Developing Inference Model to Diagnosis of Primary Immunodeficiency Diseases in Protégé

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Received: 03 Jan. 2016; Revised: 03 Aug. 2016, Accepted: 04 Aug. 2016

Primary immunodeficiency diseases (PIDs) are a genetically heterogeneous group disorders that affect distinct components of both humoral and cellular arms of the immune system (1,2). Overlapping signs and symptoms of these diseases is a challenge for diagnosis and treatment (3,4). Awareness of the symptoms and considering the possibility of PID in differential diagnosis help to rapid recognition and more appropriate treatment (2,5). Timely recognition and treatment reduced mortality and increased lifespan and quality of life of the patients (6). Memorization of all effective criteria to diagnosis is difficult, so developing a computerized program based on diagnosis criteria, improves significantly the quality of care (7,8).

To develop the inference model to the diagnosis of PIDs, ontology has been used in this study. The study focused on eight common diseases of PIDs include Common Variable Immune Deficiency (CVID), X-Linked Agammaglobulinemia (Bruton's) (XLA), Selective IgA Deficiency (SIgA), CD40L deficiency, UNG deficiency, Isolated immunoglobulin (Ig) G Subclass deficiency, Specific antibody deficiency (SAD) with normal Ig concentrations and normal numbers of B cells, Transient Hypogammaglobulinemia of infancy (THI) with normal numbers of B cells. Based on clinical guidelines and medical literature in PID (9), we designed a checklist to extract and classified most important signs and symptoms, family history, and laboratory data for eight main type of primary antibody deficiencies (PADs). To evaluate the quality of checklist, data for 100 cases in a different type of PADs were tested. Using frame-based ontology modeling to create the inference model and "Noy and McGuinness" method to develop the inference model. "Noy and McGuinness" method includes seven stages (10). Below we describe each stage of the method:

1. Determine the domain and scope of the ontology: The aim of this study is developing inference model that could help physicians in order to diagnose the PIDs according to the symptoms of patients. The study area is eight PIDs and its signs and symptoms.

2. Consider reusing existing ontologies: Today, rarely an ontology is developed from scratch (11). In this study to develop the ontology, we used expert's knowledge.

3. Enumerate important terms in the ontology: Make a list of related terms that are expected exist in an ontology, is the first step of ontology definition (11). In this study, a list of related terms include the eight PIDs and their symptoms, signs, and lab data to diagnosis them, were extracted from clinical guidelines and medical literature under the immunodeficiency specialist opinions.

4. Define the classes and the class hierarchy: After identifying related terms, these terms should be classified as a hierarchy (11). "Class" defines a set of instances that have common features (12). In this study, classes were determined based on this definition.

5. Define the properties of classes: Properties are links and relationships between classes (11)."Object type

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Property" is used for the relationship between instances of two classes. "Data type Property" define data type values for instances (12).

6. Define the facets of the properties: Facets could be defined for properties. Properties can have different facets describing the value type, allowed values, the number of the values (cardinality), and other features of the values the property can take (10).

7. Create instances: Instances are considered as members of a class (12). An ontology with instances of classes constitutes a knowledge base (10). Defining an instance of a class requires 1) choosing a class, 2) creating an instance of that class, and 3) filling in the property values (10). The eight selected PIDs are instances in this model.

The diagnosis inference model developed in 5 main classes and 24 subclasses as hierarchical. To execute the inference model use the query tab of protégé software. The symptoms are entered as input, and related diseases are returned as output. Whereas this model is developed with deterministic data and based on knowledge, the model responds correctly to the question that related knowledge exist in the knowledge base and not return any answer where the related knowledge not exist. At the end, we investigate that the model was correctly analyzed and recognized eight PIDs or not, so the inference model was tested with ten patient's record data and the model diagnosed all ten patient correctly as same as a specialist. However, it would be needed to run a test with more cases to find exact accuracy. Also integrating clinical guidelines and diagnostic criteria with information systems and electronic medical records, increase following the clinical guideline, improve the quality of patient care, ensure patient safety and reduce costs.

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