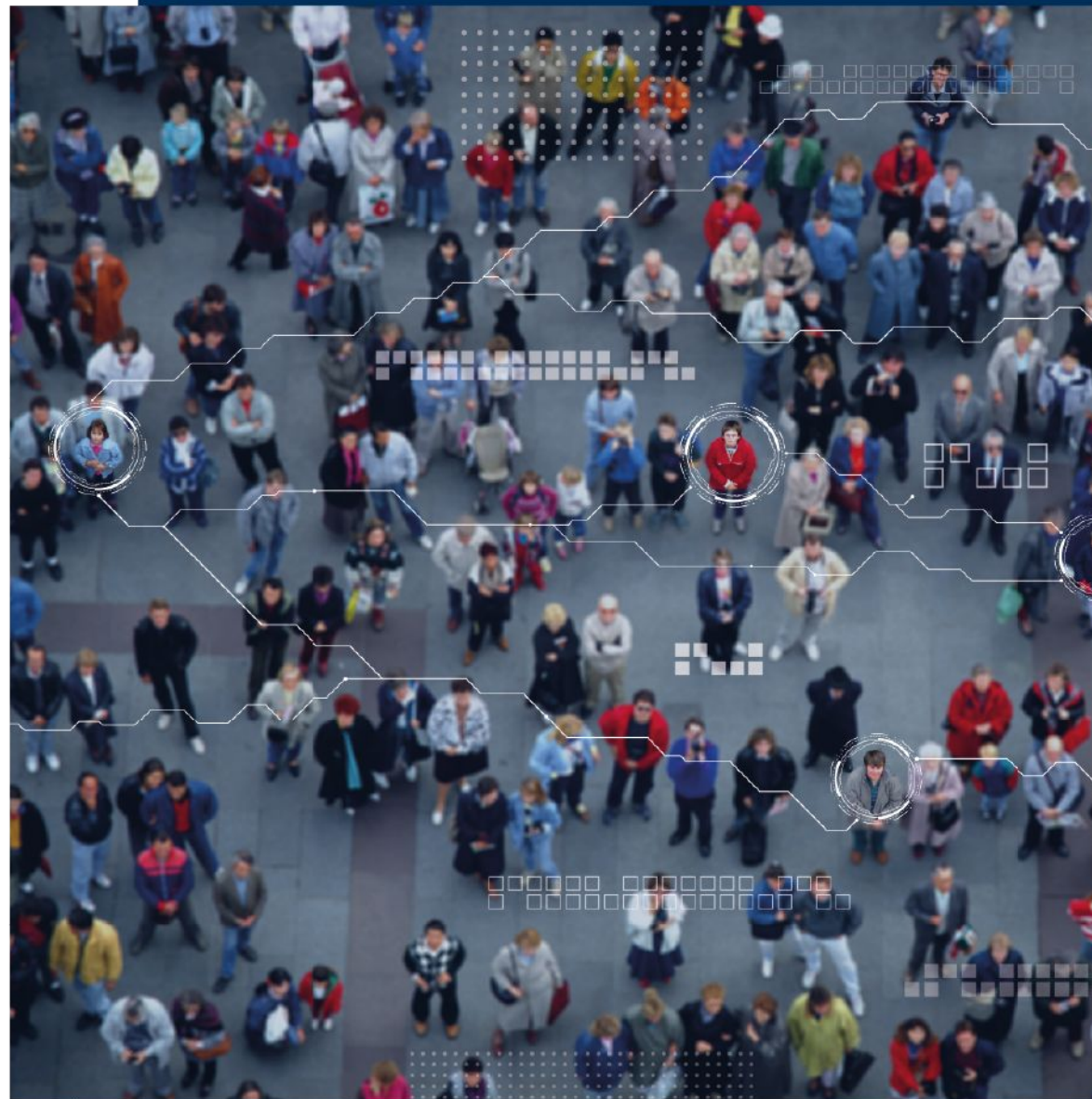


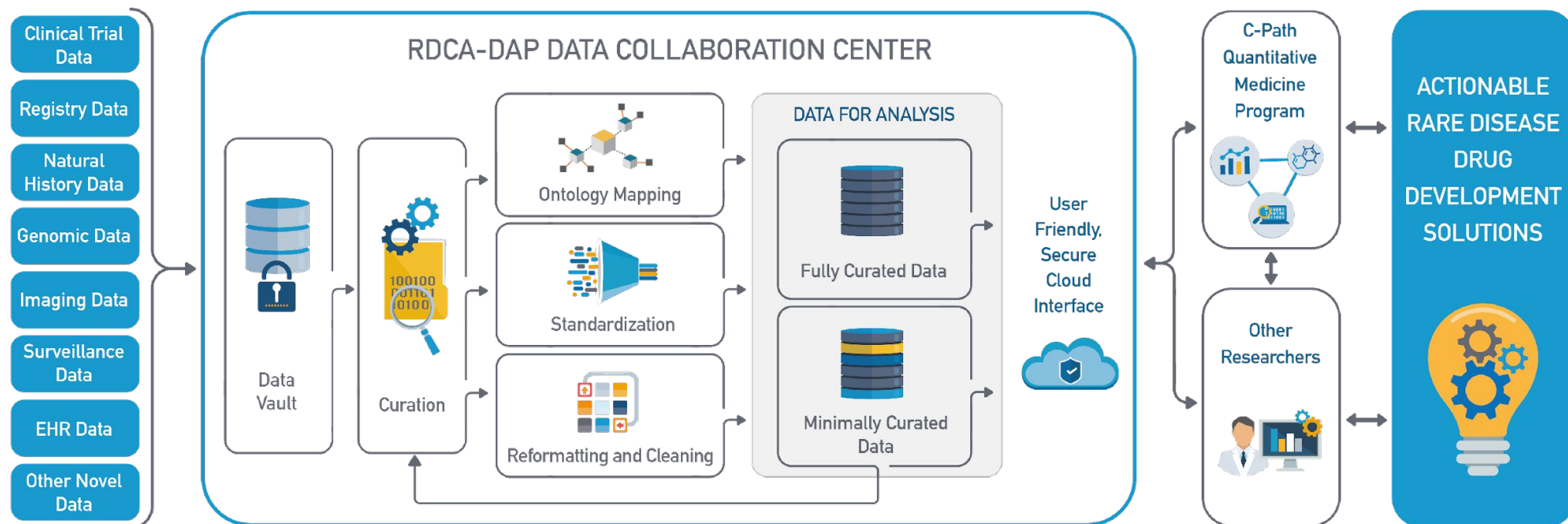


# Building an Application Ontology and Knowledge Graph for Rare Disease Patient-Level Data

Emily Hartley, Daniel Olson, Ian Braun,  
Nicolas Matentzoglu, James A. Overton,  
and Ramona L. Walls



- 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