Title

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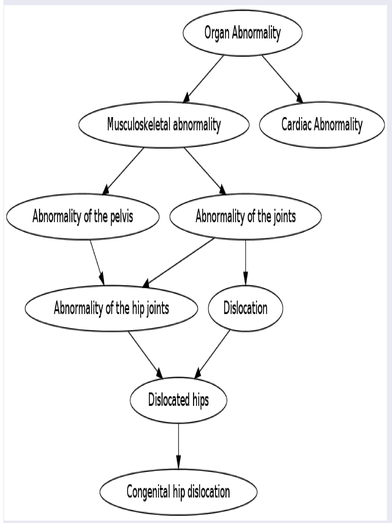
Introduction

Rare genetic disorders are caused by abnormalities in human genome. Due to different levels of expressions and influences from the environment, even patients with the same disorder may exhibit varying symptoms, which is a challenge for accurate diagnosis. However, clustering multiple patients with similar symptoms and utilizing the structural information in ontologies, we can uncover more information from patients.

Motivation



- HPO (Human Phenotype Ontology) provides structural information about phenotypes
- OMIM (Online Mendelian Inheritance in Man) annotates disorders with HPO terms
- These two resources enable us to develop more informed algorithms to analyse patients

Objective

- Cluster patients with similar phenotypes
- Perform diagnoses on patients

Methods

Clustering Method

Cluster Method

Diagnosis Method

- The objective is to measure how close a patient is to the canonical form of a disorder
- Establish an one-to-one matching between the set of patient's phenotypes and annotations of a disorder
- The cost of transforming from the real patient to the canonical patient is the measure for closeness

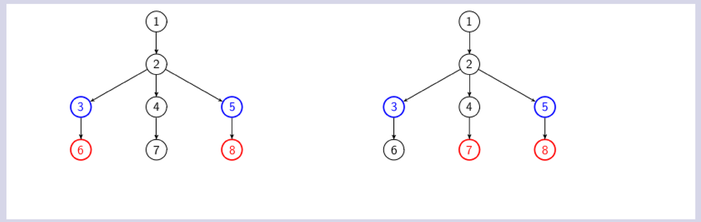


Figure 1: A Toy Ontology Example

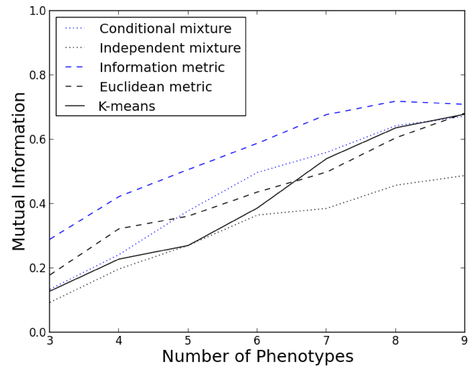


Figure 2: Comparisons of Different Clustering Methods

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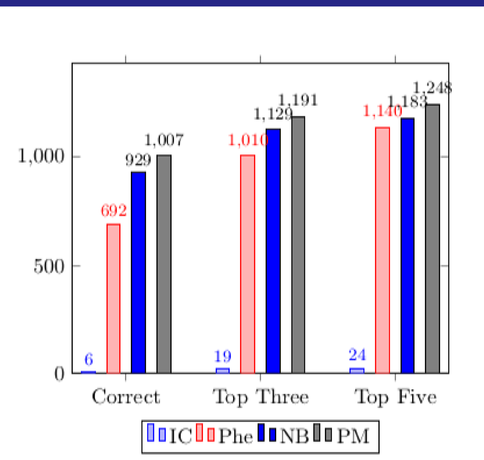

Figure 3: Diagnoses Results for 1498 Simulated Patients