BUILDING A FAMILY ONTOLOGY TO MEET CONSISTENCY CRITERIA

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For my beloved mother and father.

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ABSTRACT

Semantic web is an extension of the current web in which the existing information on the web are organized and encoded more meaningfully using ontology language, thus enabling effective communication among machines and humans. Ontology is the backbone of the semantic web that contributes to knowledge sharing among intended parties over distributed systems around the world. In the past few years, semantic web has been widely accepted by a variety of fields for better knowledge representation, communication, sharing and reasoning on the web. Now, there are existing genealogical ontologies proposed by different groups of researchers once semantic web has emerged as third generation of the web. However, existing ontologies still lack certain important concepts and properties to support the domain of family relations. This may lead to the inability of the ontology to deliver full potential of exchanging family history information among all interested parties. Moreover, existing ontologies do not employ the full potential of SWRL rules to reason the individuals within the ontology. The main aim of this research is to build a new Family Ontology which obeys the consistency criteria. Consistency checking ensures there are no contradictory concepts found within the resulting ontology. The consistency of Family Ontology will be evaluated using FACT++, HermiT and Pellet reasoners. By augmenting the additional axioms and testing the resulting ontology thoroughly using reasoner tools, the proposed Family Ontology is expected to achieve a consistency of 100%.This research is meaningful and significant to all humans since everyone has his or her own unique family history. The proposed ontology also facilitates effective and efficient communication among all intended parties since shared vocabularies and standards are employed by the proposed ontology.

ABSTRAK

Web Semantik ialah teknik terbaru yang membolehkan data pada Web zaman terkini disusun dan diaturcara secara bermakna dengan menggunakan bahasa ontologi. Ontologi struktur memudahkan komunikasi berlangsung secara efektif antara komputer dan manusia. Ontologi umpama tulang belakang bagi Web Semantik yang menyumbang kepada perkongsian maklumat antara pihak-pihak tertentu melalui rangkaian Internet di seluruh dunia. Web Semantik telah mendapat sambutan meluas dalam pelbagai bidang pada hari ini dan ia merupakan cara terbaik untuk mengekodkan data-data bagi tujuan komunikasi, perkongsian dan reasoning pada Web. Terdapat beberapa genealogi ontologi telah dicipta sejak kebelakangan ini dan kesemuanya telah dicadangkan oleh penyelidik-penyelidik berlainan apabila Web Semantik muncul sebagai Web generasi ketiga. Namun, ontologi yang sedia ada masih kekurangan konsep dan relasi penting bagi menyokong keluarga domain. Hal ini menyebabkan ontologi tidak mampu menunjukkan potensi sepenuhnya dalam perkongsian maklumat sejarah keluarga antara semua pihak. Tambahan pula, genealogi ontologi yang sedia ada tidak menggunakan fungsi peraturan SWRL sepenuhnya bagi tujuan reasoning pada individu-individu dalam ontologi. Matlamat utama kajian ini adalah untuk menghasilkan satu Ontologi Keluarga yang memenuhi kriteria konsisten. Ujian konsisten memastikan tiada konsep yang bertentangan di dalam ontologi. Konsistensi akan dinilai dengan menggunakan FACT++, HermiT and Pellet. Dengan memasukkan aksioma tambahan dan memeriksa ontologi secara teliti, Ontologi Keluarga yang dicadangkan dianggap telah mencapai konsistensi 100 peratus. Kajian ini amat bermakna dan agak penting terhadap semua manusia kerana setiap orang memiliki sejarah keluarga mereka yang unik. Ontologi yang dicadangkan ini turut membolehkan komunikasi berlangsung secara berkesan dan efektif antara semua pihak kerana kosa kata dan standard yang sama sentiasa dirujuk oleh semua pihak.

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LIST OF SYMBOLS AND ABBREVIATIONS

ABOX	-	Assertional Box
CBR	-	Case Based Reasoning
CDSS	-	Clinical Decision Support System
DL	-	Description Logic
EL	-	Expressive Language
FACT++	-	Fact Plus Plus
GEDCOM	-	Genealogical Data Communication
GENTECH	-	Genealogical Data Model
GNU	-	General Public License
HTML	-	Hypertext Markup Language
HTTP	-	Hyper Text Transfer Protocol
KACTUS	-	Knowledge About Complex Technical Systems for Multiple Use
LGPL	-	Lesser General Public License
OWL	-	Web Ontology Language
RACER	-	Renamed Abox and Concept Expression Reasoner
RBR	-	Rule Based Reasoning
RDF	-	Resource Description Framework
RDFS	-	Resource Description Framework schema
SNOMED	-	Systematized Nomenclature of Human Medicine
SPARQL	-	Simple Protocol and Resource Description Query Language
SQL	-	Structured Query Language
SWRL	-	Semantic Web Rule Language
TBOX	-	Terminological Box
UNA	-	Unique Name Assumption
W3C	-	World Wide Web Consortium

XML - Extensible Markup Language

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CHAPTER 1

INTRODUCTION

1.1 Research Background

In genetic context, a family is often regarded as a group of people who have blood relations with each other or a group of descendents from a common ancestor. Basically, a unit of family is described as living together in one household. Apart from residing in a shared physical location, they usually share many other common elements in general which include ancestors, traditions, religions, lifestyles, environments and even genes that contribute to the risk of hereditary diseases. Typically, for viewing and readability purposes, the family relationships for a unit of family over few generations can be visualized using a family tree. A family tree is a chart normally used for representing the family relations in a conventional tree structure with interconnected nodes linked together via family relations. Family history information can be utilized for various purposes. Apart from being used to trace the ancestors of a person, a doctor can also use this particular information to predict family health problems since family relations are the common factors for most of the hereditary diseases. For instance, a completed genealogical chart can be exploited or extended to support multiple kinds of functions in medical or social work. In the medical field, this goal can be achieved by annotating additional data such as medical conditions of family members who have suffered from certain diseases. By having a precise parental health history, a doctor is able to identify the risk of a person developing certain diseases at an early stage and take necessary precautions earlier to avoid and minimize the risk of those diseases [1, 2, 3].

The risk of a disease being transmitted by parents to their children becomes higher when many of the family members were affected by certain common diseases. If the family members involved are first or second degree relatives and the diseases were developed at young age, then the probability of a child inheriting the same disease as their parents will increase further [1, 2]. The importance of family medical history has long been recognized in caring a patient [4]. By examining the family medical history, a doctor is able to make quick and effective decisions on immediate actions which should be taken to minimize the risk of particular diseases. In addition, family medical history data can assist a doctor in identifying family members who have higher risk of developing certain disease, deciding whether the family members should obtain a specific genetic test, determining the type and frequency of screening tests and assess the risk of passing those diseases to their children.

However, before having a completed family medical history, the first step will be building a precise and consistent genealogical chart or family history. There were aggressive researches in recent years on genealogical ontology after the semantic web has emerged as third generation of the web but some improvements can still be made towards the existing works. Improvements can be made towards the consistency, reusability, taxonomy and inference of existing family ontologies. In semantic web, ontology is used to encode the knowledge on the web in a semantic manner. According to Gruber [5], ontology is a formal, explicit specification of a shared conceptualization. This also means that ontology codifies relevant concepts of one phenomenon into machine readable format where the encoded knowledge is understood and agreed upon by large communities in general. Moreover, recent research has found out that ontology is the most powerful tool to represent knowledge formally [6, 7].This fact is proven when there were considerable numbers of domain experts who initiated their attempts to employ ontology as their representation languages in both medical and genealogical related applications. Applications under genealogical field were clinical knowledge-based systems such as SNOMED [8], Gene Ontology [9] and National Cancer Institute Thesaurus[10].

Those initiatives have shown that the value of ontology is gradually being recognized by the public. In fact, ontology is not merely accepted widely in genealogical

and medical areas but in reality it has also been adapted in a variety of fields. For now, ontology has even become the alternative way for search engines, e-commerce web sites, WorldNet, artificial intelligence and multi-agent systems. Actually, there are multiple factors which contribute to progressive researches on ontologies and creation of ontologies for various domains. Encoding pieces of knowledge using ontology is advantageous since ontology is capable of sharing common understanding of information among different parties in a community, research group, organization and software agents across the internet. Variety of standards and heterogeneous data employed by different groups of people often turn into major obstacles for two-way communication in an efficient manner. Having common understanding also means that terminologies applied by all parties are equivalent. Refinements, modifications and discussions can always be made towards the same terminologies to cope with specific requirements. Hence, the study of encoding the family relations using ontology language is relatively important and meaningful as ontologies are capable of storing family biological relationships more efficiently. In the meantime, ontologies provide shared genealogical vocabularies and common standards for communicating the general genealogical knowledge which address fundamental issues in communicating the knowledge for the same domain among different parties.

This project is beneficial to all humans since everyone has his or her own unique family history. The advantages of this research can be enlarged to support medical fields when proposed Family Ontology is annotated with medical conditions. Therefore, this project is also significant to the healthcare environment since it shows that ontology is capable of building a more powerful and interoperable information system in the medical area. Family Ontology not only helps to store and communicate general family history knowledge conceptually and efficiently, it also supports other domain experts in transferring, processing, reusing and sharing ontology knowledge with other group of researchers. Based on the common standards and terminologies applied within the proposed Family Ontology, discussion among doctors, families and domain experts can be conducted more easily without communication barriers. Wise decisions and conclusions can always be drawn after effective communication and discussion among the key parties.

Since there are previous works available for reuse, an effort will be put on the enhancement of existing works instead of building the proposed ontology from scratch. As such, the main aim of this research will be producing a consistent Family Ontology with other additional features such as reusability, maintainability and inferencing capabilities. Consistent and high quality Family Ontology is always preferable and desirable since it allows effective sharing, transferring and reusing of common genealogical terms to be conducted more easily by all interested parties.

1.2 Problem Statement

Other than storing the family biological relationships, Family Ontology can also be used to support other important functions in different areas. For instance, Family Ontology can be mapped with Medical Ontology to produce Family Medical History Ontology. Family medical histories are very useful for a doctor in accessing the risk of a disease being passed on to their offspring and suggestions of treatments for a particular disease. However, an important prerequisite prior to a robust Family Medical History Ontology is having a precise, consistent, well-designed and complete Family Ontology. Only with a well-structured, consistent and complete Family Ontology, a computer can process, analyze, interpret and acquire the new inferred family knowledge intelligently in a shorter duration. This will definably speed up the diagnosis of a patient and improve the quality of the healthcare systems when a high quality Family Ontology is integrated with Medical Ontology to produce a more complex system.

There are existing genealogical ontologies proposed by different groups of researchers when the semantic web emerged as third generation of the web. However, existing ontologies still lack certain important concepts and properties for the domain of family relations. This may cause ontology to be unable to deliver the full potential of exchanging family history information among family members, doctors and other interested parties. Moreover, the existing ontologies still lack axioms and SWRL rules for consistency checking purposes. Consistency of ontology is fairly important as inconsistent ontology leads to misinterpretation of actual semantic meaning of the data.

Therefore, the objective of this research is to build a new Family Ontology where all required axioms, rules, new terms and properties will be embedded within the resulting ontology to support the requirements of the proposed ontology.

1.3 Objectives

The objectives of this project are as follows:

1. To build a Family Ontology that meets the consistency checking criteria.
2. To evaluate the consistency of the Family Ontology using Pellet, HermiT and FACT++.
3. To compare and analyze the results of consistency checking for the above tools mentioned in (2).

Table 1.1: List of objectives, methodologies and validation methods

Objectives	Methodologies	Validations
1. To build a Family Ontology that meets the consistency checking criteria.	Creating the family reference ontology using the guidelines provided in [11]. The ontology will be developed using the latest Protégé ontology editor version 4.3[12].	1. The validation of the results will be done using the FACT++, Pellet and HermiT. 2. The validation will include the resulting new inferred instances through the use of inference rules associated with ontology.
	Refinement of Family Ontology to confirm to the consistency metric.	
2. To evaluate the consistency of the Family Ontology using Pellet, HermiT and FACT++.	Verification and Validation (V&V) will be done using a framework for ontology evaluation [13]. The V&V will cover the ontology terms, inference rules and instances.	
3. To compare and analyse the results of consistency checking for the above tools mentioned in (2).	Verification and validation results using heterogeneous tools are compared and analysed.	

1.4 Scope

For this study, the project will develop a case study involving seventy-one (71) family members for up to three generations of relatives. However, "in-law" relations will not be included in this research. Verification and validation of proposed ontology will focus on the consistency metric only.

1.5 Dissertation Outline

This chapter presents the overview of this research and the impacts of proposed ontology towards other fields. In this chapter, we discuss the problems faced by current approach and how ontology offers a better alternative solution than traditional method. Besides, we listed out some existing genealogical ontologies with similar domain as our reference. We also state the advantages of using ontology language to model domain of family relations and the importance of having a consistent ontology. In spite of these, we also sketched out the objectives, methodologies and research scopes for this research too.

1.5.1 Chapter 2: Literature Review

This chapter provides a comprehensive review on prior researchers' works. This involves extensive comparisons on the existing tools or ontologies which offer the same functionalities as the proposed Family Ontology to be developed. The comparison will focus on the limitations, characteristics, capabilities and features of existing genealogical ontologies. Besides reviewing the internal structure, taxonomy, consistency and completeness of concepts, properties and relations for three existing genealogical ontologies, we also review a list of existing ontology reasoners in terms of their attributes. One out from three existing family ontologies which is closest to the system requirements will serve as the base for customizations.

1.5.2 Chapter 3: Research Methodology

This chapter depicts the methodology being applied in this project which consisted of four sequential steps. The four main phases are strategy design and data, followed by ontology building using relevant concepts, properties, rules and axioms before verifying the consistency of the resultant ontology and the latter ontology refinement if any bugs are discovered in the consistency checking phase. Consistency verification is a fundamental part in ontology development lifecycle since a consistent ontology eliminates false definitions and statements within the proposed ontology.

1.5.3 Chapter 4: Experimental Results and Discussions

This chapter presents the experimental results yielded once the ontology development phases were completed. The discussions revolve around the results of consistency checking using heterogeneous reasoners such as FACT++, HermiT and Pellet. The outputs of consistency checking for different reasoners were captured, compared and analyzed to support the outcomes of this research. This chapter primarily demonstrates how consistency of proposed Family Ontology can be evaluated via different ontology reasoners and how these evaluation results might vary from one another.

1.5.4 Chapter 5: Conclusion

This chapter summarizes all of the research activities that have been done throughout the entire ontology development lifecycle. The contributions of this research are listed and discussed in this chapter. Some of the possible future works are identified in order to enhance and enlarge the scope of this project to support other fields. This allows the improvements of current ontology to be carried out in the coming future in order to cope with the specific requirements of other areas.

CHAPTER 2

LITERATURE REVIEW

2.1 Introduction

Researchers from all over the world focus extensively on genealogical studies as family history has a very close relationship with human health. According to the definition from Oxford, genealogy is the study of family history including the study of who the ancestors of a particular person were. The efficiency of a healthcare system needs to be improved so that more and more patients can be cured in a shorter duration, without compromising the quality of services at the same time [14].However, effective recommendations on treatments or precautions to patients can only be made when precise, consistent and accurate parental history data is given to a doctor [15, 16, 17]. Hereditary diseases have long attracted public concern. This is because people nowadays have become more health conscious. With family history data, preventions can be taken earlier to minimize the risk of genetic diseases. In order to obtain the family history, there exists a need for a tool that can aid people in constructing their own family tree before medical conditions can be annotated to those family history data.

Previously, there were some good efforts initiated from other researchers in building the applications which assist people in building their own family tree. This includes "*My Heritage Family Tree Builder 7.0"*[18] and *"Family Echo"* [19]. As time passes, researchers realized that there is a communications gap between machines and humans as most machines have been designed to be machine-readable instead of machine-understandable. One of the greatest challenges faced by today's web is a lack

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