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# Research on the Application of Semantic Network in Disease Diagnosis Prompts Based on Medical Corpus

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**ABSTRACT-** Portion of the causes of medical errors in outpatient clinics are incorrect treatment resulting from misdiagnosis. Misdiagnosis between diseases is often caused by similar and indistinguishable symptoms. Currently, disease knowledge and related symptom words that are prone to misdiagnosis are scattered in various medical literature or open online databases. Therefore, it is possible to merge symptom words of related diseases, build an ontology based on the semantic relationship between symptoms, and associate the association between diseases and symptoms. This project finally established the "Disease Symptoms" Semantic Network (DSSN). Using DSSN as a basic data set can serve as prompts for diseases that are easily misdiagnosed, assisting doctors in accurately diagnosing diseases. This plays a significant role in reducing the misdiagnosis rate.

**KEYWORDS-** Artificial Intelligence; Semantic Network; Ontology; Misdiagnosis.

## I. INTRODUCTION

In hospital practice, approximately 10% of adverse events are caused by misdiagnosis[1]. A minor misdiagnosis may lead to a prolonged recovery period for the patient, while in severe cases, incorrect therapeutic intervention may bring life-threatening risks and become a major cause of medical accidents and medical disputes. In clinical diagnosis, the complexity of the disease and the dynamic changes in the disease often lead to discrepancies between the doctor's initial diagnosis and the actual disease. With the development of technology that combines images and data[2], advanced medical imaging technologies such as endoscopy, magnetic resonance imaging (MRI) and X-ray computed tomography (CT) play an important role in accurate medical diagnosis[3], and electronic medical records provide a rich source of diagnostic information[4], and even deep learning algorithms are being attempted for early detection of diabetic retinopathy (DR)[5][6][7] and also and machine learning can also play an important role in the field of stroke prediction[8]. However, preliminary data show that the overall misdiagnosis rate of clinical diseases still continues to be in the range of 10% to 15%[9].The main determinant of misdiagnosis is the combination of similar symptoms. Given that symptoms constitute the cornerstone of clinical diagnosis, diseases susceptible to misdiagnosis often share commonalities in their symptomatology. A comprehensive repository of knowledge pertaining to interconnected diseases and their associated symptoms is dispersed across diverse literary works, reference materials, and open-access online databases. Consequently, the amalgamation of pertinent knowledge sources is imperative to construct an exhaustive "disease-symptoms" knowledge system. The provision of guidelines for potential misdiagnoses assumes profound significance in the enhancement of clinical diagnostic precision.With the burgeoning informatization in medicine and the proliferation of Internet knowledge bases, great progress has been made in the field of biomedical knowledge representation. Firstly, the structured representation and exploration of biomedical knowledge has been significantly promoted. As a key method for structured knowledge representation, ontology proposes a clear formal specification of shared conceptual models, with the main goal of realizing knowledge sharing and reuse[10]. Significantly, ontologies have been established in key domains, including gene ontology[11], disease ontology[12], and human phenotype ontology[13].Secondly, the representation and discovery of unstructured medical knowledge have witnessed significant progress in recent years. A large amount of biomedical information and knowledge is now disseminated on the Internet in semi-structured and unstructured formats, including academic papers, medical textbooks, case reports, and more. Researchers integrate disease ontology and symptom ontology by constructing the relationship between disease and symptoms[14]. Some progress has also been made in solving the problem of data resource linking[15].However, challenges remain, particularly in the realm of providing cues for misdiagnosis. Firstly, mainly anatomy-centric and lack semantic interconnections between concepts, thus failing to reflect similarities between symptoms. Secondly, the relationship between symptoms and diseases remains stored in unstructured text, without extraction for structured representation. Furthermore, the relationship between symptoms and diseases is not strictly one-to-one, incorporating additional conditions such as common and rare differences. Most importantly, existing medical knowledge representation systems do not encompass information relevant to differential diagnosis between diseases—an area prone to misdiagnosis. Differential diagnosis knowledge typically resides in documents like diagnostic and treatment manuals, yet to be expressed in a structured format within computer systems. This limitation prevents the direct application of knowledge about diseases that are prone to misdiagnosis.

## II. RELATED WORK

The Semantic Web[16] is a concept proposed by Tim Berners-Lee, the founder of the World Wide Web, in 2001, which refers to a network of linked data. The Semantic Web is an expansion and extension of the World Wide Web, which enables semantic interconnection of data on the Internet so that its semantics can be automatically understood by machines. The Semantic Network inherits the knowledge expression capabilities of its predecessor, Semantic Network, and improves semantic interoperability and reasoning capabilities. Since the Semantic Web was proposed, many applications such as FOAF and TrueKnowledge have rapidly emerged in the scientific and commercial fields. According to Google's Semantic Web document search engine Swoogle, Swoogle has compiled millions of indexes for Semantic Web documents in the form of RDF, RDFS and OWL. Not only scientific research organizations such as biomedical sciences and earth sciences are actively involved in the development of the Semantic Web, and industry leaders such as Oracle, Vodafone, Google, Facebook, Amazon.com, Adobe and Yahoo have also invested heavily in smart network technology.The relationship between the Semantic Web and Ontology is very close. It can be said that Ontology is the core of the Semantic Web. The Semantic Web mainly provides a mechanism for semantic representation of information, with the ultimate goal of realizing information sharing and semantic interoperability. Therefore, the Semantic Web needs to be guided by ontology to manage the sharing and reuse of knowledge. Ontology is a standard conceptual system in the Semantic Web that is easy for computers to W3C has defined the OWL layer standard in the Semantic Web as ontology language. Ontology can provide the main technology for interconnecting resources in the Semantic Web and is the key to realizing the Semantic Web.

In summary, this article constructs a disease-symptom semantic network, which includes disease ontology DO, symptom ontology and the misdiagnosis-prone relationships between diseases, and evaluates this semantic network through an example in medical diagnosis. It serves as a reminder for easy misdiagnosis.

## III. CONSTRUCTION OF SYMPTOM ONTOLOGY

Establishing a Disease-Symptom Semantic Network (DSSN) involves integrating disease ontology, symptom ontology, and differential diagnosis knowledge. The process for constructing a symptom ontology unfolds through the following steps:

- ***Corpus Acquisition***: Acquire a symptom text corpus from authoritative medical domain knowledge bases such as SYMP, Wikipedia, Mayo Clinic, and Cleveland Clinic.
- ***Symptom Vocabulary Identification***: Identify words describing symptoms within this corpus to compile an extensive set of potential symptom vocabulary candidates.
- ***Semantic Similarity Computation***: Calculate semantic similarity metrics between symptom words in the candidate set to quantify their conceptual relatedness.
- ***Symptom Ontology Formation:*** Merge symptom words with significant semantic similarity to establish a refined symptom ontology, thus encapsulating a comprehensive representation of symptomatology within the semantic network.

### A. Corpus

The existing Internet landscape boasts a robust and openly accessible repository of comprehensive human disease knowledge, functioning as an unparalleled "open source" that is both efficient and user-friendly. This study utilizes SYMP, Wikipedia, Cleveland Clinic, and Mayo Clinic as principal corpora. SYMP, characterized as an anatomy-centric symptom ontology, encompasses a spectrum of 936 symptoms. Wikipedia serves as an expansive repository, housing encyclopedic knowledge pages for all diseases based on ICD-10. Each disease entry is enriched with comprehensive descriptions of both common and rare symptoms. The Cleveland Clinic and Mayo Clinic, preeminent medical institutions in the United States, amalgamate medical services, academic research, and teaching. Their respective websites have knowledge bases containing disease-related diagnosis and treatment information, combined with descriptions of symptoms of various diseases. In this study, the symptom vocabulary from SYMP is utilized, coupled with web crawler technology, to extract text containing disease symptom descriptions from Wikipedia, Cleveland Clinic, and Mayo Clinic. This aggregated corpus is the foundation for symptom identification.

### B. Symptom Words Identification

Several contemporary biomedical annotation tools, including NCBO annotator[17] and MetaMap[18], have demonstrated commendable accuracy in annotating terms associated with diseases and symptoms. Nonetheless, within the chosen corpus for this study, a large number of related terms differ from existing ontologies related to diseases and symptoms, and some are not even discretely composed of separate words. Consequently, Therefore, traditional annotation tools cannot fully recognize these terms. In pursuit of a comprehensive and precise extraction of all symptom vocabulary, this paper actively divides the text corpus into structured components (ontology) and unstructured components (text). There are two processes for identifying symptom words:

1. For the structured corpus, it is directly incorporated into the symptom vocabulary candidate set;

2.For the unstructured corpus, the text undergoes processing through the Porter Stemmer algorithm[19] to isolate words with the same root as the symptom word candidate set.

Subsequently, the English word similarity calculation algorithm[20], based on WordNet[21], is used for feature extraction and value computation, facilitating the identification of words in the text that have high semantic similarity with the candidate set. This multistep approach ultimately identifies all symptom words in the text. Figure 1 illustrates the symptom words extracted from the symptom description segment of Wikipedia for Pneumonia. At this juncture, the paper has accumulated an expanded symptom vocabulary candidate set, totaling 2171.

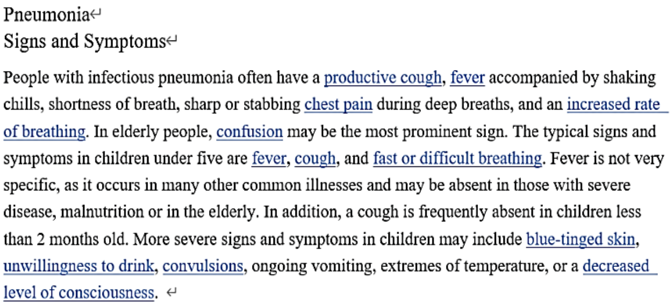
Pneumonia
Signs and Symptoms
People with infectious pneumonia often have a productive cough, fever accompanied by shaking chills, shortness of breath, sharp or stabbing chest pain during deep breaths, and an increased rate of breathing. In elderly people, confusion may be the most prominent sign. The typical signs and symptoms in children under five are fever, cough, and fast or difficult breathing. Fever is not very specific, as it occurs in many other common illnesses and may be absent in those with severe disease, malnutrition or in the elderly. In addition, a cough is frequently absent in children less than 2 months old. More severe signs and symptoms in children may include blue-tinged skin, unwillingness to drink, convulsions, ongoing vomiting, extremes of temperature, or a decreased level of consciousness.

Figure 1: Symptom word recognition results

### C. *Synonymous Symptom Words Merging*

Divergent terminologies may be employed across various corpora to delineate identical phenomena. For instance, the symptom "paralysis" may be expressed as "numbness" or "palsy". To establish a cohesive and standardized symptom description, this paper consolidates synonymous symptom vocabulary. This amalgamation transpires through a two-step process: ① Within the symptom corpus, distinct descriptions of the same symptom often feature annotations within brackets, as exemplified by "difficult swallowing (dysphagia)"; hence, the initial step involves identifying all such synonyms within the text. ② Subsequently, utilizing the English vocabulary similarity calculation algorithm based on WordNet[21], the paper computes similarity values between words, employing this metric to pinpoint symptom description words possessing identical or akin semantics in the lexicon. These identified synonymous symptoms are then merged to foster a harmonized symptom vocabulary.

### D. *Symptom Ontology Construction*

The existing symptom ontology, SYMP, is based on anatomical framework, categorizing symptoms into distinct domains such as abdominal, cardiovascular, digestive, nervous, and urinary systems. These symptoms are intricately closely with anatomical nouns, such as terms like "abdominal cramps" and "chest congestion". In contrast, the impending construction of the symptom ontology in this study adopts a hierarchical framework founded on the semantic relationships between symptom words.In the previous section of this paper, synonymous symptoms were merged to produce aliases (Xref) for symptom words. These synonymous symptoms are represented as nodes within the newly constructed symptom ontology. A semantic "is-a" relationship is then established between words describing similar symptoms based on their roots, other lexical features, and semantic category features. For instance, "Swelling" is documented as "bloating (swelling)" in the symptom ontology, with subcategories (synonyms) such as puffiness, edema, abdominal swelling, postprandial bloating, and etc. Figure 2 shows the hierarchical structure of concepts in the symptom ontology

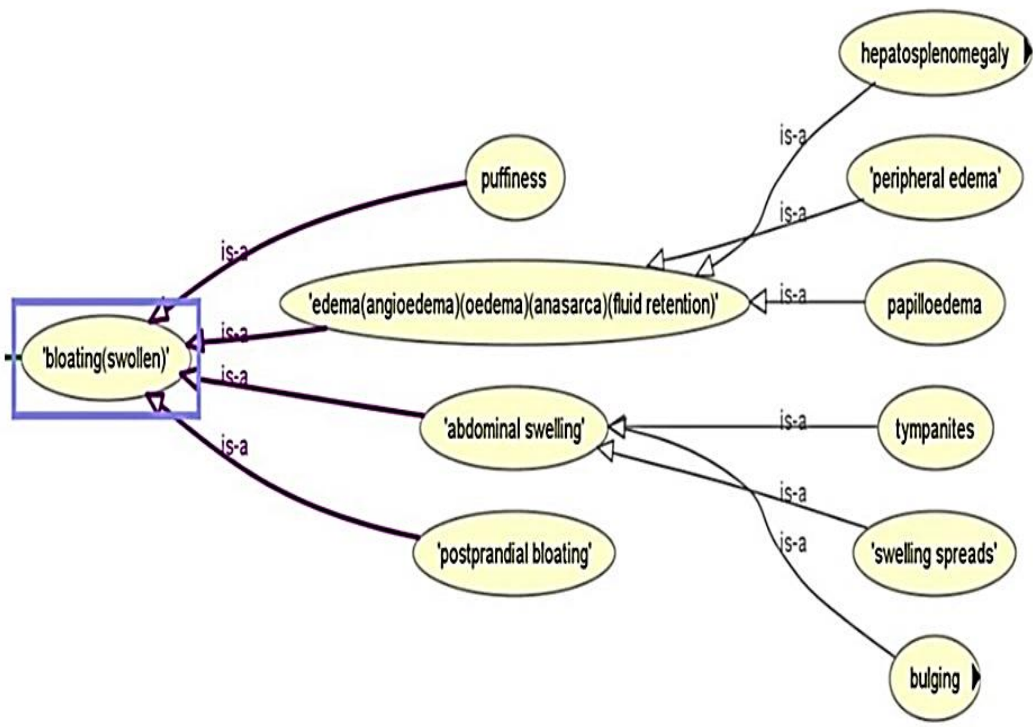


Figure 2: Hierarchical structure among concepts in symptom ontology

## IV. CONSTRUCTION OF DISEASE-SYMPTOM SEMANTIC NETWORK

### A. *Disease-symptom Relationship Extraction*

The manifestation of symptoms is often linked to specific diseases, making the disease-symptom relationship a crucial reference point in clinical diagnosis. The extraction method for the disease-symptom relationship is described as follows:

- Utilize crawler technology to acquire text describing symptoms of a particular disease from Wikipedia, Cleveland Clinic, and Mayo Clinic corpora.
- Employ the expanded symptom vocabulary candidate set to identify all symptom words within the obtained text.
- Establish the "has symptom" relationship between symptoms and diseases by associating the extracted symptom words with specific diseases. Figure 1 illustrates the extraction of symptom words from the symptom description text of Encephalitis in Wikipedia, establishing a linkage between encephalitis and its symptoms.

The relationship between diseases and symptoms is not strictly one-to-one. Some diseases may present common symptoms alongside rare ones. Beyond establishing the basic "has symptom" relationship, this paper goes further to extract and incorporate the frequency of symptoms into the disease-symptom semantic network. The method needs to obtain words describing symptom frequency from the corpus text, with over 10 such words identified, as presented in Table 1. These frequency-describing words are then located in the corpus text, and symptom words in the same sentence are extracted based on the expanded symptom word candidate set. This process determines the frequency of symptom occurrence in a particular disease, facilitating the establishment of a nuanced relationship between the disease and symptoms.In total, 379 frequency words were extracted from Wikipedia, Mayo Clinic, and Cleveland Clinic, as outlined in Table 1. Collaborating with clinicians, symptoms are categorized based on frequency words into three groups: "common symptoms" for those described with terms like "most," "most common," "common," "usually," "often," and "≥10%"; "general symptoms" for those described with terms like "sometimes," "less commonly," "less often," and "3%~10%"; and "rare symptoms" for those described with terms like "occasionally," "rare," and "≤3%".

Table 1: Results of frequency extraction

| Symptom Frequency | Counts | Total |
|---|---|---|
| most | 35 | 337 |
| most common | 33 | |
| common | 56 | |
| usually | 55 | |
| often | 120 | |
| ≥10% | 38 | |
| sometimes | 20 | 29 |
| less commonly | 3 | |
| less often | 3 | |
| 3%-10% | 3 | |
| occasionally | 8 | 13 |
| rare | 3 | |
| ≤3% | 2 | |
| Total | 379 | 379 |

### B. *Acquisition and Establishment of Easily Misdiagnosed Relationships Between Diseases*

The knowledge that diseases are prone to misdiagnosis (differential diagnosis) forms the key to constructing a disease-symptom semantic network. Misdiagnoses of diseases have been known to lead to adverse consequences, particularly in instances where non-serious conditions escalate due to delayed treatment opportunities. Given the key role of misdiagnosis knowledge in disease diagnostics, this paper incorporates such information into the disease-symptom semantic network.In order to outline the relationships between diseases prone to misdiagnosis, the classic diagnostic and treatment manual "Current Essentials of Medicine"[22] is selected as a knowledge source. This manual comprehensively outlines "diagnostic key points" and "differential diagnosis" information for 561 common diseases. Using this resource, the paper establishes relationships indicative of easy misdiagnosis between diseases. Subsequently, these relationships and the associated differential diagnosis information are integrated into the disease-symptom semantic network. In this semantic network, diseases are interlinked based on their susceptibility to misdiagnosis, creating a knowledge base for misdiagnosis prompts in medical diagnosis.

Figure 3 illustrates this process using migraine as an illustration. Commencing with the migraine page in the selected book, the "Differential Diagnosis" text is employed to identify diseases prone to misdiagnosis, such as cluster headache, meningitis, subarachnoid hemorrhage, giant cell arteritis, among others. The descriptive details of their differential diagnosis are then derived from the "Essentials of Diagnosis" text. Ultimately, this information on easy-to-misdiagnosis relationships and differential diagnoses is seamlessly incorporated into the disease-symptom semantic network.

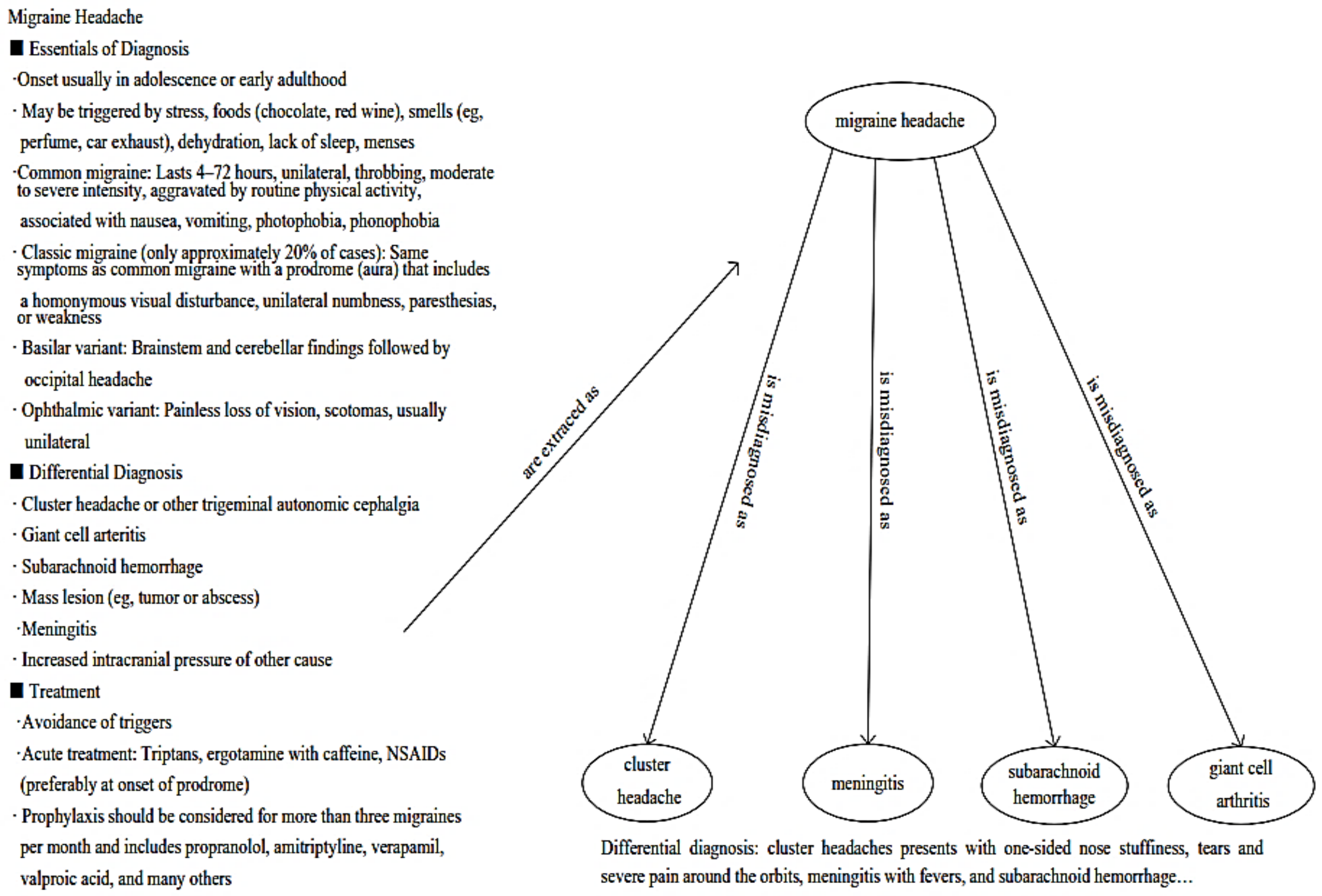


Figure 3: Acquisition and construction of misdiagnosed diseases and differential diagnosis of migraine headache

### *C. Establishment of Disease-Symptom Semantic Network*

This paper utilizes Protégé[23] to construct a "Disease-Symptom Semantic Network (DSSN)". Initially, the foundational concepts within the disease-symptom semantic network are acquired. The disease vocabulary from the disease ontology and the symptom vocabulary from the symptom ontology serve as the fundamental concepts. These concepts are input into Protégé, where they are instantiated as basic classes. Subsequently, the relationships between these concepts are defined, with the establishment of relationships primarily accomplished through object attributes.OWL category definitions specify categories as disease and symptom vocabularies. Object attributes, which signify relationships between classes, are a key focus of this article. Between diseases and symptoms, five distinct object attributes are defined: "has symptom(A)", "has symptom: common (B)", "has symptom: general (C)", "has symptom: rare(D)", and "is misdiagnosed as (E)". Each object attribute possesses a specific scope and domain, as outlined in Table 2.

Table 2: Description of Object Properties

| Object Properties | Domain and Range |
|---|---|
| has symptom | Domain:"Disease" and Range:"Symptom" |
| has symptom:common | Domain:"Disease" and Range:"Symptom" |
| has symptom: general | Domain:"Disease" and Range:"Symptom" |
| has symptom: rare | Domain:"Disease" and Range:"Symptom" |
| is misdiagnosed as | Domain:"Disease" and Range:"Disease" |

Following the above OWL definitions, this paper successfully constructs a disease-symptom semantic network grounded in semantic concepts and their relationships. This semantic network contains 1001 disease words, 2380 symptom words, and a total of 3381 concepts. The network encapsulates relationships such as disease-symptom associations, easily misdiagnosed connections between diseases, and differential diagnosis knowledge. A segment of the established DSSN network is depicted in Figure 4. This semantic network vividly portrays relationships between diseases susceptible to misdiagnosis, along with the similarities and disparities in symptoms among these diseases. Figure 5 illustrates the easily misdiagnosed diseases migraine headache and cluster headache, elucidating the distinctions in symptoms between the two.

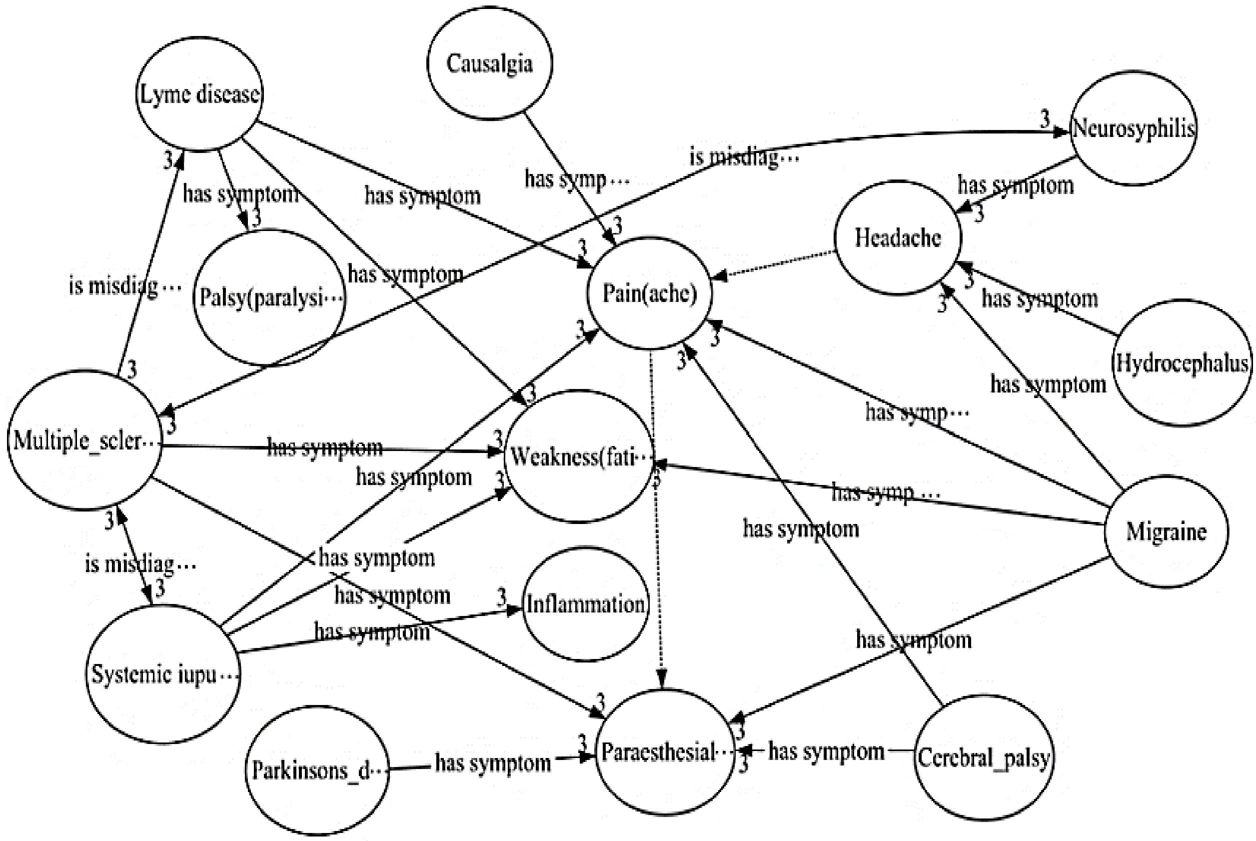


Figure 4: Disease-Sympton Semantic Network

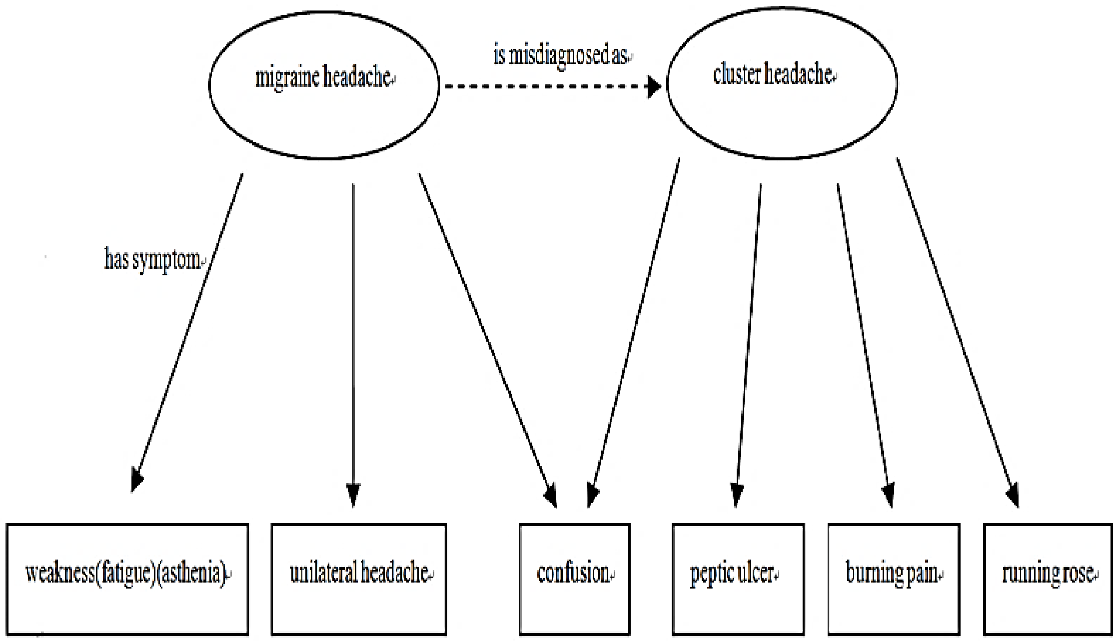


Figure 5: Example of misdiagnosed disease in DSSN

## IV. APPLICATION OF DSSN IN MISDIAGNOSIS PROMPTS

Misdiagnosis, primarily stemming from the confusion of similar symptoms, underscores the significance of differential diagnosis in averting and mitigating diagnostic errors. A clear understanding of distinct symptoms and the knowledge of differential diagnoses among diseases susceptible to misdiagnosis are crucial for prompt and accurate disease differentiation. Leveraging the disease-symptom relationship, the disease-disease misdiagnosis relationship, and differential diagnosis knowledge within the Disease-Symptom Semantic Network (DSSN), this semantic structure can effectively prompt against misdiagnoses in medical practice.Appendicitis, a prevalent surgical emergency, frequently experiences misdiagnosis and inappropriate treatment in clinical scenarios. The term "negative appendectomy" refers to the removal of the appendix in cases where inflammation is absent, and it indicates rates as high as 40%[24][25]. This paper takes appendicitis as an example to illustrate the practical application of DSSN in misdiagnosis prompts within medical diagnosis. By integrating a misdiagnosis prompt module into clinical auxiliary diagnosis and treatment systems based on DSSN, clinicians can enhance diagnostic accuracy and reduce misdiagnosis probabilities.
In a hypothetical use case, if a physician initially diagnoses a patient with appendicitis, the misdiagnosis prompt module can be invoked to lower the likelihood of misdiagnosis. Figure 6 demonstrates symptoms associated with appendicitis in DSSN, including right lower abdominal pain, fever, vomiting, constipation, and periumbilical pain, along with diseases prone to misdiagnosis such as enteritis, pancreatitis, cholecystitis, and ectopic pregnancy. These symptoms and potential misdiagnosed diseases serve as prompts for clinicians during diagnosis.
Subsequently, Figure 7 illustrates the visual display of common symptoms (abdominal pain, vomiting, and fever) and unique symptoms (constipation, pain in the lower right abdomen, and pain around the navel for appendicitis; diarrhea and diffuse pain for gastroenteritis). This information aids clinicians in making a differential diagnosis tailored to the patient's specific circumstances. In summary, the misdiagnosis module built on DSSN provides a visual representation of diseases and their easily misdiagnosed symptoms, offering valuable misdiagnosis tips for clinicians and thereby reducing the likelihood of diagnostic errors.

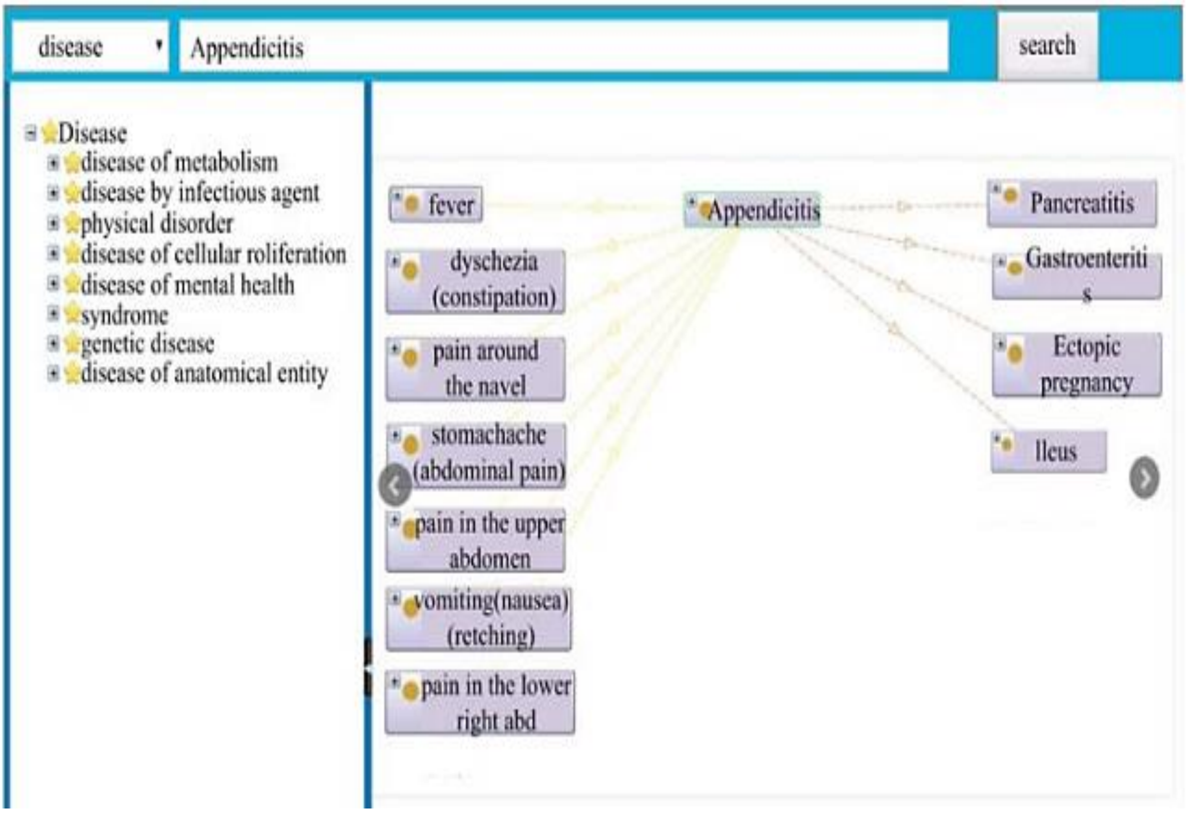


Figure 6: Symptoms of appendicitis and its misdiagnosed diseases

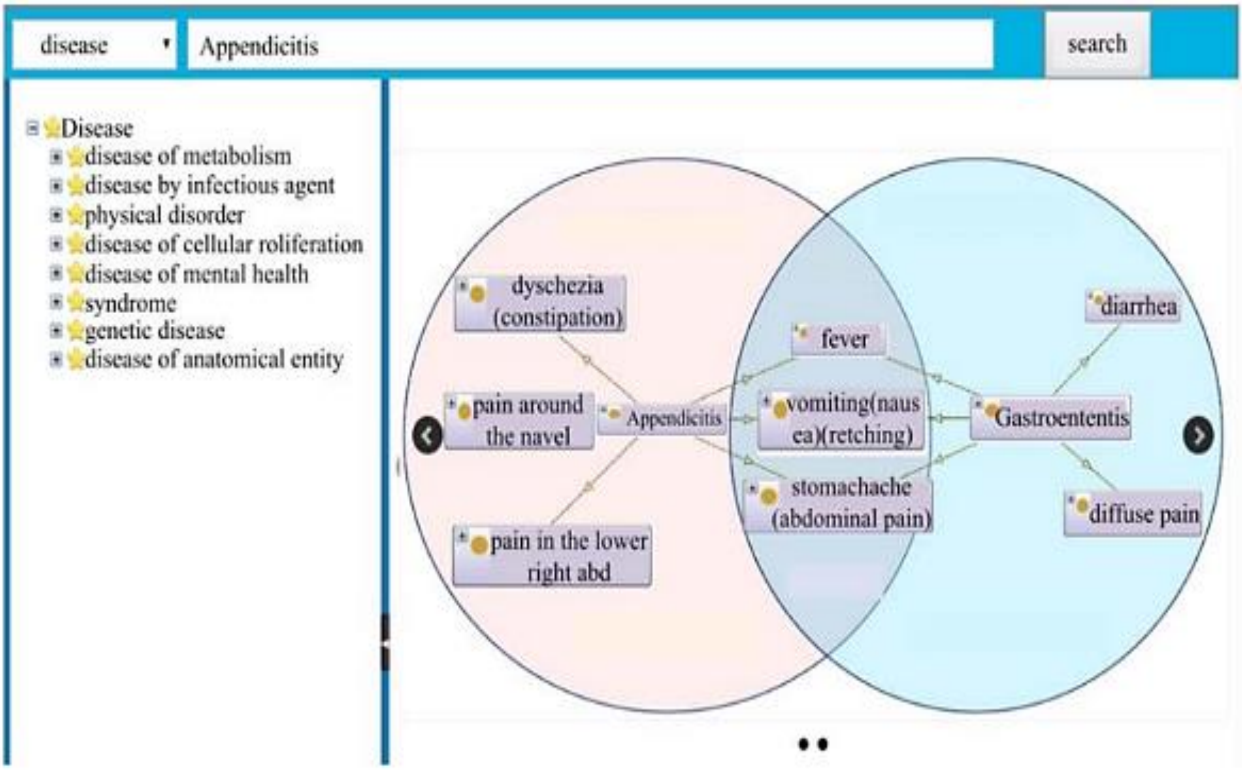


Figure 7: Same symptoms and unique symptoms between appendicitis and gastroenteritis

## V. CONCLUSION

With the development of deep learning algorithms and the Semantic Web, in addition to entities being recognized by computers[26], it is also a development trend for data to be slowly understood by machines [27][28]. In the field of clinical diagnosis, misdiagnosis is a pervasive challenge predominantly attributed to diseases with overlapping symptoms. Extensive knowledge concerning the easily misdiagnosed relationships between diseases and the nuanced differences in symptoms across diverse ailments is abundantly scattered throughout various medical literature[29][30]. To systematize and represent this wealth of information, this paper introduces the construction of a Disease-Symptom Semantic Network (DSSN). By employing natural language processing and text mining techniques across multiple medical knowledge bases, an expanded symptom vocabulary candidate set is derived. This set encompasses disease-symptom relationships, easily misdiagnosed connections, and inter-disease knowledge. These intricate relationships and knowledge are then encapsulated within the structure of a semantic network.

As bioinformatics advances and the range of relevant data sources expands, future research endeavors will focus on enriching DSSN with additional biomedical features to enhance its semantic accuracy and comprehensiveness. In addition, combining the different symptoms of the disease at different times with DSSN can further visualize the data[31][32] and improve the diagnostic accuracy. Concurrently, efforts will persist in developing tools related to DSSN, aiming to facilitate broader applications within the domain of clinical diagnosis.

## CONFLICTS OF INTEREST

The authors declare that they have no conflicts of interest

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