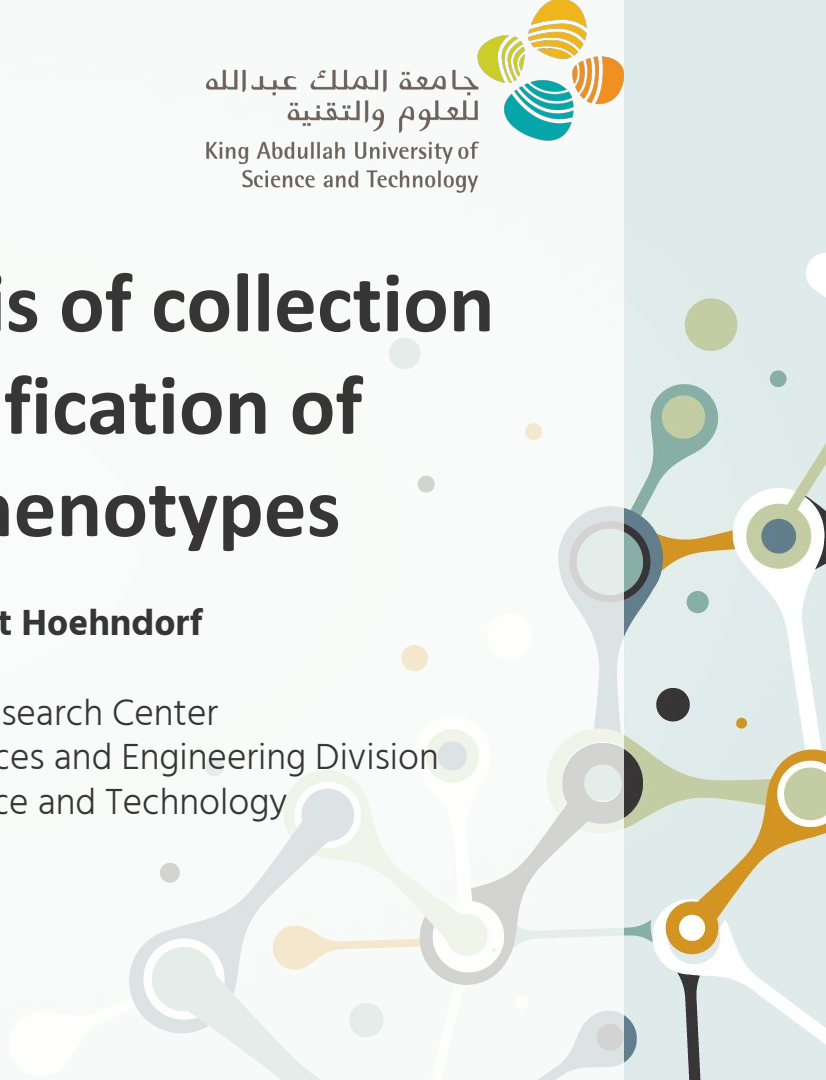#7

Ontological analysis of collection improves classification of cardinality phenotypes
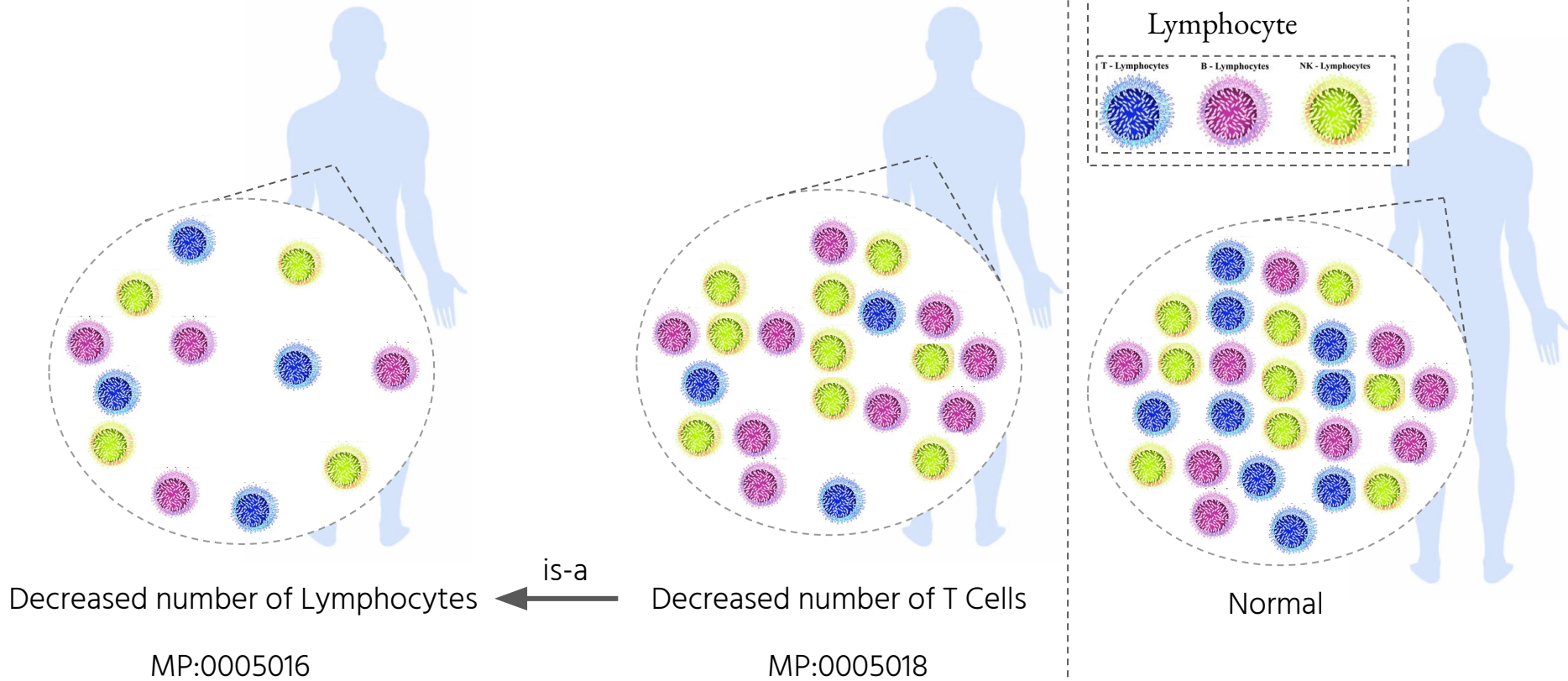
Sarah M. Alghamdi, Robert Hoehndorf

Computational Bioscience Research Center
Computer, Electrical & Mathematical Sciences and Engineering Division
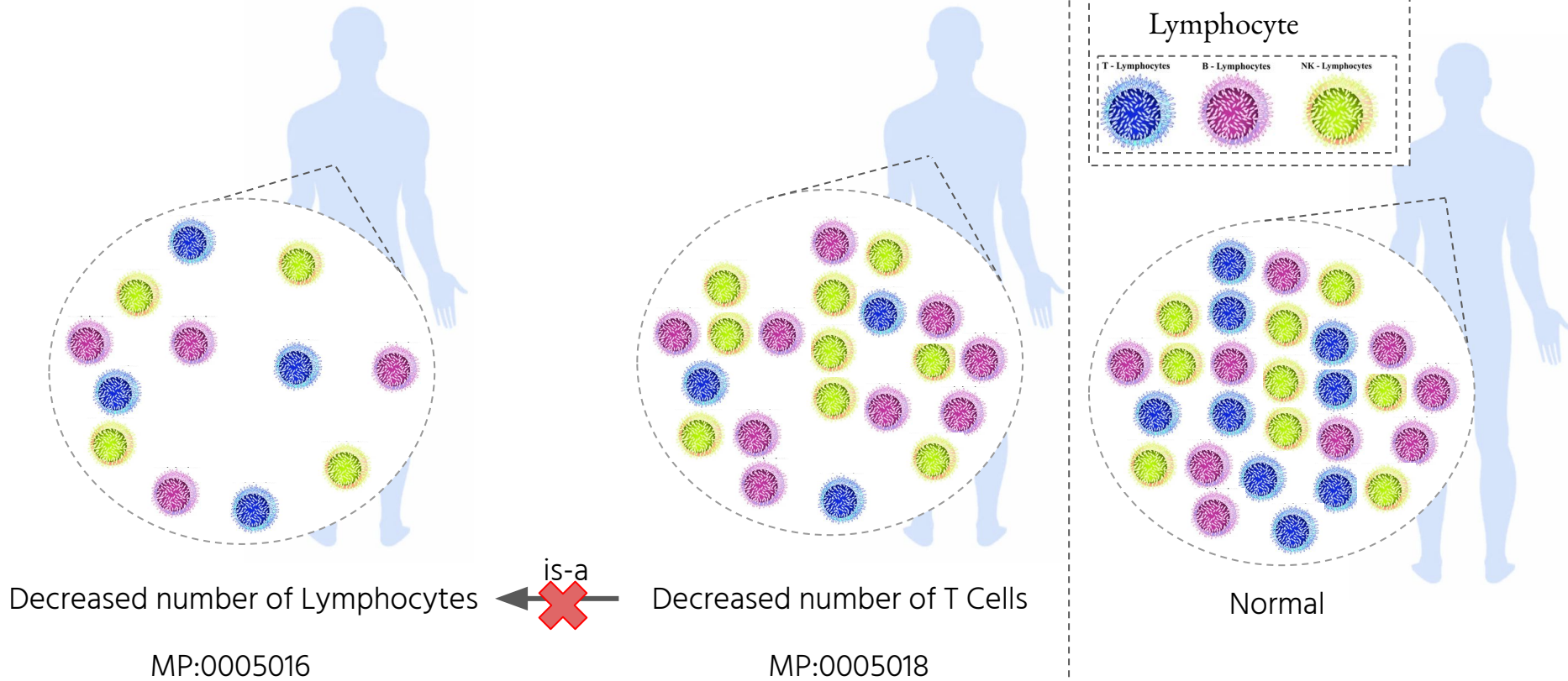King Abdullah University of Science and Technology

Motivation

- Phenotype data is critical for understanding the biological mechanisms of a disease.

- Many ontologies cover the domain of phenotypes for specific organisms: HPO, MP, ...

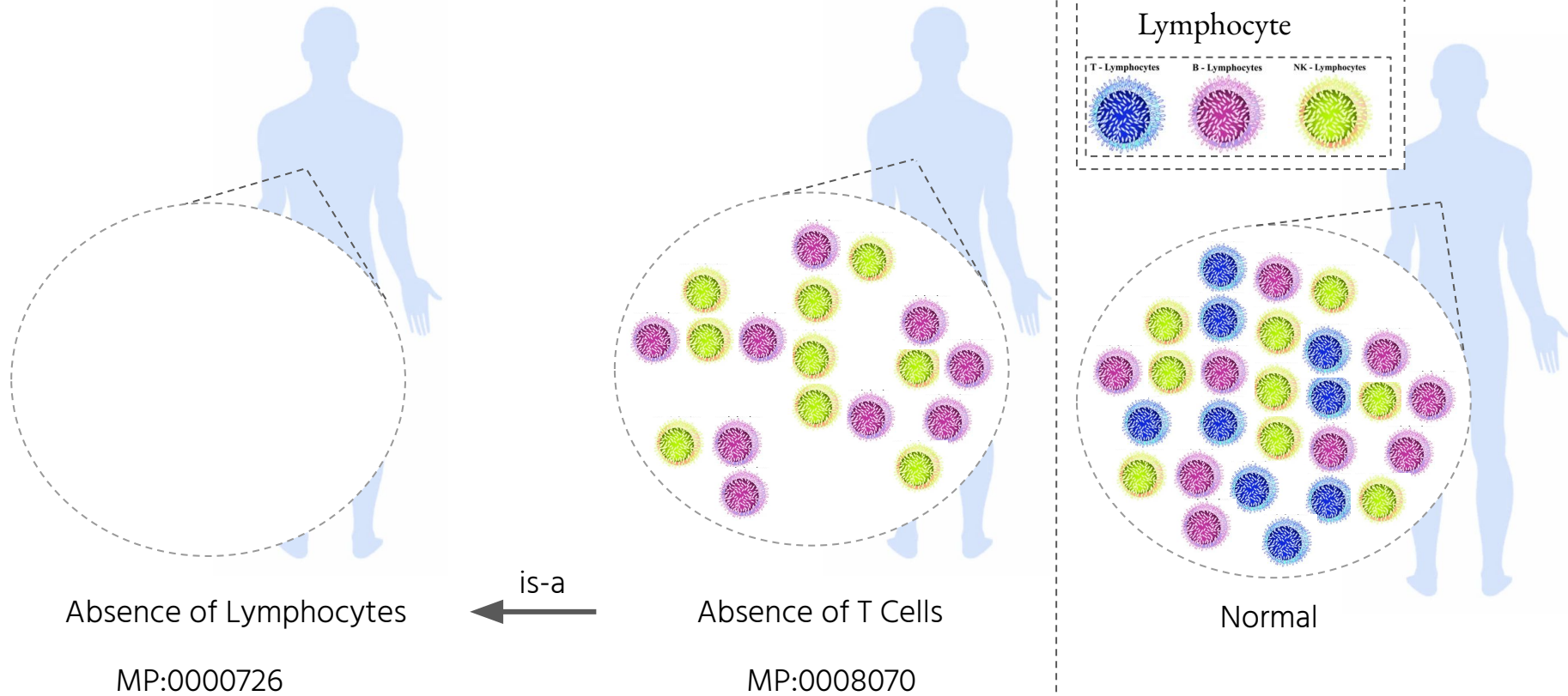- However, those ontologies have some potentially incorrect inferences
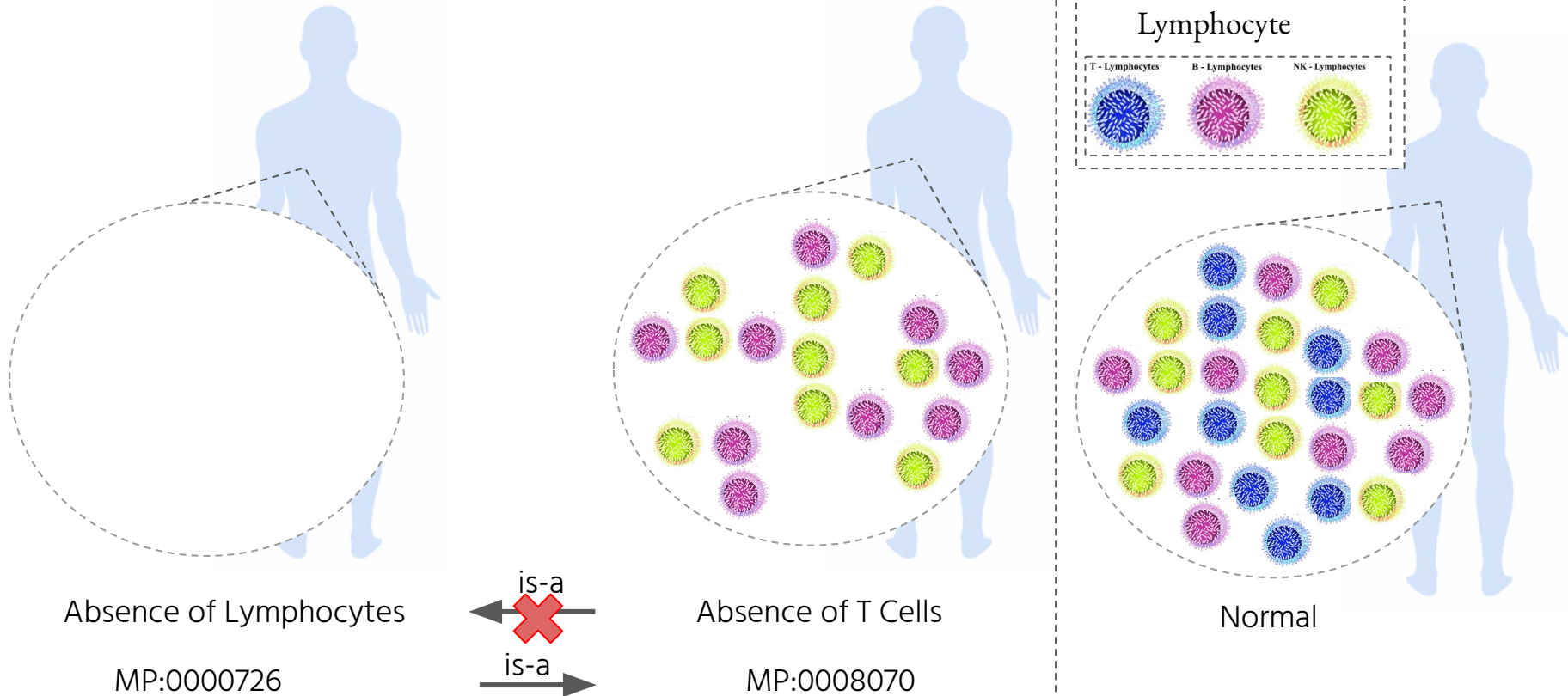
Motivation

Decreased number of Lymphocytes

MP:0005016

is-a

Decreased number of T Cells

MP:0005018

Normal

Lymphocyte

T - Lymphocytes   B - Lymphocytes   NK - Lymphocytes

Motivation

Lymphocyte

T - Lymphocytes   B - Lymphocytes   NK - Lymphocytes

Decreased number of Lymphocytes

is-a

Decreased number of T Cells

Normal

MP:0005016

MP:0005018

Motivation

Absence of Lymphocytes ← is-a ── Absence of T Cells

Normal

MP:0000726

MP:0008070

Motivation

Lymphocyte

T - Lymphocytes   B - Lymphocytes   NK - Lymphocytes

Absence of Lymphocytes

MP:0000726

is-a

is-a

Absence of T Cells

MP:0008070
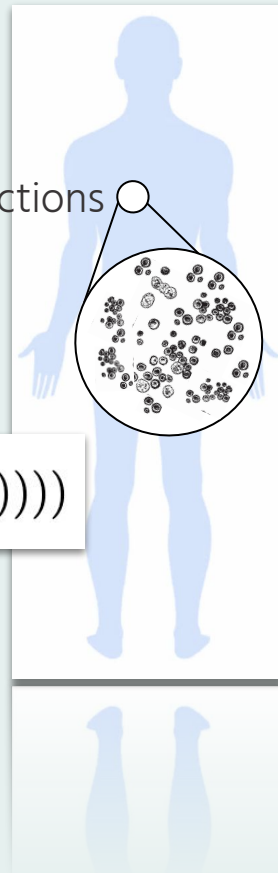
Normal

Cardinality Phenotypes

- We propose reformulating cardinality phenotypes to describe abnormalities of collections

- FOL:

$$?CX(x) \leftrightarrow (\exists y(Body(y) \wedge (\forall a(?X(a) \wedge part\_of(a,y)) \rightarrow member\_of(a,x))))$$
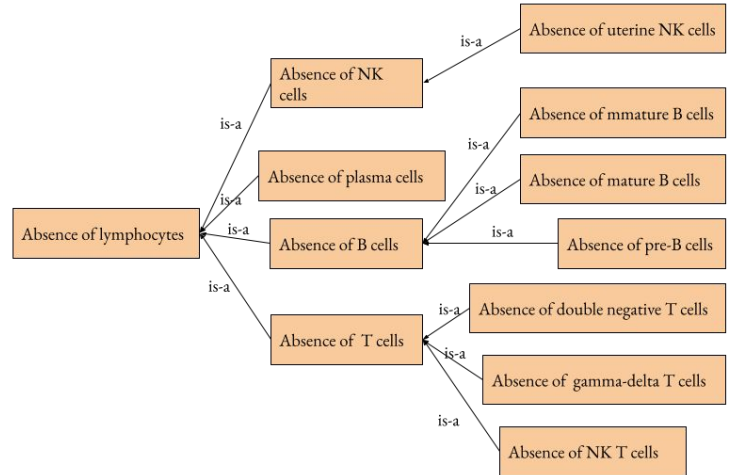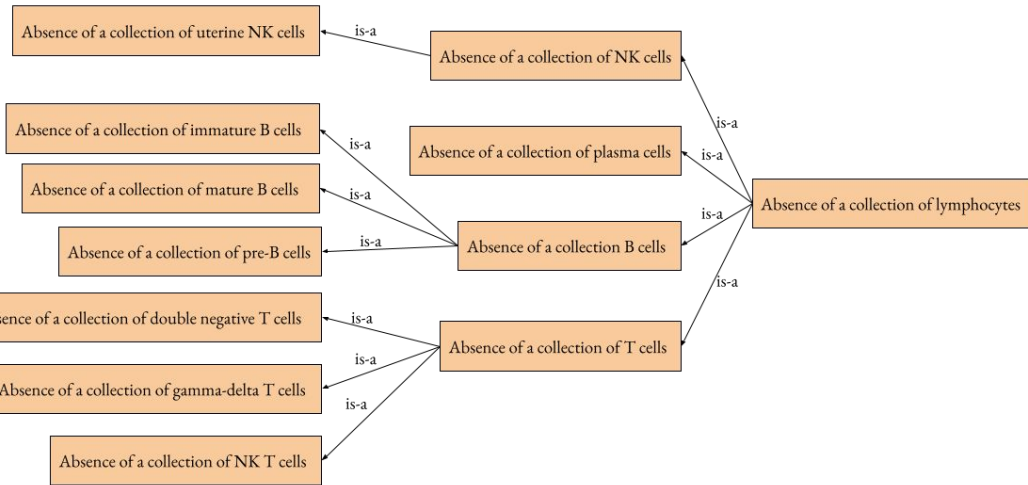
- OWL DL for collection of cells:

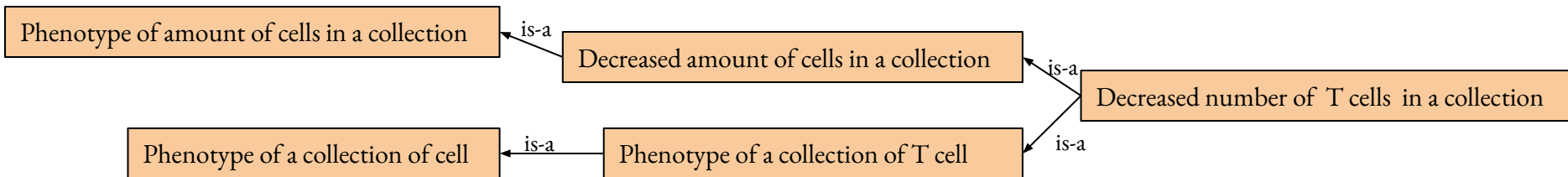$$?CX \sqsubseteq \forall member\_of.?X$$

Results & Evaluation

Results & Evaluation

Using collections
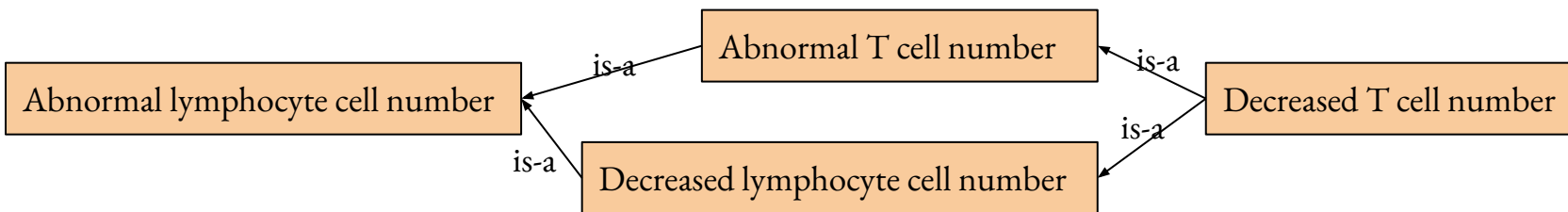
Phenotype of amount of cells in a collection ←is-a Decreased amount of cells in a collection ←is-a Decreased number of T cells in a collection

Phenotype of a collection of cell ←is-a Phenotype of a collection of T cell ←is-a

Not using collections

Abnormal lymphocyte cell number ←is-a Abnormal T cell number ←is-a Decreased T cell number

Decreased lymphocyte cell number ←is-a

#7

Come to the poster #7 for more details