Evaluation and Evolution of NAOnto – An Ontology for Personalized Diabetes Management for Native Americans

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***Abstract*—Ontology has been proposed to use in various healthcare domains to represent and manage knowledge, integrate heterogeneous data, assist natural language processing, facilitate machine learning, and support intelligent medical decisions. Although there are many ontologies defined, their adoption in real-world applications is limited, because their quality is unknown. The quality of the ontology directly determines its use and reuse in real-world applications, which is especially important for the medical domain. To assure its quality, an ontology should be evaluated with a set of comprehensive and systematic metrics and approaches. In this work, we studied the ontology evaluation problem in the medical domain. Specifically, we present the evaluation framework to evaluate, NAOnto, an ontology we developed for personalized diabetes management of Native Americans. The framework consists of two important parts of the evaluation, namely verification, and validation. We adopt software quality standard metrics to verify ontology as a software application while applying the ontology to real applications to validate its contained knowledge. Based on the evaluation results, the proposed ontology has gone through an active evolution process to address the discovered issues/problems. The proposed framework demonstrates how ontology evaluation can be practiced in the healthcare domain to assure the quality of ontologies.**

Keywords—knowledge engineering, ontology evaluation, ontology development, ontology quality

I. Introduction

Native Americans (NA) including American Indians and Alaskan Natives (AI/AN) have the highest prevalence of diabetes (14.7%) than any other racial group in the United States [1]. Besides genetic reasons, environmental and behavioral issues also contribute to this health disparity of NA. The lifestyle changes had a negative impact on the NA health [2]. Relocation removed NA from their usual food sources and the active lifestyle that requires hunting and gathering. NAs nowadays turn to food with a low intake of fibers, higher dietary fat along less energy expenditure. Moreover, poverty limits their access to medical providers and nutritious foods such as fresh fruits and vegetables or whole grain carbohydrates, which are often more expensive than commodity goods like flour or shortening. Furthermore, AI/ANs have a lower health literacy rate due to their cultural beliefs, communication styles, and language barrier [3].

Diabetes is a long-lasting disease that is difficult to cure. To deal with this disease, patients need to manage their stress levels, adopt healthy eating habits, and active lifestyles. This should be done in a personalized manner. As NA patients face special issues that vary from other ethical groups, a personalized management strategy specifically for the NA community is required. To model the personalized knowledge about the condition of NA patients and the intervention plan for them, the conceptualization of user profile and intervention has been used for this purpose[4]. This information can be used to make individualized plans to manage their diabetes. To store the knowledge in an extendable and computable format, we adopt ontology to generate formal representations of entities and relationships between them. The ontology we developed, NAOnto, composes of three sub-ontologies: The User Profile Ontology (UPO), the Diabetes Management Ontology (DMO), and the Food and Nutrition Ontology (FNO). It has been used to create a personalized tool for the NAs to manage their diabetes [5].

The construction of the NaOnto ontology has been greatly benefitted by the comprehensive evaluation process. This process provides our ontology engineers with an opportunity to iteratively improve the quality of the ontology from various aspects. Moreover, as NAOnto has been successfully utilized in our personalization systems and is getting mature, we believe it should be ready for use in other applications. The lack of systematic methodologies for evaluating ontologies can be an obstacle to their application in the industry[6]. For this purpose, we must evaluate and benchmark it to ensure a smooth transference. As no global standards are determining how good an ontology is, we have evaluated our ontology on different aspects with various metrics. In this paper, we present the strategies we used to evaluate and improve the proposed ontology.

The rest of the paper is organized as follows. Section II surveys related work on ontology evaluations. Section III reviews our proposed NAOnto ontology. Section IV describes our proposed methodology in detail. Finally, in Section V, we provide conclusions and future work directions.

II. Related Work

The whole purpose of Ontology Evaluation is to make it worth sharing and reusing among the community members[7]. Ontologies can be evaluated as a whole unit test falling in the following three categories.

The first category is the gold standard-based evaluation e.g. [7]–[9]. The ontology under consideration is compared against a gold standard reference in the domain. For example, Alani et. al [10] propose four measures to evaluate different representational aspects of the ontology with the gold standard. The second category is task-based evaluation, e.g. [11]–[13]. In this approach, the ontology is evaluated based on its suitability and utility in specific tasks/applications. For example, Zhang et al. [14] evaluate their SHKB (Semantic Healthcare Knowledge Base ontology using an application and compare the results to a reference ontology. Data-driven evaluation, e.g. [15], [16], compares an ontology to the data available in the domain. This approach extracts the corpus of the domain and counts the number of terms that overlap between the ontology and corpus or uses a vector space representation of the terms both in corpus and the ontology and measuring the fit between ontology under consideration and the corpus. Patel et al. [16] extract concepts and relations from the ontology and feed them to a document classification tool to evaluate and rank the ontologies. The fourth category is user-based evaluation [17]. This approach considers user feedback about the ontology to determine the quality of the ontology. For example, Supekar presented a method to evaluate the data in the ontology based on the “peer reviews” provided by the actual users of the ontology.

Due to the complexity involved in ontology, it is more useful to focus on the evaluation of different levels of ontology rather than considering ontology as a whole unit. Depending on the purpose different metrics allow ontologies to be evaluated on different levels. Metrics provide various types of statistical information about the knowledge contained in the ontology which can help in determining the goodness of the ontology. Tartir et. al [18] propose metrics schema metrics to evaluate the richness, width, depth, and inheritance of the ontology; instance metrics to measure the class richness, cohesion, average population, connectivity, relationship, and readability of the ontology. Based on the software engineering approach, Jones et al. proposed a suite of metrics [19] to assess the syntactic, semantic, pragmatic, and social aspects of ontology quality. Various tools are available in the domain for evaluating several aspects of the ontology such as OOPS!, OntCheck, OntoMetrics, OntoAnalyser, OntoGenerator, S-OntoEval, OntoClean, ONE-T, etc. [20]–[24].

Hlomani et. al [17] classified the evaluation of ontology under two categories namely quality and correctness. Similarly, Gomez-Perez et al. classify ontology evaluation as ontology verification and ontology validation. Ontology verification focuses on the correct construction of ontology whereas validation signifies if the correct ontology was constructed [5]. These categories address various aspects of the ontology such as Vocabulary/Lexical aspect, semantics, context, syntax, structure aspects [25]–[29]. These aspects are evaluated with the help of different metrics available in the literature. Ultimately these results are aggregated to generate qualitative results describing the quality of the ontology. Gangemi et al. described three types of metrics to measure the different aspects of the ontology [21]:(1) Structural metric- measures the syntax and formal semantics. (2) functional metric- assesses the conceptualization specified by the ontology. (3) Usability-Profiling metrics – focus on the communication context of an ontology. Ouyang et al. proposed and improved three metrics such as coverage, cohesion, and coupling based on the semiotic framework for ontology evaluation [30]. Yao et al., In another paper, adopted the software practices to build metrics to define and validate the cohesiveness of the ontology [31].

In summary, different mechanisms have been proposed to evaluate different ontologies for different purposes. There are no standard methods or procedures to evaluate an ontology. Ontology evaluation methods should be chosen depending on what kind of ontologies are being evaluated and for what purpose, for example, different mechanisms should be considered for evaluating a newly developed ontology or choosing an appropriate one for a particular application from multiple ontologies. Therefore, in this paper, to evaluate our ontology, we present our approach to evaluation, including how we choose the aspects of ontology to be evaluated; and the multiple set of criteria to be evaluated, and the right tools to be used.

III. Background on NAOnto

We have designed NAOnto [5], which is the first AI biocultural ontology defined in the literature to the best of our knowledge. It was designed and developed through a multi-phased iterative and incremental methodology. The interdisciplinary social and scientific nature of this research requires the use of an integrated approach. We developed this ontology through a collaborative process that includes domain experts, and ontology engineering experts. As there are many synergies between software engineering and ontology development, we adopt ideas of the systems development life cycle of software engineering to our ontology development life cycle. In our design, we divide the ontology development cycle into six work phases including scope definition, knowledge acquisition, specification, conceptualization, implementation, and evaluation. The ontology engineers, developers, and domain experts plan for, develop, implement, evaluate and deliver ontology based on these six phases. The ontology development process is recurring in cycles, as each work phase is cyclically and incrementally repeated. At each new cycle, the ontology is further revised and refined.

Fig. 1 shows part of the high-level ontology. Its major part contains patients’ profiles including health, preference, culture, social-economic status, and context information of the environment where the patient stays. This type of information provides evidence for personalization. The ontology also includes concepts and relations about wellness and lifestyle such as food, nutrition, and physical activity. the conceptualized ontology is implemented using a formal knowledge representation language, OWL (Web Ontology Language).

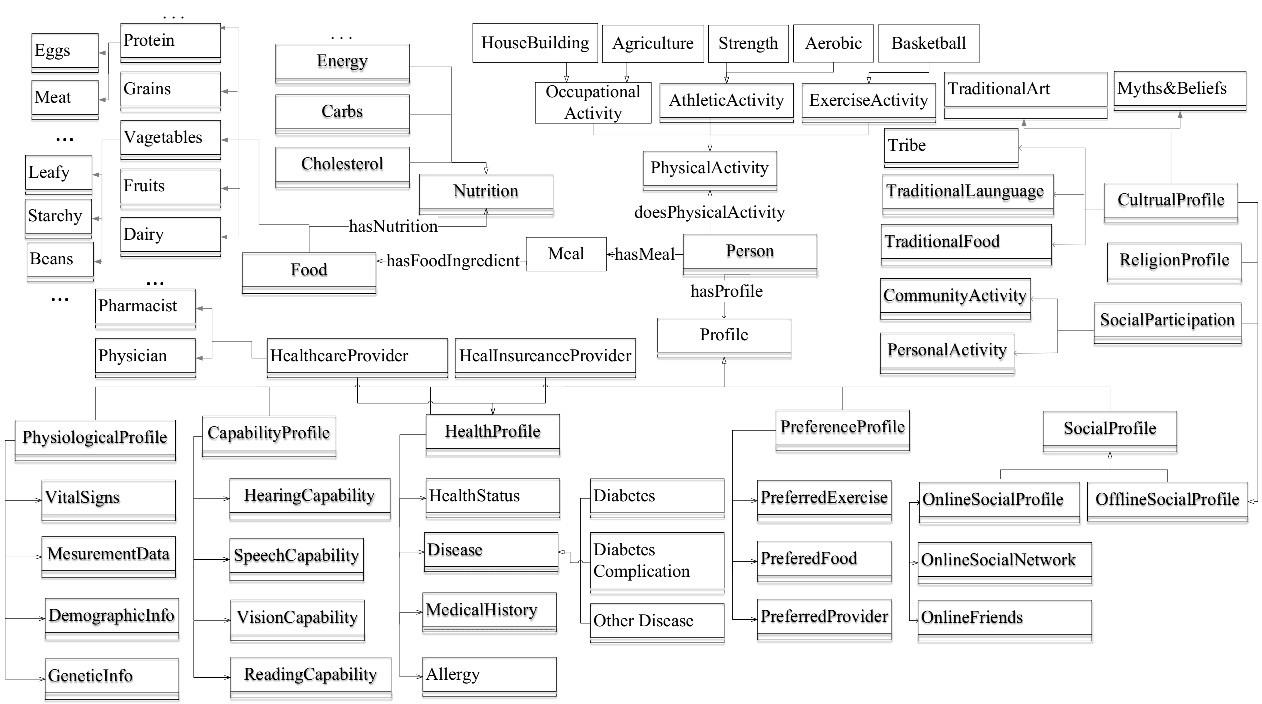

Fig.1. Part of the high-level ontology of NAOnto

IV. Evaluation Framework

Evaluation of NAOnto consists of two parts, namely, verification and validation. Verification is to test if the ontology is being built correctly and the knowledge represented by the ontology is actually in alignment with the software artifact requirement. We treat ontology as a software entity and try to verify that it has no problems in structure, operability, logical consistency, etc.

A. *Verification*

We adopt the Ontology Quality Evaluation Framework (OQuaRE) framework [32] for evaluating the quality of ontologies based on the standard ISO/IEC 25000:2005 for Software product Quality Requirements and Evaluation known as SQuaRE [33], as ontology engineers have adapted software quality standards to measure ontology quality. The defined metrics give us a better idea about ontology as a software application. This model measures an ontology in terms of the following characteristics: functional adequacy, reliability, operability, maintainability, compatibility, transferability, performance efficiency and quality in use, and structural features of ontologies. Each characteristic has multiple sub characteristics that are evaluated based on various defined metrics as well as experts. Fig. 2 illustrates the major categories and sub-categories for OquaRE-based evaluation.

We compared these evaluation metrics of our ontologies with the state-of-the-art ontologies, such as Measurement Units Ontology (MUOVOCAB), Unified Code for Units of Measure (UCUM), Gist Units of Measure Ontology (GISTUM), SCIUNITS, QUDV_SI, OpenMath, UNIPATO, and WURVOC [32][34][35][36][37]. These existing ontologies have been used as benchmarks for ontology evaluation: MUOVOCAB was developed to exploit semantics in mobile environments; UCUM intended to include all units of measures being contemporarily used in international sciences, engineering, and business; GISTUM was designed primarily for business use; SCIUNITS was for scientific units for physical science application; QUDV_SI is for quantities, units, dimensions, and values; OpenMath is for mathematical objects; UNIPATO is for representing metrical units; WURVOC represents units of measure and related concepts and.

In the following, we present our evaluation results in terms of these metrics. The values calculated using the metrics described in Fig. 2 were ultimately used to calculate quality values for Modularity, Reliability, Structural quality, and Functional Adequacy, etc. The values calculated using these metrics were transformed into the range 1 to 5, where 1 meant not acceptable, 3 as minimally acceptable, and 5 as exceeds the requirements.

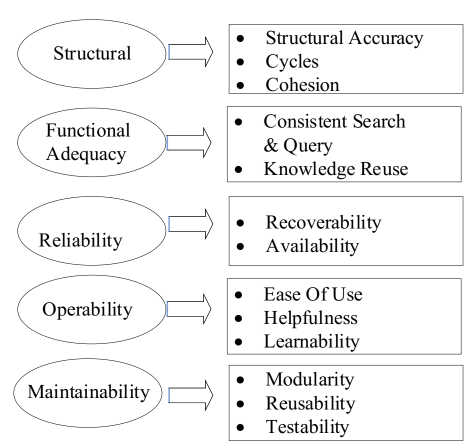

Fig. 2. Major verification metrics

1) Structural evaluation

Ontology structure is important to evaluate manually constructed ontologies. It pertains to the evaluation of the formal structure of the ontology, accounting for ontology quality factors such as consistency, formalization, redundancy, or tangledness. (Gangemi et al. [28]). Fig. 3 shows the comparison of our ontology with other reference ontologies in terms of the structure of the ontology. We can see that the three versions of our ontology perform well in terms of structural quality compared with the state of the arts.

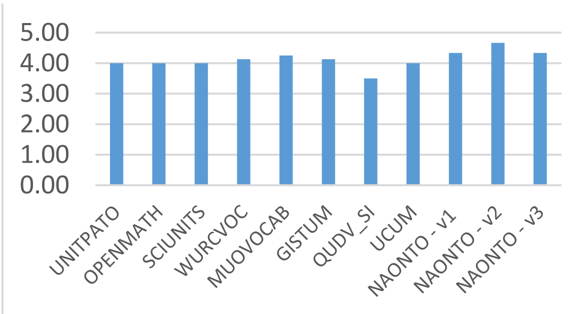

Fig. 3. Comparison of the structural evaluation results

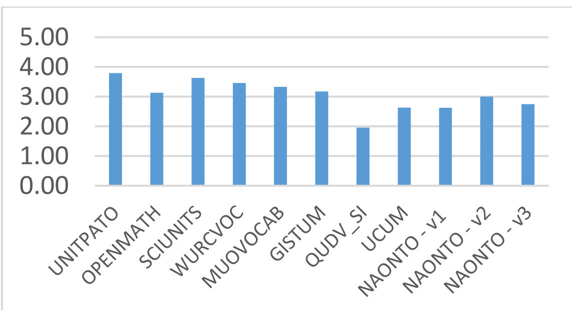

Fig. 5. Comparison of reliability results

2) Functional Adequacy

Functional adequacy measures ontology's capability to provide concrete functions[38]. It involves the measure of the vocabulary, consistency of search and query, and capability to represent knowledge, etc. Fig. 4 shows the result of the functional adequacy evaluation. Again, functionality-wise, NAOnto performs well compared with other benchmark ontologies.

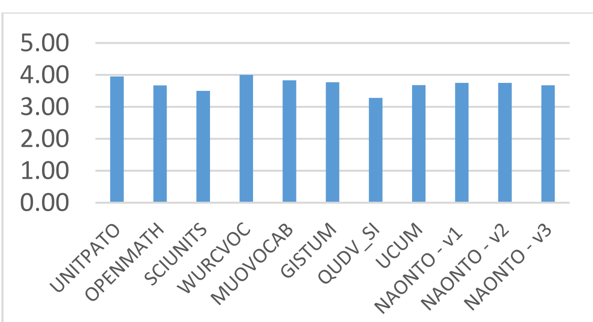

Fig. 4. Comparison of the functional adequacy results

3) Reliability

The reliability of an ontology is determined by the quality of the ontology to maintain its performance under stated conditions[39]. The OQuaRE framework utilizes recoverability and availability as sub characteristics and related metrics. Fig. 5 compares the reliability of our ontology and others. We can see that NAOnto is comparable with the others.

4) Operability:

It evaluates the effort needed for use of the ontology, in terms of helpfulness, ease of use, and learning[26]. Fig. 6 compares the Operability of our ontology and others.

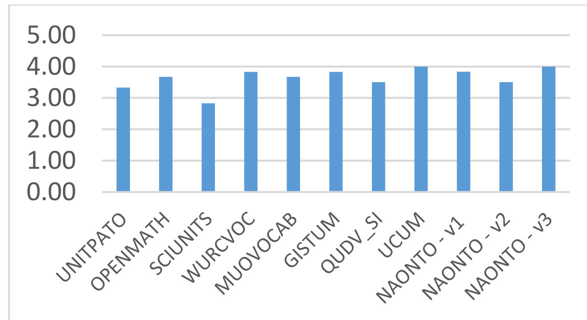

Fig. 6. Comparison of operability results

5) Maintainability:

To create a maintainable ontology, we need to determine that if the ontology is modular, implying any changes in a part of ontology should minimally affect other components of the ontology[40]. Reusability of the ontology was another factor that was considered which means a part of the ontology can be reused in more than one ontology. Fig. 7 shows the maintainability of different ontologies. Reasoning-based evaluation

In addition, reasoners were employed to provide a syntactical verification of the ontology. We have employed reasoners including Fact++, HermiT, Pellet [41]. All the inconsistent or incoherent axioms were discovered and corrected with the help of the mentioned reasoners and the Onto Debug tool.

B. Validation

Validation of the ontology means if the right ontology is built. The ontology with the data i.e., the knowledgebase needs to be evaluated with respect to its actual use. Here we discuss assessing our ontology not semantically or structurally but evaluating the information the ontology contains. Validating the ontology with the help of experts gives the end-users as well as community members the confidence to accept the ontology. Domain experts were involved continuously with the ontology engineers to guide them through the development of the ontology[42]. We tried to query some of the competency questions that we wanted as an absolute requirement that needed to be fulfilled by our ontology, and we were able to fetch meaningful results[43]. Finally, application-based validation of the ontology was done with the help of the mobile application built with integration of this ontology, we created a variety of use cases and after performing multiple tests we found the results to be satisfactory.

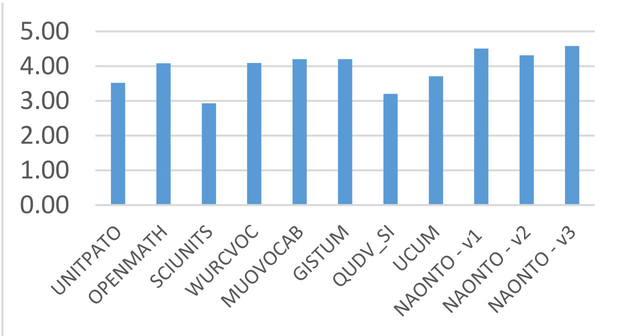

Fig. 7. Comparison of maintainability results

1) Competency Questions.

We employ competency questions to providing ontology engineers with a simple means to verify requirements' satisfiability by either knowledge retrieval or by entailment on its axioms and answer checking. Competency question also helps to evaluate the comprehensiveness and completeness of the developed ontology. With the help of domain experts, in our case, physicians and tribe leaders, we have designed a list of competency questions based on a set of existing reference documents related to native Americans and diabetes self-management. We categorized the competency queries based on the concepts related to the domain of foods, exercise, education, and native American patient profile. We implement the query with SPARQL [44] and test whether the ontology can answer the given queries. To answer the questions, we may also need to populate the ontology with individuals. For example, we pose a query:

"*Locate diabetes patients in Mandan tribe of North Dakota who do not have any health insurance and income is lower than 25k per year.*"

```
SELECT ?user
WHERE {
 ?user a :Person .
 ?user :hasHealthProfile ?userHealthProfile.
 ?user :hasSocialProfile ?userSocialProfile .
?user :hasHealthcareProvider ?userProviders .
 ?userHealthProfile :hasDiabetes ?userDiabetes.
 ?userSocialProfile :hasIncome ?userIncome.
 ?userProvider hasHealthInsuranceProvider ?userInsurance.
 ?userSocialProfile hasTribe ?userTribe .
FILTER
(
?userDiabetes =:Type2      &&
?userIncome =:25kLess      &&
?userInsurance=:None       &&
?userTribe =:Mandan
)}
```

In another example, we pose a query:

"*Find all people with type1 diabetes who are above the age of 50 with low hearing and reading capacity, and low education and low-income level.*"

This query can be represented using our ontology in SPARQL Query format:

```
SELECT ?user
WHERE {
?user a :Person .
?user hasHealthProfile ?userHealthProfile;
 ?user hasSocialProfile ?userSocialProfile .
?user hasCapabilitylProfile ?userCapabilityProfile .
?userHealthProfile :hasDiabetes ?userDiabetes.
? userSocialProfile :hasAge ?userAge.
? userCapabilityProfile:hasHearingCapabilityLevel ?hearinglevel .
? userCapabilityProfile:hasReadingCapabilityLevel ?readinglevel .
? hasSocialProfile:hasEducationLevel   ?educationlevel .
?userSocialProfile :hasIncome ?incomelevel.
FILTER
(
?userDiabetes =:Type2                                  &&
?userAge>50                                            &&
?hearinglevel=:RockConcert
&&                          ?readinglevel=:FrustrationReading
&& ?educationlevel=:PrimaryEducation                   &&
?incomelevel=:25kLess
)}
```

When these SPARQL queries are applied to our knowledge base, individual entities that satisfy the constraints of the queries can be retrieved.

2) Use Cases

In order to check if our ontology is capable of fulfilling the desired goals, such as information retrieval, semantic query, and personalized recommendations, we made the ontology run through a set of test cases and usage scenarios with the help of a mobile application. Based on this ontology, we have developed a personalized healthy lifestyle recommendation system for native American diabetes patients [5].

The following scenario demonstrates that a system can utilize the proposed ontology to makes personalized recommendations. The patient in the case is Steven Lunde, a 37-year-old Native American male living in the Mandan tribe in the state of North Dakota. His detailed health information is stored in our database (in the form of Triplestore) and listed in Table I.

TABLE I. USER BASIC PROFILE INFORMATION

Property	Value
Age	37
Gender	Male
Height	6' 7"
Weight	201lb
Waist	38"
Allergies	NA
Smoker	Yes
Drinker	Yes
Diabetes[1]	Type 2
Hip Size	1.05
Blood Pressure	150/95
Chronic Diseases	Hypertension, Diabetes
Physical Activity[2]	Sedentary
Tribe	Lower Sioux
Education	Some High School
Income	25k or less

1:(Diabetes type: Type1, Type2, Gestational or Other)
2:(Physical Activity levels: very active, active, low active and sedentary)

Using the information listed above, a personalized recommendation can be provided by applying semantics-based rules on the stored ontology to infer new knowledge. According to the Institute of Medicine of the National Academics, meal cholesterol should be in the range of 250 to 325 mg per day for men and 180 to 205 mg per day for women. The following SWRL rule can be applied to Steven's profile information and infer if the meal satisfies the cholesterol constraints.

```
Person(?user) ^
hasMeal(?user, ?meal) ^
hasCholesterol(?meal,?chol) ^
hasEnergy(?user, ?energy) ^
hasEnergyRequirement(?user, ?er) ^
swrlb:divide (?mealPer, ?energy,?er) ^
swrlb:multiply(?higherLimit, ?mealPer,325) ^
swrlb: multiply (?lowerLimit, ?mealPer,250) ^
swrlb: greaterThanOrEqual (?chol, ?higherLimit) ^
swrlb: lessThanOrEqual (?chol, ?lowerLimit) ->
isRecommended(?meal,false)
```

In another example, based on the American Heart Association guidelines, People with diabetes should have their blood pressure under 130/80 mm Hg. People with diabetes have a risk of hypertension which increases the chances of other diabetes-related diseases such as kidney disease and cardiovascular diseases. Now assume Steven chooses to check his blood pressure, we can apply this rule to check if Steven should be concerned.

```
Person(?user) ^
hasSystolic(?user,?sys) ^
hasDiastolic(?meal,?dia) ^
swrlb:greaterThan(?sys, 130) ^
swrlb:greaterThan(?dia, 80) ->
cautionRecommended(?user,true)
```

Applying ontological rules on Steven's ontology profile, the reasoner can figure out that if a meal should be recommended for Steven. Based on his profile information and dynamic context information, the system can provide various healthy recommendations and medical guidelines specific to him.

C. Evolution

Ontology NAOnto has gone through multiple rounds of revisions based on the many evaluation results and changing user requirements. These revisions induce changes to the ontology, and we need to keep the changing ontology healthy in different characteristics such as Structural, Functional Ability, Maintainability, Reliability, Operability, Transferability, Compatibility. Therefore, we keep monitoring and evaluating the ontology all the time during our development and usage of the ontology. For example, as shown in Fig.3-7, we have three different versions of NAOnto. In the first version, we found that the Reliability and Functional Adequacy were not satisfactory. We then revised the ontology by controlling the vocabularies: for example, we added more attributes for classes. This requires researching more about the domain knowledge that could be incorporated to make the classes attribute rich. Similarly, adding enough instances of classes would be helpful to represent the query results. Reasoners such as Hermit and fact++ were used to check and remove inconsistencies and unsatisfactory classes/axioms from the ontology. We used the OOPS tool to find the major pitfalls in ontology. There were three levels of pitfalls that were recognized by OOPS: critical, important, and minor. All the pitfalls were manually removed from the ontology. For example, missing domain and range pitfall, synonyms classes pitfall, etc. After improving upon various aspects related to Functional Adequacy and Reliability, we found that the overall mean of all the characteristics plummeted. Therefore, another iteration was required in which we made further changes to the sub characteristics, and we were able to obtain satisfactory results for individual characteristics as well as the overall mean.

V. CONCLUSIONS

Lack of techniques, metrics, and tools to evaluate ontologies and ensure their quality impedes ontology's deployment and usage in the healthcare domain. In this paper, we present our framework to evaluate a healthcare ontology, NAOnto, we developed previously. Through the evaluation process, we have identified best practices, metrics, and tools in ontology evaluation, and demonstrated how to create an ontology evolution process to ensure ontology quality across its whole lifecycle. Specifically, we showed that our ontology can be used in applications with the same purpose for creating, querying, exploiting knowledge. The framework enables ontology engineers to detect faults in the ontology and helps them to determine if the ontology is suitable to be reused to build other ontologies. This evaluation framework ensures that NAOnto is designed well and behaves correctly for a personalized healthcare system. Eventually, it improves the quality of ontology development and maintenance.

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