OntONeo: The Obstetric and Neonatal Ontology

Fernanda Farinelli[a, b, c, 1], Mauricio Barcellos Almeida[a,d], Peter Elkin[b,d] and Barry Smith[c,d]
[a] School of Information Science, Federal University at Minas Gerais, Belo Horizonte, MG, Brazil
[b] Department of Biomedical Informatics and [c] Department of Philosophy, University at Buffalo, Buffalo NY, USA
[d] National Center for Ontological Research, University at Buffalo, Buffalo NY, USA
[1] Corresponding Author: fernanda.farinelli@gmail.com

***Abstract* - This paper presents the Obstetric and Neonatal Ontology (OntONeo). This ontology has been created to provide a consensus representation of salient electronic health record (EHR) data 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 genomics disciplines and 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 on the ontology development principles of the Open Biomedical Ontologies Foundry, including reuse of reference ontologies wherever possible. We describe the structure and coverage domain of OntONeo and the process of creating and maintaining the ontology.**

Keywords* - *Biomedical Ontology, Obstetric and Neonatal Ontology, electronic health records, BFO, OBO Foundry

I. INTRODUCTION

Electronic health records (EHRs) are tools for communication of medical data among health professionals. They serve as evolve repositories of information about the physical and mental state of patients and also about the state of health of the patient's 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 change of health state.

A. An EHR Case Study

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

However, the healthcare facilities themselves are autonomous and they have established many different information systems (ISs) to deal with their EHR data, built around many different standards, terminologies and conceptual models. This thus requires a great deal of effort to unify EHR data at the national levels to ensure continuity of care for both mother and child.

In what follows we propose a strategy to overcome this lack of interoperability using a solution based on a 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 Fig. 1 and Fig. 2 [5; 10]. Information pertaining to all of these stages is recorded in the EHR. Medical care during these stages is divided 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 baby and placenta [2]. Clinical care in each stage calls upon different medical specialties. The course of pregnancy calls upon different lab tests and imaging procedures, as well as immunizations, screening and other healthcare-related processes [2-4; 6].

Embryology is the discipline responsible for studying the human embryo and the embryogenesis process which is the stage of human development initiated by fertilization and extending through the first eight weeks of development. During embryogenesis the division and differentiation of cells occurs so that by ten weeks after fertilization almost all organs have developed [2; 8].

After childbirth, each newborn is subjected to a series of clinical encounters. The childhood period involves 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 the Intensive Care Unit [10].

Women's medical care involves several medical specialties. During childhood she is monitored by pediatricians. In her adolescence, she starts her care with a gynecologist who she will visit periodically. When a woman becomes pregnant she is monitored by an obstetrician, who cares for her also during the postpartum period [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, the two specialties are joined together under the heading Obstetrics and Gynecology (OB/GYN) [2].

Both obstetricians and gynecologists are required to know about human anatomy at successive stages of development and to understand the physical changes in the mother from her birth until menopause. In addition, the obstetrician should understand the physical changes occurring in each stage of human development including the embryonal and fetal stages which occur during prenatal period [2-6; 8; 9].

Obstetric care also relies on data regarding family history, concerning both health and social behavior. Genetic defects deriving from both maternal and paternal lineage are of equal significance. In addition to genetics, family members will often exhibit other common factors that influence health, including environment, educational level, behavior, and lifestyle habits such as dietary and physical activity [2-4; 6].

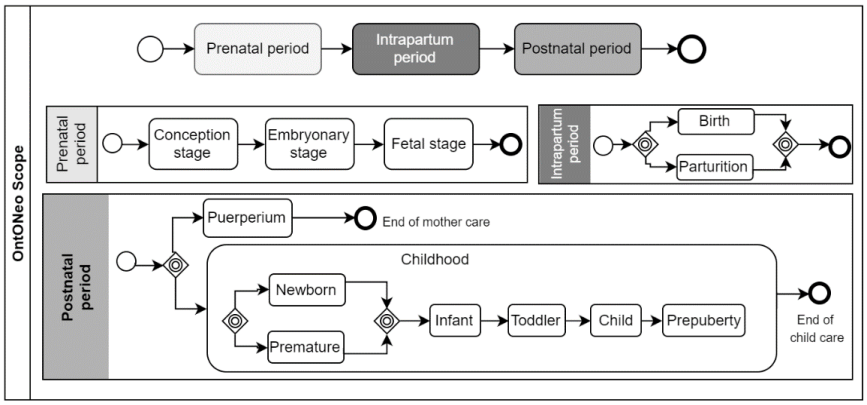

Fig. 1. Stages of pregnancy and child development (By authors)

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)

III. The Obstetric and Neonatal Domains of OntONeo

We focus specifically on EHRs involved in care activities involving the mother, the developing offspring, and the newborn child. These stages, depicted in Fig. 1, are also called the prenatal, intrapartum and postnatal periods of care. The scope of OntONeo was established on the basis of the assessment of a set of EHR standards – both Brazilian and international – and also from the EHR systems adopted by different hospitals. Some interviews with Brazilian and North American obstetricians were also conducted to identify the workflow of women's health clinics and the information needs of the medical practitioners involved. In the future we will conduct interviews also with pediatricians.

Some standards used are: 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; 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: embryology of human beings; anatomy of the mother, embryo, fetus and child; general gynecology and obstetrics specialties; and pediatrics and neonatology.

In our analysis of the above-cited reference documents, we identified a set of basic types of information that are common across different EHRs independently of specialty. For example, data about the health care facility, the health care provider (physician, nurse, etc) and the health care consumer (patient) demographic information. Moreover, at each medical encounter, the healthcare provider gathers vital signs data (e.g. body temperature, blood pressure, heart rate, and respiratory rate).

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 desirable family health history combines information from three kinship generations on both the mother's and (where possible) the father's side [2; 4].

Finally, the OntONeo coverage domain includes those types of clinical information that are related to the patient's medical history, both the pregnant mother, the embryo/fetus, and the 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 are required to know embryology and embryogenesis [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. The results enable tracking of anatomical abnormalities or birth defects of the baby.

Human anatomy and physiology and human development (from birth through adolescence) are knowledge required by pediatricians and by those involved in neonatal care. 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 childbirth EHR in the case of the newborn and of the pediatric EHR in the case of childhood care.

C. *Gynecology and Obstetrics in OntONeo*

The gynecological domain of OntONeo comprehends data gathered from the evaluation of the mother's health during her life such as for example 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].

In case of the obstetrics domain, OntONeo needs to represent data gathered during the course of pregnancy such as screening, testing, physical and labs exams, immunizations, signs, and symptoms. The obstetrics domain also involves data relating to the postnatal care of the mother – for example, relating to breastfeeding and psychological symptoms and irregular bleeding – as well as data gathered during the intrapartum period [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 to monitoring and care by neonatologists. Typically a newborn infant is subjected to a physical exam after 24 hours. In addition, the child has a routine of pediatrics appointments, exams, screening tests, immunizations, and so forth [5]. After the second month of birth, a baby will need monitoring regarding her own health, leading in due course to the involvement of pediatricians.

IV. Methodology for building OntONeo

We used 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 existing data or models but rather to represent (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 aims to develop 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 ontology of OntONeo, which is widely currently accepted in use in the medical and biological domain [7]. In addition, with the aim of fostering interoperability among the existing biomedical ontologies and taking advantage of previously developed ontologies, the we decided to reuse others ontologies from the OBO Foundry.

OntONeo is being developed in an iterative and incremental lifecycle model. It means that the ontology is gradually built through various iterations in which each iteration scope is previously defined. Each iteration brings about a piece of the ontology which increases the ontology with new entities and relations.

Our work on developing OntONeo proceeds on three conceptual levels before the formalization. First, we define the scope of the iteration based on the OntONeo scope. Second, we draw as far as possible on existing ontologies, we build a consensus vocabulary for tagging the different sorts of data, covering: i) the basic biomedical sciences of human anatomy and embryology; ii) medical specialties like gynecology, obstetrics, pediatrics, and neonatology. These terminological resources allow one to understand how data are recorded in previously developed ontologies. Third, we draw new terms and relations required in OntONeo scope. New terms can be created in existing ontologies due to the commitment of OntONeo developers to collaborate with OBO Foundry.

Considering the scope of OntONeo, we reviewed ontologies from the OBO Foundry using the Ontobee portal searching for collections of terms relevant to the OntONeo domain. We also searched for related biomedical ontologies in Bioportal. The ontologies from which we selected terms for reuse are listed in Table 1.

After the iteration scope definition, we established a list of main terms to be represented. Then, to perform the second developing step, we search on Ontobee [14] and Bioportal the terms on our list. When a search returns more than one result, we used criteria for selection such as i) is the ontology in question adherent to the OBO Foundry principles?; ii) does the class have a definition?; iii) how many times the term was previously reused?; iv) the status at OBO foundry according to legend (F-*Foundry*, L-*Library*, N-*Not specified*). We used OntoFox and Protégé to import the needed terms and axioms into the OntONeo OWL file.

At the third step we provide a sketch of how the selected classes within the scope of OntONeo will be organized. We generate each version of sketch using the tool CMap Collaborative Ontology Environment. Our results are described in section V.

As OntoNeo recognizes the importance of reusing terms, one can argue how we intend to update the ontology. We understand that an ontology, as a system, federated database or even a book, will certainly become obsolete and in some point of future will require updates. As happens in system development, this will be done as required, but up to then, our ontology continues to work with the classes and relations developed so far.

TABLE I. 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 health care, 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 ensure access to the different bodies of data collected in different EHRs relating to the prenatal, intrapartum and postnatal periods of care.

To promote maximum flexibility OntONeo is divided into loosely coupled modules designed to meet specific needs. OntONeo is thus a suite or collection of open ontology modules relating to different parts of the relevant EHRs, of which the three most important are:

- *OntONeo-Core*: collects the basic information required by all aspects of Obstetric and Neonatal care.
- *OntONeo-Documents*: focuses on the 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 Fig. 1 and Fig. 2, we incorporated terms from related biological process of Gene Ontology. It´s worth noticing that the intermediary terms were omitted (Fig. 5). The corresponding terms are part of *OntONeo-Core*.

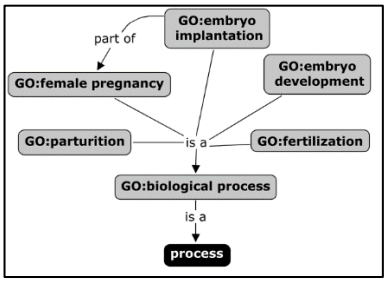

Fig. 3. Biological proccess related to the course of pregnancy

B. The OntONeo representation of the EHR

Each EHR includes a general part that contains general information, independent of medical specialty, such as patient demographic information and vital signs data. Each medical specialty has specific information about the care provided, so we can associate a specific EHR corresponding to each specialty. Each EHR contains what we can think of as separate sections where specific subjects are treated. Thus each specialist EHR contains sections relating to e.g. menstrual history, physical examination, family history, pregnancy history, demographic information. In addition, each section groups items relating to specific fields in corresponding forms (e.g. emergency contact information, health care consumer identification, and health care facility identification). Finally, we have single data items related to a data set or to a document (e.g. age at menarche, personal name, age of appearance of nipples).

To cover the corresponding forms and documents we incorporated terms from the Information Artifact Ontology

(IAO). For IAO, an EHR or *medical record* is an information content entity classified as a subtype of document, and both the *obstetric medical record* and the *pediatric medical record* are subtypes of *medical record*. A document has parts, and we classified the sections of the *medical record* accordingly. The set of information in a *medical record section* we classified as an IAO data set and each datum or field of data grouped in a set of information we classified as data item, as illustrated in Fig. 4 and Fig. 5. The corresponding terms are part of *OntONeo-Documents* and they represent what BFO calls generically dependent continuant entities which are related to specifically dependent continuants (such as the pattern of an actual signature or of an actual paper document) or independent continuants (for instance the author of the signature, the subject of a record). For example, a *medical record* document is about an organism (i.e. health care consumer or patient).

In the sketch diagrams below, the entities in black are entities from BFO, gray represents reusable entities from OBO Foundry ontologies, and white represents entities new to OntONeo.

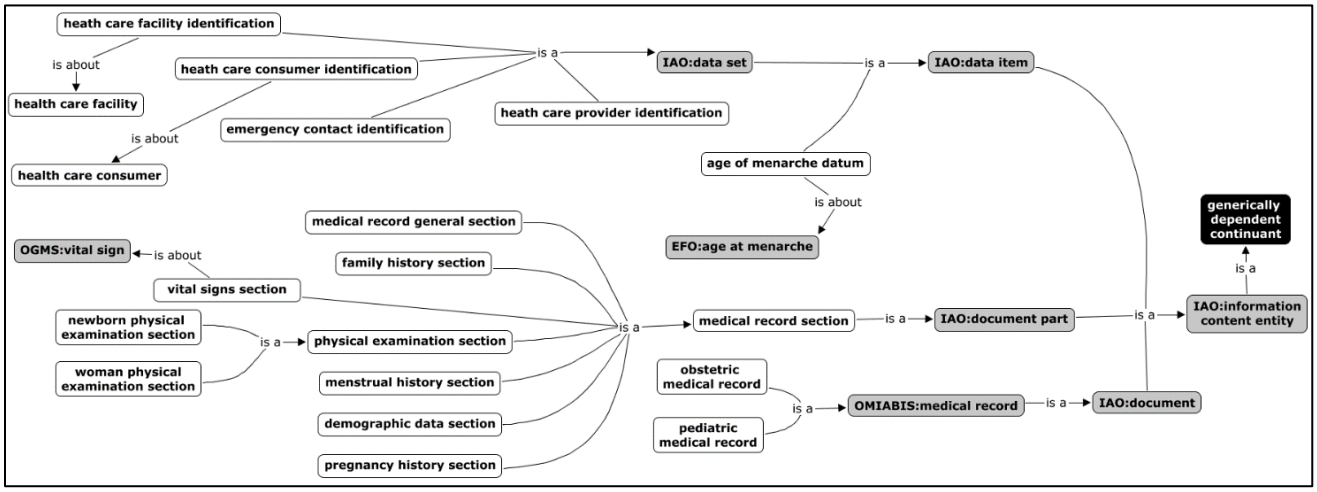

Fig. 4. Electronic health record template representation of OntONeo (By authors)

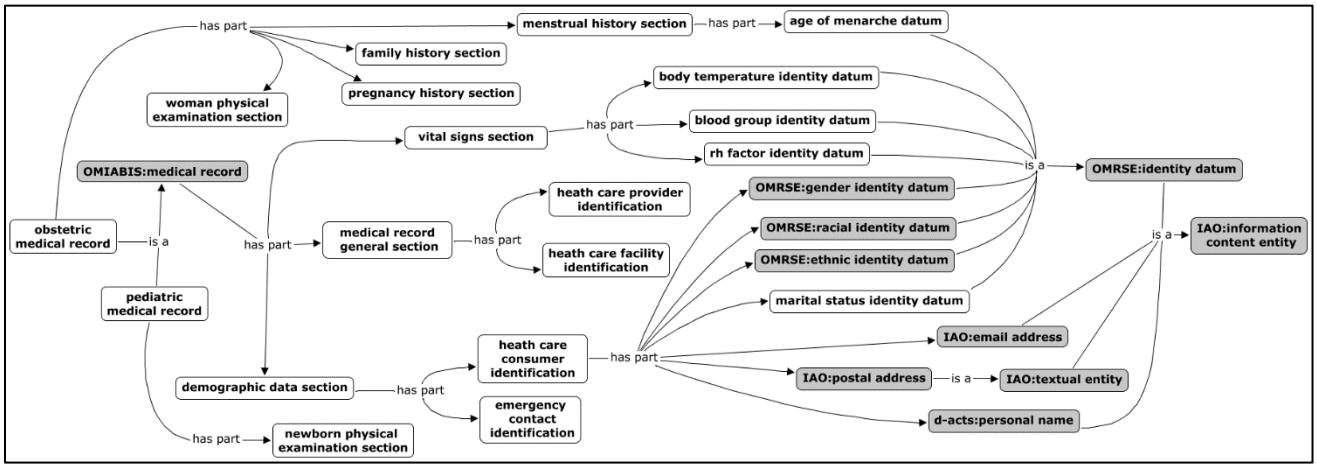

Fig. 5. Electronic health record parts and sections representation of OntONeo (By authors)

C. *The Anatomy and Embryology domain of OntONeo*

We next considered that type of EHR that records information about a woman's physical examination in each medical appointment. A physical pelvic exam is used in obstetrics and gynecology to detect signs of disease in the reproductive organs of the mother. Thus an ontological representation of female anatomy and physiology is required in order to provide a single semantic interpretation of her body structures. For this purpose we have imported terms and relations from the Foundational Model of Anatomy (FMA), 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 imported terms and relations from the Gene Ontology (GO) project. 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 pregnancy. Some data from the prenatal EHR are related to the mother, some to the fetus/embryo, reflecting the fact that anomalies and congenital diseases may become apparent 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), reflecting phenomena described in embryology textbooks such as: i) *blastocyst has disposition to adhere to wall of uterus realizable by the implantation process. If the implantation fails or is interrupted the woman will not be pregnant. So, the wall of uterus has disposition to be adhered by blastocyst;* ii) *after fertilization, the zygote tends to suffer divisions by the process known as cleavage.*

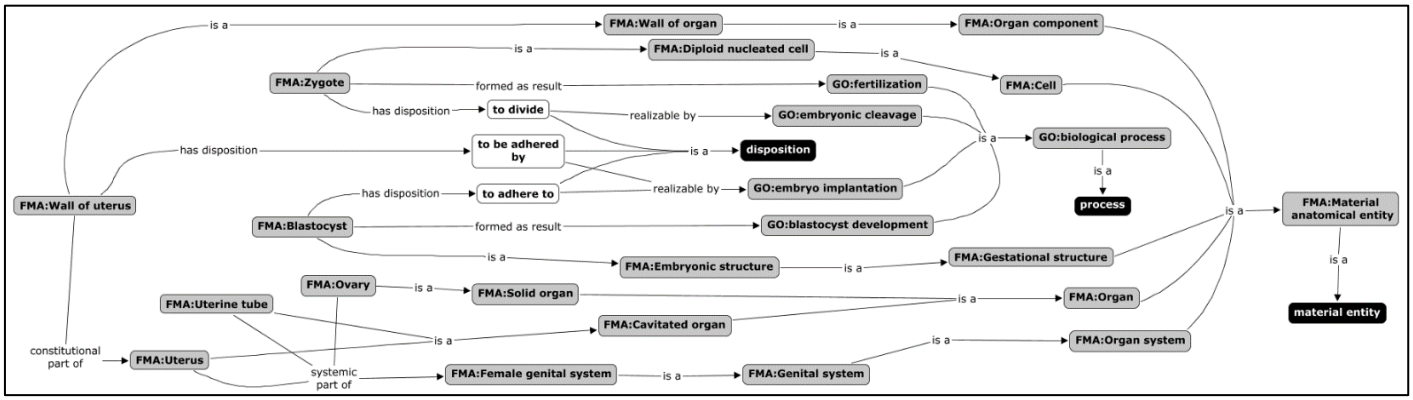

Fig. 6. Extract of the FMA and GO ontology representing the female reproductive system and parts of embryogenesis(By authors)

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 the pregnant woman, and of her child from fetus through the newborn, infant and toddler stages. We also described the methodology that has been used, which is founded in ontological realism.

Since OntoNeo is developed using examples from specific EHRs as starting point, it might be argued that the result will be unsuitable for in other sorts of context. However, given the method used to develop the ontology its content is focused on the representation of general entities, which means that it can be used in a wide range of situations within a domain. 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 of representing a specific context will be organized in a specific level of the ontology. We follow the governance, versioning and update practices adopted by the ontologies in the OBO Foundry.

OntONeo is a project in the early stages of development, and the current version can be found at *http://ontoneo.wordpress.com*. We view ontology development is an interactive process and each successive version will be subjected to validation by representatives of different communities of specialists, including physicians and other healthcare professionals.

Research in OntONeo is we believe justified by the lack of formal representation in the obstetric and neonatal domain. Within this domain, OntONeo provides a needed specialized vocabulary that is projected to include a formal representation more comprehensive than other currently available ontologies such as FMA, GALEN, and UMLS. OntONeo will contribute to the interoperability of information about the different stages of pregnancy, of anatomy, embryology, and other disciplines involved in childbirth and development. In addition, it will facilitate the understanding of how such information can be organized in EHRs for purposes of healthcare.

ACKNOWLEDGMENTS

We thank CAPES and CNPq for financial support to the primary author. This work is also supported in part by the NIH NCATS under CTSA award Number UL1TR001412.

REFERENCES

[1] R. Arp, B. Smith, and A.D. Spear, *Building ontologies with basic formal ontology*, Mit Press, 2015.
[2] C.R.B. Beckmann, F.W. Ling, W.N.P. Herbert, D.W. Laube, and R.P. Smith, *Obstetrics and Gynecology*, Lippincott Williams & Wilkins, 2014.
[3] F.G. Cunningham, K.J. Leveno, S.L. Bloom, C.Y. Spong, J.S. Dashe, B.L. Hoffman, B.M. Casey, and J.S. Sheffield, *Williams Obstetrics*, McGraw-Hill Education, New York, 2014.
[4] A.H.N. DeCherney, Lauren; Laufer, Neri; Roman, Ashley S. , *CURRENT Diagnosis & Treatment: Obstetrics & Gynecology*, 2013.
[5] W.W. Hay, M.J. Levin, R.R. Deterding, and M.J. Abzug, *CURRENT Diagnosis & Treatment: Pediatrics*, McGraw-Hill Education, New York, 2014.
[6] B. Hoffman, J. Schorge, J. Schaffer, L. Halvorson, K. Bradshaw, and F. Cunningham, *Williams Gynecology, 2e*, McGraw Hill Professional, New York, NY, 2012.
[7] M.R. Kamdar, T. Tudorache, and M.A. Musen, A Systematic Analysis of Term Reuse and Term Overlap across Biomedical Ontologies, *Semantic Web journal* (2009).
[8] W.J. Larsen, *Human embriology*, Churchill Livingstone, 2001.
[9] D.A. Morton, K.B. Foreman, and K.H. Albertine, Chapter 14. Female Reproductive System, in: *The Big Picture: Gross Anatomy*, The McGraw-Hill Companies, New York, NY, 2011.
[10] J.M. Rennie, *Rennie and Roberton's Textbook of Neonatology*, Churchill Livingstone Elsevier, Edinburgh, 2012.
[11] . Smith, et al. BFO 2.0 Specification and User's Guide, in, 2015, p. 97.
[12] B. Smith, et al.The OBO Foundry: coordinated evolution of ontologies to support biomedical data integration, *Nature Biotechnology* **25** (2007), 1251-1255.
[13] B. Smith and W. Ceusters, Ontological realism: A methodology for coordinated evolution of scientific ontologies, *Applied ontology* **5** (2010), 139-188.
[14] M.C. Xiang Z, Ruttenberg A, He Y, Ontobee: A Linked Data Server and Browser for Ontology Terms, in, 2011, pp. 279-281.