PRESENTER: Rowdy de Groot

INTRO:

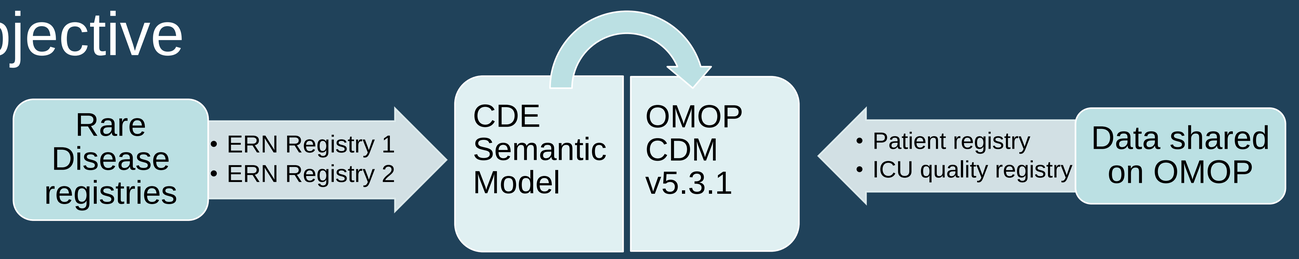
- 6000 rare diseases affecting 30 million Europeans.
- Rare disease research hampered by sensitive nature, sparse, scattered and heterogeneous.
- European Joint Programme on Rare Diseases (EJP RD) → **FAIR principles**
- The EJP RD CDE semantic model:
  - Ontological model
  - Common data elements (CDE)
  - Reusable ETL pipeline
- Manual mapping CDE semantic model → OMOP CDM

METHODS:

- EJP RD CDE semantic data model
  - Design principles of the Semanticscience Intergrated Ontology
  - NCIt - 21, SIO - 8, HPO - 8, DUO - 6, OBI - 4, EDAM - 2, ICO - 1, OBIB - 1, OMIABIS - 1
- OHDSI OMOP CDM
  - OMOP CDM V5.3.1
- Mapping of terms
  - Manual mapping
  - Athena
  - Hackathons
  - Mapping relations
    - OWL
    - RDFS
    - SKOS
- Data transformation
  - Load RDF into a triplestore
  - Use SPARQL to generate OMOP CDM compatible tables

*We made an **ETL pipeline** to convert rare disease data (CDE) to **OMOP CDM***

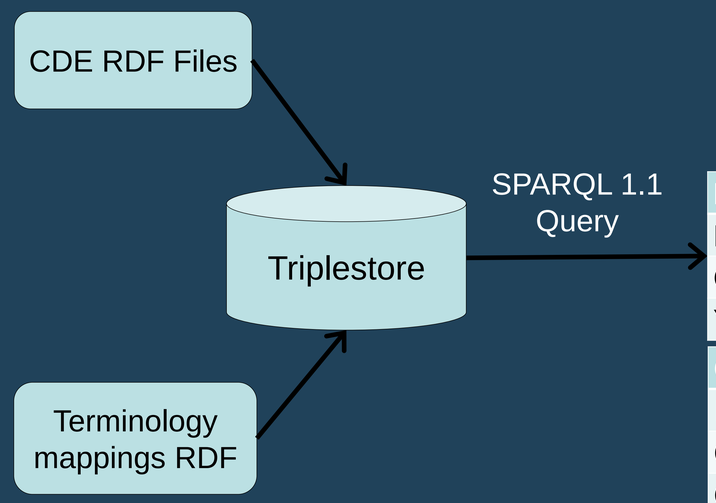
Objective



Methods

Terminology mapping	Data transformation	Apply on use-case
• Manual mapping • SSSOM representation of mapping results • Curation with ontology experts	• Load RDF into a triplestore • Use SPARQL to generate OMOP CDM compatible tables	• Apply the mapping to an ERN registry

ETL Pipeline



PERSON	value
Person_id	420
Gender_concept_id	8532
Year_of_birth	1969

OBSERVATION	value
Person_id	420
Observation_concept_id	4163894
Observation_source_value	Lost to follow-up

RESULTS:

- Terminology Mapping
  - 52 CDE terms to 30 Athena terms
  - SNOMED CT - 21, LOINC - 6, OMOP - 2, HCPCS - 1
  - SKOS → relatedMatch
- ETL Pipeline
  - Use SPARQL to generate OMOP CDM artefacts
  - Use curated mappings for transformation

- **Future work:**
- Finalize and validate with use-cases from rare disease registries that collaborate with the EJP RD

- **Available resources:**
- Link to the EJP RD website
- Link to the CDE semantic model on GitHub

Rowdy de Groot, Nirupama Benis, Pablo Alarcón, Rajaram Kaliyaperumal, Marco Roos, Ronald Cornet, Núria Queralt-Rosinach

Amsterdam UMC Universitair Medische Centra

OHDSI

EUROPEAN JOINT PROGRAMME RARE DISEASES