The TOP Framework: A Next-generation Phenotype Repository

JRG "Terminology- and Ontology-based Phenotyping"

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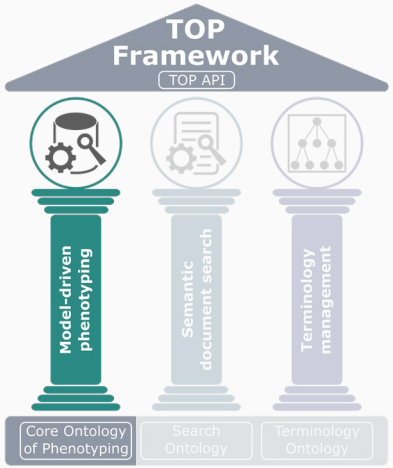
Overall Aims of the TOP JRG

1. Contribute to the **ontological foundation** of phenotyping
2. Develop a standardised phenotype **modelling language**
3. Combine phenotype models with data sources in the MII, enabling **search queries**
4. Integrate **external terminologies**
5. Enable **document search** in unstructured documents
6. Provide **user-friendly tools** for next-generation phenotyping

Next-generation phenotyping combines the **semantic and structured (standardised)** definition of phenotypes with the **creation of phenotype algorithms**. – Mo et al. [1].

Ontological Foundation

Phenotypes



- Phenotypes are observable (combinations of) characteristics of an organism [2]

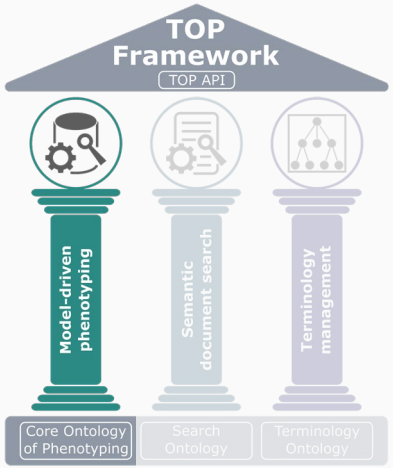
Two classes of phenotypes:

Basic phenotypes

directly/indirectly
observable or measurable

eye color, blood pressure, insuline medication

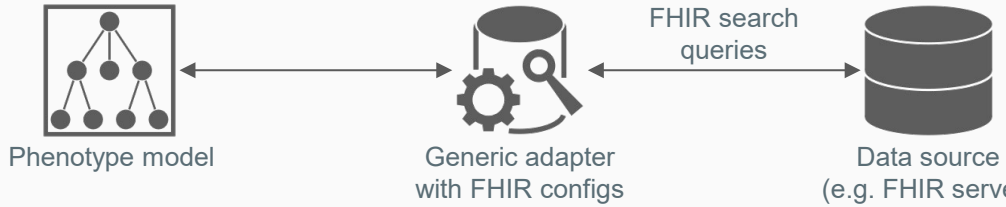
Phenotype Models



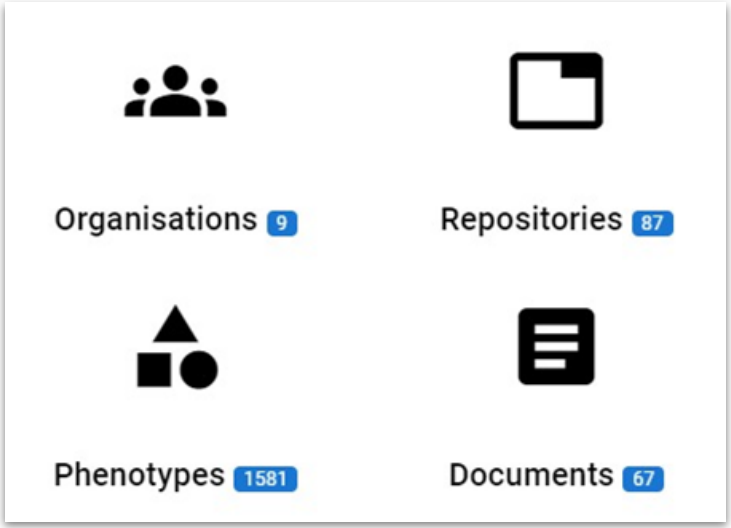
Purposes:

- Detection of diseases
- Risk assessment
- Recruitment for clinical studies
- Individualised patient care and treatment

- Actual data is not stored in the framework!
- Models can be translated into query languages
- Data sources are accessible via adapters:
  - **generic adapters** with configurations (e.g., OMOP, POLAR, FHIR Search)
  - **specific adapters** (e.g., LIFE study)



Model-driven Phenotyping With the TOP Framework

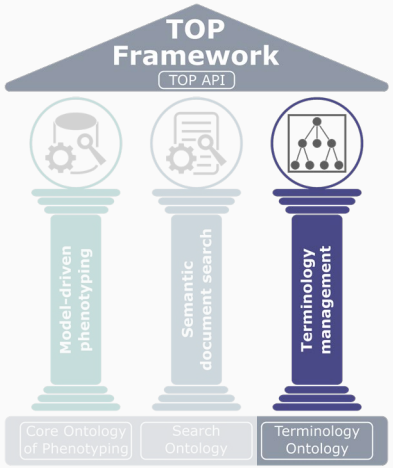


Key functionalities:

- Collaborative web application for non-IT staff
- Development of computable phenotype models using standardised terminologies
- Efficient data exploration and analysis through a standardised query interface [3]
- Sharing and reusing phenotype models and their components [4]

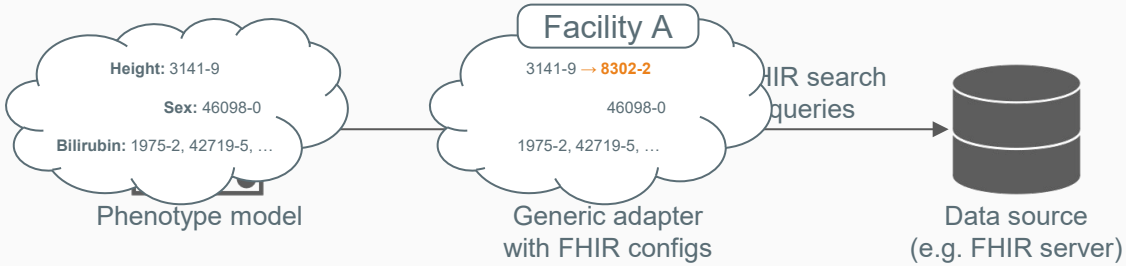
TOP Framework has the potential to enhance knowledge discovery, efficiency, and collaboration within clinical research.

Terminology Management

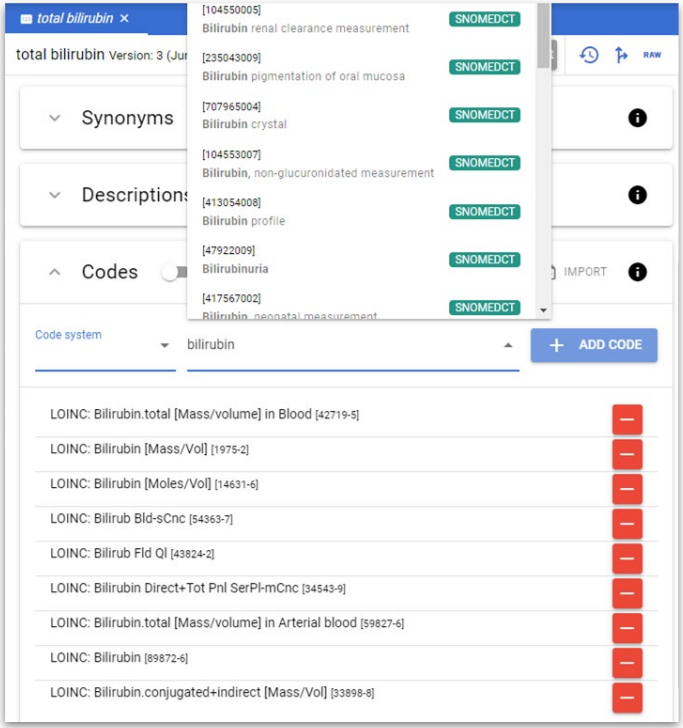


- Terminologies are stored in the **Ontology Lookup Service**
  - Medication: ASK, ATC, PZN
  - Diagnosis: ICD-10
  - Observation: LOINC, SNOMED CT
  - Procedure: OPS

- TOP Framework provides search widget for OLS codes
- Codes are used in phenotype models to enable interoperability with data sources



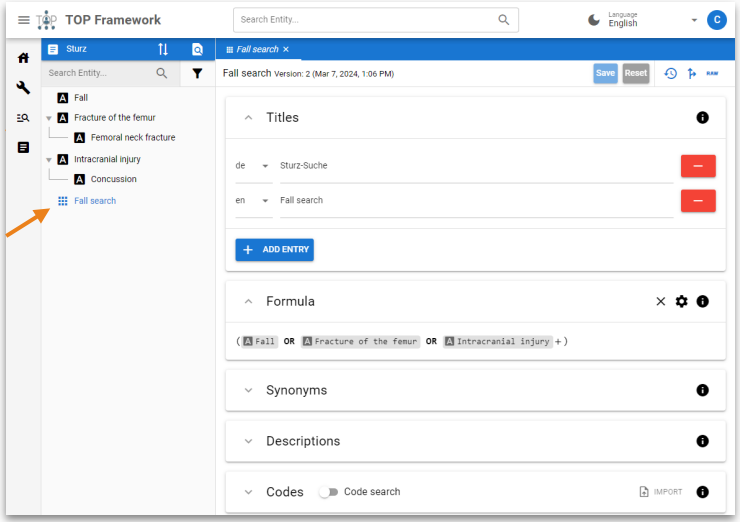
Terminology Management



Future work:

- Handling of subtrees
  e.g. in ICD-10:
  - `P05–P08`: Disorders of newborn related to length of gestation and fetal growth
    ⇒ 15 codes
- Comparison of terminology versions [5]
  - Integration into the TOP Framework
- Terminology updates may affect models!

Semantic Document Search



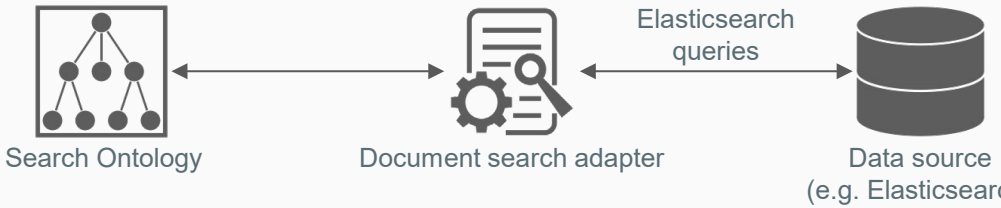
- Ontology-based modelling of text search queries with concept classes

Two classes of search concepts:

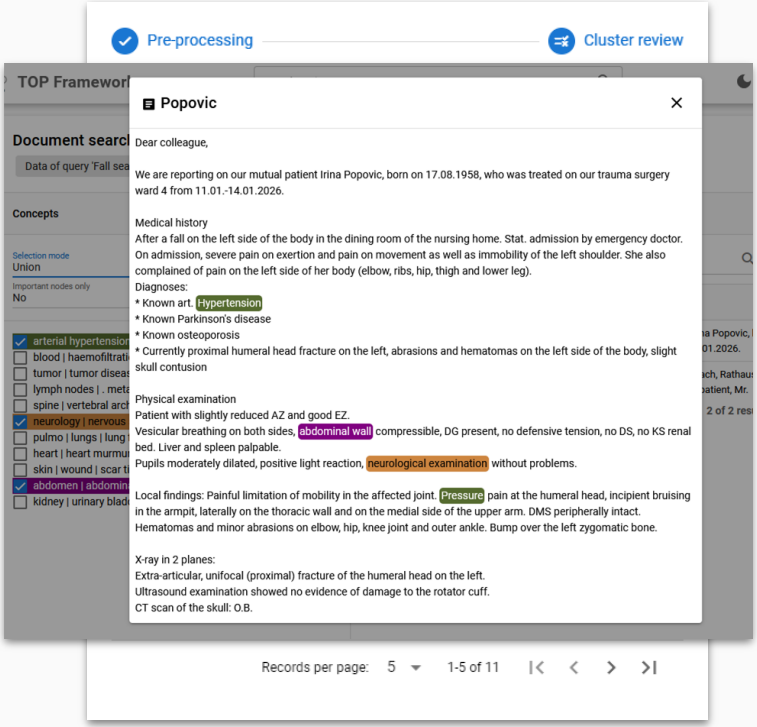
Single concepts

representing **sets of search terms**, organised **hierarchically**

Fall, Intracranial injury



Semantic Document Search



- **Concept clusters** are the actually realized concept graphs in the TOP Framework (Graph DB in background)
  - Queries can be enhanced with graph algorithms
- Curation of concept clusters
  - Clusters are created **completely automatically**
  - Reasonable clusters are selected manually

Search ontology and concept clusters can be combined or used individually to search for documents.

Conclusion

- We addressed challenges in standardising phenotype models for building a next-generation phenotype repository.

- The TOP Framework, built upon the COP and TOP API, empowers researchers with **standardised models** and facilitates **efficient data exploration and analysis**.
- It can be **integrated into the MII infrastructure** for streamlined data access.
- Real-world applications demonstrate the **potential to support clinical research** by enhancing efficiency, collaboration, and knowledge discovery.

Publications

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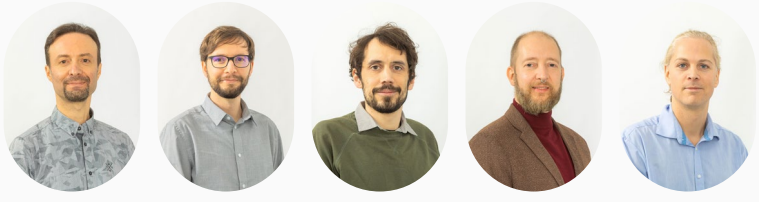

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