StrokePEO: Construction of a Clinical Ontology for Physical Examination of Stroke

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***Abstract*—Clinical ontology is a standardized medical knowledge representation model that facilitates the integration and analysis of a large amount of heterogeneous electronic health record (EHR) data. Using ontologies to represent clinical terms can improve data integration to build robust and interoperable medical information systems. To date, there is no ontology existing to represent the medical knowledge for physical examination of stroke, which has inhibited the stroke physicians to make full use of clinical information captured in EHR data to understand stroke patient's health status and plan effective medication and rehabilitation treatment. In this research, we co-design with two stroke clinical specialists a stroke clinical ontology "StrokePEO" using advanced natural language processing and deep learning techniques to extract terms and their relationships from real clinical case records provided by a tertiary hospital in China. We apply the W3C Resource Description Framework (RDF) data model to represent these clinical terms and relationships, and successfully store all case data in a graph database with StrokePEO. Our experiment results suggest that our methods and the output of StrokePEO can be applied in various medical contexts that require extraction of medical knowledge from free text for decision making. These include, but not limited to, physical assessment, drug and rehabilitation treatment outcome evaluation, medication effect analysis, and patient risk prediction.**

***Index Terms*—Electronic health records, clinical ontology, stroke, physical examination, term relationship extraction**

I. INTRODUCTION

In today's age of information and big data, electronic health records (EHR) are being created and collected at an unprecedented rate in medical setting [1]. As the volume of data has grown exponentially, so has the scope and depth of the stored EHR data, which often include patient demographics, diseases, diagnoses, symptoms, medications, treatments, and other health service data. Therefore, observational electronic health record data is a large treasure chest that waits to be explored and utilized. However, there are also many flaws, such as incomplete, inconsistent or incorrect data, and insufficient data details and missing data in EHR, data from different information systems owned by different healthcare providers can be very different. Therefore, EHR data needs to be handled with special caution to ensure their appropriate use to generate high performing algorithms. It is important to ensure safe and ethical use of EHR that will improve patient safety, healthcare quality and efficiency. To avoid ambiguity and ensure data quality, a standardised data representation that can be recognized by both machines and humans is needed. An ideal data representation needs to standardize knowledge in the relevant health domain and can facilitate analysis and integration of heterogeneous data from diverse data sources. This calls for ontology.

An ontology is a systematic representation of domain knowledge, composed of concepts, attributes, and relationships in a hierarchical structure [4]. Concepts are atomic domain terms, connected by the semantic relationships between each other. Attributes are supplements to concepts, improving the coverage and expressiveness of ontology.

In the medical domain, ontology has successfully supported many important application scenarios, including precision medicine [1], [5], clinical decision support systems [4], [6], recommender systems [7], [8]. Using ontology to represent clinical terms can standardise data and enable data integration. Thus, ontologies have been applied to build robust and interoperable medical information systems, meeting the needs of reusing, sharing, and transmitting medical data, and provide statistical aggregation based on various semantic standards [9].

There are many challenges that are yet to be resolved in clinical ontology research. Among them, insufficient disease coverage, *i.e.*, a lack of high-quality annotated databases for certain diseases, *e.g.* stroke, remains the biggest obstacle to the advancement of research and applications of clinical ontologies [9]. Literature [10]–[15] suggests that none of the publicly available stroke ontologies have modeled the information related to physical examination of stroke.

Physical examination is a key step in stroke diagnosis. Through physical examination at the time of admission, doctors can obtain a preliminary understanding of the patient's stroke condition. Based on this, they will further prescribe complex diagnostic tests and treatment plans, *e.g.* medication or rehabilitation treatment. Stroke rehabilitation can reduce or remove the direct pathogenic impact factors for stroke, *e.g.*, ischemia or cerebral hemorrhage. Image tests, such as neuroimaging, are typically aimed at identifying the pathogenic areas but cannot determine whether a patient has recovered from stroke. Only detailed physical examination can provide a comprehensive assessment of the recovery status of various physical functions of the patient.

In this research, we construct a clinical ontology dedicated to stroke physical examination, called "StrokePEO", which focuses on stroke assessment. Different from the existing ontologies for stroke [10], [11], our source data comes from the real clinical case records of the Third Affiliated Hospital of Sun Yat-sen University, Guangzhou, China. The ontology schema, term classes, relationships, *etc*. are co-designed and validated by two stroke specialists. The terms and relationships in the StrokePEO are represented in the Resource Description Framework (RDF) data model [16]. The annotated dataset is used for training, evaluating and testing of the deep learning-based term relationship extraction methods. Experiments show that our approach can effectively mine clinical terms and relationships critical for stroke physical examination. We conduct ontology integration, including term alignment and linkage with other ontologies, to enhance the robustness, consistency and scalability of the StrokePEO in stroke ontology research.

The contributions of this paper are as follows.

- We contribute a clinical ontology StrokePEO dedicated to stroke physical examination. StrokePEO provides an essential component for the construction of large stroke knowledge graph, complements the mainstream stroke ontology research and facilitates the development of AI-based diagnosis and recommendation systems.
- We contribute method and approach for engaging the domain experts - clinical specialists - into co-design the ontology StrokePEO, and various advanced natural language processing (NLP) and deep learning technique to extract the terms and relationships from raw clinical record data to construct the StrokePEO.
- We integrate StrokePEO with globally recognized stroke ontologies, *e.g.* Stroke Ontology (STO) [10] and National Institutes of Health Stroke Scale Ontology (NIHSS) [11].

The rest of the paper is organized as follows. In Section II, we review the related work in stroke ontologies and the technologies for constructing a medical ontology. In Section III we provide detailed description of our approach to construct the StrokePEO. Section IV presents the dataset and the experiment results. Finally, the paper concludes in Section V.

II. Related Work

In this section, we summarize the existing stroke ontologies and the technologies for constructing medical ontologies from natural language. The existing research commonly breaks the task of constructing a medical ontology into four key steps, namely text preprocessing, term extraction, relationship extraction, and ontology integration. Below we summarize the existing technologies for each sub-task.

A. Existing Stroke Ontologies

From the world's largest biomedical ontology portal "BioPortal" [17], we find two public stroke ontologies. The first is Stroke Ontology (STO) [10]. It has 1,712 classes, 69 instances and 35 properties, covering the knowledge of stroke as suggested by expert review. Currently, it is the largest, most comprehensive and most internationally recognized stroke ontology. The other is National Institutes of Health Stroke Scale Ontology (NIHSS) [11], which has been linked to STO as a subclass of the "Scales" class. It focuses on quantitative assessment of stroke severity, including 18 classes, 106 instances and 22 properties.

Some academic research on stroke ontology is available. Townsend *et al.* [12] firstly designed a Neural Motor Recovery Ontology "NeuMORE" to represent the stroke patients' neuromotor function recovery status. Teresa *et al.* [13] built a Stroke Diagnostic Ontology (DStrokeOnto), which contains 456 classes, 77 restrictions and 233 properties. It contributes the formalized medical knowledge for stroke diagnosis. Radhi *et al.* [14] created an ontology to represent knowledge for upper limb stroke rehabilitation in the patient information system. This ontology overcomes the problem of information inconsistency from various assessments. Soonhyun *et al.* [15] proposed a stroke medical ontology based on brain anatomies, lesions and stroke-related disease, aiming to assist the AI-based stroke prediction system.

The literature suggests a lack of effort to construct a comprehensive stroke physical examination ontology. This motivates our research to focus on developing a specific StrokePEO ontology to represent stroke physical examination as a complement to the Stroke ontology research field.

B. Text Preprocessing

The first step to construct a domain ontology from text is data preprocessing. This can be achieved by applying the common method of natural language processing (NLP) [9] for text parsing. Several successful NLP tools provide mature functions to accomplish these tasks.

The Natural Language Toolkit (NLTK) [18] is an open source platform that provides general text preprocessing capabilities such as sentence segmentation, word tokenization, stemming, part-of-speech (POS) tagging, parsing, and semantic reasoning. FreeLing [19] is another widely used library that supports high-level NLP parsing functions such as word sense disambiguation and semantic role labelling.

Unlike English, Chinese words usually consist of more than two Chinese characters, so special word tokenization methods are required. Jieba [20] is a widely recognized Chinese word tokenization module that provides functions such as word segmentation and part-of-speech tagging. It supports customized dictionaries which is quite helpful for specific domain text processing. HanLP [21] is a multilingual NLP library that is primarily designed for Chinese text processing. It offers deep parsing functions including semantic dependency parsing, constituency parsing, semantic role labeling and abstract meaning representation (AMR) parsing.

C. Term and Relationship Extraction

The basic unit of an ontology is often represented in the form of triples, where two associated terms (classes) are described as $<$ term 1, relationship, term 2 $>$. The main task during the construction of a medical ontology is to extract terms and relationships from unstructured data.

In the early years, people used manual extraction to collect relevant terms through experts according to certain rules. However, due to the high cost of manual extraction, automatic term extraction has become a research hotspot, known as "named entity recognition (NER)". Recently, deep learning based NER approaches are widely used and have achieved high accuracy in mining the terms from raw text. A mainstream deep learning model for the NER tasks of medical information is BiLSTM-CRF [22].

The extracted entity relationships can be classified into two categories, *i.e.*, the hierarchical relationships and the non-hierarchical relationships [9]. The hierarchical relationships are always between the same entity type, mainly meaning "is-a" or "part-of" relation. The non-hierarchical relationships often fall between different types of entities, indicating Entity 1 as "has-attributes" and Entity 2 as "has-properties". Since the types of entities in the medical field are relatively limited (mostly diseases, symptoms, treatments, medicines, *etc.*), the relationship types to be extracted between two entities are usually predefined, and then the extraction task becomes a classification problem. Thus, deep learning based classification models are suitable for relationship extraction.

D. Ontology Integration

Ontology integration is the process of organizing the high-level knowledge obtained from different sources and involves data integration, disambiguation, reasoning verification, updating and other steps under the same framework specification.

Ontology integration can be subdivided into intra-class alignment and ontology linkage with other ontologies. Intra-class alignment determines whether classes in multi-source heterogeneous data point refer to the same object in the real world by considering instances and their attribute similarity. Ontology linkage starts from "ontology matching", *i.e.*, matches the semantic similarity of classes in one ontology with those in the other ontologies [23]. As an ontology grows in size and becomes more complex in structure, the classes, attributes, entities and their interrelationships are also taken into consideration. In the medical field, Dieng-Kuntz *et al.* [24] converted medical databases into medical ontology, and then used semi-automatic language tools for semantic extraction from other text corpora, extended and completed ontology building manually, using heuristic rules.

Although there are some meaningful attempts (*e.g.* [25]), it still requires a lot of manual processing to integrate ontologies in the medical field; therefore, further research is required to develop effective technology for efficient ontology integration in this setting.

III. Methodology

In this section, we illustrate in detail the key steps we take in constructing the StrokePEO. Following the seven-step approach of ontology construction recommended by a Stanford research group [27], we use Protégé [26] to build the StrokePEO *i.e.*, to determine scope, consider reuse, enumerate terms, define classes, define properties, define constraints and create instances. We apply the appropriate technologies to accomplish the task at each step. *i.e.*, text preprocessing, ontology schema definition, schema constrained term relationship extraction, term alignment and ontology integration.

A. Text Preprocessing

We apply a series of NLP techniques to preprocess the raw clinical text data. These include unifying format, removing the staleness, sentence segmentation and word tokenization with POS tagging.

Unlike the common article sentence, the structure of medical record text usually does not have a complete and standard syntactic structure, but lists multiple subject-predicate phrases in a sentence. This inhibits the effective application of the semantic dependency parsing method to process these clinical records. Therefore, instead of the common practice for English in using "period" as the delimiter, we use "comma" or "semicolon" as the delimiter to divide sentences. The resultant segmented sentences are short in length, but still contain single or multiple terms to form the triples of "subject, predicate and object" (SPO).

To tokenize Chinese words, we adopt the Jieba [20] package with self-defined dictionaries. Two dictionaries are imported to help enhance the accuracy of word tokenization. The first is a dictionary named "THUOCL-medical" [28] produced by Tshinghua University, with medical words and their frequency annotated. The other is annotated by us and reviewed by clinical experts, to handle special terms with POS tagging suitable for stroke physical examination records.

B. Ontology Schema Definition

Through in-depth analysis of the structure and concepts of stroke terms, we develop a schematic ontology representation

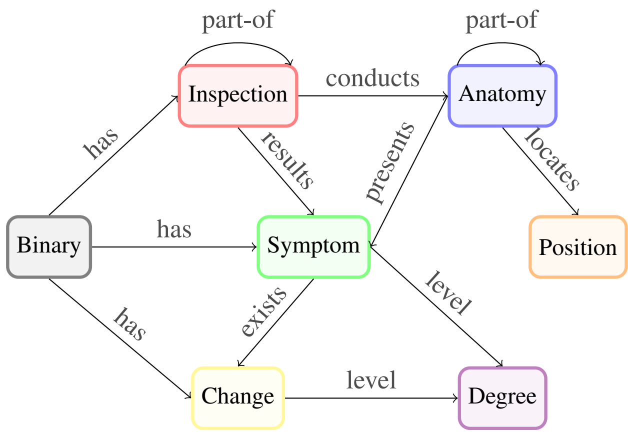

Fig. 1. The schematic representation of our StrokePEO.

and represent the terms using the Resource Description Framework (RDF) data model [16]. Its atomic data format is called RDF triple, which consists of three entities in the form of "subject, predicate, object" to show the semantic statement of "term 1 has relationship with term 2".

Specifically, in our StrokePEO, triples are composed of fine-grained terms and relationships, to express the knowledge as precisely and as accurately as possible. To construct our StrokePEO, we define seven classes of terms, *i.e.*, Anatomy, Inspection, Symptom, Position, Binary, Change and Degree. Twelve relationships are defined among the term classes. The detailed ontology representation schema is shown in Fig. 1.

C. Extraction of Constrained Term Relationships

To construct the StrokePEO, we mine useful terms and their relationships from a large amount of raw clinical EHR text data, using two hot research techniques in the field of text mining, *i.e.*, Named Entity Recognition (NER) and Relation Extraction (RE). With the continuous development of machine learning and deep learning to reach maturity level, many mature NER and RE algorithms are now publicly available.

We co-define with the two stroke specialists in our team the ontological representation of each concept for stroke physical examination. Due to that a sentence usually contains multiple RDF triples, we apply multi-relational classification for model training and prediction. We first classify the relationship constraint. Then we put the same term into different classes in accordance with the relational constraint in a relevant sentence to resolve the ambiguity of semantic relationships expressed by the same term in different context.

We apply the TensorFlow-based Entity and Relation Extraction model [29], a schema-based pipeline entity-relation extraction model. This model has achieved excellent performance comparable to the SOTA model in the "2019 Language and Intelligence Challenge" [30] and has been widely recognized by high popularity stars in GitHub. Different from other models that first perform NER and then extract relationships, this model first trains a multi-class model to predict all possible relationships in a sentence, and then combines the prediction results and applies sequence labeling algorithm to capture the terms within the relationship triples. To improve the model accuracy, instead of using the original model, we adopt a more advanced Chinese embedding named Chinese Pre-trained BERT with Whole Word Masking (ROBERTA_wwm_large_ext) [31]. It significantly outperforms the standard BERT embedding for our entity relationship extraction task. We discover as broadly and comprehensively terms as possible, resulting in many terms with similar or even the same semantic meaning.

D. Term Alignment

The purpose of this step is to unify the synonymous terms into one standardised term to ensure atomicity of the concept classes in the constructed StrokePEO. To improve accuracy, we combine the open source Chinese synonym tool "Synonyms" [33] with the word2vec model to process the clinical data. As both models have fully learned the context information embedded in the adjacent and distant words during training, they can infer, to a large extent, the original meaning of words and their relationships.

We use these two models to obtain the ten most similar terms for each extracted term, respectively. After filtering out terms with different term classes, the remaining terms are marked as synonyms of the standardised term. Finally, clinicial experts are called upon to validate accuracy of the machine-generated thesaurus.

E. Ontology Integration

We set up the scope of the StrokePEO ontology as the diagnostic physical assessment of stroke patients in clinical setting to address this gap in Chinese stroke ontology. Based on the systematic review of existing ontologies and previous research work, our StrokePEO can be recognized as a complement to the research field of stroke ontology, which can be directly integrated into the current most authoritative Stroke ontology (STO) [10], under the "Stroke-Diagnosis-Evaluation of stroke-Physical Examination" class.

The clinical experts in our team expect the StrokePEO to have the ability to be integrated with other stroke ontologies to meet the needs of real-world applications and research. For example, when there is a clinical requirement to assess the severity of a patient's stroke condition, the clinicians usually use the international standard NIHSS (National Institutes of Health Stroke Scale) [11]. In order to align with the NIHSS international standard, we integrate the two ontologies, StrokePEO and the NIHSS ontology.

As mentioned in the literature section, there is no fully automatic ontology fusion algorithm in medical domain; therefore, manual fusion has to be conducted in this project. The NIHSS is composed of 11 classes, including consciousness level, eye movement, motor arm and leg, speech, *etc*. It has less classes than our StrokePEO. Thus, with the guidance and quality control of the clinical experts, we manually match the classes of StrokePEO with those in the NIHSS ontology. This integration mainly consists of two tasks, one is to match the "Inspection"

TABLE I
THE STATISTICS OF THE ANNOTATED TERMS AND RELATIONSHIPS.

Subject Term Type	Relationship	Object Term Type	Count
Inspection	results	Symptom	85,893
Inspection	conducts	Anatomy	11,968
Anatomy	presents	Symptom	15,888
Anatomy	locates	Position	24,096
Binary	has	Symptom	10,393
Binary	has	Inspection	26,657
Binary	has	Change	386
Symptom	exists	Change	3,219
Symptom	level	Degree	7,187
Change	level	Degree	893

class in StrokePEO with the classes in NIHSS, and the other is to match the "Symptom" class in StrokePEO with the value set in NIHSS. After integration, the resulted StrokePEO will afford people even without professional training to acquire a quantitative assessment score of a patient's stroke condition by observing the patient's clinical manifestations.

IV. Experiments

To efficiently extract the terms and relationships from the large amount of text data, we apply the advanced deep learning-based techniques to automatically recognize the terms and classify their relationships in each sentence. As supervised learning requires a batch of data annotated with correct labels to train the algorithm, we first introduce our approach to acquire the annotated dataset, and then report the setting and performance of the two algorithms used for term extraction and relationship classification. Finally, we evaluate the quality of the constructed StrokePEO.

A. Dataset

The study dataset is collected and labeled from the clinical case records of physical examination results for stroke patients from the Third Affiliated Hospital of Sun Yat-sen University, China. The definition of ontology schema, including the classes of terms and relationships are all guided and approved by two stroke experts. The dataset contains 89,351 annotated samples, and are randomly split into training set, evaluation set and test set at a ratio of 4:1:1. Each annotated sample is composed of the raw text and lists of SPO ("subject, predicate, object") triples to show the terms and relationships. As a pipeline model, both term extraction and relationship classification algorithms are trained on the same dataset. Therefore, we add the term type into the SPO triples, indicating the subject type and object type. For example, the sentence "右侧肢体肌力5级" is extracted into three RDF triples, *i.e.*, (subject: "肢体", subject_type: "Anatomy", predicate: "locates", object: "右侧", object_type: "Position"), (subject: "肢体", subject_type: "Anatomy", predicate: "conducts", object: "肌力", object_type: "Inspection"), and (subject: "肌力", subject_type: "Inspection", predicate: " results", object: "5级", object_type: "Symptom"). Table I shows the statistics of the annotated terms and relationships in the dataset.

TABLE II
RELATIONSHIP CLASSIFICATION RESULTS

Output Results	Count	Total Numbers	Accuracy (%)
Correct	8125	8228	98.7482
Superset	52	8228	0.6320
Subset	13	8228	0.1580

B. Relationship Classification Results

We conduct a multi-class classification model to predict the possible relationships in a sentence. The input of this model is raw texts from training samples, which are first tokenized and embedded by a BERT layer. We have found that, replacing the BERT embedding with the ROBERTA embedding [31] has led to much better performance. The embedding sequences are then passed to the multi-class classifier, which outputs the predicted set of possible relationships in the text.

To evaluate the accuracy of relationship classification, we compare the predicted relationships with the golden set. If the predicted relationship matches the golden set, it is marked as "Correct". If the output set is equal to or greater than the golden set, it is marked as a "Superset". Finally, a "Subset" result indicates that the output set contains only a part of the correct relationships in the golden set. Table II shows the results. As it can be seen that, the classification algorithm can effectively identify all possible relations in sentences with more than 98% accuracy. In a few cases of inaccurate predictions, partially correct relationships can also be identified with a small number of redundant or missing predictions.

C. Term Extraction Results

We run a sequence labelling model to extract the terms from the input text, *i.e.*, the relationship classification results. First, the model converts a training sample with multiple labels into multiple samples, so that the mapping between the original text and label in each sample is one-to-one relationship. Then the predicted subject and object terms are constrained by the classified relationships to suit their types.

To evaluate the performance of the term extraction algorithm, we calculate the accuracy of the predicated SPO triples. A "correct SPO" indicates that the predicated SPO triples are exactly the same as the golden set regarding the terms, types and relationships. We also report the number of predicted SPO triples and the number of SPO triples in the golden set. Finally, we evaluate the performance of the term extraction model using the common metrics, including Precision (P), Recall (R) and F1-score ($F1$), which are defined by:

$$P = TP/(TP + FP)$$
$$R = TP/(TP + FN)$$
$$F1 = (2 \times P \times R)/(P + R),$$

where TP is the number of true positives, FP is the number of false positives, and FN is the number of false negatives.

TABLE III
TERM EXTRACTION RESULTS

Correct SPO num	14,114	Submitted SPO num	14,567
Golden set SPO num	15,086		
Task	Precision (%)	Recall (%)	F1-score (%)
Term extraction	96.89	93.56	95.19

The detailed results of term extraction are shown in Table III. For all of the evaluation metrics, the larger the values, the better the algorithm performs.

V. CONCLUSION AND FUTURE WORK

For the first time, this study has developed and validated a clinical ontology "StrokePEO" for physical examination of stroke using real clinical case record data. We have applied multiple NLP techniques to preprocess the raw text records and have adopted advanced deep learning techniques to successfully extract the terms and relationships pertaining to physical examination of stroke. Our approach and the resulted StrokePEO ontology provide the useful machine learning model and base for the further development of diverse clinical decision support systems that generate knowledge from rich clinical text. These include, but not limited to, physical assessment, drug and rehabilitation treatment outcome evaluation, medication effect analysis, and patient risk prediction.

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