CliniPhenome: Clinical and Phenotypic Annotation Database

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Background

- Advancement in clinical diagnostic processes encompassing techniques akin genome/exome sequencing is governing generation of data in heaps.
- The aim of this amelioration is not only effective diagnosis, conjointly also to make the data sharable for research, to accelerate progress in biomedical sciences from bench to bedside to community.
- For translational research, it's imperative that researchers work simultaneously with the clinicians for interpretation and application.
- Need embodiment of the clinical and phenotypic annotations with the vocabulary that is interpretable by both the clinical staff as well as the researchers.
- **BRiDGEIRIS project -** Aims to develop a big data platform that can be used for sharing genomic and clinical data and support discovery studies in clinical genomics.

Goals

- A major aspiration of the project is structuring a database that supports patients data against the genetic diseases focused upon by the genetic centers and support for disparate phenotype with clinical ontologies.
- Proposal of a generalized clinical schema design along with a merged ontology schema (Phenotype and Clinical) that provides cross-referencing possibilities.
- Introduction of controlled vocabulary and data structure that answer for the prevalent problem of plain text storage.

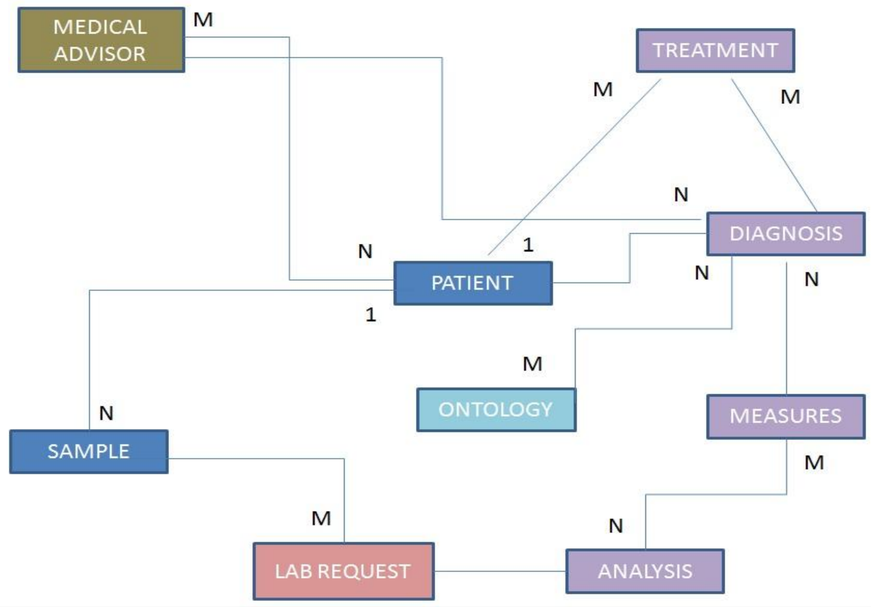
Research Outcome

- Development of a PHP based MySQL application termed *CliniPhenome* that supports clinical and phenotypic measures for a patient with any kind of disease.

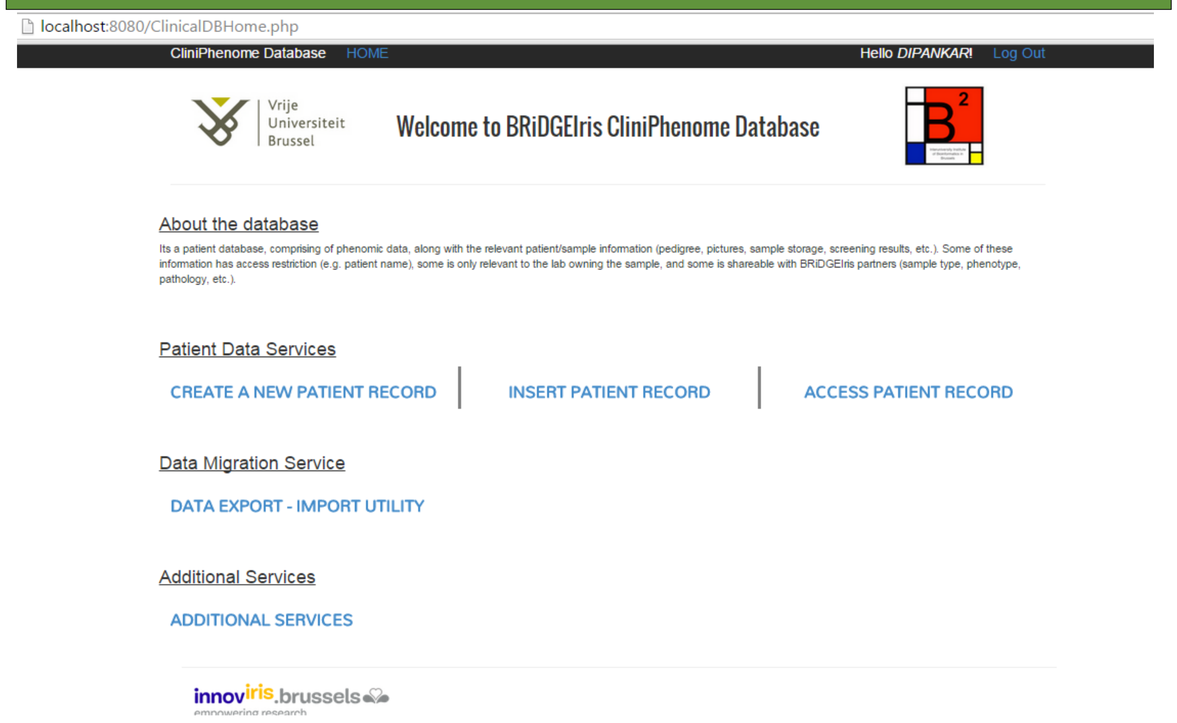
CliniPhenome Key Aspects

- *CliniPhenome* supports patient clinical and phenotypic detailed information.
  - ❖ Generalized schema design to support measures for all the diseases.
  - ❖ Support for **phenotypic ontology** - HPO, OMIM, Orphanet; and **clinical ontology** – LOINC; SNOMED and ICD-10 (to be integrated) for annotation purpose.
  - ❖ Interactive user interface for data handling for the medical staff and researchers.
- Secured data access: 3 kinds of users (Administrator, Privileged, Restricted) with tailored rights.
- De-identified data exchange/migration utility that allow data sharing between hospitals/centers.

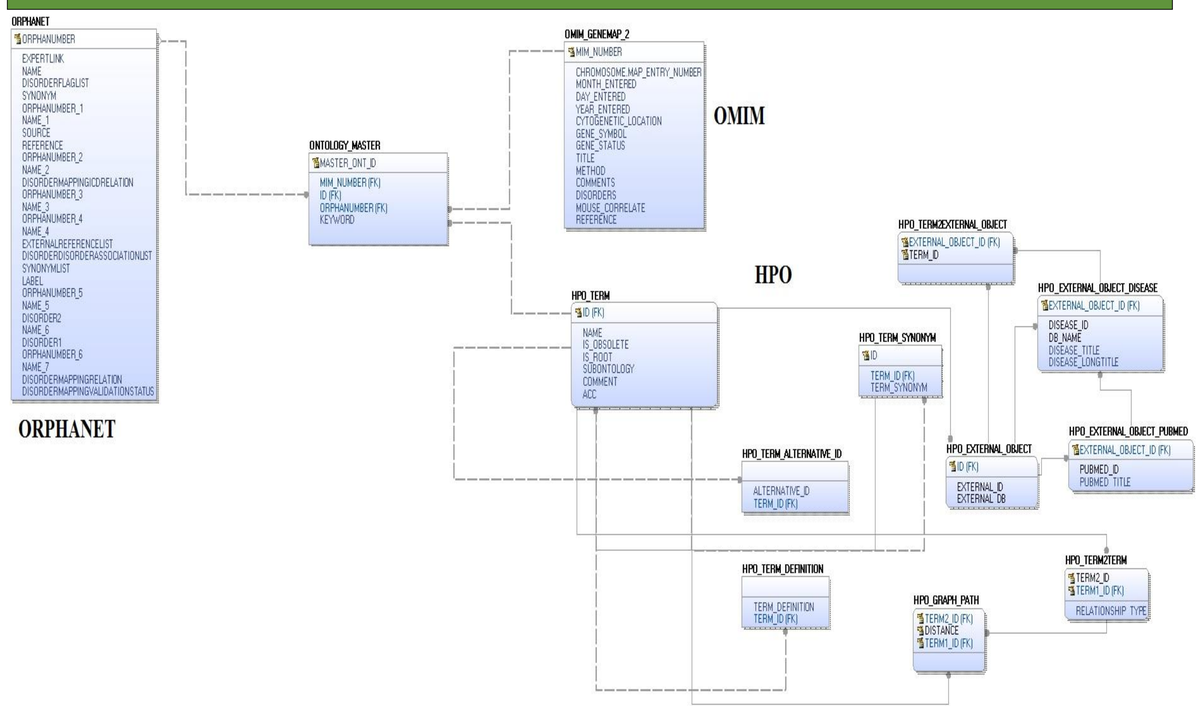
Data Flow Relationship



Sneak Peek of CliniPhenome



Merged Phenotype Ontology Schema



Future Direction

- Further, the project intention is to agile the application interface of *CliniPhenome* for equipping users (clinicians and researchers) with customization feature.
- Also, as part of Big data solution of BridgeIRIS project, *CliniPhenome* is being made scalable and being worked upon is its integration with genomic database.

Acknowledgement

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