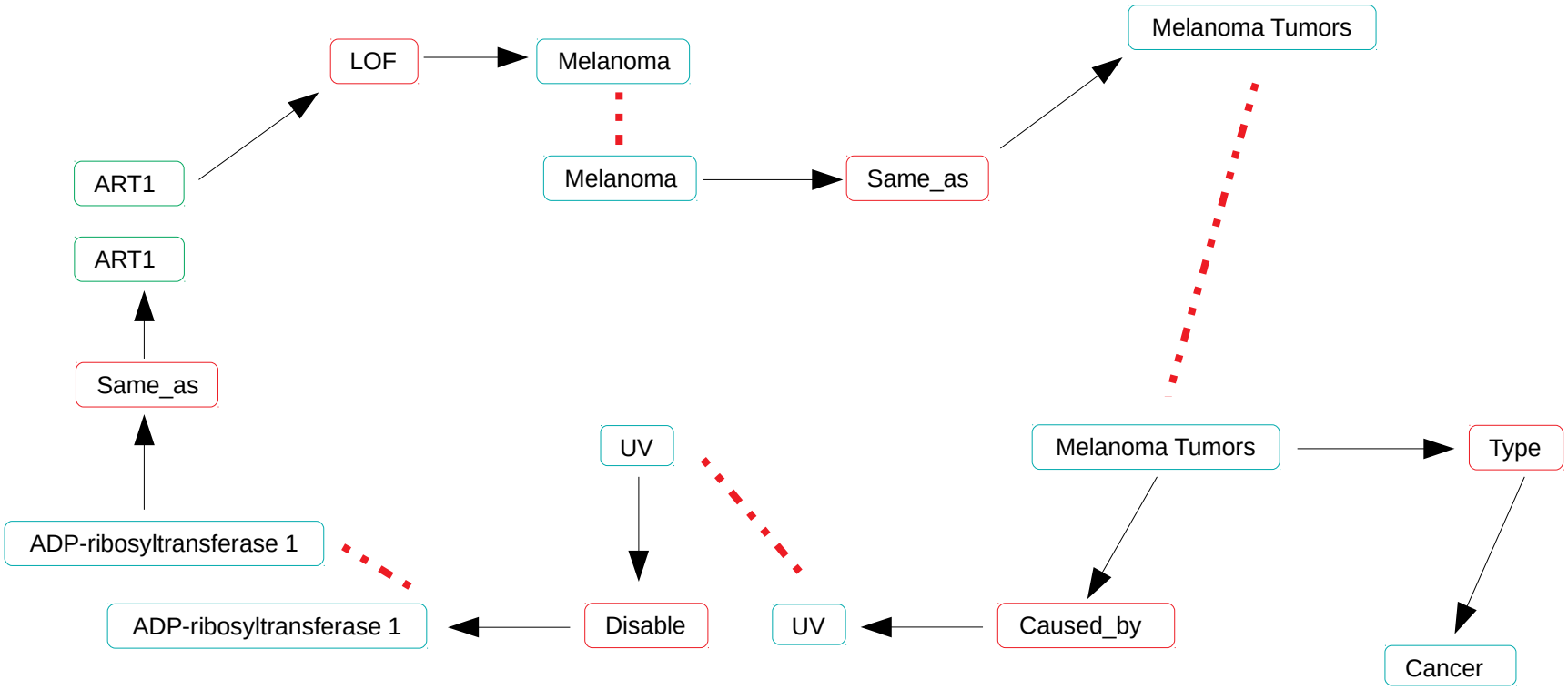
The linked open data

- Linked open data example



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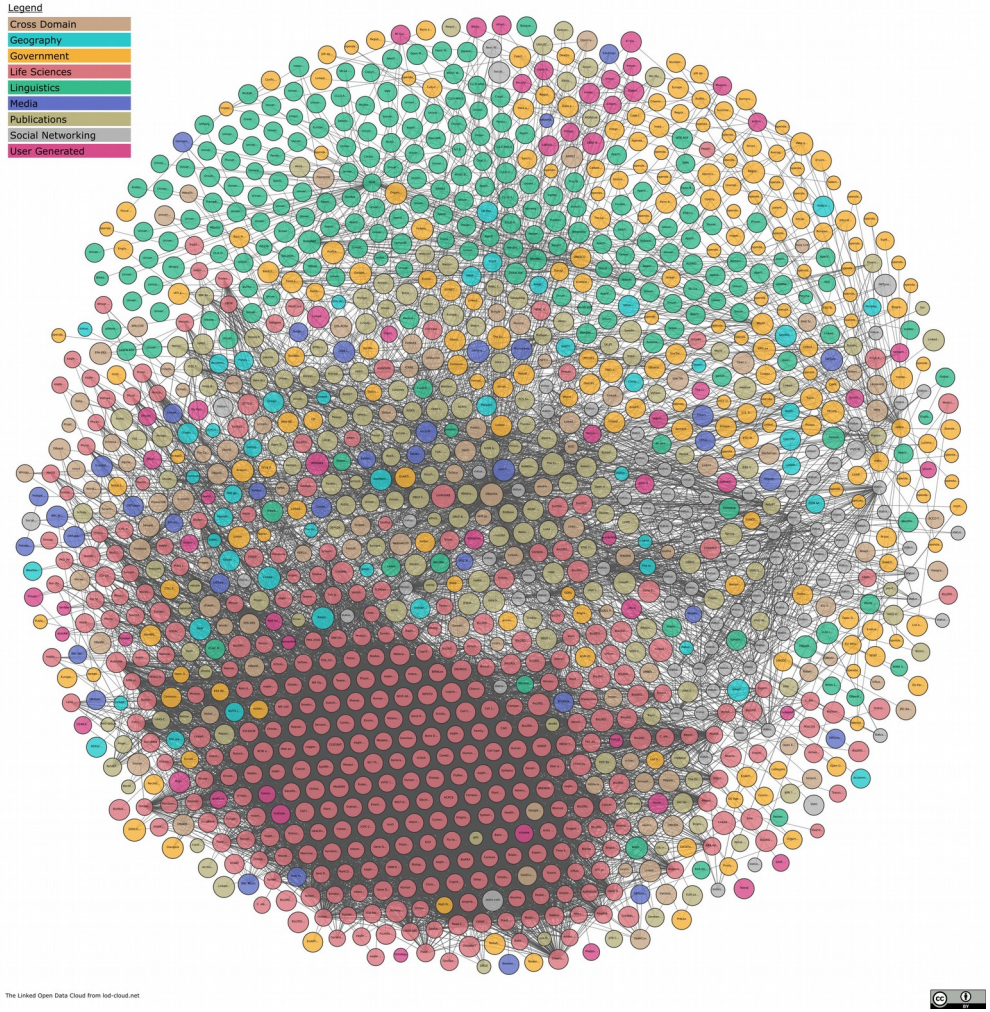
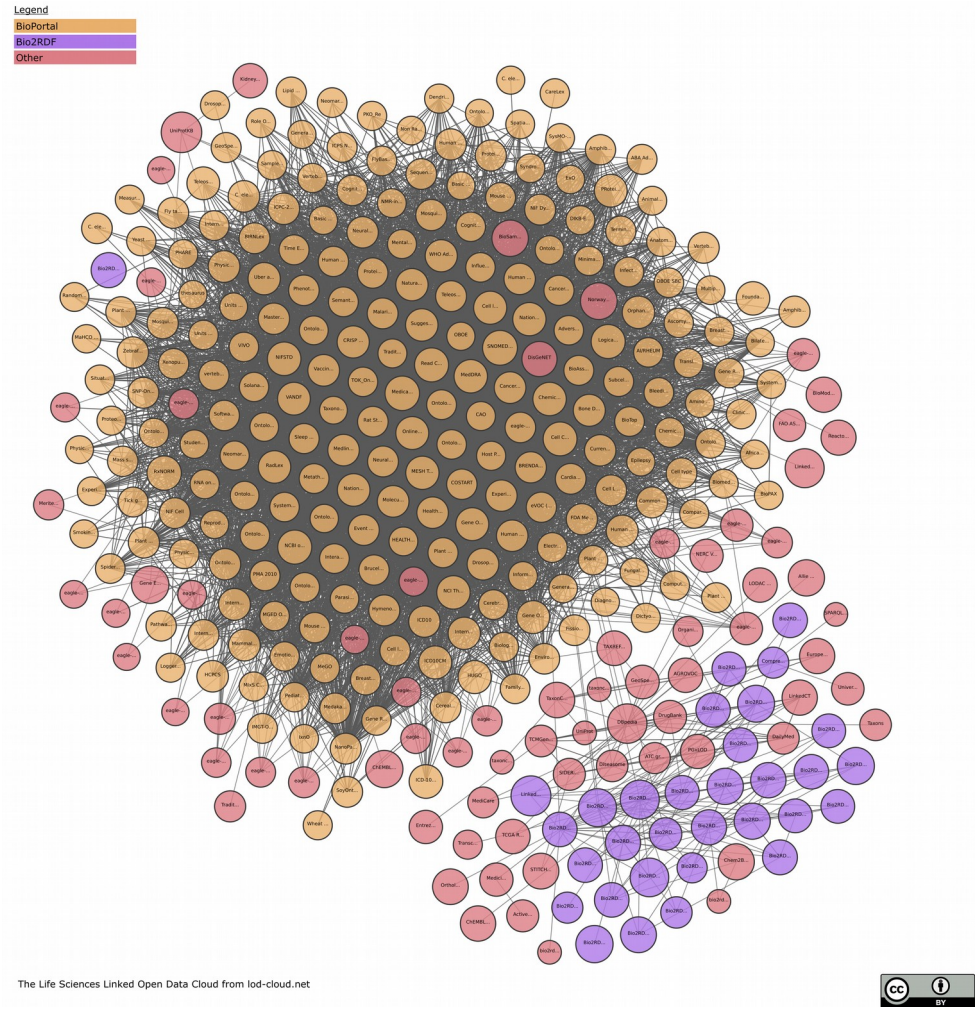


The life sciences data cloud



Basics of the web

- Web structure:
    Server vs. Client

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- Moving from pages to resources
    - Interactive web, Web 2.0 or semantic web

Semantic Web

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Semantic Web (SW) is an extension of the World Wide Web that uses the **Resource Description Framework (RDF)** and **Web Ontology Language (OWL)**, among other standards, to make the Internet machine-readable.

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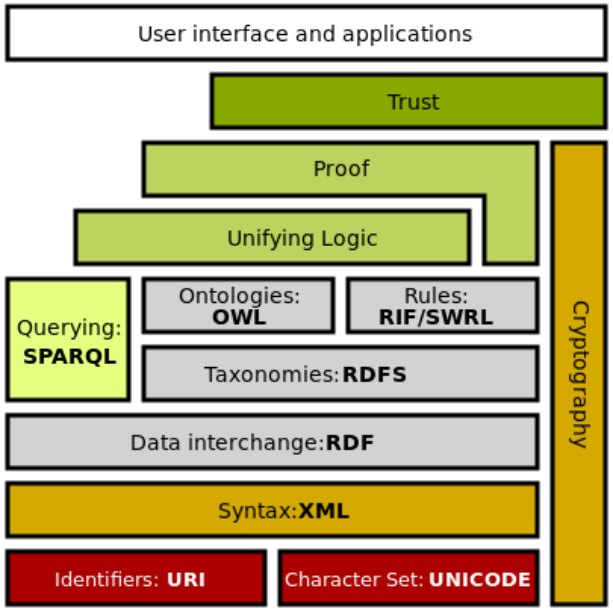

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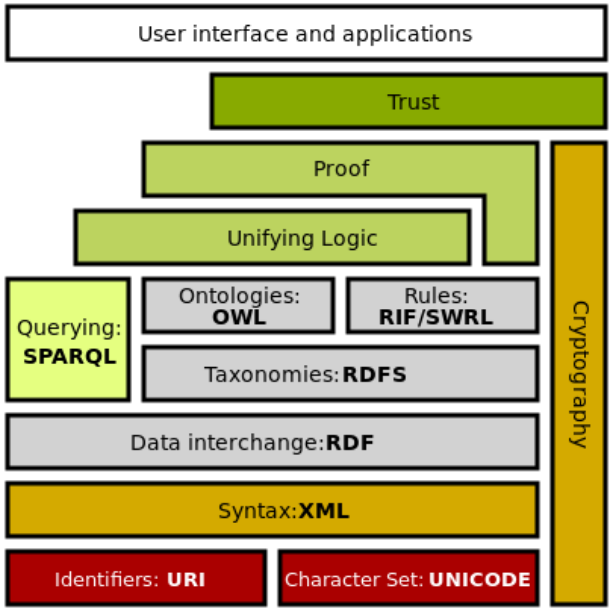

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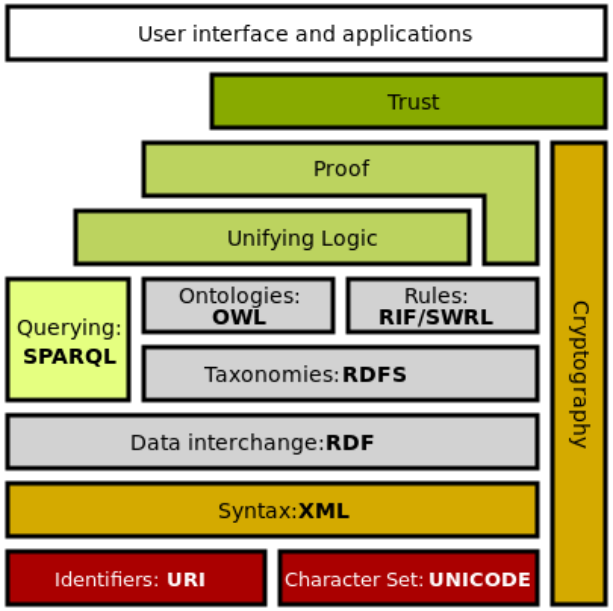

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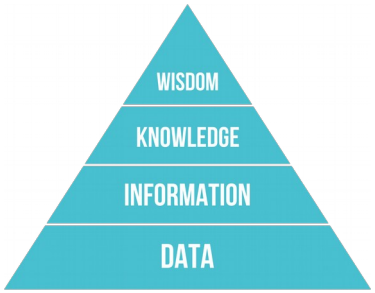

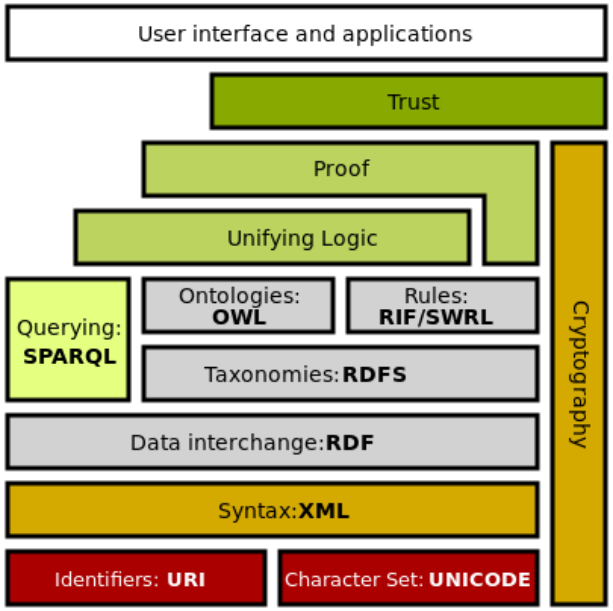

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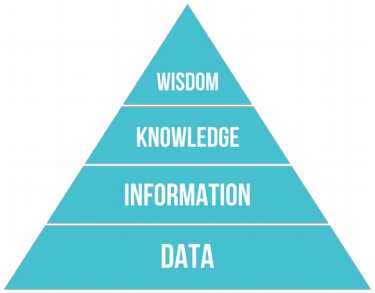

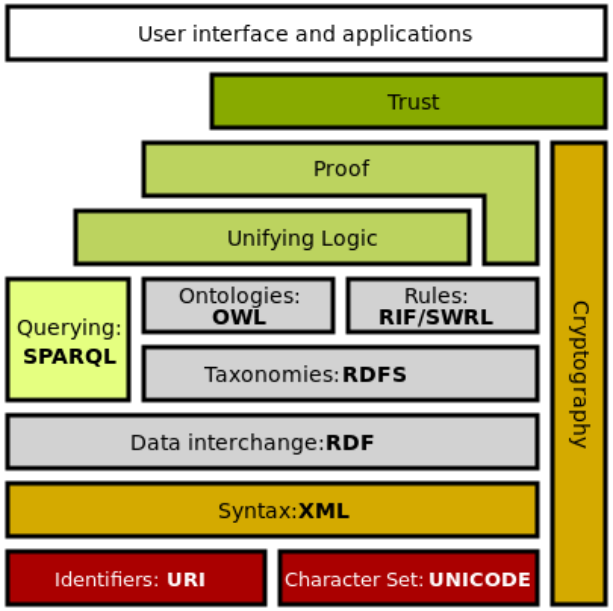

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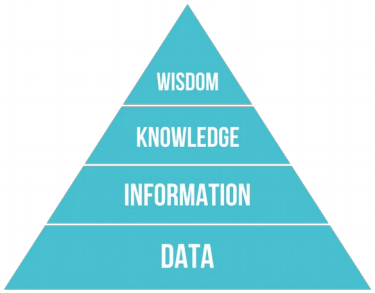
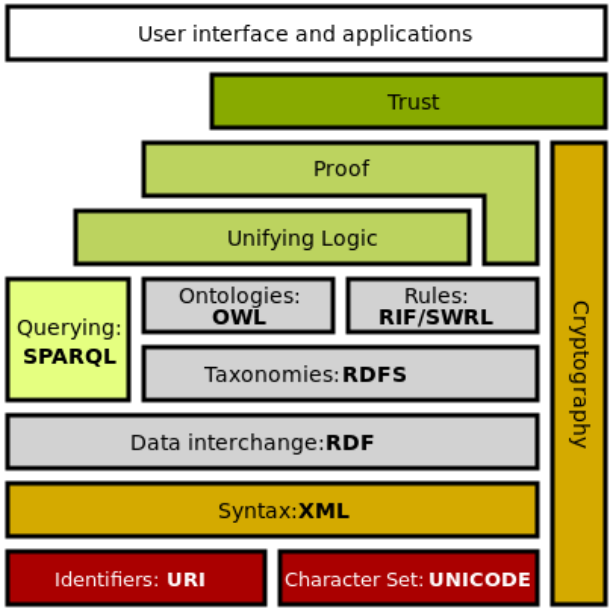




Why?

- Presenting knowledge about data
- Allowing data integration from data silos
- Introduce intelligence to systems

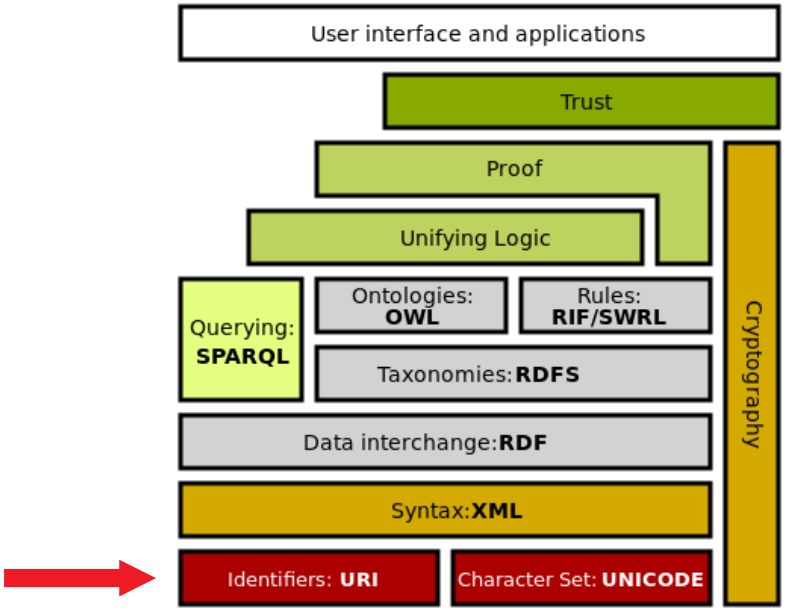
Semantic Web Standards

URI:

What is a Resource?

URL → URI → IRI

Physically located → conceptually identified → conceptually identified in all languages



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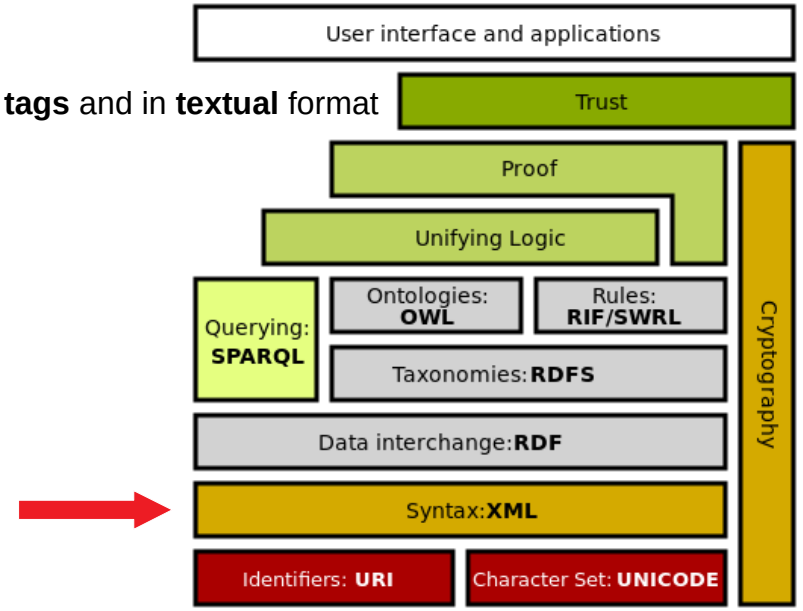
XML:

Open family of languages represent **structured** data using **tags** and in **textual** format

Rules:

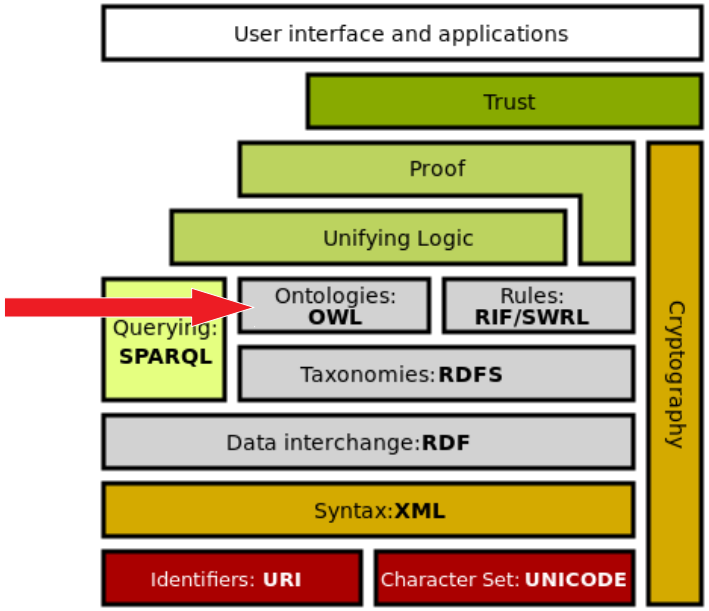
- Only one root <root> </root>
- Opening with closing <Gene></Gene>
- no tag begin with number or xml
- Case sensitive <Gene> != <gene>
- Order matters <Gene> <nucl> </nucl></Gene>
- Tags may have attributes <Gene inherited='true' />



Semantic Web Standards

OWL:

OWL provides a rich vocabulary to add semantics and context and allow reasoning and inference



Ontology

- **Ontology is**
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- **Benefits of an ontology in Biomedical research? (And why they are important)**
  - Data integration
  - Language processing via domain vocabulary
  - Defining the precise meaning of classes
  - Automated processing

Ontology Continued

- **Ontology as a set of:**
  - Definitions
  - Terms and their synonyms
  - Relationships

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- **OBO : ChEBI** Access via : ‘https://github.zhaw.ch/agha/D-Heath’

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[Term]
id: CHEBI:60871
name: selenium(2+)
def: "The selenium ion with two positive charges." []
synonym: "Se(2+)" RELATED [UniProt:]
synonym: "selenium dication" RELATED [ChEBI:]
synonym: "Se2+" RELATED [SUBMITTER:]
synonym: "Se" RELATED FORMULA [ChEBI:]
synonym: "[Se++]" RELATED SMILES [ChEBI:]
synonym: "InChI=1S/Se/q+2" RELATED InChI [ChEBI:]
synonym: "InChIKey=MFSBVGSNNPNWMD-UHFFFAOYSA-N" RELATED InChIKey [ChEBI:]
is_a: CHEBI:60250
is_a: CHEBI:30412

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is_a: CHEBI:36914
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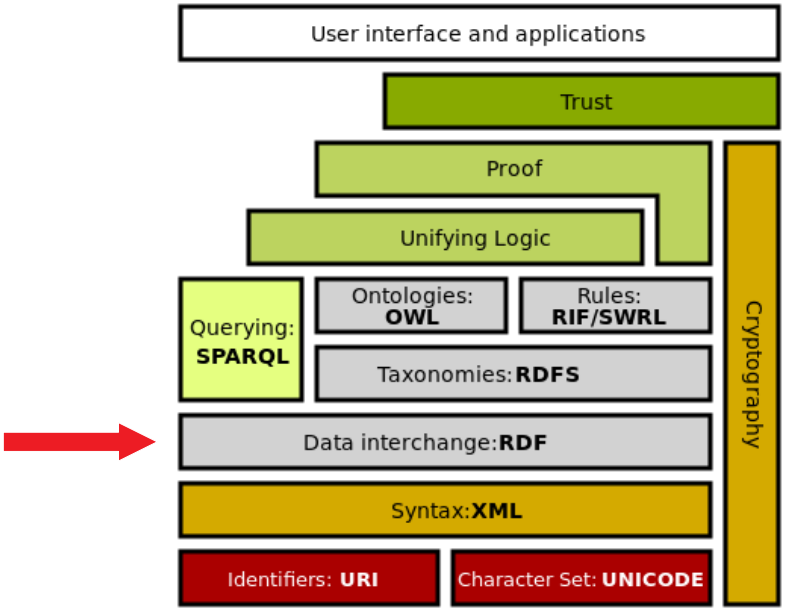
- **UMLS:**
  - Metathesaurus
  - Semantic network
  - Specialized Lexicon

Semantic Web Standards

RDF:

RDF is a **graph-based data model** and the set of **syntax** that allows us to write **description** about the resources on the web and to exchange them. It presents data in the **triple format** and gives it structures and unique identifiers so that data can be easily linked



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Triple structure: (subject, predicate, object)

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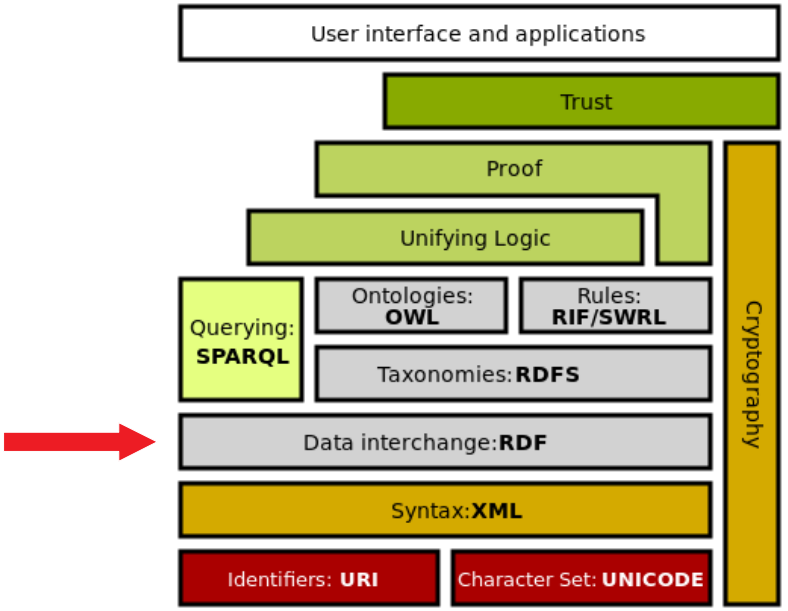
Predicates are labeled

Predicates are directed

RDF is a graph model



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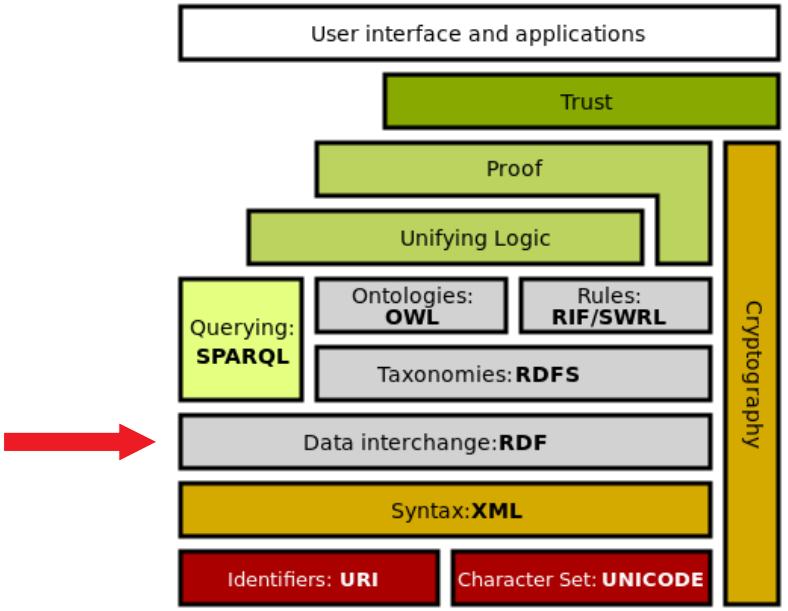
RDF serialization:

XML, N-triple, Turtle, TriG, JSON-LD



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- Storing data in form of triplets: (Subject, Predicate, Object)
  e.g. (ART, LOF, Melanoma_Tumors)
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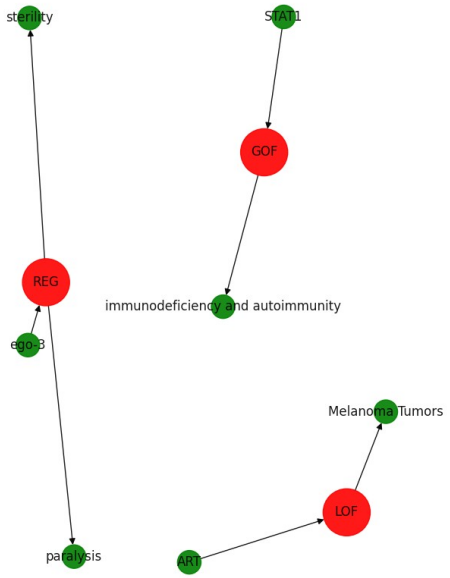

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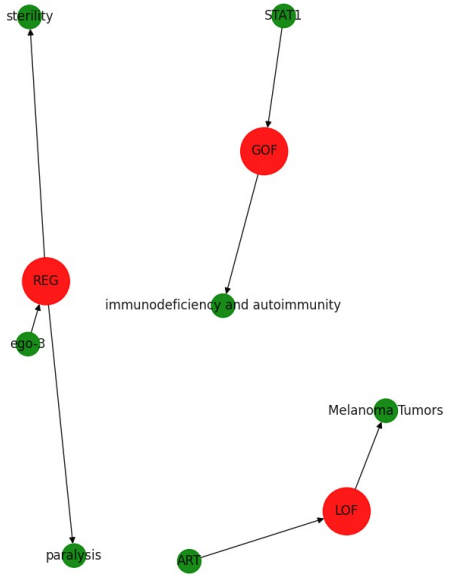

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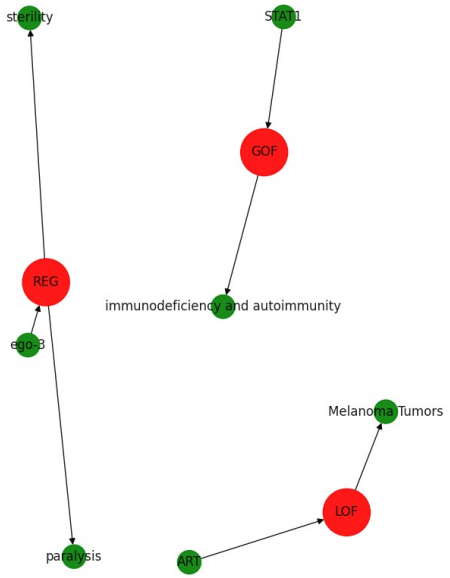



```
SELECT ?gene ?relation
WHERE {
        ?gene ?relation Melanoma_Tumors .
}
```

Subjects, Predicates, Objects

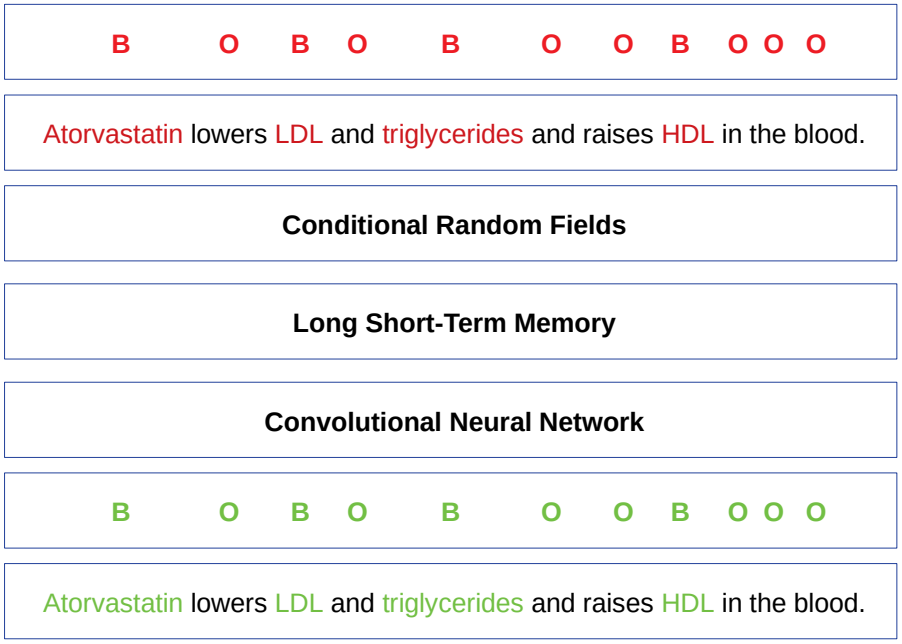
Named Entity

Relationship

NLP components:

- Named Entity Recognition (NER)
- Named Entity Disambiguation (NED)
- Relation Extraction (RE)

Named Entity Recognition (NER)



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$$F1 = 2 \cdot \frac{Precision \cdot Recall}{Precision + Recall}$$

Named Entity Disambiguation (NED)

Atorvastatin lowers LDL and triglycerides and raises HDL in the blood.

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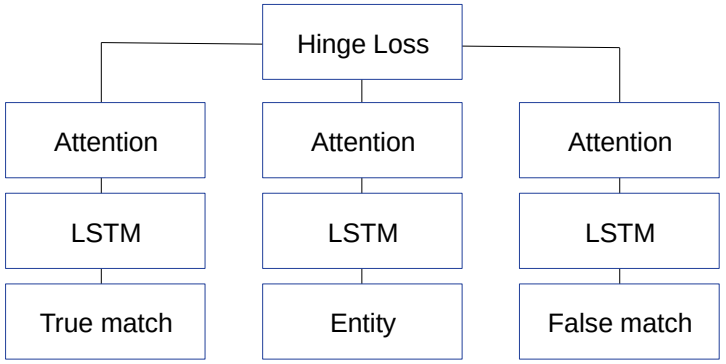

Aghaebrahimian, A., Cieliebak, M.(2020), Named Entity Disambiguation at Scale, **ANNPR**, Winterthur, Switzerland

Relation Extraction (RE)

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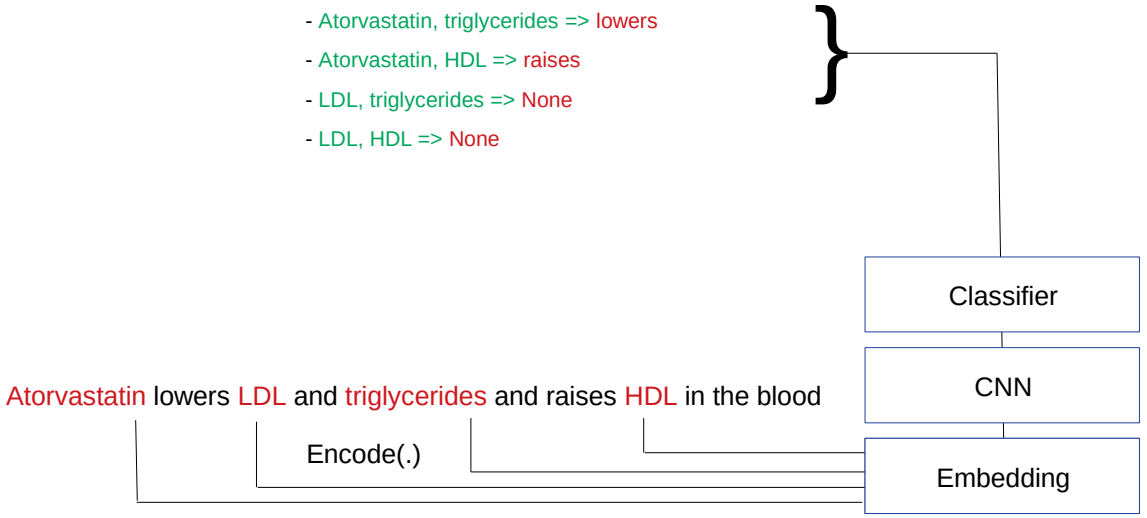
Single hop RE:

- Atorvastatin, LDL => lowers
- Atorvastatin, triglycerides => lowers
- Atorvastatin, HDL => raises
- LDL, triglycerides => None
- LDL, HDL => None



Aghaebrahimian, A. and Jurcicek, F., (2016), Open-domain Factoid Question Answering via Knowledge Graph Search, **NAACL**, San Diego, USA

Knowledge Graph

Collection of billions of triplet graph structures known as **Assertion** modeled in the **RDF** model

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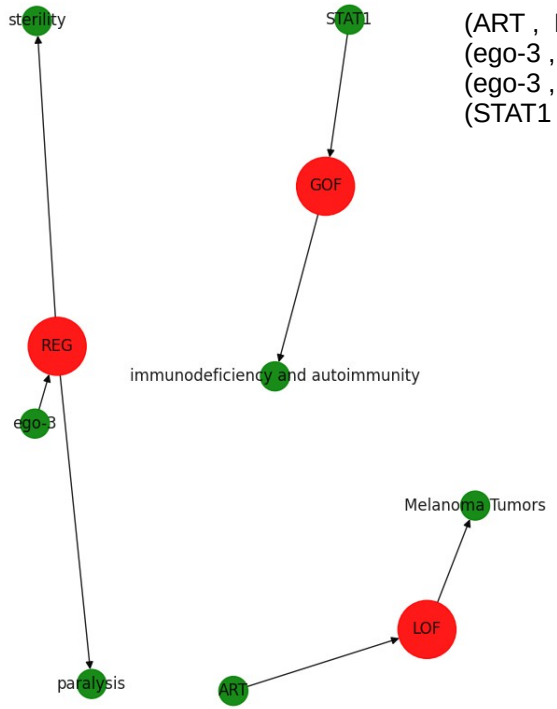

(ART , LOF, Melanoma Tumors)
(ego-3 , REG , paralysis)
(ego-3 , REG , sterility)
(STAT1 , GOF , immunodeficiency and autoimmunity)

Knowledge Graph

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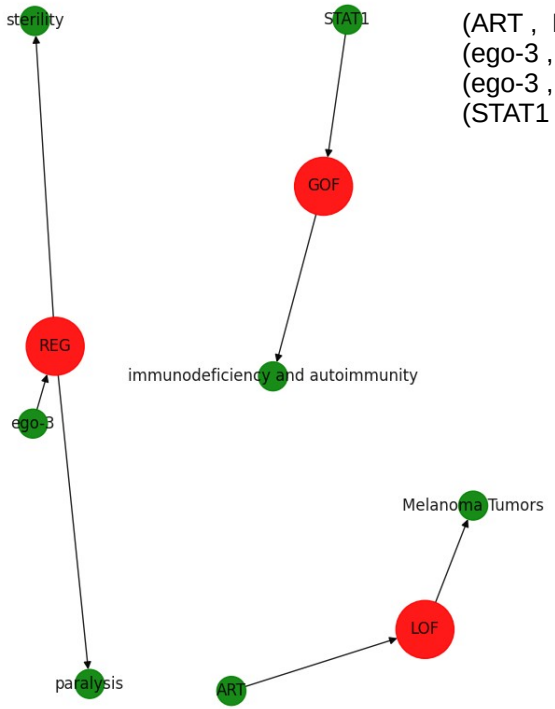

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Which genes are related to paralysis?
How STAT1 impacts the immune system?

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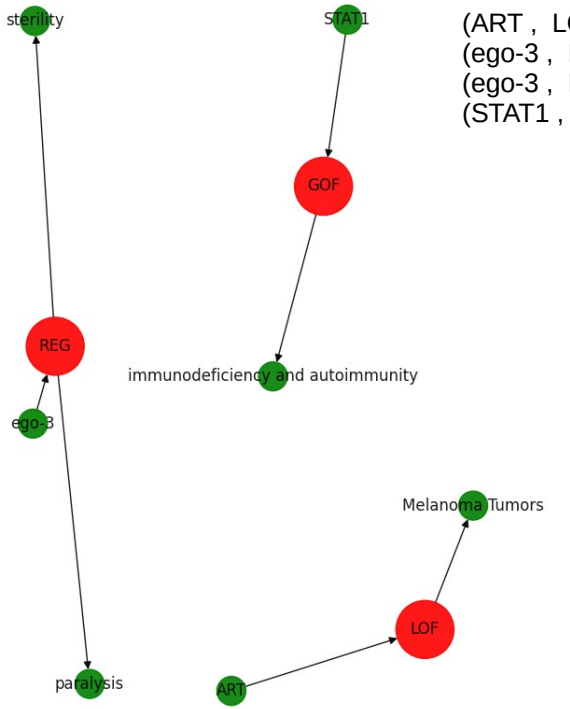

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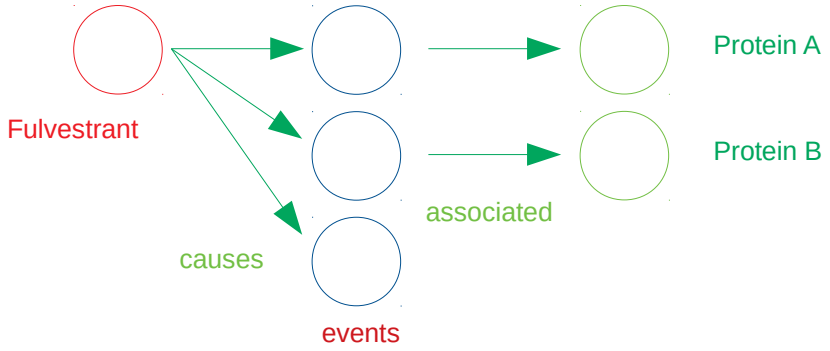
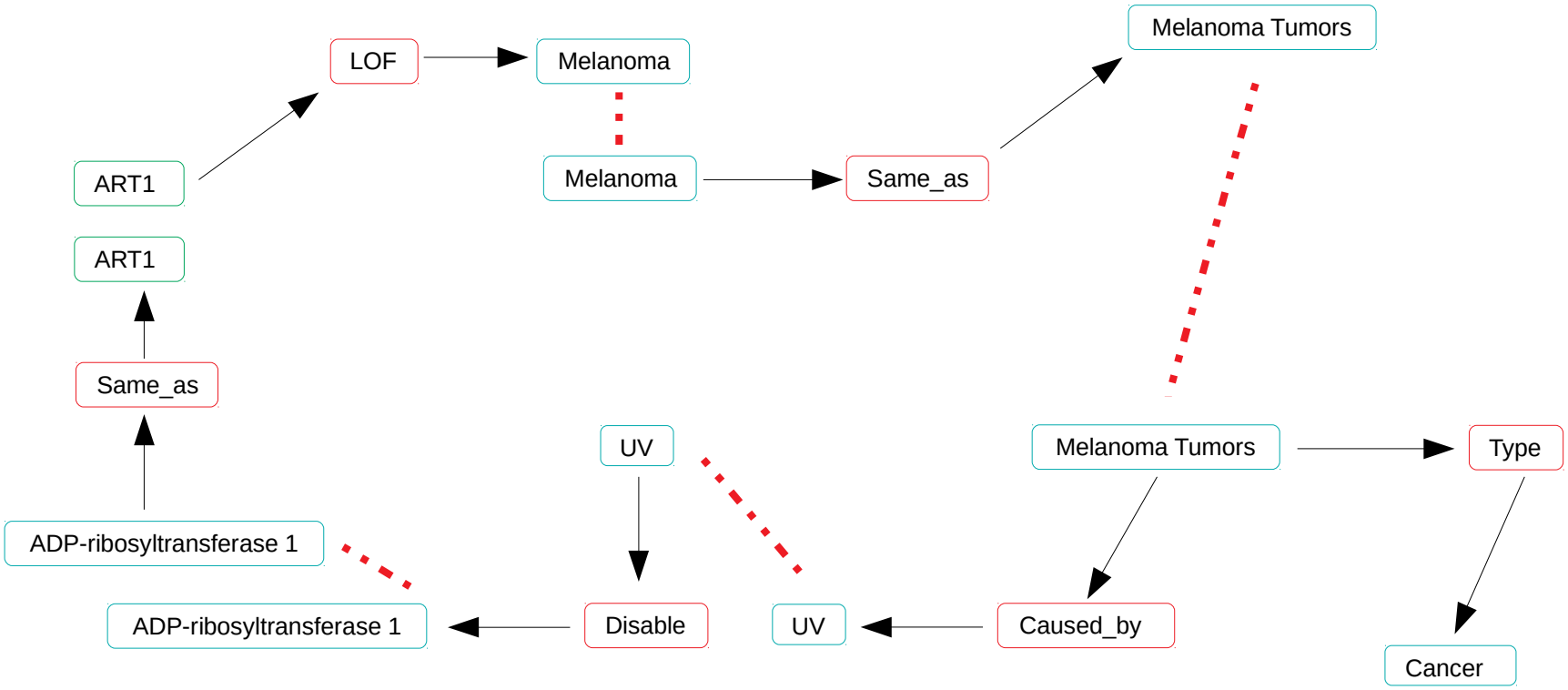
Which genes are related to paralysis?
How STAT1 impacts the immune system?

What proteins are associated with adverse events caused by Fulvestrant?



The linked open data

- Linked open data example



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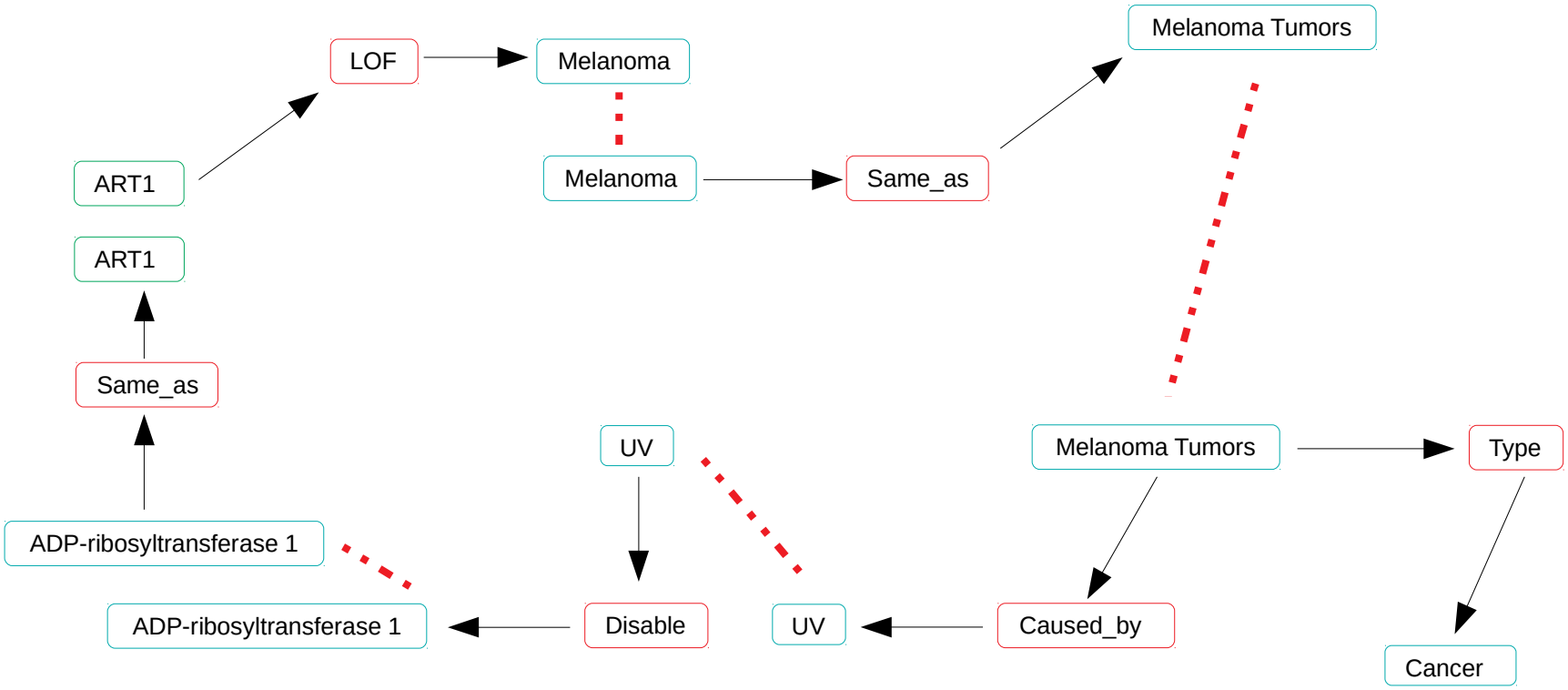


Question: How do we know that the dotted entities are the same entities.

Semantic Web tools

- RDFa:

Extracting triples from HTML pages via markups

https://rdfa.info/play/

- Gleaning Resource Descriptions from Dialects of Languages (GRDDL):

Algorithms instead of markups

<link rel="transformation" href="http://www.w3.org/2000/06/dc-extract/dc-extract.xsl" />

- JSON for Linked Data: JSON-LD

Attaching context to JSON files

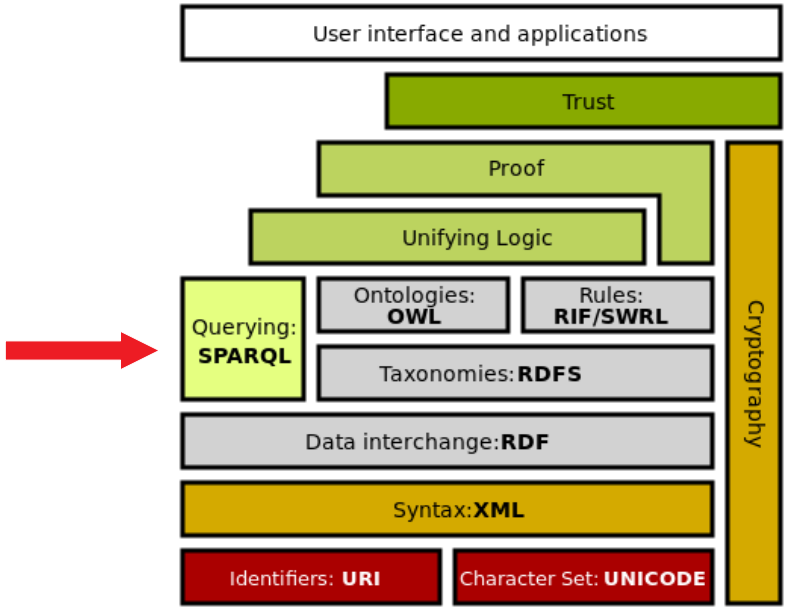
- R2RML: Transforming tables to RDF

SPARQL

W3C standard

SPARQL Protocol And RDF Query Language

Lab work: https://bit.ly/3wjyHpf



Life Sciences RDF data and SPARQL Endpoints

A SPARQL endpoint gets queries and returns their results using HTTP protocol

- Generic
  - http://sparql.org/sparql.html
  - http://demo.openlinksw.com/sparql
- Specific
  - Dbpedia
    - https://dbpedia.org/sparql
  - SIB Swiss Institute of Bioinformatics
    - UniProt: http://sparql.uniprot.org
    - neXtProt: http://snorql.nextprot.org
  - EBI European Bioinformatics Institute:
    - BioSamples, BioModels, ChEMBL, Expression Atlas, Reactome, Ensembl
    - https://www.ebi.ac.uk/rdf/services/sparql
  - NCBI National Center for Biotechnology Information:
    - PubChemRDF (rdf only, no SPARQL endpoint)
    - https://pubchem.ncbi.nlm.nih.gov/rdf/
  - http://sparql-playground.sib.swiss/