# Building an Application Ontology and Knowledge Graph for Rare Disease Patient-Level Data

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## 1. Introduction

This poster describes the process of developing an application ontology, the Critical Path Ontology (CPONT), and building a knowledge graph from Observational Medical Outcomes Partnership (OMOP) structured data. The integrated knowledge graph is part of the Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®) [1] and will support clinical development in rare diseases.

## 2. Methods and Results

We used the Ontology Development Kit [2] to set up the CPONT development environment. We then used ROBOT [3] to import modules from multiple OBO ontologies. We imported curated mappings defined in OMOP2OBO [4] as well as a list of our own mappings of OMOP vocabulary terms from the patient-level data to terms in the OBO ontologies. To create a knowledge graph, we used a python script to load the patient-level records into a ROBOT template file and used ROBOT to convert the template into OWL instance-level triples. This OWL file is merged with cpont.owl, and lastly, ROBOT query is used to run SPARQL queries on the combined OWL file. CPONT is available at https://gitlab.c-path.org/c-pathontology/critical-path-ontology. The knowledge graph will be available to approved users through the RDCA-DAP portal.

## 3. Conclusion

To our knowledge, this will be the first widely available knowledge graph to encompass patient-level clinical trial and electronic health record data for rare diseases.

## 4. Acknowledgements

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## 5. References

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