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Ontologizing health systems data at scale: making translational discovery a reality

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OHDSI Community Call - September 25, 2023

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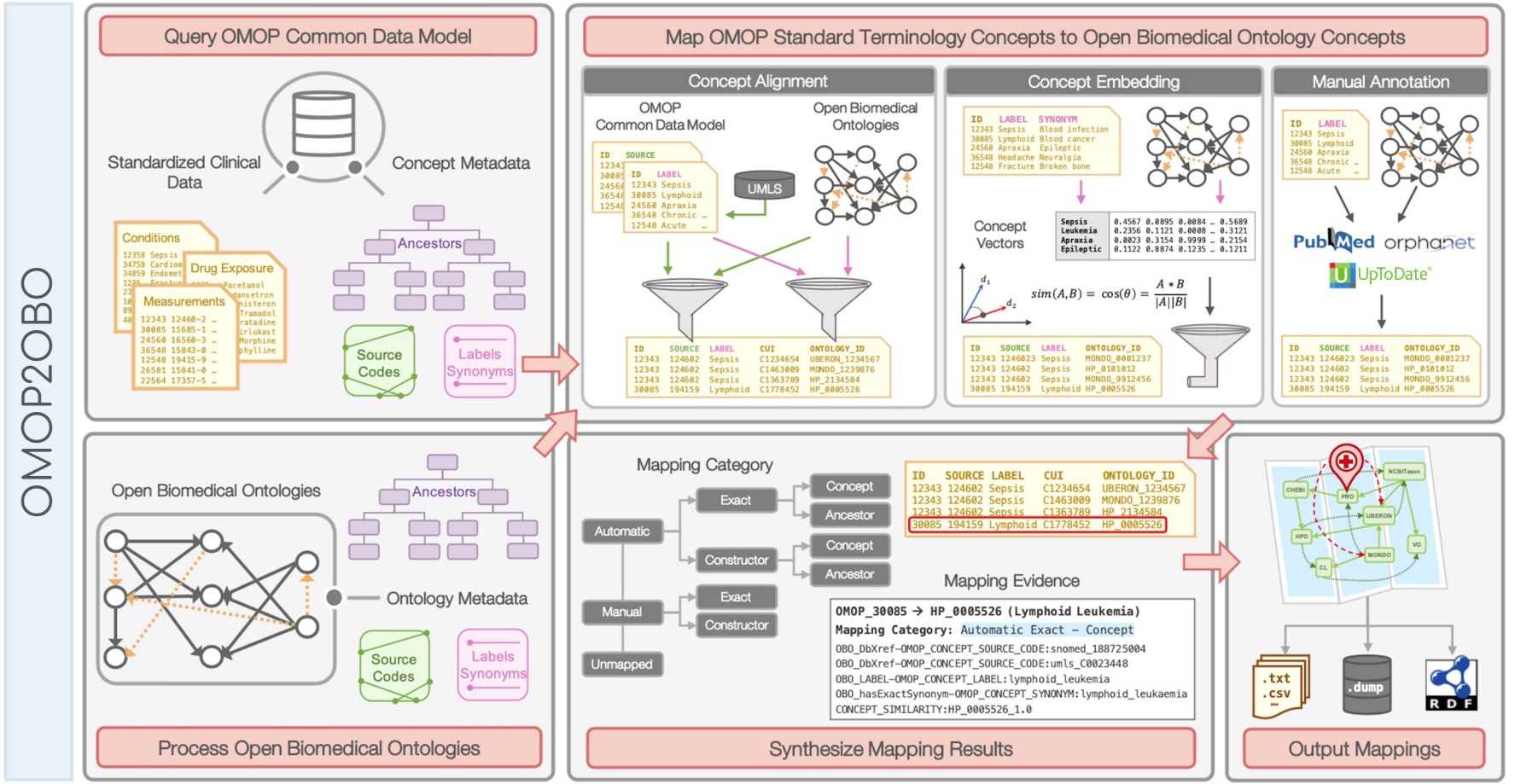
Background

- Deep phenotyping, or "the precise and comprehensive analysis of phenotypic abnormalities in which the individual components of the phenotype are observed and described"[1], requires timely synthesis of multiple types of patient data[2-3]
- Common data models solve many challenges of standardizing electronic health record (EHR) data but are unable to semantically integrate all of the resources needed for deep phenotyping[4]
- Open Biological and Biomedical Ontology (OBO) Foundry ontologies provide computable representations of biological knowledge and enables the integration of heterogeneous data[5]

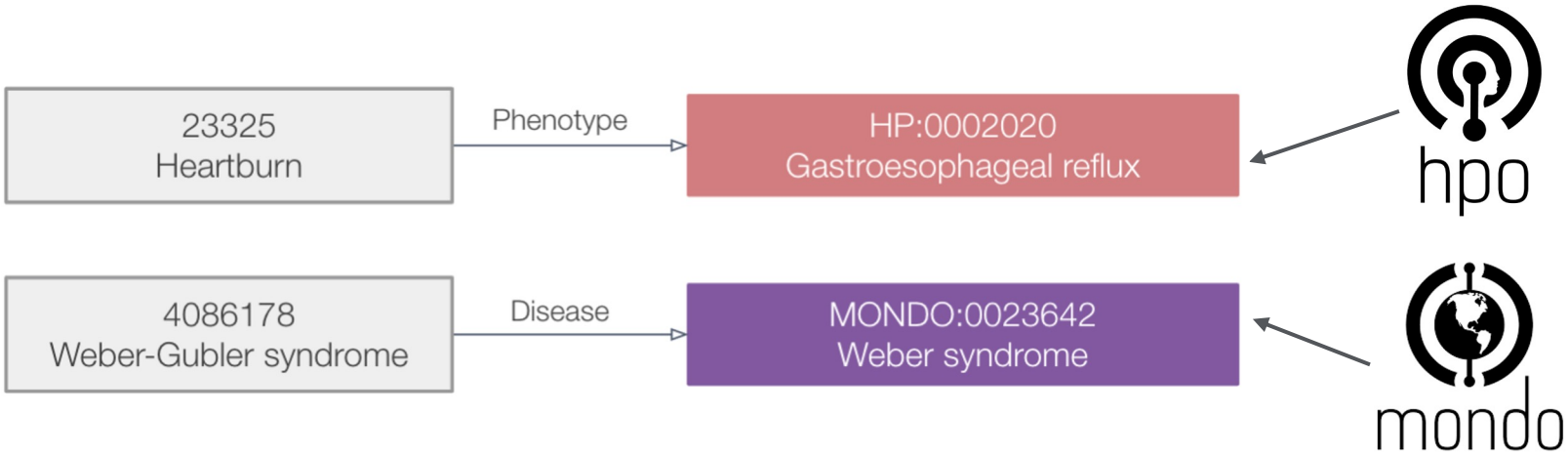
Challenge: Mapping EHR data to OBO ontologies requires significant manual curation and domain expertise

[1]PMID22504886; [2]PMID:26536218; [3]PMID:32335224; [4]PMID:24071798; [5]PMID:25863278

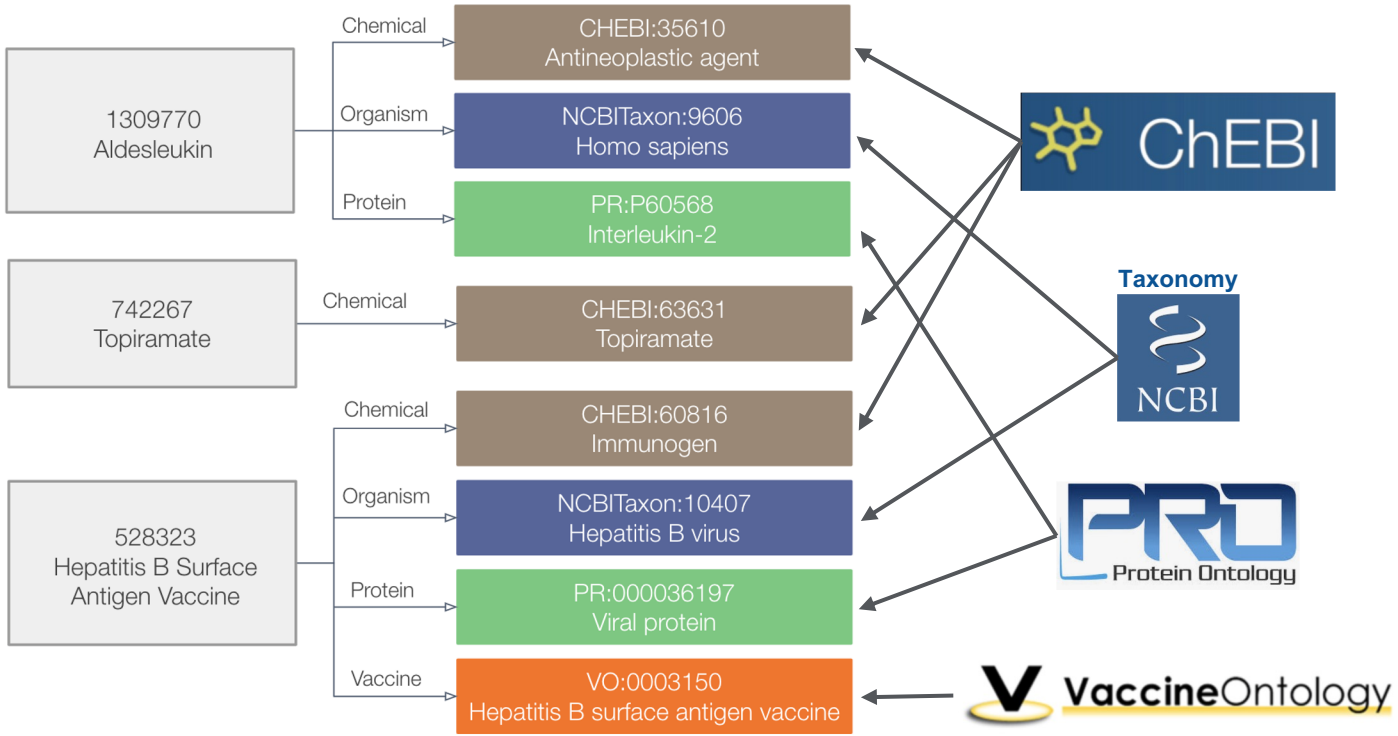
Objective: Develop an algorithm that generates clinically meaningful and biologically relevant mappings between standard OMOP vocabularies and Open Biomedical Ontologies



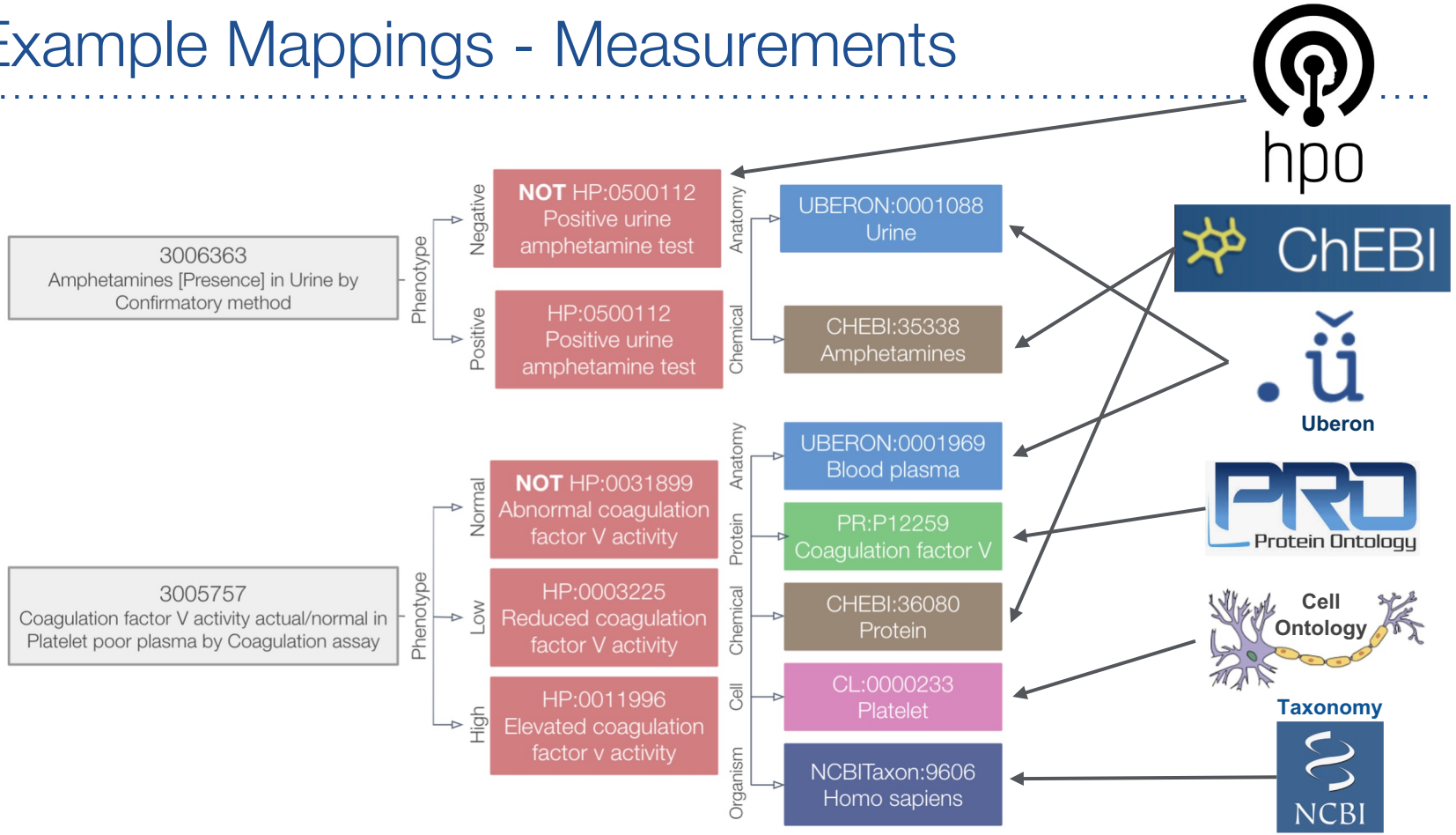
Example Mappings - Condition Occurrences

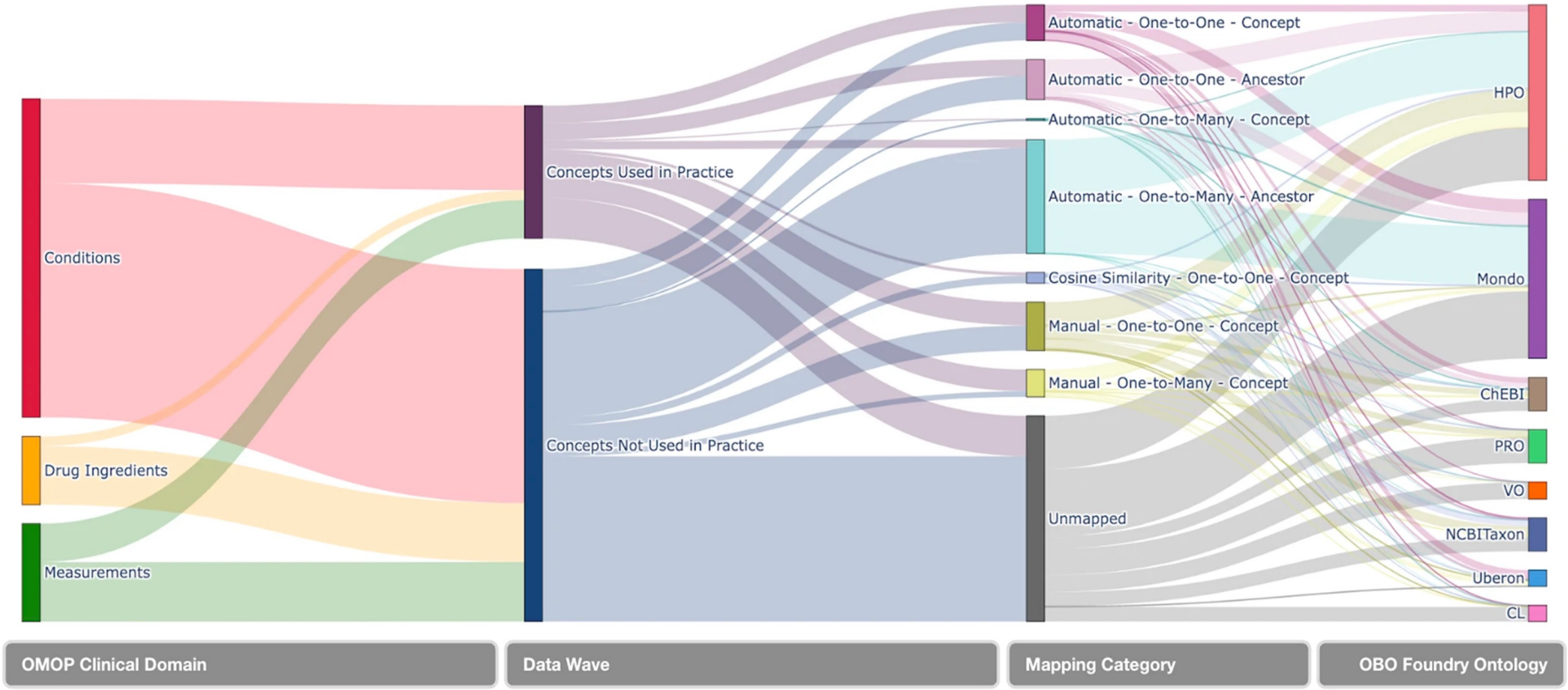


Example Mappings - Drug Exposure Ingredients



Example Mappings - Measurements





Using OMOP2OBO, we built mappings for 92,367 conditions, 8615 drug ingredients, and 10,673 measurement results to ontology concepts representing **9636** diseases, **6309** phenotypes, **83** anatomical entities, **2704** organisms, **4261** chemicals, **132** vaccines, and **272** proteins

Evaluation

Accuracy

10 domain experts manually reviewed 20% of the most challenging manual OMOP2OBO mappings

- Correct Mappings: 73.9% (condition), 70.7% (drug ingredient), and 92.9% (measurements)

Generalizability

OMOP2OBO mapping concept coverage in 24 Independent hospitals (OHDSI Concept Prevalence Study)

- Coverage: 99.5% of conditions, 99.9% of drug ingredients, and 68% of measurement results

Clinical Utility

Compare OMOP2OBO mappings to validated manual mappings when used to identify patients with rare genetic diseases using AoU data

- Query Performance: OMOP2OBO mappings identified 99.3% of the patients identified by the validated manual mappings using fewer codes and one-third of the query time

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Blake Martin, MD


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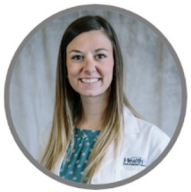
Katy Trinkley, PharmD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Organizations

OHDSI
OBSERVATIONAL HEALTH DATA SCIENCES AND INFORMATICS

Children's Hospital Colorado

HEALTH DATA Compass

PEDSnet
A Pediatric Learning Health System

NIH U.S. National Library of Medicine

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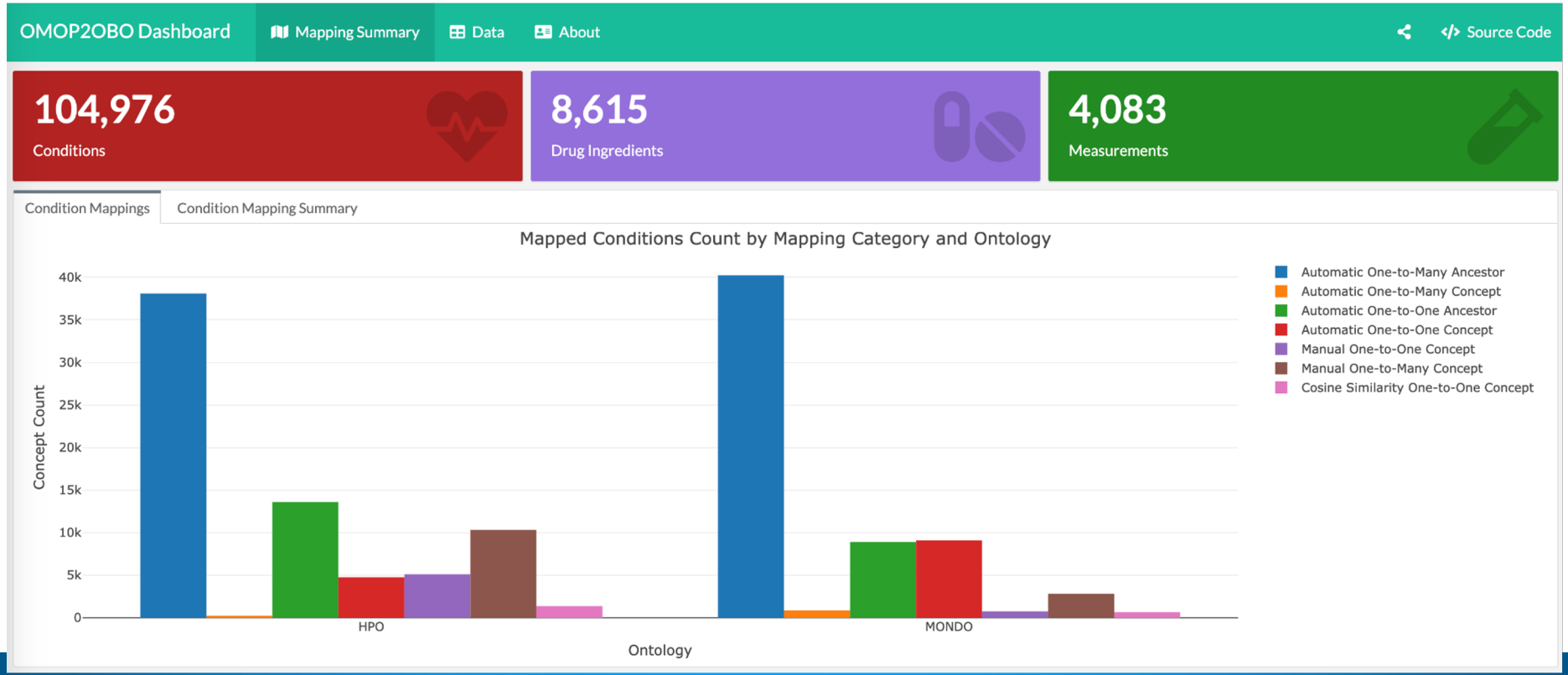
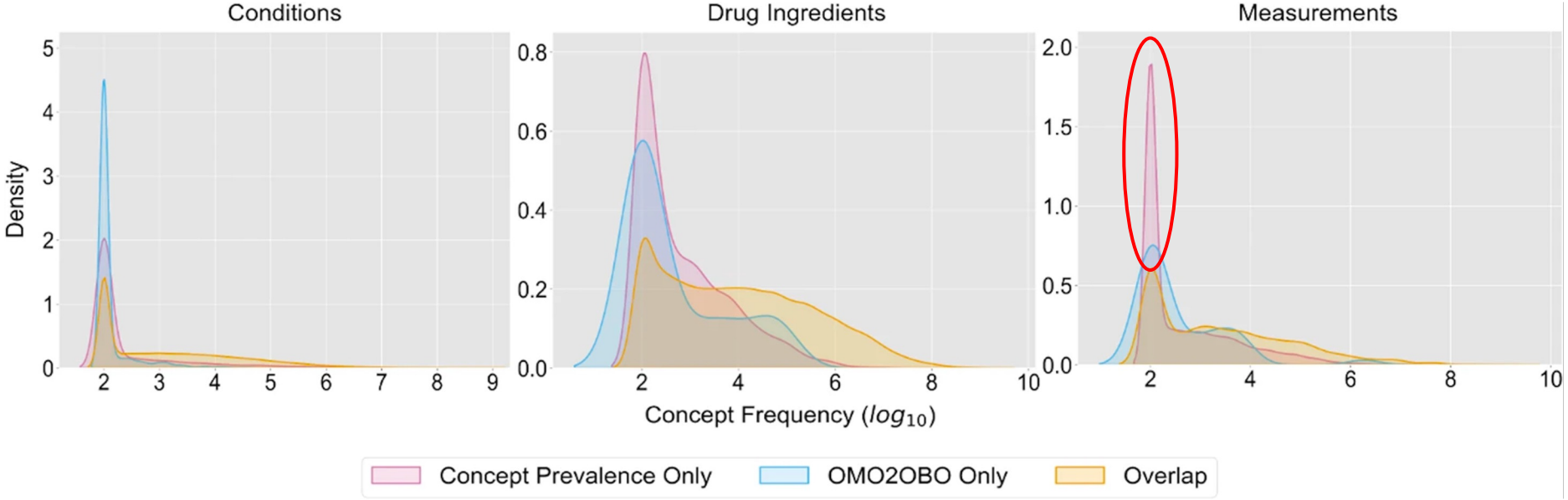
Dashboard

Conditions: https://doi.org/10.5281/zenodo.6774363
Drug Ingredients: https://doi.org/10.5281/zenodo.6774401
Measurements: https://doi.org/10.5281/zenodo.6774443



http://tiffanycallahan.com/OMOP2OBO_Dashboard

Evaluation


Conditions
Drug Ingredients
Measurements
Density
Concept Frequency (log_{10})
Concept Prevalence Only
OMO2OBO Only
Overlap