Anna Bernasconi

PhD candidate, 4th year. Advs: **S. Ceri, A. Campi**
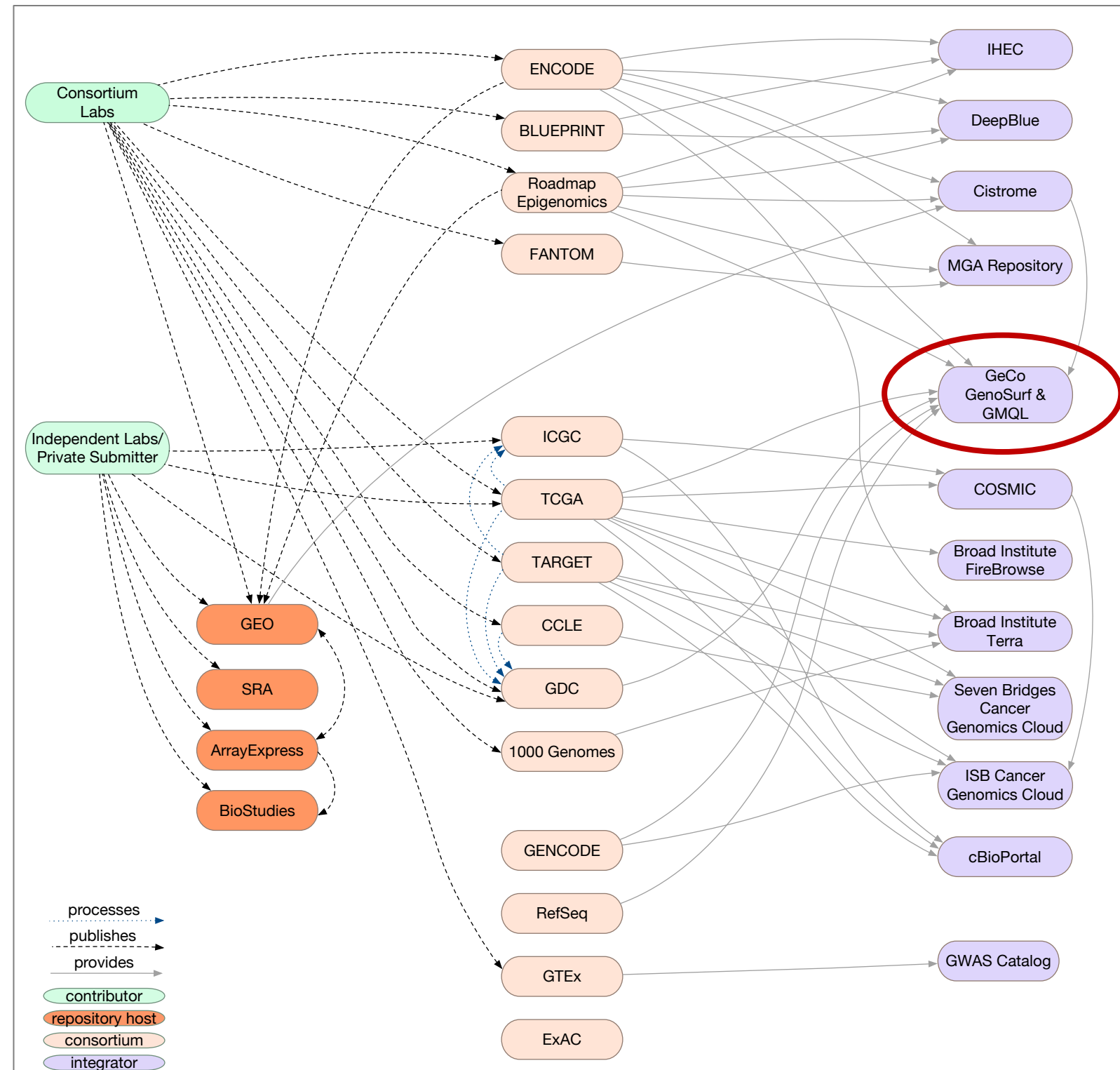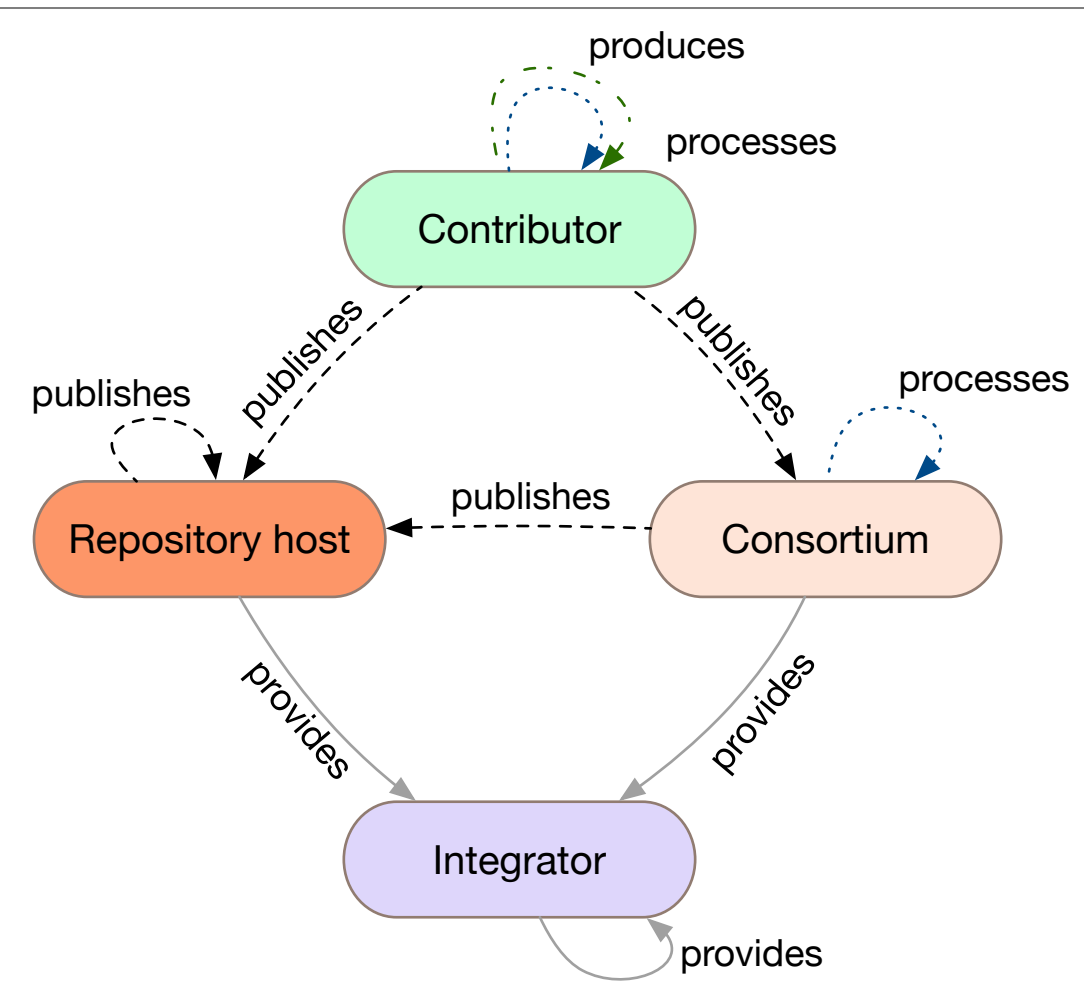Special thanks to: Arif Canakoglu
Thesis submission: October 2020

Building a repository of genomic datasets: modeling and integration of (meta)data for interoperable search and analysis
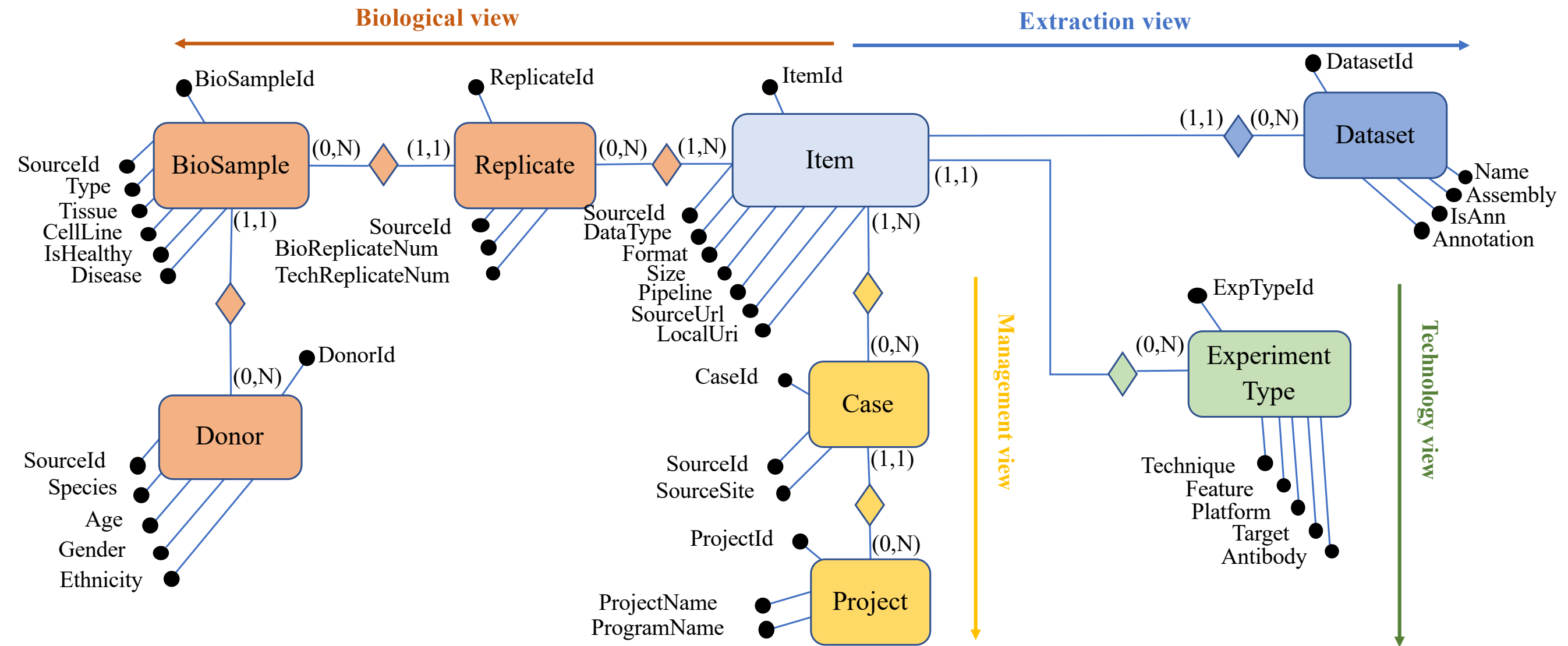
Relevant publications/submissions

- **A. Bernasconi**, S. Ceri, A. Campi, and M. Masseroli. "Conceptual Modeling for Genomics: Building an Integrated Repository of Open Data." In *International Conference on Conceptual Modeling (ER2017).*

- **A. Bernasconi**, A. Canakoglu, M. Masseroli and S. Ceri. "META-BASE: a Novel Architecture for Large-Scale Genomic Metadata Integration". Under review @ *IEEE/ACM Transactions on Computational Biology and Bioinformatics.*

- **A. Bernasconi**, A. Canakoglu, A. Colombo and S. Ceri. "Ontology-Driven Metadata Enrichment for Genomic Datasets". In *International Conference on Semantic Web Applications and Tools for Life Sciences 2018.*

- **A. Bernasconi**, A. Canakoglu, and S. Ceri. "From a conceptual model to a knowledge graph for genomic datasets". In *International Conference on Conceptual Modeling (ER2019).*

- A. Canakoglu, **A. Bernasconi**, A. Colombo, M. Masseroli and S. Ceri. "GenoSurf: Metadata driven semantic search server for integrated genomic datasets". In *DATABASE – The Journal of Biological Database and Curation.*
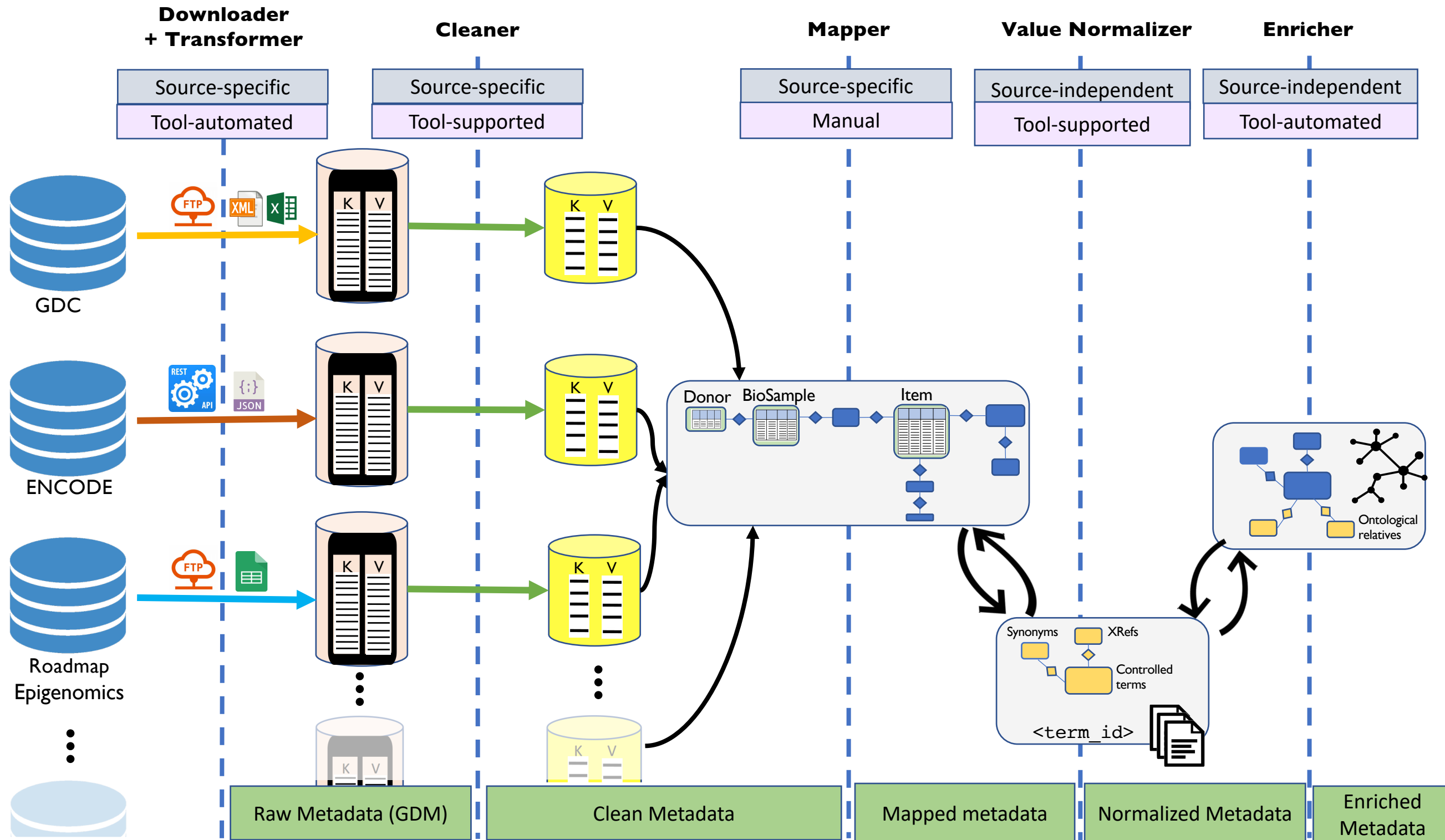
Taxonomy of Genomic Data Players

The Genomic Conceptual Model

Anna Bernasconi, Stefano Ceri, Alessandro Campi, and Marco Masseroli.
"Conceptual Modeling for Genomics: Building an Integrated Repository of Open Data."
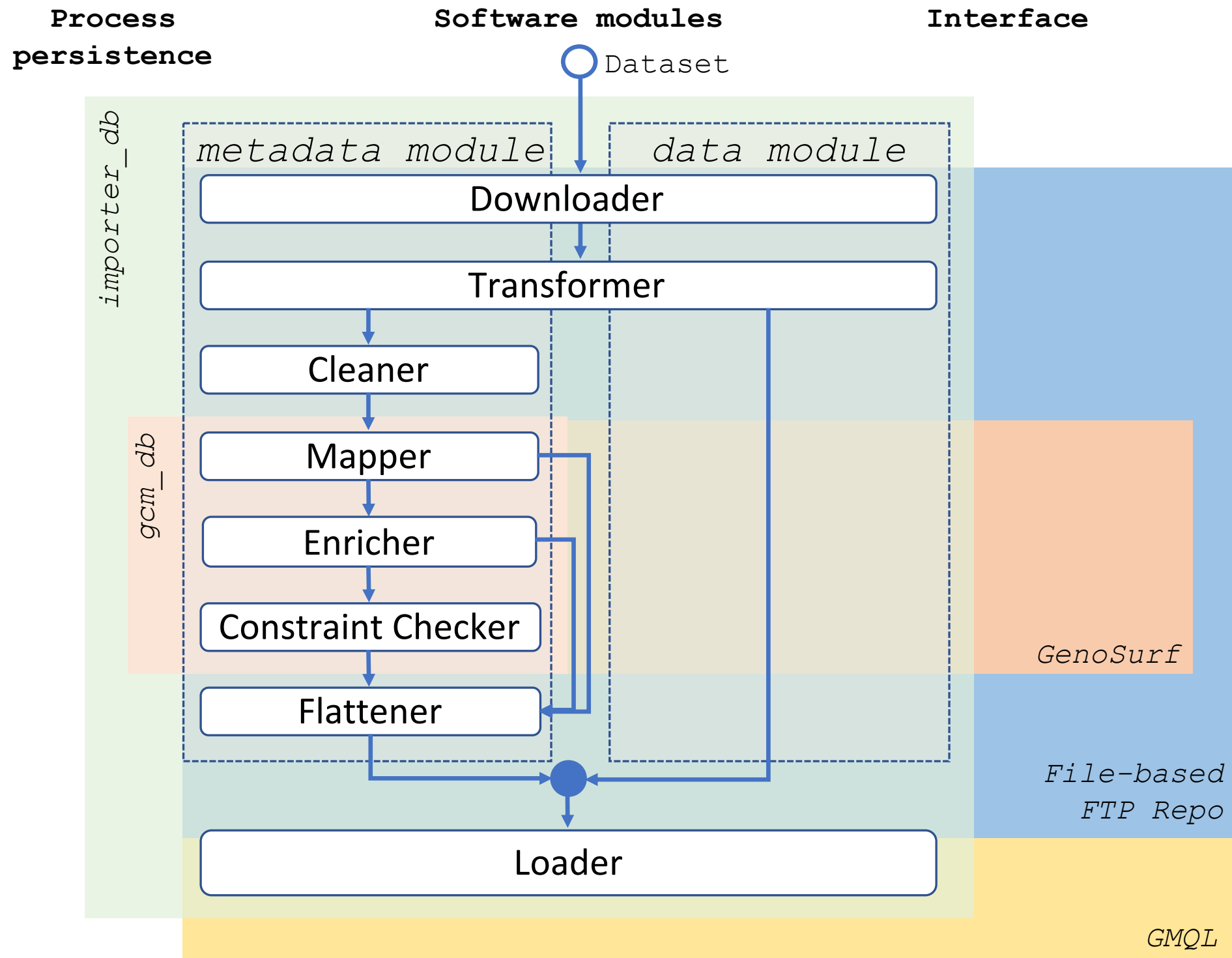In *International Conference on Conceptual Modeling (ER2017)*.
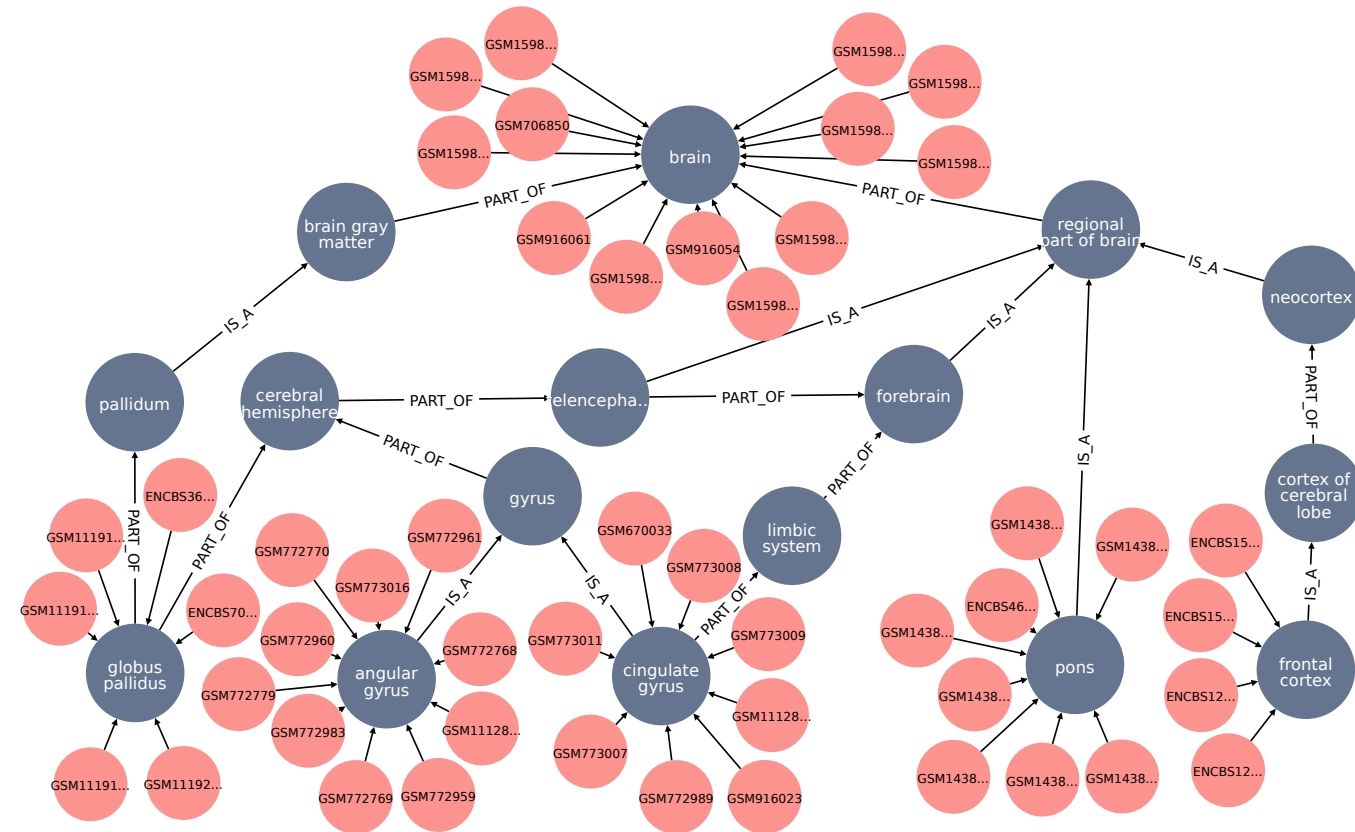
The Data Integration Pipeline

Anna Bernasconi, Arif Canakoglu, Marco Masseroli and Stefano. Ceri.
"META-BASE: a Novel Architecture for Large-Scale Genomic Metadata Integration".
Under review @ *IEEE/ACM Transactions on Computational Biology and Bioinformatics*.
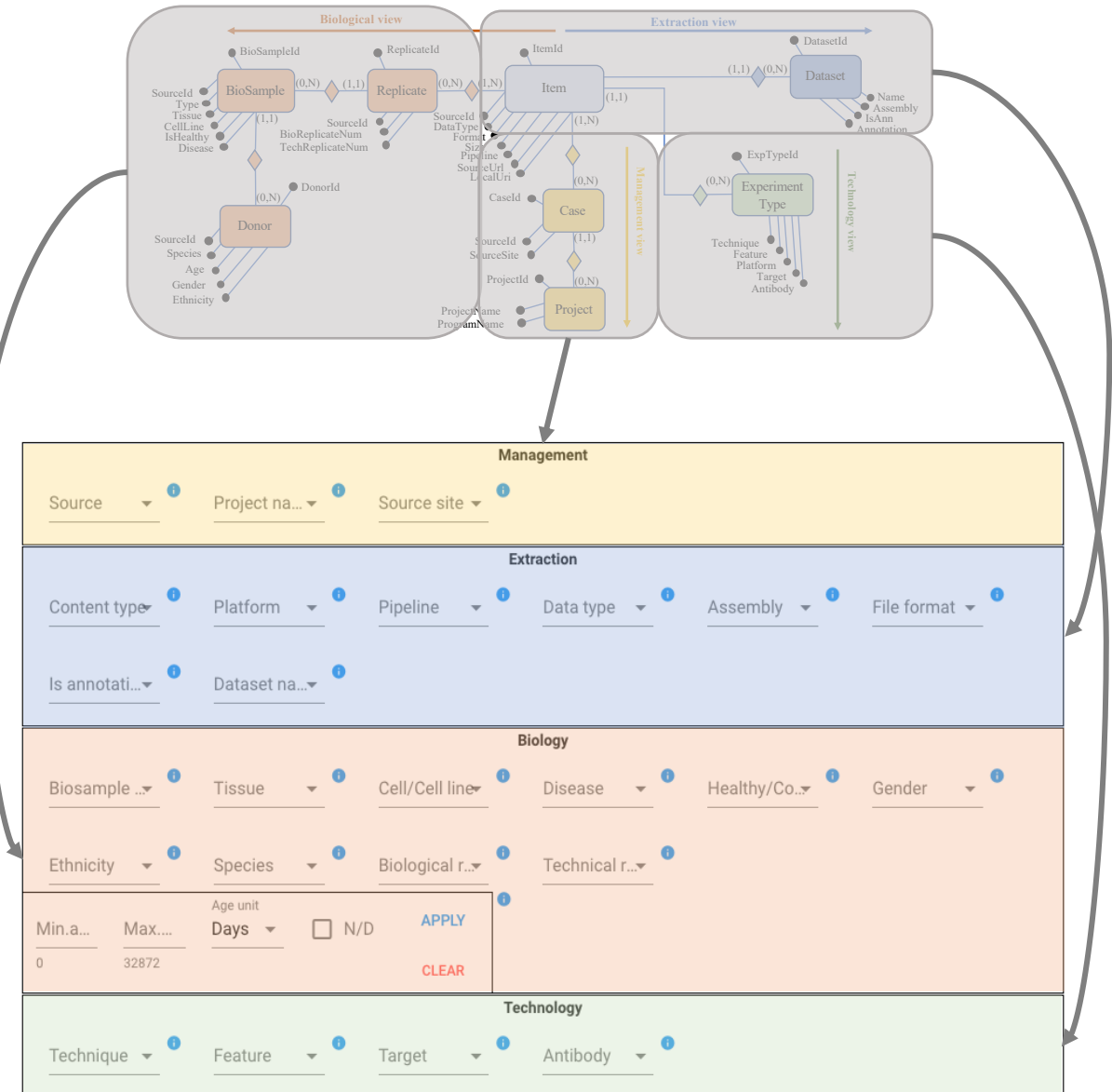
Importer software implementation

Interfaces: GeKnowGraph and GenoSurf

Anna Bernasconi, Arif Canakoglu, and Stefano Ceri. "From a conceptual model to a knowledge graph for genomic datasets".
In *International Conference on Conceptual Modeling (ER2019)*.

Arif Canakoglu, **Anna Bernasconi**, Andrea Colombo, Marco Masseroli and Stefano Ceri. "GenoSurf: Metadata driven semantic search server for integrated genomic datasets".
In *DATABASE — The Journal of Biological Database and Curation*.

Data Quality in Genomic Data Integration
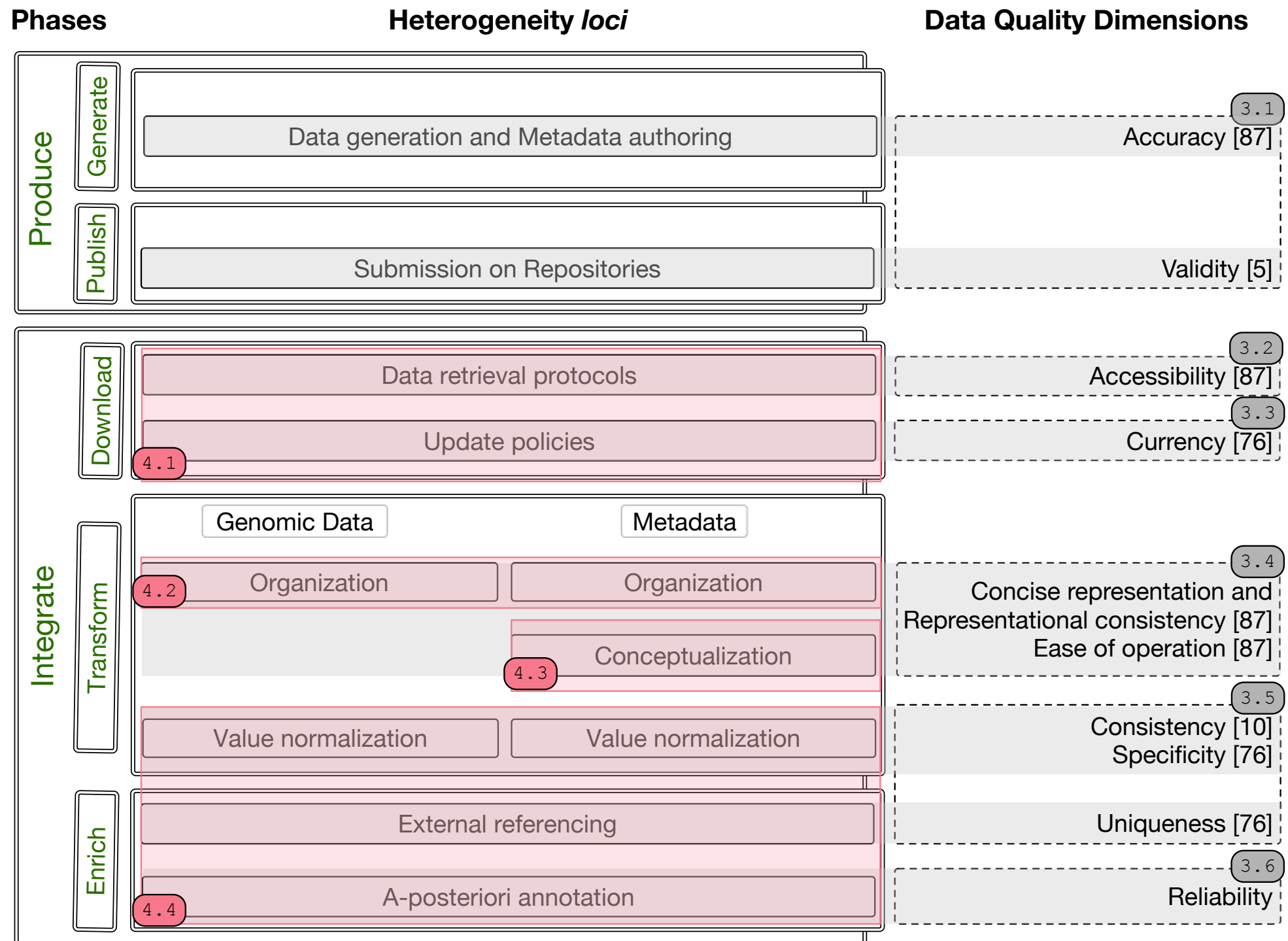
Repository's current data sources coverage

National Genomic Data Initiatives: A Worldwide Update

Brazil
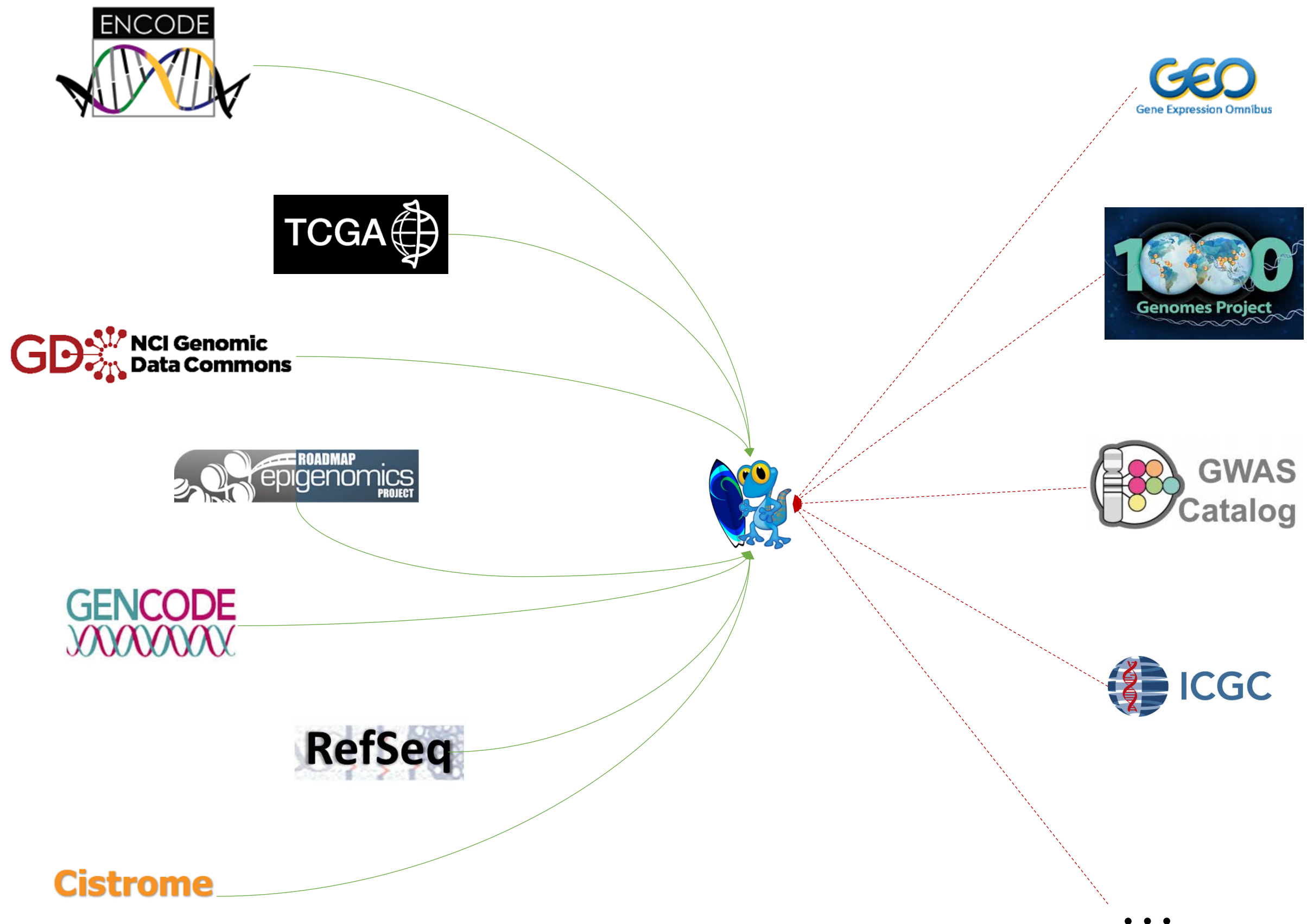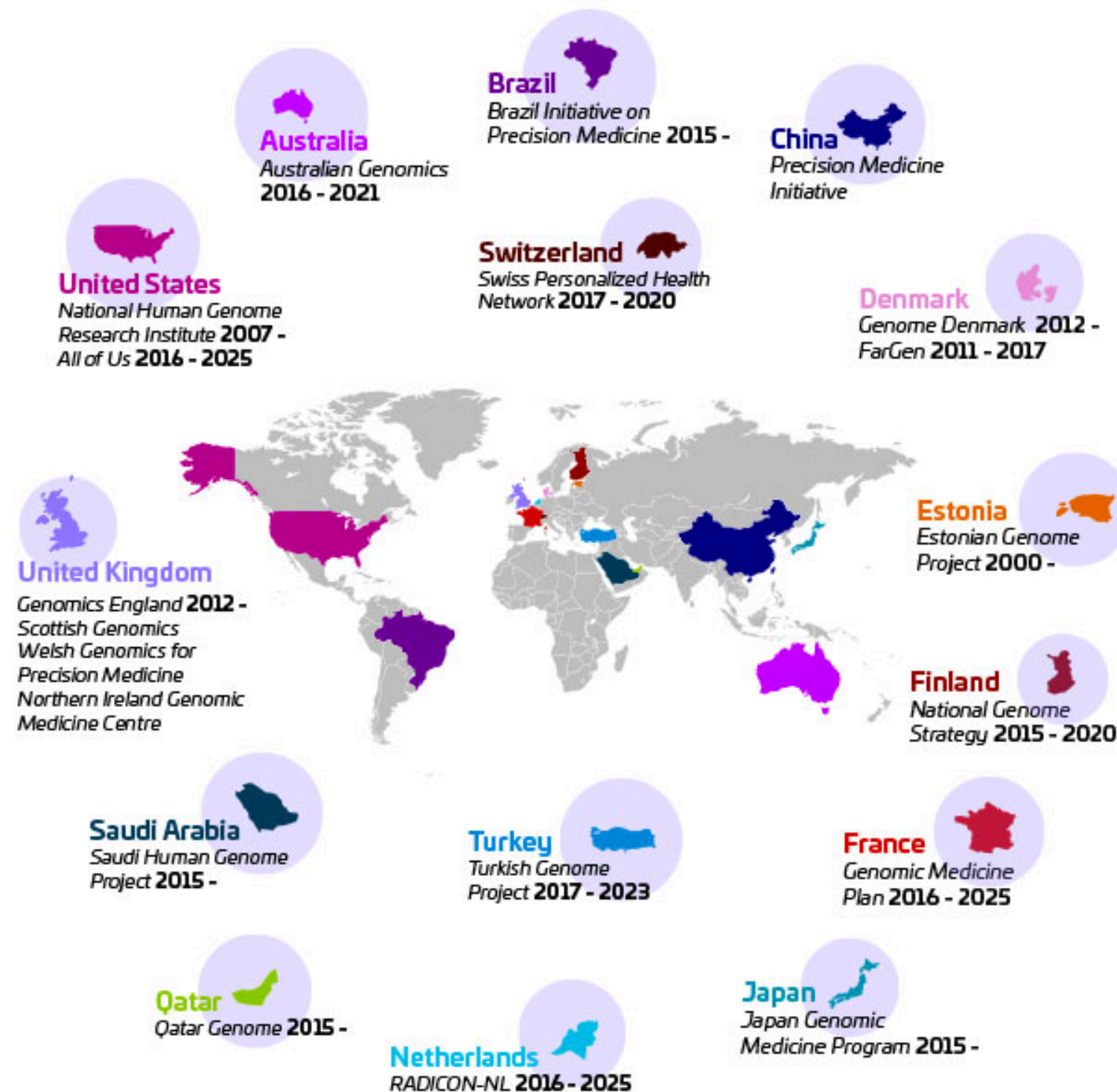Brazil Initiative on Precision Medicine **2015 –**

China
Precision Medicine Initiative

Australia
Australian Genomics **2016 - 2021**

Switzerland
Swiss Personalized Health Network **2017 - 2020**

Denmark
Genome Denmark **2012 –**
FarGen **2011 - 2017**

United States
National Human Genome Research Institute **2007 –**
All of Us **2016 - 2025**

Estonia
Estonian Genome Project **2000 –**

United Kingdom
Genomics England **2012 –**
Scottish Genomics
Welsh Genomics for Precision Medicine
Northern Ireland Genomic Medicine Centre

Finland
National Genome Strategy **2015 - 2020**

Saudi Arabia
Saudi Human Genome Project **2015 –**

Turkey
Turkish Genome Project **2017 - 2023**

France
Genomic Medicine Plan **2016 - 2025**

Qatar
Qatar Genome **2015 –**

Netherlands
RADICON-NL **2016 - 2025**

Japan
Japan Genomic Medicine Program **2015 –**

Source: https://www.bio-itworld.com/2019/08/12/national-genomic-data-initiatives-worldwide-update.aspx

Thank you for your attention!