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Prototype Expert System for Gout Diagnosis on an Outpatient Basis

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Abstract. Gout is a systemic disease that is caused by the deposition of monosodium urate crystals in various tissues which leads to inflammation in them. This disease is often misdiagnosed. It leads to the lack of adequate medical care and development of serious complications, such as urate nephropathy and disability. The current situation can be improved by optimizing the medical care provided to patients, which requires searching for new strategies in terms of diagnosis. One of these strategies is the development of an expert system for providing information assistance to medical specialists which was a purpose of this study. The developed prototype expert system for gout diagnosis has knowledge base including 1144 medical concepts and 5 640 522 links, intelligent knowledge base editor and software which helps practitioner make the final decision. It has sensitivity of 91,3% [95% CI, 89,1%-93,1%], specificity of 85,4% [95% CI, 82,9%-87,6%] and AUROC 0,954 [95% CI, 0,944-0,963].

Keywords. Knowledge base, Expert system, Clinical decision support system, Gout

1. Introduction

Gout is a rheumatic disease prevalence of which in the world according to various studies is estimated to be up to 6,8% [1,4] of population (Australia, 2015), in Russian Federation this figure is around 1-2% [1,2]. The pathogenesis is related to the deposition of monosodium urate crystals in various tissues of the body mostly in joints in patients with hyperuricemia associated with environmental and/or genetic factors. This causes an inflammation in patient's tissues and/or tophi development [2] which can be a reason for development of various complications of the disease and comorbid conditions leading to patient's disability.

In 2015 American College of Rheumatology and European League Against Rheumatism developed new three-step classification criteria for gout diagnosis –

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ACR/EULAR, 2015 [4]. On the first step it required at least one episode of swelling, pain or tenderness in a peripheral joint or bursa [2,4]. On the second step ACR/EULAR, 2015 recommend using the golden standard in the gout detection - polarizing microscopy of synovial fluid or tophus [2,4]. But this study is not widespread, for example in the United Kingdom crystal-proven diagnoses in gout are limited to 18%, in Spain – 32% [7]. In Russian Federation using of the polarizing microscopy has limited, so practitioners can use it just in 1 case out of 10 during gout diagnostics or a similar disease. On the third step criteria recommend assessing the clinical state of patient mostly based on the more than 50 different medical concepts with varying degrees of confidence. Such huge number of clinical signs which practitioners must operate with during diagnostics lead to increase of medical errors.

The situation can be changed for the better integrating the clinical decision support system, in particular expert systems. These systems can be defined as software collecting and analyzing information about patient's disease and influencing the practitioner's decision-making during examining a patient, diagnosing, prescribing treatment to reduce errors and improve the quality of medical care [6]. The aim of this study is to develop prototype expert system for gout diagnosis and validate it using medical records.

2. Methods and Materials

The group of experts (rheumatologists, therapists) consisted of 3 highly qualified specialists of the Department of Faculty Therapy named after academician A.I. Nesterov, Pirogov Russian National Research Medical University. The criteria for the selection of experts were the following: academic degree, the position held, work experience in the field of rheumatology for more than 8 years, the presence of cited publications.

Prototype expert system for gout diagnosis was developed using Microsoft Visual Studio 2019, C# programming language. As a technical solution to store knowledge base we used Neo4j graph database.

Validation of the prototype expert system for gout diagnosis was based on the 2183 electronic medical records provided by Tyumen Department of Health which were converted into format of structured clinical documents (HL7 CDA2.0). 1001 medical records were about gout patients (gout group) and 1182 medical records were about patients with septic arthritis, pyrophosphate arthropathy, reactive arthritis, rheumatoid arthritis, psoriatic arthritis, and other diseases which clinical symptoms are similar to gout symptoms according to experts' group (non-gout group). Medical records about gout patients were collected over a period from 01.01.2017 to 01.05.2021. Medical records of the second group were collected over a period from 01.08.2021 to 30.09.2022.

Each medical record includes depersonalized patients' information. There are patient's age and gender, results of doctors' consultation (patient's complaints, anamnesis of life, anamnesis of the disease, objective status, diagnosis and recommendations), results of laboratory studies (complete blood count (CBC), biochemical analysis of blood, ESR (sed rate) or CRP (C-reactive protein)), and instrumental studies (ultrasound and x-ray examinations, computed and magnetic resonance tomography of affected joints). None of medical records has the results of polarizing microscopy, but this was not restriction for data collection.

3. Results and Discussion

3.1. The development of the prototype expert system for gout diagnosis

Prototype expert system for gout diagnosis contains two components: knowledge base (KB), including 1144 medical concepts and 5 640 522 links, and computer program incorporating problem solver. The development of the KB started with primary formalization of the subject area by nomenclature of medical terms and logical schemas for gout diagnosis [5]. These information objects were used to increase effective interaction between knowledge engineer and expert group allowing to exclude various technical issues. Nomenclature of medical terms comprises 495 main concepts and 679 synonymic terms which are used in system like a reference to the main concepts and can be unlimitedly expanded. There are 40 schemas which are used to convey the logic of primary gout diagnosing (ICD-10: M10.0, ICD-11: FA25.0).

Our team developed intelligent KB editor. It helps knowledge engineer to create and maintain the KB according to the information objects. This tool consists of 4 modules: processing textual sources module (splitting them up into semantic fragments and extracting medical terms), module for working with nomenclature of medical terms, knowledge base architecture module and knowledge base editor module. We used KB editor to fill the KB using nomenclature of medical terms and logical schemas.

KB architecture was designed using combination of two methods of knowledge representation (semantic nets and frames), ontology approach and elements of fuzzy logic [5]. Architecture describes 7 types of concepts bound by 11 types of links. Each of them has a set of attributes. One of the attributes is confidence factor (CF) which describes experts' confidence in how unambiguously symptoms, syndromes or both identify the disease in the range of 0 to 100% [3]. We used them in links, connecting concept types "Symptom", "Syndrome" and "Disease". Filling CF was carried out by our team in three steps. At first step experts determined the value of CF for connection between one concept which type is "Symptom" or "Syndrome" to the concept which type is "Disease". At the second step expert defined sets of the concepts (types "Symptom" or "Syndrome") which is sufficient to make a diagnosis according to ACR/EULAR, 2015. At the third step using linear interpolation knowledge engineer and experts determined intermediate value of CF for different sets concepts (types "Symptom" or "Syndrome").

Computer program (CP) uses algorithm which identifies gout with certain confidence and generate diagnostic recommendations to practitioner who will make the final decision. CP consists of three modules: "Pre-processing of input data", "Determination of the syndrome\diagnosis" and "Generating diagnostic recommendations". The first module analysis structured input data such as electronic health record or structured clinical documents and collect clinical parameters which can be used in KB and transform them into certain format. The second module includes problem solver to find the most appropriate link with CF using the results of the first module. The third module also includes problem solver which define minimum number of clinical parameters which expert system will recommend practitioner to screen to make a diagnosis of gout.

3.2. Validation of the prototype expert system for gout diagnosis

Provided medical records contain signs of patient's disease detected by practitioner and final clinical diagnosis made by him\her. None of them has a result of polarizing microscopy which is highly recommended in ACR/EULAR, 2015. Our team exclude 165 medical records from the gout group and 293 from non-gout group because of the lack of information to make a diagnosis. There were only 38 medical records from the gout group with sufficient clinical information to diagnose gout according to ACR/EULAR, 2015.

Prototype expert system for gout diagnosis analyzed medical records and defined against each of them the CF of gout presence in the patient which were identified by the experts. Then we used ROC-analysis to estimate prototype and define the ROC point with the highest sensitivity and specificity. So, when threshold of CF is 67.44% the prototype expert system for gout diagnosis has sensitivity of 91,3% [95% CI, 89,1%-93,1%], specificity of 85,4% [95% CI, 82,9%-87,6%] and AUROC of 0,954 [95% CI, 0,944-0,963].

It is assumed that prototype integrating in hospital information system will help practitioner to diagnose gout. The prototype will recommend practitioner to examine necessary clinical signs, to order laboratory and instrumental studies or to schedule a consultation of rheumatologist in accordance with the ACR/EULAR, 2015. This approach will allow to improve the quality of medical care by speeding up the diagnosis of gout and providing medical care in a timely manner.

4. Conclusions

The prototype expert system for gout diagnosis was developed in collaboration with experts and validated using 1725 depersonalized medical records (without excluded records from the sample) provided by Tyumen Department of Health with sensitivity of 91,3% [95% CI, 89,1%-93,1%], specificity of 85,4% [95% CI, 82,9%-87,6%] and AUROC of 0,954 [95% CI, 0,944-0,963].

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