# PRINCIPLES OF BIOMEDICAL ONTOLOGIES & IMPORTANCE OF SEMANTIC INTEROPERABILITY FOR DATA INTEGRATION

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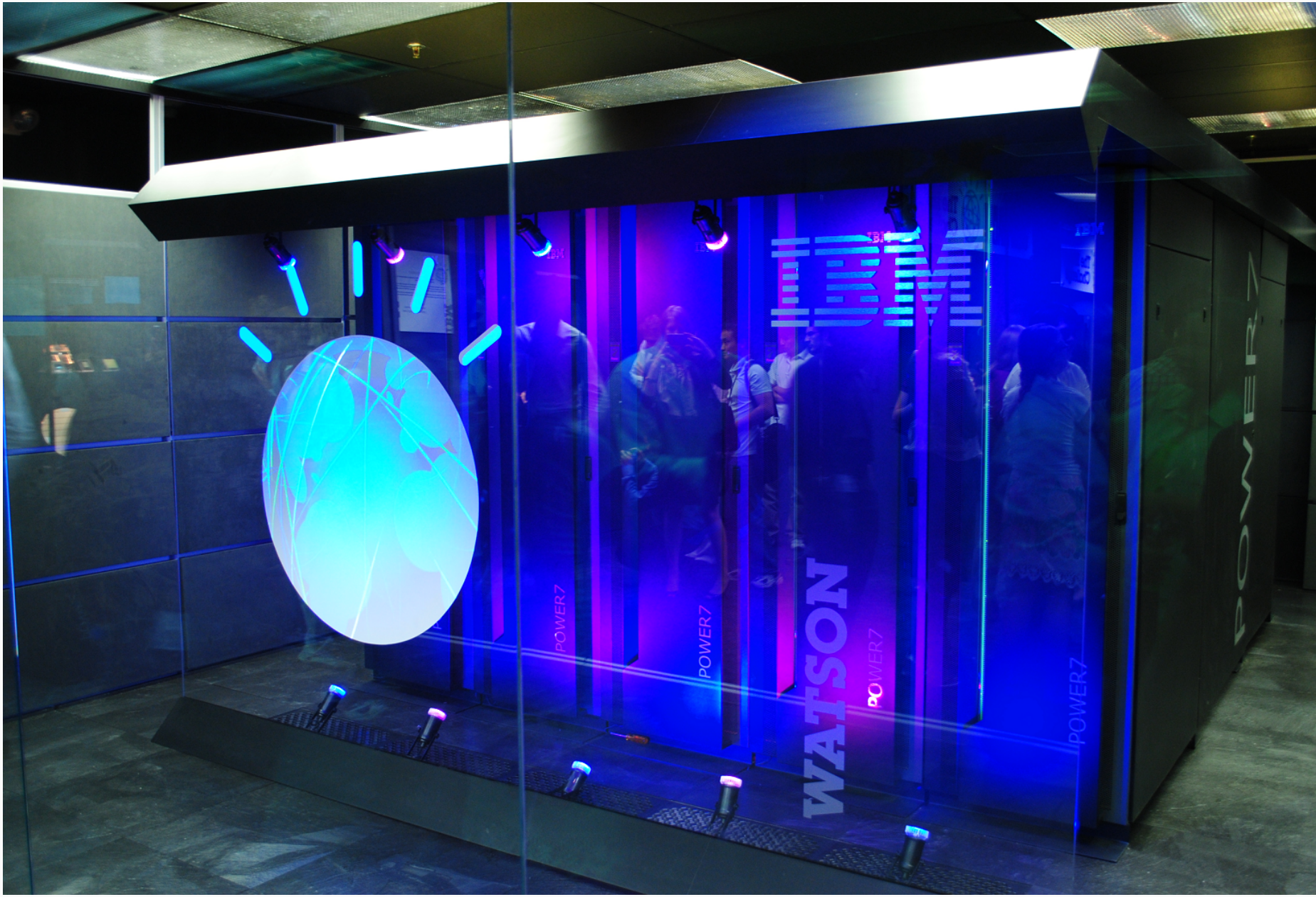
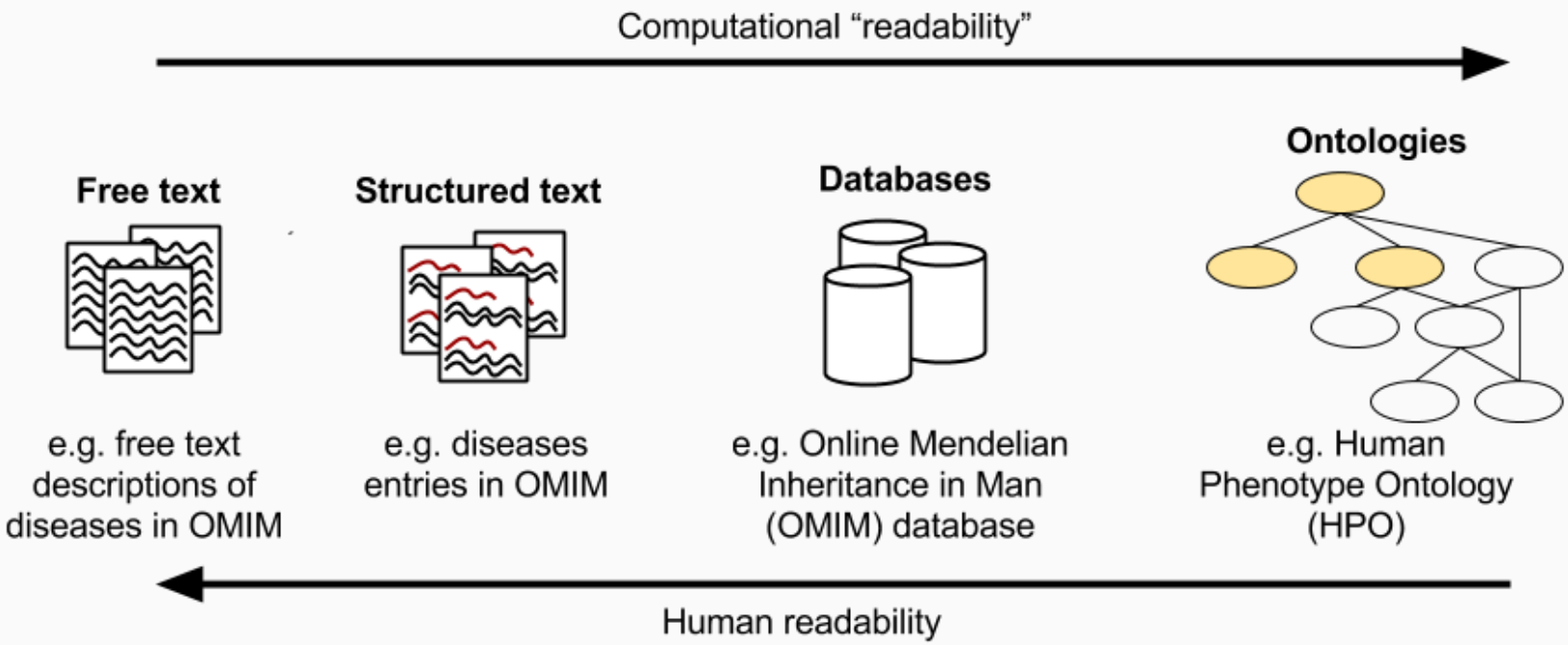



Source: https://commons.wikimedia.org/wiki/File:IBM_Watson.PNG, author: Clockready

# COMPUTATIONAL "READABILITY"



# OUTLINE

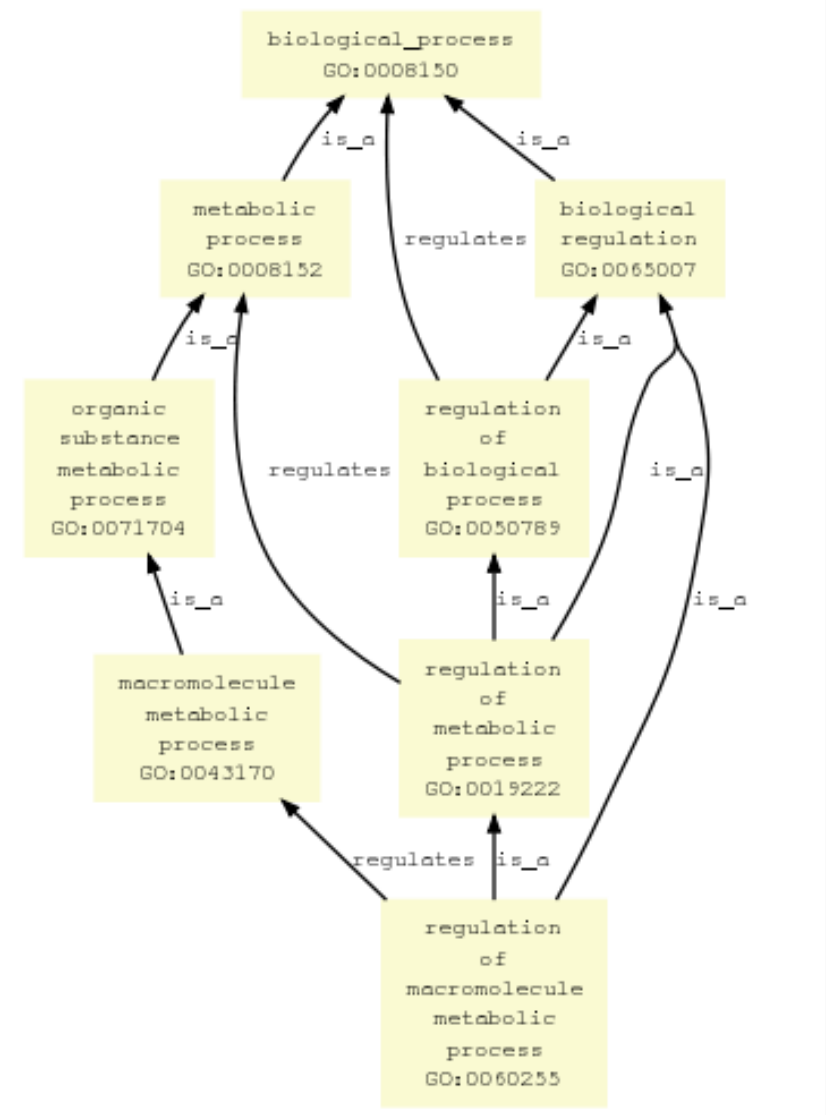
1. Ontologies in detail
2. Examples of medical resources
3. Ontologies and data integration

# A LITTLE BIT ABOUT MYSELF

- Senior PostDoc at KCL
- used ontologies for text and data mining, data integration
- worked with model organism data in the past (International Mouse Phenotyping Consortium project)
- now utilising data from electronic health records (EHRs)

# Ontologies in detail

# BEST WAY TO IMAGINE ONTOLOGY: GRAPH




Source: Generated with old Ontology Lookup Service (OLS)

## Definition of e.g. *regulation of biological process*

**Class relations**

**Equivalent to:**

- biological regulation and *regulates* some biological_process

**Subclass of:**

- biological regulation
- *regulates* some biological_process

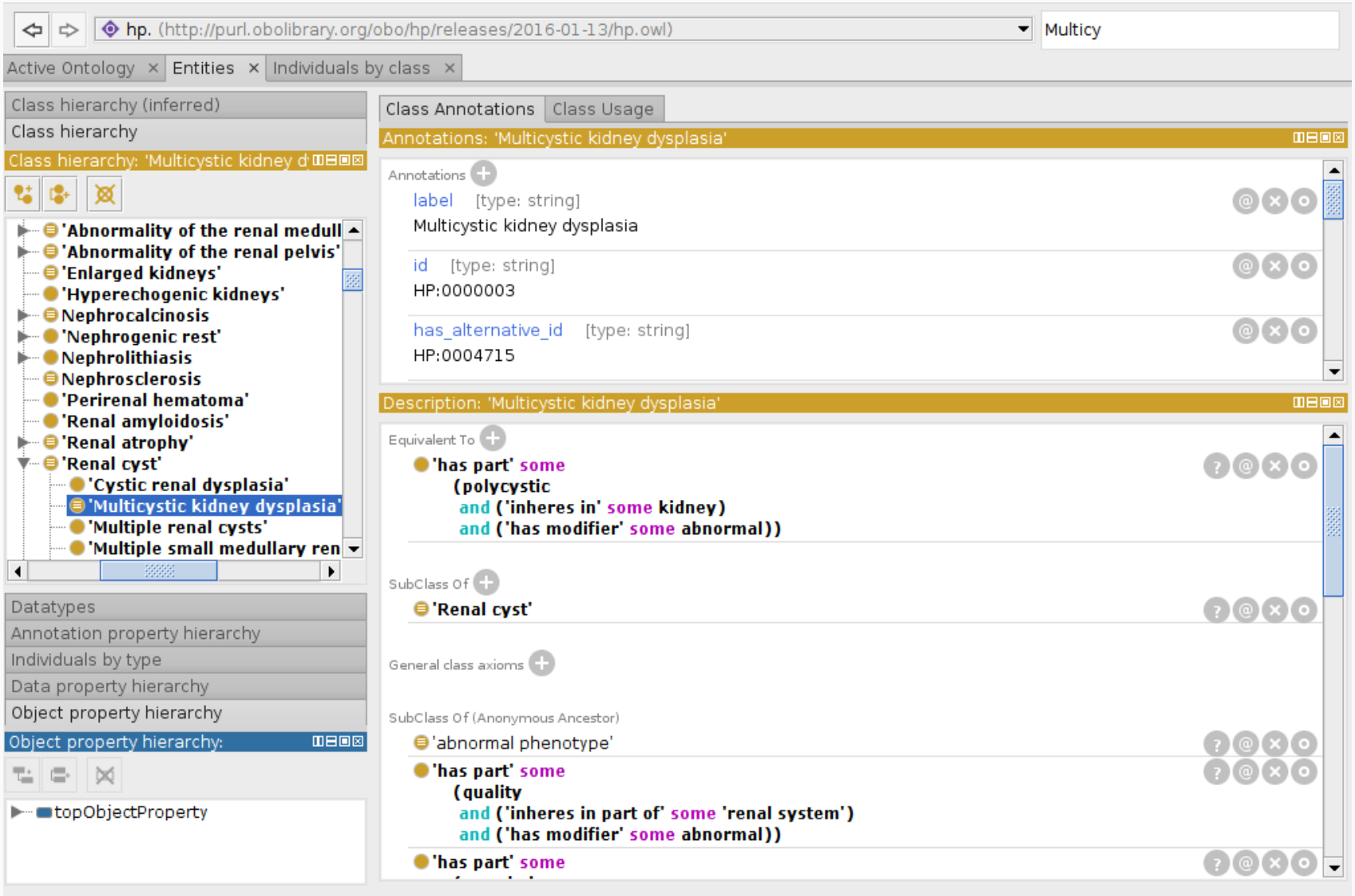
Source: Generated with new Ontology Lookup Service (OLS)

# Open Biomedical Ontologies (OBO) format

```
[Term]
id: HP:0000003
name: Multicystic kidney dysplasia
alt_id: HP:0004715
def: "Multicystic dysplasia of the kidney is characterized
by multiple cysts of varying size in the kidney and the absence of a
normal pelvocaliceal system. The condition is associated with ureteral
or ureteropelvic atresia, and the affected kidney is nonfunctional."
[HPO:curators] comment: Multicystic kidney dysplasia is the result of
abnormal fetal renal development in which the affected kidney is
replaced by multiple cysts and has little or no residual function. The
vast majority of multicystic kidneys are unilateral. Multicystic
kidney can be diagnosed on prenatal ultrasound.
synonym: "Multicystic dysplastic kidney" EXACT []
synonym: "Multicystic kidneys" EXACT []
synonym: "Multicystic renal dysplasia" EXACT []
xref: MeSH:D021782 "Multicystic Kidney Dysplasia"
xref: UMLS:C0345335 "Multicystic Kidney Dysplasia"
is_a: HP:0000107 ! Renal cyst
```

ftp://ftp.geneontology.org/pub/go/www/GO.format.obo-1_4.shtml

# WEB ONTOLOGY LANGUAGE (OWL)




https://www.w3.org/TR/owl-features/, http://protege.stanford.edu/

# Example of medical resources

# HUMAN PHENOTYPE ONTOLOGY

- ~11k concepts
- ~115k annotations for hereditary & ~4k for common diseases
- reports phenotype abnormalities
- several groups working on its extension
- initial version generated from OMIM
- pre-dominantly used in research projects, e.g. in Deciphering Developmental Disorders (DDD) project

`http://human-phenotype-ontology.github.io/`, `http://www.ddduk.org/`

# SYSTEMATIZED NOMENCLATURE OF MEDICINE–CLINICAL TERMS

- SNOMED CT
- ~316k concepts
- aims to cover all aspects of healthcare (e.g. drug events, environmental factors, ...)
- it is multilingual
- pre-dominantly used in clinical environments, e.g. EHRs

http://ihtsdo.org/fileadmin/user_upload/doc/download/doc_StarterGuide_Current-en-US_INT_20141202.pdf?ok

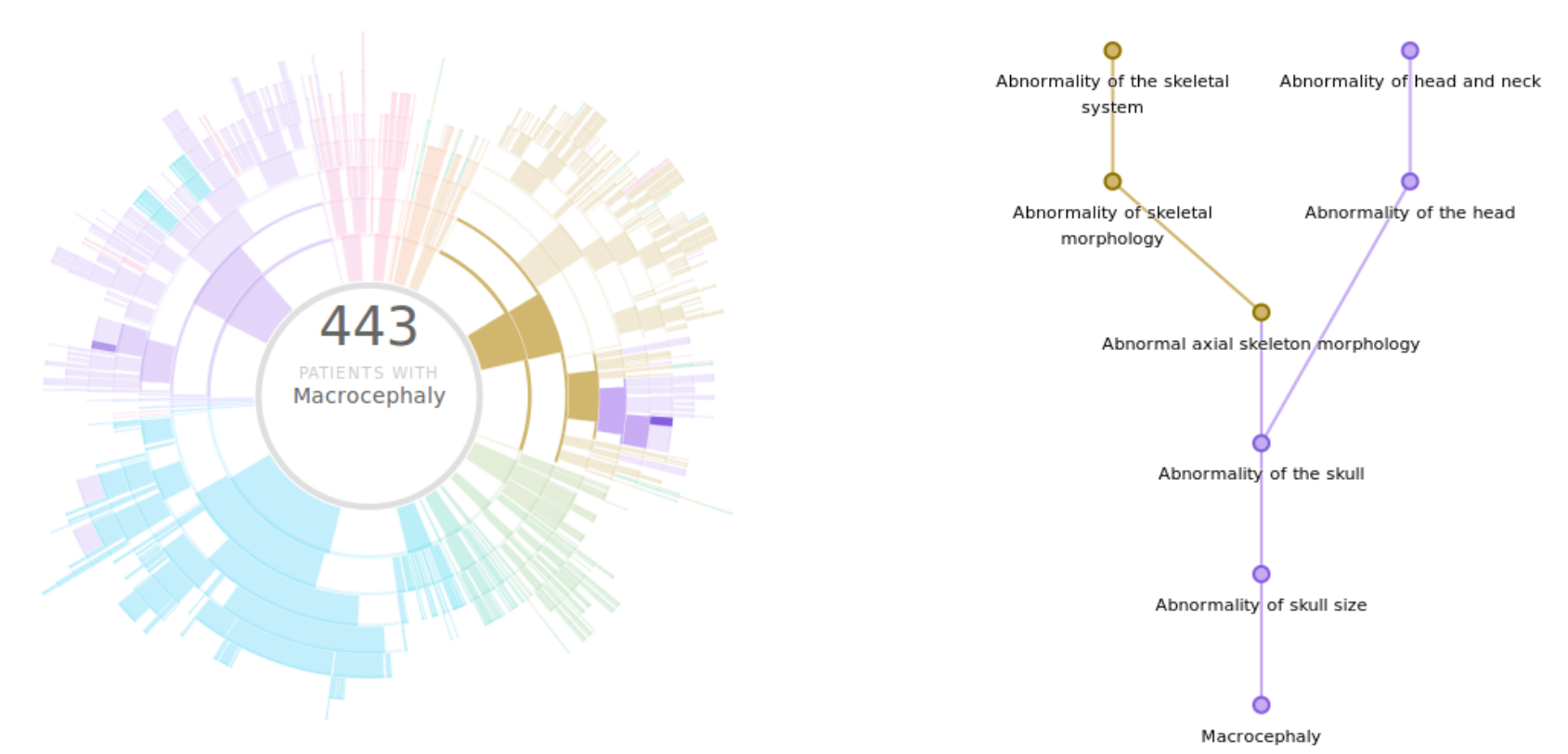
# OTHER RELEVANT RESOURCES

- Logical Observation Identifiers Names and Codes (LOINC)
- Unified Medical Language System (UMLS)
- International Classification of Diseases (ICD 9/10)
- Medical Subject Headings (MeSH)

https://loinc.org/, https://www.nlm.nih.gov/research/umls/, http://www.who.int/classifications/icd/en/, https://www.nlm.nih.gov/mesh/

# Ontologies and data integration

# DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources




https://decipher.sanger.ac.uk/

# DECIPHER - PATIENT VIEW

Patient: 865

Overview | Genotype 2 | Phenotype 5 | Karyotype | Citations 0 | Contacts

**Legend**

**Copy-Number Variants** Gain Loss

**Chromosome 2**
**Copy-Number Variants**


**Chromosome 22**
**Copy-Number Variants**

**Legend**

**Copy-Number Variants** Gain Loss

Patient: 865

Overview | Genotype 2 | Phenotype 5 | Karyotype | Citations 0 | Contacts

Patient 5 | Mother 0 | Father 0

Patient phenotypes

| Phenotype | Observation | Code |
|---|---|---|
| Abnormally low-pitched voice | present | HP:0010300 |
| Autism | present | HP:0000717 |
| Intellectual disability | present | HP:0001249 |
| Macrocephaly | present | HP:0000256 |
| Obesity | present | HP:0001513 |

# PHENOGRAMVIZ TO SEE GENES THAT COULD IMPLICATE PHENOTYPE

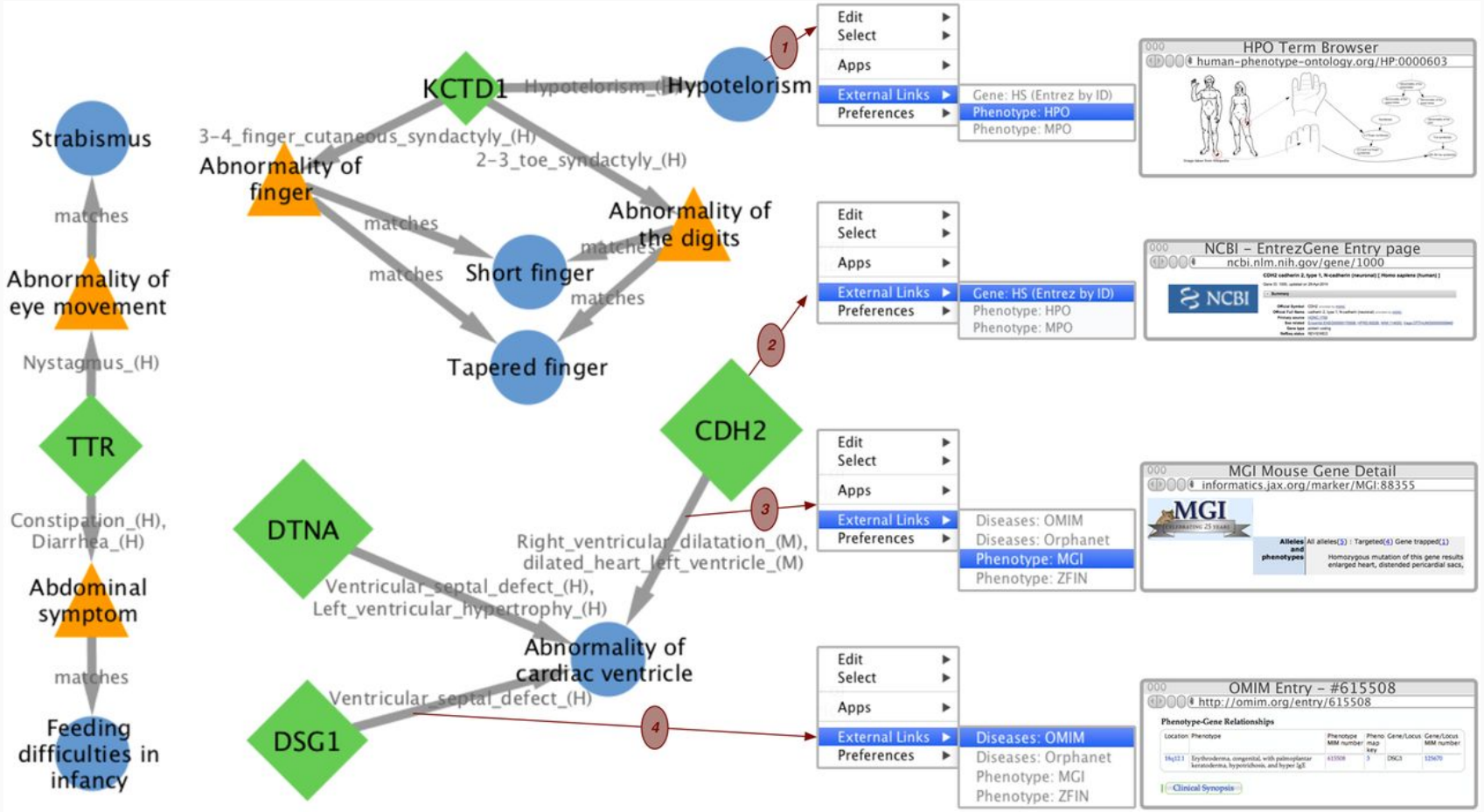
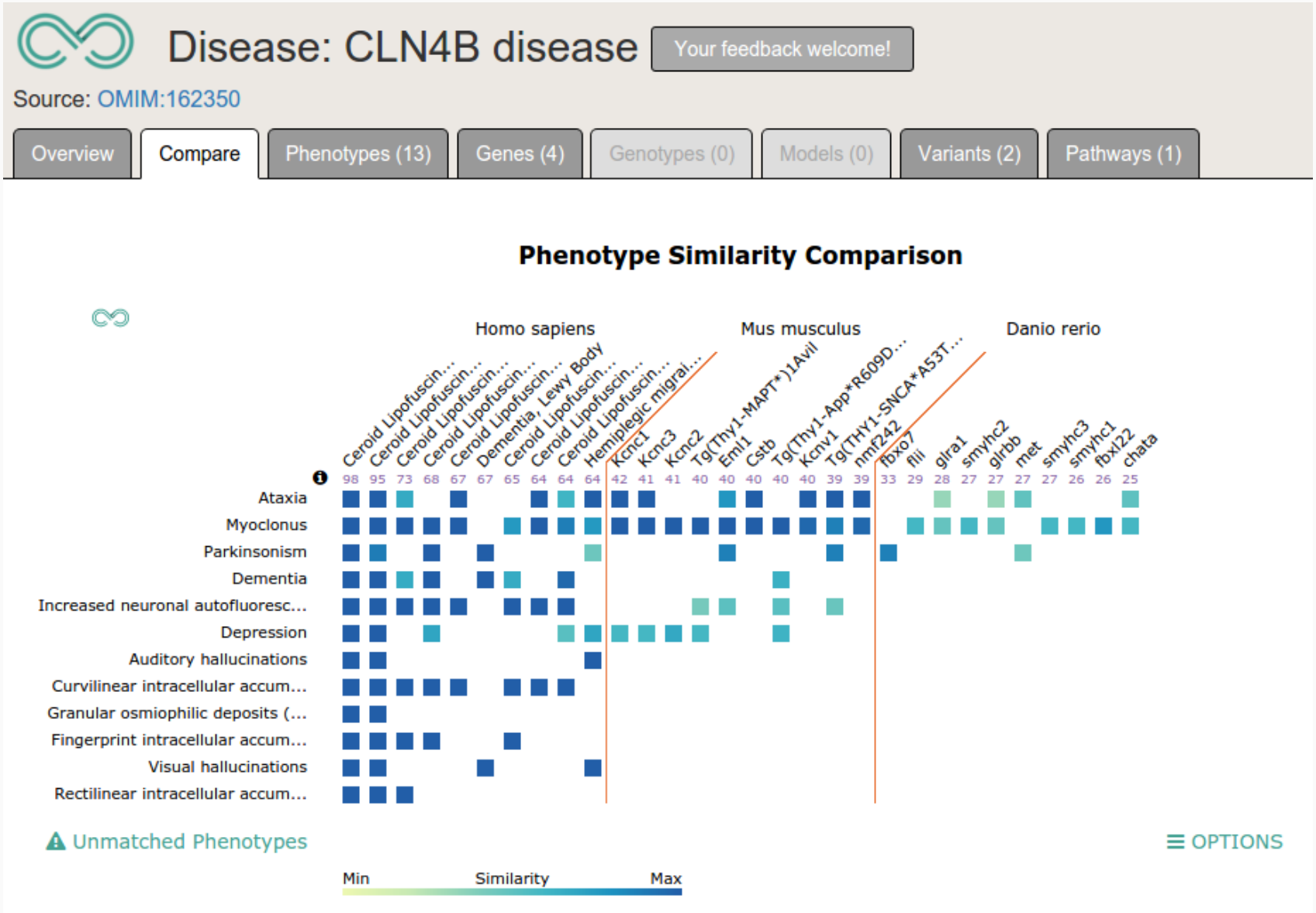



Figure taken from: K'öhler, Sebastian, et al. "Clinical interpretation of CNVs with cross-species phenotype data." Journal of medical genetics 51.11 (2014): 766-772.




http://monarchinitiative.org/disease/OMIM:162350

# SUMMARY

Ontologies ...

- define meaning of concepts
- have a hierarchy on which concepts are sorted
- are defined with formal languages
- can help data visualisation
- applied for semantic similarity measures