AN EXPERT SYSTEM FOR THE DIAGNOSIS OF IRRITABLE BOWEL SYNDROME

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Abstract

For a long time functional digestive disorders (FGIDs), including irritable bowel syndrome (IBS), were described based on what they are not, without considering them real entities. Although IBS evolution is benign, it has major effects on the quality of life, it determines a higher number of days of sick leave or days when work activities had to be interrupted, thus increasing the expenses for the society. Our system, developed using CORVID Exsys, is called IBS Expert and addresses both patients and physicians. Answering some simple questions, patients will receive information about their diagnosis, together with additional recommendations. Non-expert physicians will have rapid access to the algorithm of IBS diagnosis, and could use recommendations for future investigations. This expert system is based on the information provided by the most complete collection of data about FGIDs, created by an international group of experts, who elaborated Rome III criteria. The system consists of three sub-systems, including a set of eighteen questions and the diagnosis of IBS, as well as additional recommendations, established using confidence variables. The evaluation of the system consisted of verification (determining the accuracy of the results to ensure the delivery of diagnosis and recommendations as it was intended), validation of knowledge (met through the use of the gold standard in the field) and assessment of the impact on users, either patients or physicians (measuring overall satisfaction and the user effort score in using the system). The system is available online at www.gastroterra.ro and can be accessed by using any Java-enabled device, from standard computers to mobile devices.

Keywords: irritable bowel syndrome, expert system, evaluation of the system.

Introduction

For a long time functional digestive disorders (FGIDs), including irritable bowel syndrome (IBS), were described based on what they are not, rather than real entities. IBS is a functional disorder, without pathophysiological changes and we can only know that a patient has this syndrome based on his or her own reporting [1]. For many physicians the diagnosis of IBS is still based on the observation that no structural changes are observed during patients' examinations.

A study conducted in 8 Western Europe countries has showed that in adults the prevalence of IBS is 11.5%. Ninety percent of IBS patients were consulted in primary care, 17% in a hospital (5% several times) and 69% used medication for this condition. Among IBS patients, 19% were diagnosed after the first consultation, and 56% after 1 to 5 medical visits [2]. This study shows that IBS is a burden for the healthcare system, through repeated use of medical resources and unnecessary investigations for a disorder that does not influence life expectation. Although IBS evolution is benign, it has major effects on the quality of life, it determines a higher number of days of sick leave or days when work activities have to be interrupted, thus increasing expenses for the society [2].

Given the results of Hungin *et al.*, and knowing that FGIDs represent almost a half of the gastroenterological consultations in primary care, internal medicine and gastroenterology [1], we developed an expert system for the diagnosis of IBS.

An expert system is a computer application that can be successfully compared with a human expert. The purpose of an expert system is to obtain, similar to human experts, results related to difficult activities or for activities for which a solving algorithm doesn't exist. An expert

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system consists of three major components: the knowledge base, the working memory and the inference engine [3]. The knowledge base is a structure that contains all the specific knowledge provided by human experts, which tends to remain constant during the system's operation. The knowledge base is usually created in the development phase of the system, aiming at covering all situations that may occur, and it is typically updated in line with the developments in the field of expertise. It can be built by engineers, with the support of experts, by experts in the field or using learning methods of the system, generally based on examples. The working memory contains transient knowledge that changes significantly during the system use. Working memory contains all user data, both initial input and interim results. The inference engine is the operational component of an expert system that gets solutions and/or recommendations by applying the knowledge base to the working memory and deriving the results by changing the latter. For the development, maintenance and use of the system, the following interfaces are required: 1. interface with the developers - used to modify important aspects of the system such as search strategy or the selected variables; 2. interface with experts - used for acquiring new knowledge and changing existing ones; 3. user interface - used to initialize the working memory to transmit control signals to inference engine, receive results and display them (figure 1).

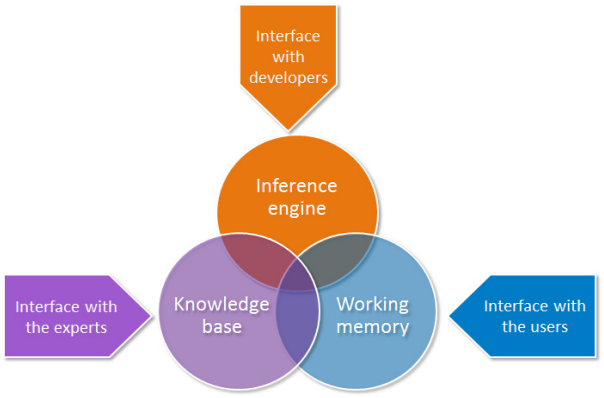

Figure 1. The components of an expert system.

Our system, called IBS Expert addresses both patients and physicians. Answering some simple questions, patients will receive information about their diagnosis, together with additional recommendations. Non-expert physicians will have rapid access to the algorithm of IBS diagnosis, and could use recommendations for future investigations. Many patients with IBS never consult their physician, either because of lack of time, or because they consider that their symptoms are not important or they got used to them [2]. This type of patients could use our system to identify the presence of a so called "red flag" that imposes urgent investigations. On the other hand, this system addresses patients scared of a more severe condition, which consult every year for the same chronic symptoms; they could also use our system to identify the presence of a "red flag" that requires consultation. Given this aspects, the questions were formulated to be understood by a wide audience. This expert system can also be used by general physicians, internists, non-experts gastroenterologists, who are not familiarized with FGIDs, allowing an easy and rapid identification of IBS patients.

The system was developed using the platform Exsys CORVID 3.1.1, it uses Java applets, being very accessible from any location and any device connected to the Internet (figure 2).

Figure 2. Home screen of the IBS Expert System.

The knowledge base

Medicine is not an exact science, hence when developing an expert system one should perform multiple iterations to determine the current knowledge of experts. Rome Foundation and the "Rome process" have the purpose to update the knowledge on FGIDs including IBS through an international scientific effort making it easier to reference it as gold standard.

The latest Rome criteria were published in 2006, also known as Rome III criteria. According to Rome III, the IBS definition is based only on clinical data, because nowadays it is considered that IBS and organic disorders may coexist. The most important step in diagnosing IBS is taking the medical history. The decision for additional tests such as erythrocyte sedimentation rate, complete blood count or lower gastrointestinal endoscopy is based on additional data such as the age of the patient, the duration of the symptoms, familial history of gastrointestinal disorders, presence of associated symptoms or signs (the so called "red flags") [4]. A study published in 2010 showed that, by contrast with experts in IBS, a very important proportion of non-expert physicians (primary care physicians, but also gastroenterologists) considered the IBS diagnosis to be an

exclusion diagnosis (72% vs. 8%, p<0.0001), resulting in more diagnostic tests, and double the expenses of experts per patient [5].

This expert system is based on the information provided by the most complete collection of data about FGIDs, created by an international group of experts, who elaborated Rome criteria. The current knowledge about IBS is the result of years of research worldwide, and the Rome criteria are widely used for the selection of patients by any group of researchers.

The system consists of three sub-systems, including a set of eighteen questions. Transposing these questions in the system was done by defining static variables, easily identifiable in the application because they contain the prefix *static*, a number that indicates the subsystem (0, 1, 2), a number (not necessarily consecutive) indicating the question and a suggestive name - for example: *static04TimpDebut*. A first set of questions is designed to identify patients who meet clinical criteria for IBS, namely: pain/abdominal discomfort recurring at least 2-3 days/ month in the last 3 months, associated with at least two of the following criteria: 1. improvement of pain/discomfort with defecation; 2. occurrence of pain associated with a change in defecation frequency; 3. occurrence of pain associated with a change in the form or appearance of the stool. In addition, in order to differentiate IBS from other transient digestive symptoms, one should consider the chronicity of symptoms. In IBS, symptoms must have first appeared at least 6 months before diagnosis. The presence of pain at least 2-3 days per month in the last 3 months is required to demonstrate disease activity [4].

Some of these questions are essential for the diagnosis of IBS. Thus, we can say that there is no IBS without abdominal pain or discomfort. Given this observation. the absence of pain excludes the diagnosis of IBS, and the user will be directed to the exit screen. The diagnosis of IBS also includes a time criterion. The symptoms must be present for at least 6 months, their chronicity supporting the functional character. The frequency criterion (at least 2-3 days per month) is also required [4]. The absence of any of these two criteria exclude the diagnosis of IBS.

Although IBS coexists with other digestive organic diseases, many experts exclude this diagnosis in the presence of inflammatory bowel disease (IBD) or colon tumors. Diverticulitis can cause symptoms similar to IBS, but often the symptoms do not improve after treatment, the patient having a component of irritable bowel. Our system includes three questions concerning the patient's medical history: whether he/she had had a colonoscopy, which is the result of this investigation and whether the treatment of the disease outlined in colonoscopy determined disappearance of symptoms. Rome III criteria do not clearly state that the presence of the organic diseases mentioned above exclude the diagnosis of IBS. In IBS patients, the colonoscopy is rarely normal. For example, a prospective study of 466 subjects with symptoms suggestive of IBS with constipation, without alarm features, showed that 18.2% of patients had hemorrhoidal disease, 14.6% polyps and 8.8% had colic diverticulosis [6]. Another population-based study conducted in the USA showed that IBS is associated with an increased chance of colonic diverticulosis (OR = 1.8, 95% CI 1.3 to 2.4), but not diverticulitis. The authors concluded that IBS and diverticular disease may be connected [7]. Based on the literature, namely looking at the exclusion criteria of various studies [6] in the presence of colitis (IBD, ischemic colitis, drug induced, infectious, microscopic), colon tumour, colonic diverticulosis (when symptoms resolved after treatment) the diagnosis of IBS was excluded, while in the presence of colonic diverticulosis without improvement of symptoms with treatment or other disorders (included in our questions in the category of *not applicable* - for example polyps, hemorrhoidal disease, angiodysplasia, melanosis coli, etc.) the diagnosis of IBS was not excluded [6,7].

Patients with celiac disease have similar symptoms to IBS patients, therefore we also included 2 questions to determine if the user had a previous gastroscopy, or a diagnosis of celiac disease. In the presence of celiac disease, we excluded the diagnosis of IBS [4,6].

Several studies showed that the Rome criteria have a good accuracy in the diagnosis of FGIDs, including IBS, and the exclusion of organic diseases, with a sensitivity of 89% and a specificity of 71% [8]. Given these observations, experts do not recommend further investigation when all the criteria are met. The presence of "alarm symptoms" or a family history that increases the risk of organic disease (adenomatous polyps, rectocolonic tumour, etc.) impose further investigations. In this context, the rest of the questions (namely 7) had to identify the presence of „alarm symptoms". The presence of any „alarm symptom" is followed by a recommendation to seek specialized advice and future investigations.

The Inference Engine

The diagnosis of IBS is established using a confidence variable [9]. In this program the cut-off value for the confidence variables is 300, value above which it is considered that a diagnosis or referral are necessary. Two methods are used to determine the final value of these variables – adding intermediate values (based on users' responses) or by taking the maximum value, and once the maximum value has been assigned to a variable, no other changes will decrease its value below the cut-off level. For easier future developments, the diagnosis of irritable bowel syndrome is generated using a collection variable, *collRecomandari*. The logical structure of the system is contained in the logic block *Anamneza* (figure 3), and the recommendations are determined in another logical block *Recomandari.*

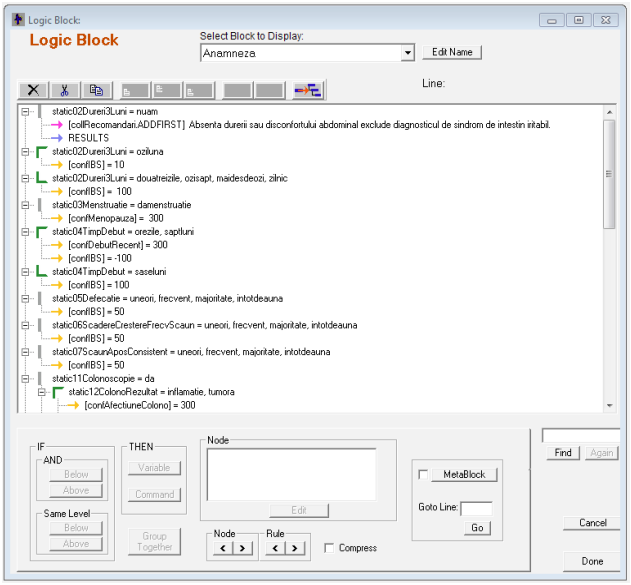

Figure 3. IF-THEN structures in the logic block *Anamneza.*

The control unit ensures the run of the logical block *Anamneza* and *Recomandari* (figure 4), in the end displaying the results (the system verifies if the confidence variables reached the 300 cut-off value, displaying the results accordingly).

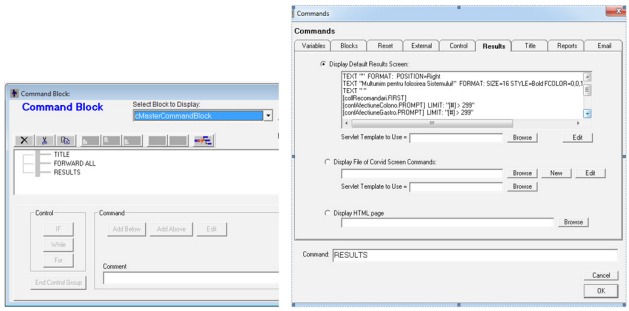

Figure 4. The control unit (on the left) and assessing whether the cut-off value of 300 for two confidence variables (*ConfAfectiuneColono, ConfAfectiuneGastro*) was met.

The figure 5 is an example of the display of recommendations.

Figure 5. Example of results (diagnostic and recommendations returned by IBS Expert System).

System's evaluation

The evaluation of the system consists of:

• Verification – determining the accuracy of the results to ensure the delivery of diagnosis and recommendations as it was intended.

• Validation of knowledge – is met through the use of the gold standard in the field, the Rome III criteria.

• Assessing the impact on users, either patients or physicians [10].

Regarding the systems' verification, the diagnosis of IBS is the most complex subsystem. Controlling for equivalent responses we generated 65 variants of responses combinations (for answers with the same impact on *confIBS* variable, we generated a single combination). We followed the correct encoding of Rome III criteria and we determined the final value of *confIBS*, checking if the final result is the one expected or not (figure 6). From this point of view the system operates correctly.

Regarding the subsystem relating to medical history, questions are clear, and we evaluated two IF and THEN clauses. In the case of a condition incompatible with IBS diagnosis, the decrease of the variable *confIBS* by 100 points will make impossible the diagnosis of IBS, because as shown in the above table, the maximum value from the first subsystem is 350 (and the threshold is 300). The subsystem regarding the "alarm symptoms" generates

douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	niciodata, nustiu	200	uneori, frecvent, majoritate, intotdeauna	250	niciodata, nustiu	250
douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	niciodata, nustiu	200	uneori, frecvent, majoritate, intotdeauna	250	uneori, frecvent, majoritate, intotdeauna	300
douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	uneori, frecvent, majoritate, intotdeauna	250	niciodata, nustiu	250	niciodata, nustiu	250
douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	uneori, frecvent, majoritate, intotdeauna	250	niciodata, nustiu	250	uneori, frecvent, majoritate, intotdeauna	300
douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	uneori, frecvent, majoritate, intotdeauna	250	uneori, frecvent, majoritate, intotdeauna	300	niciodata, nustiu	300
douatreizile, ozisapt, maidesdeozi, zilnic	100	da	100	saseluni	200	uneori, frecvent, majoritate, intotdeauna	250	uneori, frecvent, majoritate, intotdeauna	300	uneori, frecvent, majoritate, intotdeauna	350
douatreizile, ozisapt, maidesdeozi, zilnic	100	nu, na	100	orezile, saptluni	0	niciodata, nustiu	0	niciodata, nustiu	0	niciodata, nustiu	0
douatreizile, ozisapt, maidesdeozi, zilnic	100	nu, na	100	orezile, saptluni	0	niciodata, nustiu	0	niciodata, nustiu	0	uneori, frecvent, majoritate, intotdeauna	50
douatreizile, ozisapt, maidesdeozi, zilnic	100	nu, na	100	orezile, saptluni	0	niciodata, nustiu	0	uneori, frecvent, majoritate, intotdeauna	50	niciodata, nustiu	50

Figure 6. Example of combinations created to determine the final value of the confidence variable *confIBS*. In yellow are the combinations that reached the cut-off value of 300, necessary for the diagnosis of irritable bowel syndrome.

an output message if any of the seven questions are answered such as to indicate a potential organic disorder, and we consider that it also operates correctly.

To establish the impact of this system on users, we used two indicators, the satisfaction in using the system and the effort required to use the system [11]. Satisfaction in using the system measures the overall satisfaction. The question was: *On a scale of 1 to 5, where 1 means very dissatisfied and 5 means very satisfied, how satisfied are you with IBS Expert system*? Response options for this question were: 1 - very dissatisfied, 2 - dissatisfied; 3 - neither satisfied nor dissatisfied; 4 - satisfied; 5 - very satisfied.

The measurement of the effort score in using the system is a safer way to determine users loyalty, leading to repeated use, which is one of the important elements to ensure the acceptance and adoption of the system by the users. Comparing overall satisfaction and ease of use of the system, the latter can be considered a better predictor for repeated use of the system [11]. The question we used was: *On a scale of 1 to 5, where 1 means very difficult and 5 is very easy, how simple was to use the system and get the final recommendations?* Response options for this question were: 1 - very difficult, 2 - difficult, 3 - neutral, 4 - easy and 5 - very easy.

By applying a questionnaire containing the above two questions on ten people we achieved a maximum score both on satisfaction and on effort score. With the publication of the system on the Internet, the measurement of these two indicators may be extended to a larger sample.

Future developments

As shown in one of the first medical expert systems applied in medicine in Cluj [12] further developments come naturally, the author being aware of them as soon as he/she considers the work completed. Some immediate areas for improvement are:

• Creating two different operating modes, "patient" and "physician". In this way system messages could be customized, more complex for the physician, without the risk of confusion for the patient.

• Development of a section dedicated to family history, and the inclusion of further recommendations.

• Evaluation of the system in a clinical setting to determine the impact with respect to the objectives set at the beginning of the paper.

The system is available online at www.gastroterra.ro and can be accessed by using any Java-enabled device, from standard computers to mobile devices.

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