OntONeo: The Obstetric and Neonatal Ontology

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***Abstract* – This paper presents the Obstetric and Neonatal Ontology (OntONeo), an ontology in development to provide a consensus representation of salient electronic health record (EHR) data for both mother and child and to serve interoperability of the associated data and information systems. More generally, it will serve interoperability of clinical and translational data, for example deriving from clinical trials. Interoperability of EHR data is important to ensuring continuity of care during the prenatal and postnatal periods for both mother and child. As a strategy to advance such interoperability we use an approach based on ontological realism and the ontology development principles of the Open Biomedical Ontologies (OBO) Foundry, including reuse of reference ontologies wherever possible. We describe the structure and coverage domain of OntONeo, the process of creating and maintaining the ontology.**

Keywords* – *Biomedical Ontology, Obstetric and Neonatal Ontology, Electronic Health Records, EHR, BFO, OBO Foundry

I. Introduction

Electronic health records (EHRs) are tools for recording and communicating medical data among health professionals. They serve as repositories of information about the physical and mental state of patients and patients' families as these evolve over time. Easy access to EHR data is crucial to ensuring continuity of care when patients move from one healthcare provider to the next because of changes in health state.

A. An EHR Case Study

Our example case concerns how continuity of care for women and newborns is addressed in the Brazilian Unified Health System, which guarantees a woman's entitlement to reproductive planning, pregnancy, childbirth and postpartum care. The Brazilian healthcare structure involves facilities established by governments at the federal, provincial, state and local municipal levels. The Brazilian Unified Health System uses its SisPreNatal system to gather information about prenatal care from healthcare facilities at all these levels.

However, the healthcare facilities themselves are autonomous and have established multiple information systems to deal with their EHR data, which are built around differing standards, terminologies and conceptual models. This non-standardized process thus requires a great deal of effort to unify EHR data at the national levels to ensure continuity of quality care for both mother and child.

In what follows we propose a strategy to overcome this lack of interoperability using a solution based on formal ontology. We describe both the OntONeo ontology and the development method we have used to build it.

II. Background: Prenatal and Postnatal Care

The course of pregnancy, childbirth and child development involves a series of stages as illustrated in Figures 1 and 2 [5; 10]. Information pertaining to all of these stages is recorded in the EHR. Medical care during these stages is organized into *prenatal*, *intrapartum* and *postnatal* periods of care. The pre- and postnatal periods are the periods before and after birth (*partum*). The intrapartum period is defined (roughly) as the period from the onset of labor to the completion of delivery of both baby and placenta [2]. Clinical care in each stage calls upon different medical specialties, different lab tests and imaging procedures, as well as different immunizations, screening and other healthcare-related processes [2-4; 6].

Embryology is the discipline responsible for studying the human embryo and the processes of embryogenesis which occur in the stage of human development initiated by fertilization and extending through the first eight weeks of development. Through the processes of division and differentiation of cells involved in embryogenesis almost all organs have developed by ten weeks after fertilization [2; 8].

After childbirth, each newborn typically passes through a series of clinical encounters involving care by neonatologists and pediatricians. Pediatrics deal with the physical, emotional, and social health of infants, children, adolescents, and young adults from birth to 18 years old (21 in the US) [5]. Neonatology is a subspecialty of pediatrics that consists of medical care for critical newborn infants, usually premature and full-term infants following discharge from an Intensive Care Unit [10].

The medical care of the mother, too, involves several distinct medical specialties, including the care of a gynecologist in her adolescence and the obstetrician, who will see to her care during the pregnancy and postpartum periods [2-4; 6]. Gynecology is the medical specialty that deals with the health of the female reproductive system and of her breasts. Obstetrics is the specialty that deals with pregnancy, childbirth, and the postpartum period. Nowadays, these two specialties are joined together under the heading Obstetrics and Gynecology [2].

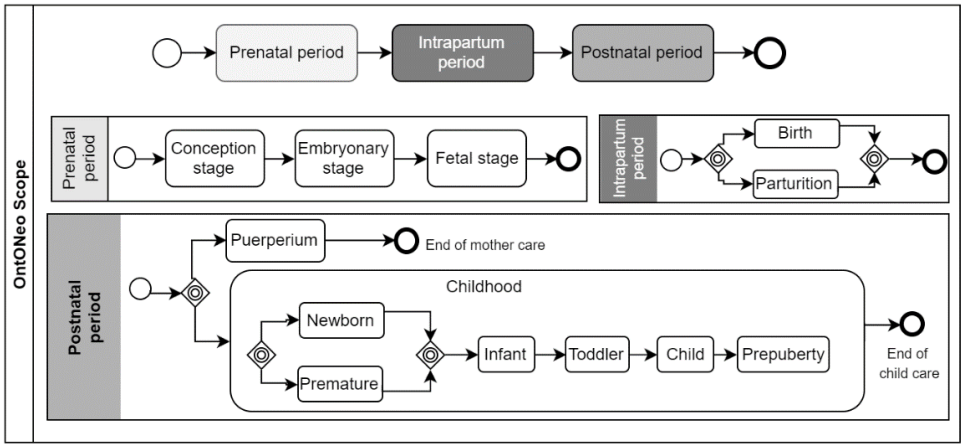

Fig. 1. Stages of pregnancy and child development (By authors)

Fertilization and prenatal period			
		Gestational Age	
Ovulation		Week 1-2	
Conception	Fertilization	Week 3	Day 1
	Cleavage		Day 2
	Compactation		Day 3
	Differentiation		Day 4
	Cavitation		Day 5
	Implantation		Day 6
Embryonary period	Gastrulation	Week 4	
	Somitogenesis	Week 5	
	Organogenesis	Week 6	
	Histogenesis	Week 7	
	Neurulation	Week 8	
Fetal period		Week 9-40	
Normal Gestation Birth			

Puerperium or postnatal period (Mother)	
Immediate puerperium	First 24 hours after labor
Early puerperium	First week after labor
Remote puerperium	Around 6 weeks after labor

Childhood (Child)		
Premature	Baby born under 36 weeks of gestation	
Newborn	1st month after birth	
Infant	2nd to 12th month	
Toddler	1st month of 2nd year to 3 years	
Child	4th year to Prepuberty period (around 10 years)	
Adolescent	Prepuberty (girls)	Around 11 years
	Prepuberty (boys)	Around 12 years
	Puberty (girls)	11th to 15th/17th years
	Puberty (boys)	12th to 16th/17th years

Fig. 2. Timeline of pregnancy and child development (By authors)

Both obstetricians and gynecologists need to possess extensive knowledge of human anatomy at successive stages of development in order to understand the physical changes in the mother from the onset of labor until menopause. Obstetricians need in addition to understand the physical changes which occur in human development from the embryonal and fetal stages onwards [2-6; 8; 9]

Obstetric care also relies on data regarding family history concerning both health and social behavior, and on data regarding genetic defects derived from both the maternal and paternal lineages. It will need to take account also of other common factors in family history that influence a newborn's health, including environment, educational level, behavior, and lifestyle habits such as diet and physical activity [2-4; 6].

III. The Obstetric and Neonatal Domains of OntONeo

Our strategy in building OntONeo focused specifically on EHRs involved in care activities encompassing the mother, the developing offspring, and the newborn child in the prenatal, intrapartum and postnatal periods of care depicted in Fig. 1. For purposes of ontology population, we reviewed not only the EHR systems adopted by different hospitals but also Brazilian and international EHR standards. We also conducted interviews with Brazilian and American obstetricians to identify the workflow of women's health clinics and the information needs of the medical practitioners involved. In the future, we plan also to conduct similar interviews with pediatricians.

The EHR standards we reviewed include: the *Woman's Health Record* and *Antepartum Record and Postpartum Form* provided by the American College of Obstetricians and Gynecologists (ACOG); the *Children's Electronic Health Record Format* provided by the Agency for Healthcare Research and Quality (AHRQ); the *Standards for the Clinical Structure and Content of Patient Records* provided by the UK Health and Social Care Information Centre and the Academy of Medical Royal Colleges; and the *National Standard for Patient Discharge Summary Information* from the Australian Health Information and Quality Authority.

A. The general clinical information in OntONeo

OntONeo is designed to support the integration and interoperation of data originating from the following medical specialties and domains: human embryology; anatomy of the mother, embryo, fetus and child; general gynecology and obstetrics; neonatology; and pediatrics.

In our analysis of the above-cited reference documents, we identified a set of basic types of information that are common across EHRs independently of specialty. Examples are: data about healthcare facility, healthcare provider (physician, nurse, etc.) and healthcare consumer (patient). At each medical

encounter, the healthcare provider gathers vital signs data (e.g. body temperature, blood pressure, heart rate, and respiratory rate) and needed demographic data.

Typically, the obstetrics EHR also includes a section devoted exclusively to recording information about the family members of the pregnant mother. A family's health history is important for determining the health risks both to the woman and her child. A usable family health history will combine information going back three generations on both the mother's and the father's side [2; 4].

Finally, the OntONeo coverage domain includes clinical information related to the medical history of both mother and child in different stages of life, including: i) immunizations, vaccines, permanent or sporadic medications; ii) existence of allergies, syndromes, chronic and previous diseases; iii) symptoms reported and diagnoses; iv) surgeries and treatments submitted, physical examinations, laboratory and images test results.

B. *Embryology and Anatomy in OntONeo*

Because anomalies and congenital diseases may arise during development of the embryo, physicians that monitor prenatal stages must have expertise in embryology [2; 8]. The pregnant mother is examined in order to detect fetal or embryonic complications, and screening exams may be performed to detect developmental anomalies. For example, a prenatal ultrasound exam will provide images of the baby, amniotic sac, placenta, and ovaries.

Because female anatomy and physiology change through puberty, post-menarche, and pre-menopause periods, knowledge about human female anatomy and physiology in these successive stages are also foundations of obstetric and gynecologic care [2-4; 6].

Furthermore, during gynecological and obstetric care, a doctor performs a physical examination of the mother to detect any changes in her body that might indicate an underlying disease. The physician's observations during these examinations are recorded in the gynecological and obstetric EHR for both mother and fetus, and similar observations form part of the EHR of the child in the newborn and subsequent stages.

C. *Gynecology and Obstetrics in OntONeo*

The gynecological domain of OntONeo provides ontology terms for annotating data gathered from the evaluation of the mother's health including i) physical examination of pelvic region and breast; ii) menstrual and contraceptive history; iii) current and history information of the sexual behavior; iv) gynecological image exams with results; and v) information on her general medical history [2; 4; 6].

The obstetrics domain of OntONeo, in contrast, provides the terms needed to annotate data gathered during the course of pregnancy such as screening, testing, physical and labs exams, immunizations, signs, and symptoms. The obstetrics domain also covers data relating to the intrapartum and postnatal care of the mother – for example relating to irregular bleeding, breastfeeding and psychological symptoms [2-4].

D. *Pediatrics and Neonatology in OntONeo*

Child development involves distinct periods, as shown in Fig. 2. Newborn infants, particularly those born prematurely or born with some syndrome or disease, are submitted for monitoring and care by neonatologists. Generally, 24 hours after birth the physician performs a physical exam of the newborn infant. In addition, the child undergoes a routine of pediatrics appointments, exams, screening tests, immunizations, and so forth [5]. After the second month of birth, the child´s healthcare monitoring are transitioned to a pediatrician..

IV. METHODOLOGY FOR BUILDING ONTONEO

We applied the methodology of ontological realism to develop OntONeo. This methodology can be summarized as the view that an adequate ontology for a domain should be constructed not in order to represent the contents of existing data or models but rather in order to represent the entities in the world described by (the terminological part of) the relevant established science. Principles to be followed in achieving this goal are summarized in [1; 13].

The development of OntONeo follows the Open Biomedical Ontologies (OBO) Foundry principles, which aim to build a set of interoperable ontologies for representation of biological and biomedical reality [12]. We employed the Basic Formal Ontology (BFO) version 2.0 [1; 11] as top-level and reused other ontologies from the OBO Foundry wherever possible.

The development of OntONeo follows an incremental modular approach, with each increment adding some new body of terms to the ontology as thus far developed. The work on each increment proceeds in three steps. First, we define the scope of the module to be added by creating a preliminary list of relevant terms. Second, we build a consensus vocabulary on the basis of this list, beginning with the examination of relevant existing ontologies. (Table 1 presents the ontologies from which we selected terms for reuse.) Third, we ingest the new terms, together with required relations, into OntONeo. Given our commitment to collaboration with the OBO Foundry, we also inform the editors of existing ontologies the creation of new terms and relations where gaps are identified.

Step one consists in the creation of a preliminary list of the terms needed for tagging data of the relevant sorts.

We perform the second (vocabulary building) step by searching on Ontobee [14] and Bioportal for the terms on our list. When a search returns more than one result, we refine our selection using the following criteria: i) does the ontology in question adhere to the OBO Foundry principles? ii) does the ontology provide a definition? iii) how many times was the term as defined in this ontology previously reused? iv) OBO Foundry status of the ontology (according to the legend F–*Foundry*, L–*Library*, N–*Not specified*).

In the third step, we use OntoFox and Protégé to import the needed terms and axioms into the OntONeo OWL file. To guide this process we create a sketch of how the selected term will interrelate within the scope of OntONeo. Successive versions of this sketch are generated using the CMap tool from the Collaborative Ontology Environment. Section V describes our results.

TABLE 1: CANDIDATE ONTOLOGIES FOR REUSE

Ontology	Contributions to OntONeo
Information Artifact Ontology (IAO)	Information artifacts and information content entities such as documents, images.
Ontology of Document Acts (d-acts)	Social acts that transfer and revoke rights and duties among people. E.g. declarations.
Ontology for General Medical Science (OGMS)	Terms of clinical medicine used across medical disciplines. E.g. Symptoms, signs.
Ontology for Biomedical Investigations (OBI)	Terms from the domain of experimentation. E.g. protocol, sample.
Ontology of Medically Related Social Entities (OMRSE)	Social entities related to healthcare, such as families, marriages, consent forms.
Gene Ontology (GO)	Terms from embryology such as fertilization, gastrulation.
Foundational Model of Anatomy Ontology (FMA)	Terms from human anatomy and development. E.g. Organ, uterus, ovary, embryo.
Human Disease Ontology (DOID)	Domain of human diseases. E.g. disease, syndrome, genetic disease, congenital.
Phenotype And Trait Ontology (PATO)	Phenotypic qualities. E.g. Color, temperature, odor, phenotypic sex (female, male).
Vaccine Ontology (VO)	Terms relating to vaccines and vaccination.
Ontology for Newborn Screening Follow-Up and Translational Research (ONSTR)	Processes and agents involved in newborn screening.

V. RESULTS: ONTONEO ENTITIES AND RELATIONS

OntONeo is being developed to support the semantic interoperability of data from EHRs in the obstetric and neonatal domains. It also aims to support access to the different bodies of data collected in different EHRs relating to the prenatal, intrapartum and postnatal periods of care.

The division of OntONeo into loosely coupled modules promotes maximum flexibility by allowing the ontology to prioritize newly identified needs. The three most important modules in the OntONeo are:

- *OntONeo-Core*: provides terms for the annotation of the basic information elements required by all aspects of Obstetric and Neonatal care.
- *OntONeo-Documents*: provides terms representing the types of documents used in Obstetric and Neonatal care such as EHRs and consent forms.
- *OntONeo-Social*: covers the domain of social entities involved in obstetric and neonatal care such as family relations and demographic information.

In this section we explain the ontological representation of some major terms used in EHRs and detail the corresponding parts of the OntONeo suite.

A. The Obstetric domain of OntONeo

To represent the stages involved in the course of pregnancy, as illustrated in Figures 1 and 2, we incorporated selected terms from GO Biological Process ontology, as illustrated in (Fig. 3). The corresponding terms are part of *OntONeo-Core*.

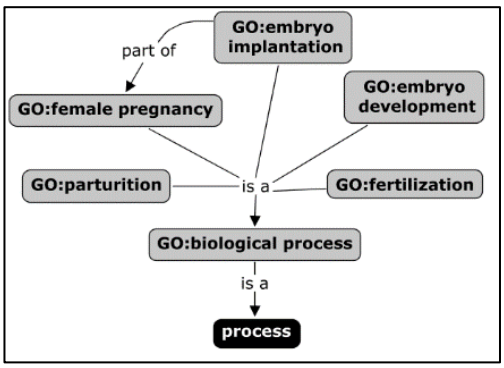

Fig. 3. Biological proccesses related to the course of pregnancy

B. The OntONeo representation of the EHR

Each EHR includes a section that contains general information, independent of medical specialty, relating to patient demographics and vital signs. In addition, each medical specialty has specific information about the care provided which enables us to associate a specific set of EHR terms with each specialty. These terms are divided into sections. Thus, each gynecology EHR contains sections relating for example to menstrual history, physical examination, family history, pregnancy history, demographic information. Some sections contain items grouped according to corresponding forms or other documents (relating for example emergency contact information, healthcare consumer identification, healthcare facility identification, age at menarche, personal name, age of appearance of nipples, and so on).

To cover the corresponding forms and documents we incorporate terms from the Information Artifact Ontology (IAO). For IAO, an EHR is a *medical record*, a subtype of *document* that is in turn a subtype of *information content entity*. *Obstetric medical record* and *pediatric medical record* are subtypes of *medical record* in their turn. A document has parts, for example, *medical record section*, and subsections such as *vital signs data section*. Certain sections have as parts specific sets of information items such as *vital signs measurement data set*. Each *data set* groups *data items*, for instance, *respiration rate measurement data item* and *body temperature measurement data item*, as illustrated in Figures 4 and 5.

The corresponding terms are included in *OntONeo-Documents* and they represent what BFO calls generically dependent continuants, entities which are concretized (for example made manifest to a study nurse) in specifically dependent continuants such as the patterns of actual signatures on actual paper documents). This module includes in addition terms referring to *independent continuants* (for example organisms, healthcare facilities) and to *roles* (for instance the roles *author of a signature*, *subject of a record*). A *medical record* document is in IAO terms *about* some *organism*. Each *measurement datum* data item is the output of some *planned process* of measurement such as *respiration rate measurement process*.

In the Figures 4 and 5 below, black signifies terms from BFO, gray represents terms reused from OBO Foundry ontologies, and white represents entities new to OntONeo.

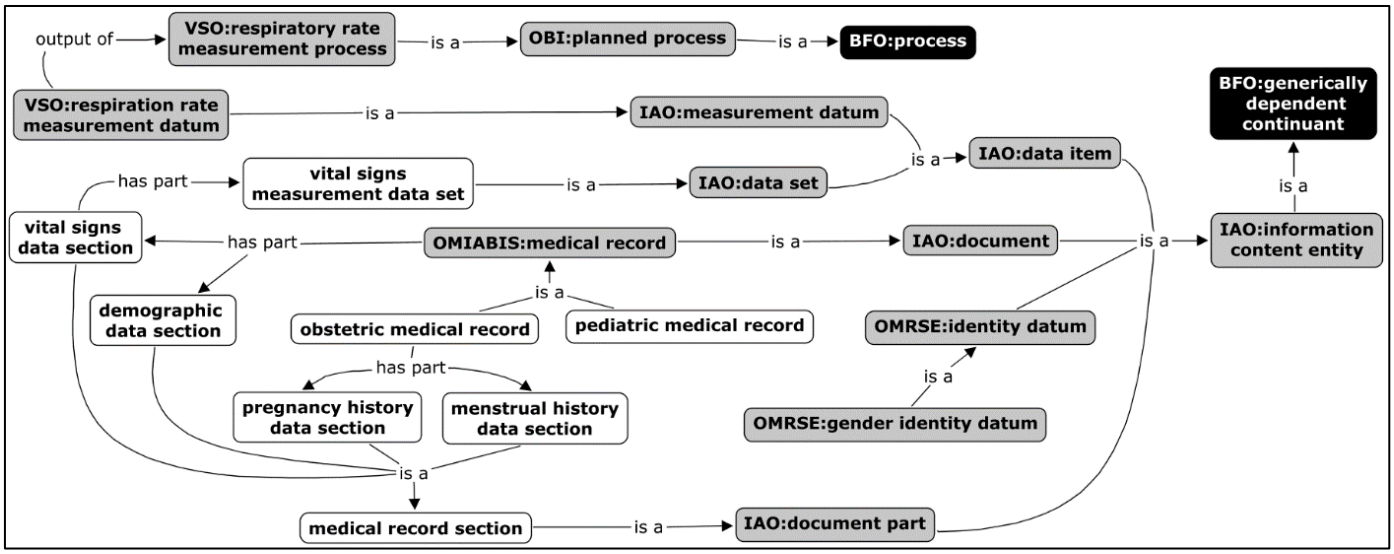

Fig. 4. Electronic health record template representation of OntONeo (By authors)

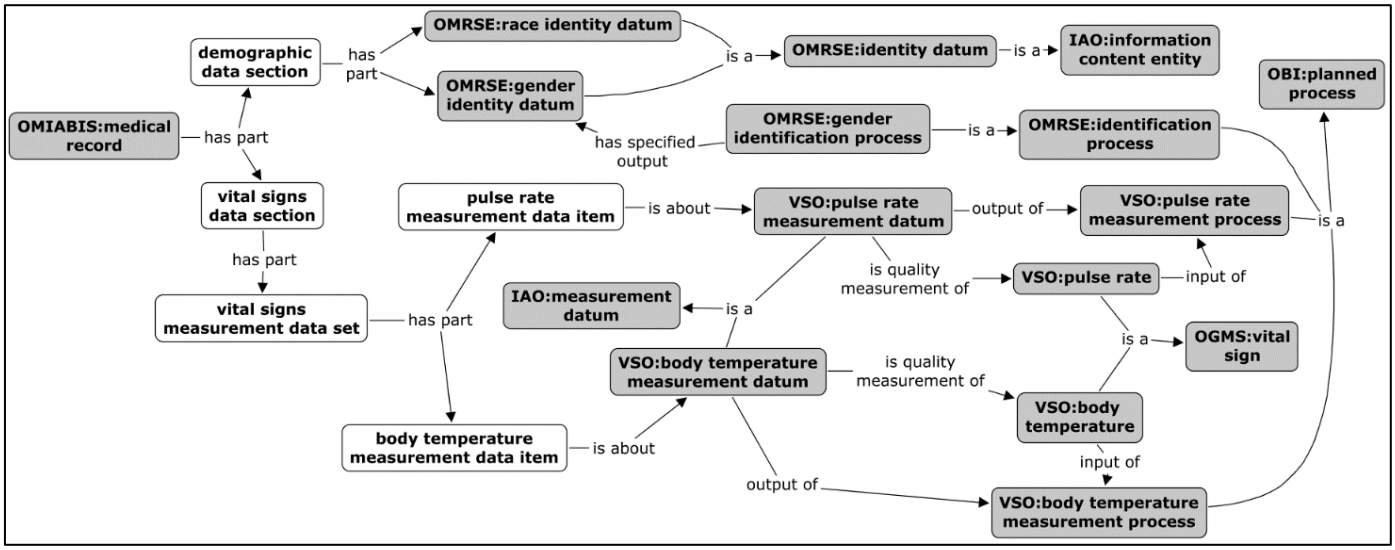

Fig. 5. Electronic health record parts and sections representation of OntONeo (By authors)

C. The Anatomy and Embryology domain of OntONeo

Next, we consider EHRs that document information deriving from the physical examination of a female. A physical pelvic exam is used in obstetrics and gynecology to detect signs of disease in the reproductive organs of the mother and an ontological representation of female anatomy and physiology is provided in OntoNeo in order to provide a single semantic interpretation of female body structures. For this purpose we have imported terms and relations from the Foundational Model of Anatomy (FMA), as shown in Fig. 6. The female reproductive system consists of the ovaries, uterine tubes, uterus, vagina, and external genitalia [9]. The corresponding terms are part of *OntONeo-Core*.

To incorporate human development stages and the embryogenesis process presented in Fig.1, we have once again imported terms and relations from the GO. We also needed to represent in OntONeo entities related to prenatal and postnatal care. Often, obstetricians examine the pregnant mother and the fetus/embryo in order to monitor the course of a pregnancy. Some data from the prenatal EHR provide information about the mother, some are about the fetus/embryo, reflecting the fact that anomalies and congenital diseases may become apparent on either side during successive stages of pregnancy.

Finally, we propose a sketch of the entities realized through the embryogenesis process and of material entities that represent the stages of embryological development (Fig. 6). These types of entities and their interrelations are described in textbooks of embryology, where we learn for example that zygote *has_disposition to_form* spindle; blastocyst *has_disposition* to_*adhere_to* wall of uterus *realized_by* implantation process; wall of uterus *has_disposition to_be_adhered_to_by* blastocyst; and so forth.

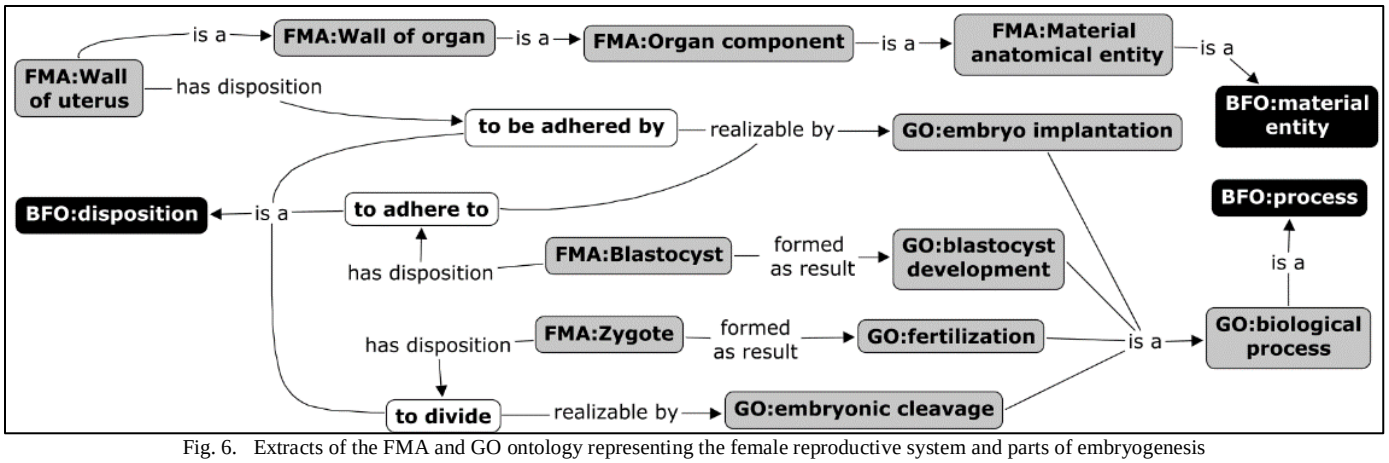

Fig. 6. Extracts of the FMA and GO ontology representing the female reproductive system and parts of embryogenesis

VI. Conclusion

We described OntONeo, an ontology in the obstetric and neonatal domain. OntONeo aims to represent the EHR data involved in the care of pregnant woman and of her child from fetus through the newborn, infant and toddler stages. We also described the methodology that has been applied in creating OntONeo.

Since OntoNeo uses examples from specific EHRs as its starting point, it might be argued that the result will be unsuitable for use in other contexts. However, given the methods adopted in its development, the ontology's coverage domain includes the representation of highly general entities in order to allow its use in a wide range of different situations. For example, in the USA the label 'Latino' is used in a way which makes no sense in Brazil. Our approach allows ontology annotations to deal with such differences in a neutral manner. Cases in which there is the need for representing a specific context will be organized in a specific level of the ontology.

OntONeo is still in the early stages of its development. The current version at any given stage can be found at *http://ontoneo.wordpress.com*. We follow the governance, versioning and update practices adopted by the ontologies in the OBO Foundry, viewing ontology development as an interactive process. Thus, each successive version will be subjected to validation by the representatives of each community of specialists, including physicians and other healthcare professionals, as well as by experts in ontology development.

We believe research in OntONeo is justified by the lack of formal representation in the obstetric and neonatal domain, for which we provide a needed specialized vocabulary that will include a formal representation that is much more comprehensive than other currently available ontologies (including FMA, GALEN, and ontologies in UMLS). OntONeo will contribute to the interoperability of information regarding the different stages of pregnancy, drawing on anatomy, embryology, and other disciplines related to childbirth and development. In addition, it will facilitate the understanding of how such information can be organized in EHRs for purposes of effective healthcare.

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