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Pattern-Based Logical Definitions of Prenatal Disorders Grounded on Dispositions

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Abstract. Biomedical ontologies define concepts having biomedical significance and the semantic relations among them. Developing high-quality and reusable ontologies in the biomedical domain is a challenging task. Pattern-based ontology design is considered a promising approach to overcome the challenges. Ontology Design Patterns (ODPs) are reusable modeling solutions to facilitate ontology development. This study relies on ODPs to semantically enrich biomedical ontologies by assigning logical definitions to ontological entities. Specifically, pattern-based logical definitions grounded on dispositions are given to prenatal disorders. The proposed approach is performed under the supervision of fetal domain experts.

Keywords. biomedical ontologies, Ontology Design Patterns, logical definitions, prenatal disorders, BFO, dispositions

1. Introduction

High-quality and reusable ontologies are fundamentals for developing relevant semantic applications [1]. In the biomedical domain, building such ontologies is a challenging task, especially as the size and complexity of the ontology increases [2]. Ontology Design Patterns (ODPs) [3] address the quality and reusability concerns by providing different types of patterns supporting ontology design [1]. They are encouraging to capture common modeling situations, help facilitate ontology development and avoid common mistakes [2]. ODPs, which do not depend on any specific representation language, are categorized into different types such as *Presentation*, *Reasoning*, *Content*, and *Structural*. This work considers *Content* ODPs (CPs) being very beneficial kind of patterns for ontology design, because they provide solutions to domain-oriented problems [3]. They aim to solve modeling issues regarding ontology content, either in the general or a specific domain of the study [4]. We are interested in CPs in the specific domain which is the modeling of logical definitions.

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Logical definitions aim to associate terms within an ontology under development with terms in external species-neutral ontological resources such as BFO², PATO³, and RO⁴. These definitions allow cross-species mapping using automated semantic reasoning and support quality control [5] and classifications (is-a/subclass relationships inferences). In biomedical ontologies, logical definitions are managed mainly in HPO⁵ [6]. Encoding logical definitions in ontologies under development is a challenging task. Thus, approaches supporting pattern-based ontology design (e.g., eXtreme Design (XD) [7,8] and DOS-DPs [9]) and extensible ontology development (e.g., MIREOT [10]) are tackled. Such approaches bring solutions to create/reuse and apply logical definitions as ontology patterns.

This work is part of the SUOG (Smart Ultrasound in Obstetrics and Gynecology) project⁶. In SUOG, an ontology-based decision support system for complex ultrasound diagnosis in obstetrics and gynecology is intended [11]. The SUOG ontology, which is under development, distinguishes two main sub-ontologies: *prenatal disorders* (e.g., cerebral midline anomaly, complete molar pregnancy, etc.) and *prenatal phenotypes* (e.g., absent right superior caval vein, absence of discrete gestational sac, etc.) that suggest one or multiple disorders. In previous work [12], prenatal phenotypes entities are defined logically by reusing quality-oriented CPs from HPO⁷ [6]. In this study, we focus on assigning logical definitions to prenatal disorders using disposition-oriented CPs.

The remainder of this paper is organized as follows. Section 2 introduces the methods. The results are presented in section 3. We discuss the study in section 4. Finally, section 5 concludes the paper.

2. Methods

Our research's main objective is to logically define prenatal disorders using CPs. To achieve the goal, we are based on eXtreme Design (XD) [7,8] to create the required ontology patterns. Besides, we envisaged that the targeted pattern(s) will be grounded on dispositions. The concept of *disposition* is addressed in some upper-level ontologies such as Basic Formal Ontology (BFO) [13,14] and the Unified Foundational Ontology (UFO) [15]. In BFO, a *disposition* (BFO:0000016) is defined as a *realizable dependent continuant inherent in some independent continuant* [16]. Two main characteristics of dispositions are exposed: 1) *dependency* - the existence of a disposition (e.g., fragility) requires a *bearer* (e.g., a material object such as glass) which is independent (i.e., to exist, a bearer is not required) and having some physical makeup features (e.g., molecular structure); 2) *realization* - a disposition is realized in a *process* (e.g., glass breaking) based on triggering conditions (e.g., the glass being forcefully pressed) [14,17]. Thereby, to exist, dispositions require some qualities inherent in the bearers (called *categorical basis* [18,19,20]) which are, or would be, causally relevant to the manifestation of a disposition [21]. Examples of categorical basis are the structure or morphology of the

²Basic Formal Ontology, <http://www.obofoundry.org/ontology/bfo.html>

³Phenotype And Trait Ontology, <https://github.com/pato-ontology/pato/>

⁴Relation Ontology, <http://www.obofoundry.org/ontology/ro.html>

⁵Human Phenotype Ontology, <https://hpo.jax.org/app/>

⁶<https://www.suog.org/>

⁷Human Phenotype Ontology, <https://hpo.jax.org/app/>

disposition's bearer. In the medical domain, *allergy* is an example of a disposition inherent in specific components of the immune system of an organism and *allergic reaction* represents the realization of allergy [22].

In the SUOG ontology, prenatal disorders are prescribed as “malformations” or “anomalies” affecting fetal organs. The ontological analysis of disorders is debatable in the biomedical ontology engineering community. In this study we follow Scheuermann [23] and Rohl [19] approach that considers *disorders as physical basis of dispositions*. Based on these perspectives, the ontological requirements in SUOG and a list of Competency Questions (CQs) are defined. Examples of CQs are: (CQ1) what dispositions having prenatal disorders as material basis? (CQ2) What anatomical structure is the bearer of such dispositions? (CQ3) What qualities are the categorical basis of such dispositions? (CQ4) What is the bearer of the categorical basis? (CQ5) The disposition is realized in what process? (CQ6) Exist any participants in the process?

In the following, we give a formalization of the specifications of dispositions applied in the context of prenatal disorders using some conventional logical operators of first-order logic such as \rightarrow , \exists , and \wedge . In formalization, unary predicates are represented in bold, and relations are in italic and x , y , and z are variables. First, the specification of prenatal disorders as material basis of dispositions is formalized.

prenatal disorder(x) $\rightarrow \exists y$ (**disposition**(y) \wedge *is material basis for*(x,y))

The specification of dispositions' bearers, representing the affected anatomical structures associated to disorders, is formalized as follows.

disposition(x) $\rightarrow \exists y$ (**anatomical structure**(y) \wedge *inheres in*(x,y))

The specification of dispositions' categorical basis is formalized as follows.

disposition(x) $\rightarrow \exists y$ (**quality**(y) \wedge *has categorical basis in*(x,y))

In the SUOG ontology, a prenatal disorder is *suggested* by one or more prenatal phenotypes (defined as *qualities* captured by ultrasound mechanisms). Thus, following BFO's definition of *qualities as continuants* and analysis of *processes having continuants as participants*, we consider that phenotypes participate in the realization of dispositions (i.e., processes). The specification of dispositions realization is formalized as follows.

disposition(x) $\rightarrow \exists y$ (**process**(y) \wedge *has realization*(x,y)) \wedge
 $\exists z$ (**prenatal finding**(z) \wedge *has participant*(y,z))

These formalizations are considered to create P, a disposition-oriented pattern composed of six main clauses. P includes basic categories (concepts and relations) which are reused from existent validated ontologies and variables (*var*), which will be filled during pattern application. Examples of basic concepts are *disposition*, *process*, and *quality* reused from BFO. Examples of relations are *is material basis for* and *has categorical basis in* which are specializations (in the SUOG ontology) of *is basis for realizable* (RO_0004018) and *realizable has basis in* (RO_0004017) respectively.

P: 'is material basis for' some ('bfo:disposition'
 and 'has realization' some ('bfo:process'
 and 'has participant' some var)
 and 'inheres in' some var
 and 'has categorical basis in' some (var
 and 'inheres in' some var))

3. Results

The disposition-oriented pattern is applicable to logically define the different categories, sub-categories, and specific classes of prenatal disorders. Figure 1 depicts examples of logical definitions assigned to cerebral midline anomaly and complete molar pregnancy. The application of the proposed pattern in the SUOG ontology is in progress. A total of 11 general categories and 45 sub-categories of prenatal disorders are logically defined. The generated logical definitions are verified against the CQs, evaluated, and validated by the fetal domain experts.

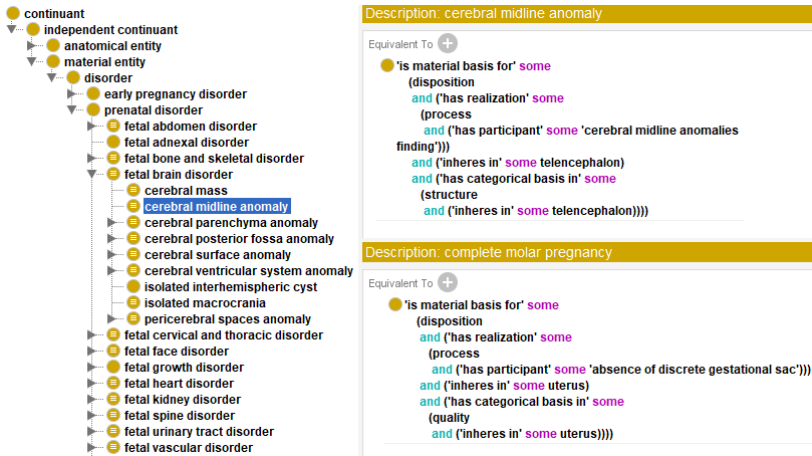


Figure 1. Examples of logical definitions assigned to prenatal disorders

4. Discussion

In this work, we have proposed a pattern-based approach for assigning logical definitions to prenatal disorders grounded on dispositions. By applying this approach, we aligned the SUOG ontology to upper-level ontological resources and enriched the ontology model semantically. Besides, using Content ODPs has simplified the ontology engineering process and permitted the extensibility of the ontology. In this study, the SUOG ontology is aligned to BFO [13]. In further work, we will analyze the definition of dispositions provided in UFO [15] and its applicability to build/enhance our pattern(s). On the conceptual level, in the proposed (general) pattern, we considered a single categorical basis for a disposition. Meanwhile, a disposition may have multiple categorical basis [17] which will be envisaged for defining the most specific prenatal disorders entities. Finally, concerning the participation of prenatal phenotypes in the realization of dispositions, the proposed pattern will be specialized to cover the cases where multiple prenatal findings are participating. For the best application of the disposition-oriented pattern(s), it is required that the fetal domain experts perform or supervise the fulfillment of variables for each prenatal disorder entity (e.g., 4 variables/disorder in P). The associated values, which are provided as csv file, represent the input data required for the automatic application of the patterns. This process risks delaying the pattern application to define the entire prenatal disorders entities (total 1132 in the current version of the SUOG ontology).

5. Conclusion

Building high-quality and reusable ontologies in the biomedical domain is challenging due to ontologies' increasing size and complexity. Ontology Design Patterns and ontology reuse approaches are tackled to overcome the challenges and simplify the ontology building process. This study discussed pattern-based logical definitions of prenatal disorders entities in the SUOG ontology. A proposed content pattern is grounded on BFO:disposition leading to promising results in modeling logical definitions.

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