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

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The Critical Path Institute is a non-profit public-private partnership with the US FDA.

The RDCA-DAP is an FDA-funded initiative with the goal of accelerating therapy development across rare diseases by providing a centralized and standardized infrastructure to support and accelerate rare disease characterization.

The RDCA-DAP houses integrated patient-level data from diverse sources across a multitude of rare diseases.
We are building an application ontology by importing terms from OBO ontologies, such as HPO, MONDO, MAXO, OBI, UBERON and NCIT.

The knowledge graph includes:

- CPONT
- HPOA disease to phenotype mappings
- MONDO - Orphanet mappings
- Synthetic version of our patient data