An Ontological Approach to Breast Cancer Screening: Risk Assessment and Personalized Testing Recommendations⋆

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Abstract

This article introduces OntoBreastScreen, an ontology-based application designed to provide personalized recommendations for breast cancer screening. The application calculates a woman's breast cancer risk using different risk models and suggests an appropriate periodicity for screening tests. The underlying conceptual model is the BRCS-Onto ontology, which provides personalized screening recommendations consistent with established healthcare guidelines.

Keywords

breast cancer prevention ontology, description logics reasoning, ontology-based application

1. Introduction

The importance of detecting and preventing diseases at an early stage cannot be denied, even more for oncological diseases. In recent years, there has been a significant shift in prevention strategies from standardized approaches to more personalized ones, that focus on patients' risk profiles and preferences. For breast cancer, strategies focus on estimating a woman's risk of developing the disease by applying different risk models [1], and performing screening tests by guidelines from recognized health organizations[2].

This paper describes the OntoBreastScreen application, an ontology-based application for breast cancer prevention. The ontology-based design of the OntoBreastScreen application provides a structured and standardized framework for representing and organizing knowledge about breast cancer risk and screening. Using the ontology as a conceptual model for the application enables flexibility for possible extensions. This approach also has the advantage that the consistency of the model can be verified by the ontology reasoner against any change in the ontology.

The OntoBreastScreen application uses the BRCS-Onto ontology, an open ontology available on the Bioportal [3]. This ontology interacts with risk calculators based on different risk models, each of which may be based on potentially distinct data, assumptions, and methodologies. The application allows clinicians to use the different risk calculators in a unified framework to obtain specific screening recommendations according to recommendation guidelines given by recognised healthcare institutions.

Additionally, the front-end of the application is generated using Ontoforms [4], a user interface generator for ontologies. It creates a structured interface for populating the woman's data into the BCSR-Onto ontology. The Ontoforms generator allows automatic regeneration of the user interface when new risk calculators or changes in recommendation guidelines are introduced. This automated process reduces the need for manual coding, improving usability and streamlining the OntoBreastScreen application development process.

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In summary, OntoBreastScreen allows clinicians to select a risk calculation model, enter a woman's data using the Ontoform generator, and classify her into a specific breast cancer risk category. The clinician can then select a recommendation guideline to follow, and the system uses reasoning to generate screening recommendations based on the BCSR-Onto ontology, which is populated with risk models, recommendation guidelines, and the woman's data. To the best of our knowledge, we do not find breast cancer prevention systems like OntoBreastScreen, based on open ontologies, that provide screening recommendations for the early detection of the disease.

2. The OntoBreastScreen application

OntoBreastScreen is based on two key components: the BCSR-Onto ontology that conceptualizes the breast cancer prevention domain, and the OntoForms application that provides OntoBreastScreen a solution to generate the application front-end from the ontology.

Figure 1 shows the implemented architecture, consisting of the Breast Cancer Recommendation System and the OntoForms System. The right side of the figure shows the Breast Cancer Recommendation System, implemented by a front-end, and a back-end application that provides services to the front-end.

The Breast Cancer Recommendation System lies on the Ontoforms System, structured by a front-end, a back-end and a triplestore. The Ontoforms front-end provides the system administrator user with a set of functionalities to upload ontology updates and add configuration settings to customize the form generated from the ontology. The Ontoforms Core API, is a back-end application that takes as input the BCSR-Onto ontology from the triplestore and a form configuration, to generate the structure that describes the user interface. This structure is of great relevance for the Breast Cancer Recommendation Api to render the user interface for the doctor. Ontoforms Core API also provides an endpoint to populate BCSR-Onto with the woman data instances entered by the doctor.

The BCSR-Onto ontology is illustrated in Figure 2. This ontology was presented in previous works [5], and has evolved recently [3]. BCSR-Onto represents knowledge about risk models, breast cancer screening guidelines for different risk levels, women's attributes and the test screening recommendations.

In the figure, ovals represent concepts, dots represent individuales and arrows represent roles (or relations). The upper segment of the diagram of Figure 2 outlines concepts such as *Model*, *Risk*, and *Guideline*, which are used by medical professionals when interacting with patients. A model predicts a high or medium risk of developing breast cancer, so a woman may be considered high risk under one model and medium risk under another.

In the lower part of the figure, women are represented by the concept *Woman* and associated properties such as *Age* and *History* to represent clinical history. These attributes serve as critical determinants for risk calculation through various models. Subclasses of *Woman* groups instances of that concept according to the risk calculated by each risk model. The concept *Recommendation* bridges the two conceptual levels, encapsulating the recommendations provided to women by applying guidelines tailored to medium and high-risk levels.

We highlight the ability of the BCSR-Onto model to derive screening recommendations using exclusively the reasoning mechanisms of description logic. The two following axioms define the properties *hasRecommendationMedium* and *hasRecommendationHigh*, which make it possible for the reasoner to infer recommendations for women:

$$hasAgeMedium \ o \ hasRange \ o \ intervalOfMedium \sqsubseteq hasRecommendationMedium$$
$$hasAgeHigh \ o \ hasRange \ o \ intervalOfHigh \sqsubseteq hasRecommendationHigh$$

where *hasRecommendationMedium* and *hasRecommendationHigh* have domain *Woman* and range the sets of recommendations given by guidelines for medium and high risk levels, represented by the concepts $Recommendation \sqcap \exists gives^{-}.(\exists forRiskLevel.\{Medium\})$ and $Recommendation \sqcap \exists gives^{-}.(\exists forRiskLevel.\{High\})$.

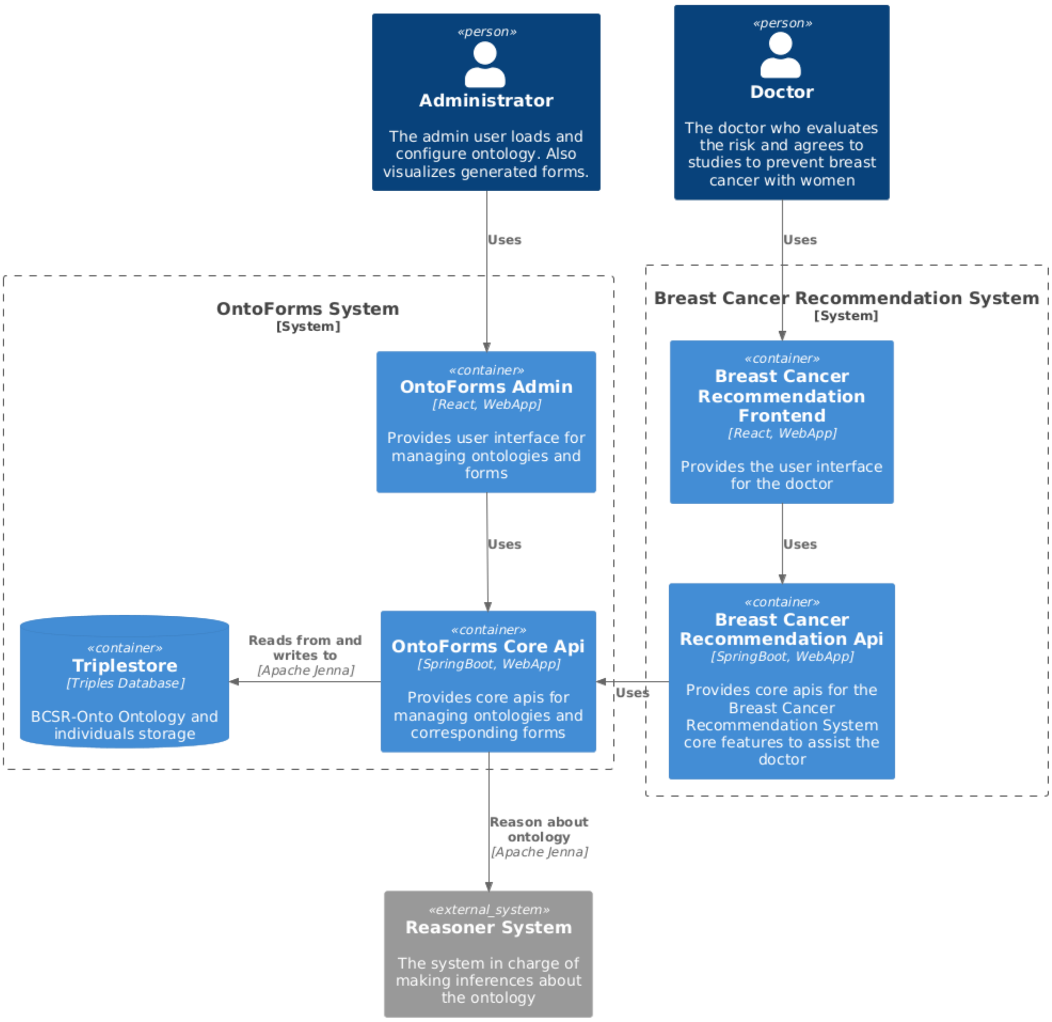

Figure 1: High level OntoBreastScreen architecture

$hasAgeMedium$ and $hasAgeHigh$ are sub-properties of $hasAge$ with range Age and domain the concepts $Woman \sqcap \exists hasRisk.(\exists hasLevel.\{Medium\})$ and $Woman \sqcap \exists hasRisk.(\exists hasLevel.\{High\})$. Instances of $hasAgeMedium$ and $hasAgeHigh$ for each woman are entailed by the reasoner once she is classified into a high or medium risk level, thanks to axioms $Woman \sqcap \exists hasAge.\top \sqcap \exists hasRisk.(\exists hasLevel.\{Medium\}) \sqsubseteq \exists hasAgeMedium.\top$ and $Woman \sqcap \exists hasAge.\top \sqcap \exists hasRisk.(\exists hasLevel.\{High\}) \sqsubseteq \exists hasAgeHigh.\top$.

$intervalOfMedium$ and $intervalOfHigh$ are sub-properties of the inverse of the $forInterval$ property with domain $RecInterval$ and range $Recommendation \sqcap \exists gives^{-}.(\exists forRiskLevel.Medium)$ and $Recommendation \sqcap \exists gives^{-}.(\exists forRiskLevel.High)$.

The set of axioms described above makes it possible that starting from a woman instance, if she is classified into a medium or high risk level and has an age that fits into an interval of a recommendation for that risk level, then the woman is connected to corresponding recommendations by the property $hasRecommendationMedium$ or $hasRecommendationHigh$.
For more details of the model of the BCSR-onto, the reader can see [5].

The OntoForms Core API [4] works on top of the BCSR-Onto ontology. It generates a form structure that corresponds to the ontology's subgraph accessible from a class marked as the main class through its declared and inferred properties. The back-end of Breast Cancer Recommendation System invokes OntoForms Core API with BCSR-Onto and the class Woman as the main class. To go across all classes accessible from the main class through outgoing properties, the OntoForms Core API algorithm works

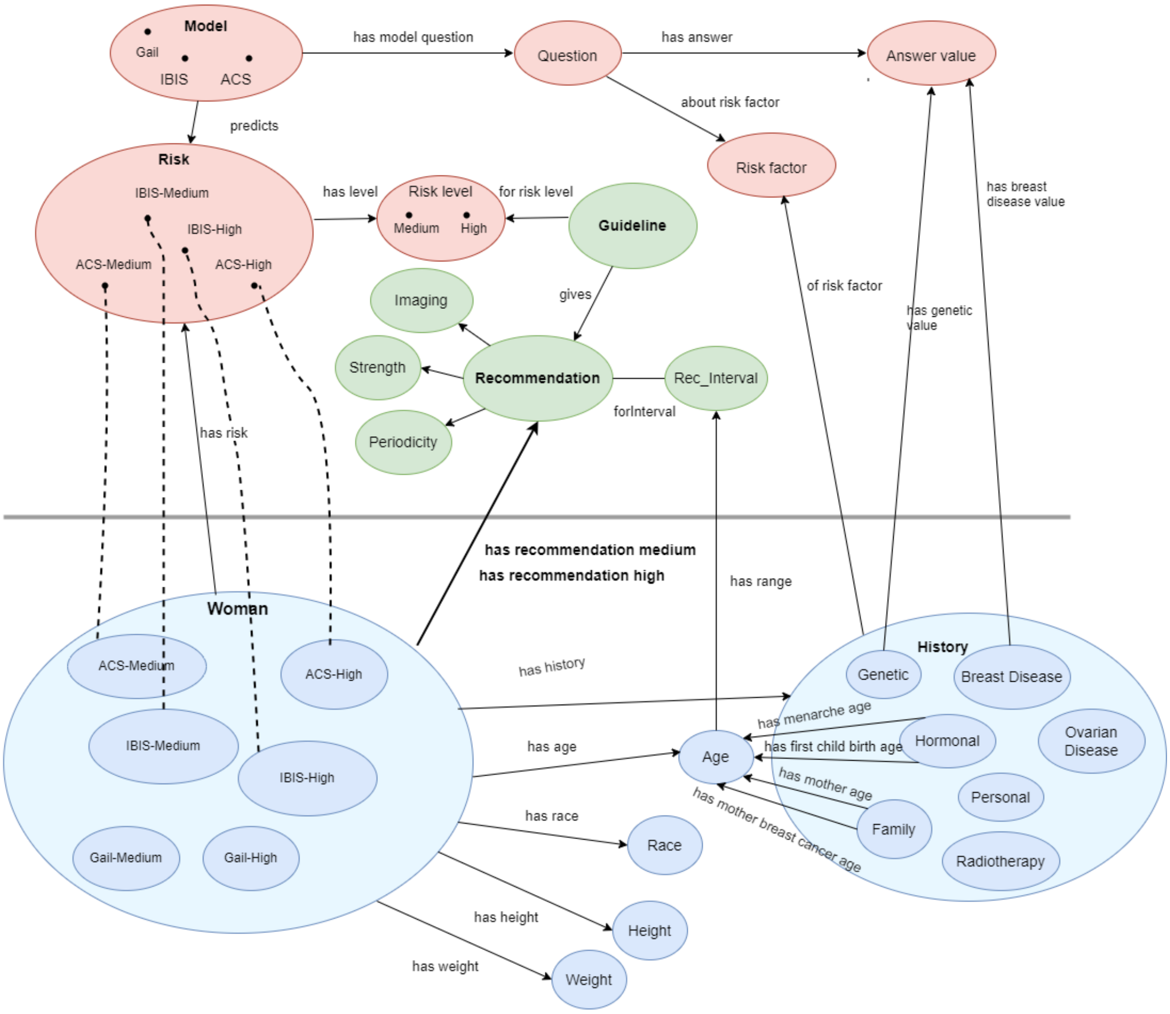

Figure 2: The BCSR-Onto ontology model

recursively by setting each reached class as the main class. Moreover, it automatically locates prompts and fields grouped in different sections for subclasses and when necessary creates some instances that are transparent to the user who enters the data.

The user interface generated from this graph structure allows the clinician to enter the woman instance and the set of instances corresponding to all the current data properties and object properties associated with her. For example, to populate the woman's medical history, the clinician must select the correct answers to questions (for a given risk model) that correspond to risk factors, such as genetic and hormonal, modeled by the subclasses of *History*. To generate the form, Ontoforms goes from the *Woman* class through the *hasHistory* property to each subclass of *History*, e.g. *Genetic*; then, *Genetic* is set as the main class to reach the *Answer value* class, which is populated with preloaded possible answers to each genetic question. In this case, the administrator user configures subclasses of History as "transparent", so that the doctor can directly visualize possible answers; then, when the doctor selects the correct answer, the instance of Genetic is automatically created and connected to the selected answer.

Figure 3 shows on the left a graph with an instance *Woman1* pointing to a *Genetic* History instance related to the selected answer "Normal test", and on the right a fragment of the form rendered using OntoForms with the values entered by the doctor for *Hormonal* and *Genetic* History. Then, when the doctor selects the correct answer, the instance of *Genetic* is automatically created and associated with the chosen answer.

We highlight that OntoForms makes use of reasoning services based on the semantics of description logics, so that besides declared axioms, entailments are also included in the generated structure.

Figure 3: a) Graph with instances of Woman1. b) Form fragment for hormonal and genetic history

3. Conclusions and future work

We introduced OntoBreastScreen, an innovative ontology-based application for breast cancer screening. Unlike other approaches, OntoBreastScreen uses description logic reasoning within the Breast Cancer Screening Recommendation Ontology (BCSR-Onto), eliminating the need for rule-based systems and providing a robust framework for generating screening recommendations. By leveraging ontologies as modeling tools, the system facilitates clinicians selecting the most appropriate screening recommendations, aligned with guidelines from recognized health organizations. Moreover, the use of Ontoforms generator for automated ontology-user interface generation facilitates maintainability by guidelines changes. This approach not only improves the maintainability and adaptability of the application but also helps ensure that it remains up-to-date with evolving clinical practices and patient needs.

While OntoBreastScreen has been validated by an expert user who is one of the authors of this work, it requires validation by independent healthcare professionals, including imaging specialists, oncologists, gynecologists, and breast cancer researchers. The next steps are starting to proceed with this validation, as it is crucial to evaluate how effectively the tool facilitates communication between doctors and patients.

We also plan to address internationalization and localization. By adapting the system to support multiple languages and cultural contexts, we aim to make it accessible in diverse healthcare settings worldwide. Another important area of future work is developing additional ontologies for related health conditions. This will broaden the scope of the system, making it applicable to other areas of preventive medicine and expanding its potential impact.

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