ENT Disease Diagnosis Using an Expert System

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Abstract. The field of medicine has witnessed a dramatic growth. However, the accurate and timely diagnosis of disease continues to be a serious clinical problem. This is particularly important for Otolaryngology/Ear-Nose-Throat (ENT) disease because ENT disorders can affect hearing, speaking, learning and many other important activities. Further, certain untreated ENT diseases can be fatal. Therefore, early diagnose of ENT diseases is vital. While ENT specialist's service is not always readily accessible, computer aided smart technologies that can assist general physicians or junior medical officers in diagnosing ENT diseases and subsequently refer complicated cases to senior ENT experts can enhance the efficacy of healthcare system. Despite the significance of computer aided ENT disease diagnosis systems, the research related this subspecialty is limited. Therefore, in this paper, we describe the research project about an ENT diagnosis expert system that can assist physicians in diagnosing ENT diseases. In particular, we will discuss in detail the development, evaluation and potential benefits of an ENT disease diagnosis expert system.

Keywords: ENT Expert System, Otolaryngology Disease Diagnosis, Expert System, Inferencing.

1 Introduction

Today, the field of medicine has improved immensely due the advancements in computing technology. Computers are involved in almost all the clinical practices. Artificial intelligent (AI) technologies assist both healthcare practitioners and patients in numerous ways. AI is a branch in computer science that can analyze complex medical data and identify meaningful relationships that can be used for clinical diagnosis and treatment.

Despite the latest advancements in healthcare, it does not always reach out to those most in need. Enormous difficulties have been faced by ordinary people in seeking medical assistance. Particularly, access to medical specialist is a critical problem(Van Doorslaer, Masseria, & Koolman, 2006). Due to the limited availability of specialist, ordinary people find it difficult to channel specialist frequently. i.e. specialist attend only critical cases and are only available in metropolitan hospitals. Thus, the accurate and timely diagnosis of disease continues to be a serious clinical problem.

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Therefore, smart technologies that can assist in early diagnosis and prevention of serious health problems can be a great relief for many patients and healthcare system in general. This is particularly important for Otolaryngology/Ear-Nose-Throat (ENT) disease because ENT disorders can affect hearing, speaking, learning and many other important activities. Further, certain untreated ENT diseases can be fatal. Therefore, early diagnose ENT diseases is vital. As discussed above specialist access is not always available. While Otolaryngology specialist's service is not always readily available, computer aided smart technologies that can assist general physicians or junior medical officers in diagnosing ENT diseases and subsequently refer complicated cases to senior ENT experts can enhance the efficacy of healthcare system.

Despite the significance of computer aided ENT disease diagnosis systems, the research related this subspecialty is limited. Therefore, in this study we have developed a research based ENT (Ear-Nose Throat) Disease Diagnosis Expert System that can assist physicians or junior doctors in diagnosing ENT diseases. In particular, we will discuss "how an ENT disease diagnosis system can be designed and evaluated?" Furthermore we discuss the potential benefits of the system. The proposed system uses rule-based inferencing in diagnosis process and adopted an approach similar the one followed by ENT experts in their diagnosis process. Therefore, this system can not only used by physicians but also to train medical students towards ENT diseases diagnosis process and patient centered healthcare systems.

The rest of the paper is organized as follows. The immediately following section provides a background on existing medical expert systems. Then we describe our ENT disease diagnosis system- Virtual Doctor followed by the implementation details of the system. Subsequent sections provide discussion and finally conclusion.

2 Background

Artificial intelligent systems in medicine, started to emerge during late 1960s and many experimental systems were developed by research laboratories. Early AI based medical applications have laid the foundation for many new, recent applications. MYCIN(Edward Hance Shortliffe, 1976), is one the popular early rule-based expert system to diagnose and treat infectious disease. INTERNIST(Miller, Pople Jr, & Myers, 1982), is also rule-based expert system designed for the diagnosis of complex problem in internal medicine. The system was capable to cover 80% of the knowledge in internal medicine. CASNET(Weiss, Kulikowski, & Safir, 1978), is an expert system based on causal-associational network and was used for the diagnosis and treatment of glaucoma. Based on the concepts of MYCIN, expert systems such as ONCOCIN(Edward H Shortliffe, 1986) and PUFF(Aikins, Kunz, Shortliffe, & Fallat, 1983) were designed to assist physicians in treating cancer and lung diseases respectively.

These first generation expert systems laid the foundation for the next generation expert systems. In recent years expert systems have integrated multimedia technologies, machine learning, artificial neural networks and fuzzy logic and genetic algorithms to

enhance the diagnosis. Examples of such systems are AI/RHEUM(Athreya, Cheh, & Kingsland III, 1998), ESTDD(Keleş, 2008) and ODPF(Chi, Street, & Katz, 2010).

AI/RHEUM(Athreya et al., 1998) is an expert system designed to diagnose, rheumatic diseases. The results from AI/RHEUM reveal 92% accuracy. The authors claim that this expert system can assist in both consultation and education. The AI/RHEUM uses criteria table paradigm for reasoning and provides conclusions at three levels of certainty- definite, probable and possible. Melek et al(Melek, Sadeghian, Najjaran, & Hoorfar, 2005) developed a neuro fuzzy based expert system for disease diagnosis. This system aims to assist physicians in their daily practices.

Expert System for Thyroid Disease Diagnosis (ESTDD)(Keleş, 2008), is a fuzzy rules based expert system for diagnosing thyroid diseases. The results reveal that the tool is able to predict a thyroid diseases diagnosis with 95.33% accuracy. Optimal Decision Path Finder (ODPF)(Chi et al., 2010) is a machine learning based expert system, which can expedite the diagnosis process and reduce the cost of diagnosis by reducing the number of diagnostic tests ordered. It is a decision support system that provides information on the disease probability, based on patient's available information. The system dynamically estimate minimum set of tests that are likely to confirm a diagnosis. The ODPF algorithm mainly uses lazy learning classifiers, confident diagnosis and, locally sequential feature selection (LSFS).

According to the literature review, medical expert systems have been developed for coronary artery diseases , thyroid diseases (Keleş, 2008), bone diseases (Hatzilygeroudis, Vassilakos, & Tsakalidis, 1997), rheumatic disease (Athreya et al., 1998), dengue (Karim, Suryaningsih, & Lause, 2009), cancer (Edward H Shortliffe, 1986), lung disease (Aikins et al., 1983), infectious diseases(Edward Hance Shortliffe, 1976) and glaucoma(Weiss et al., 1978). However, research on expert system that diagnoses ENT diseases is limited. Besides, the diagnoses of ENT diseases are based on symptoms which are often difficult identify. Many sophisticated examinations may be necessary in the diagnosis process. Thus, we aim to fill this gap by building an expert system to diagnose ENT diseases.

3 Virtual Doctor – ENT Disease diagnosis System

"Virtual Doctor" is an artificial intelligence (AI) based expert system, designed to assist physicians or junior doctors in the diagnosis of ENT related disease in the absence of ENT experts. Thus, this system can reduce the backlog created due to the busy schedules of ENT experts and enhance the effectiveness and efficiency of healthcare system. Virtual Doctor - ENT diagnosis uses rule based system for knowledge representation and has five core sub-systems which would enhance the physician's ability in reaching a diagnosis decision with confidence. Initially the symptoms are captured through the user interfaces as inputs. Then these symptoms are matched with inference rules in the knowledge base and finally a diagnosis is made by the inference engine. If the system is unable to diagnose the disease with the given symptoms or if the system is unable to identify the exact ENT disease the expert system recommends for some laboratory tests. Subsequently when these test results are

provided, diagnosis will be carried out with more accuracy. If the system is unable to diagnose the disease even after producing the laboratory test results, the system will recommend for an ENT specialist consultation. The following sections describe the five main functionalities in detail. Figure 1(a) shows the main interface of Virtual Doctor, with references to each sub-module. For example when user (i.e. physician) clicks on disease diagnosis (see figure 1(b)) he/she will be directed to disease diagnosis sub-module.

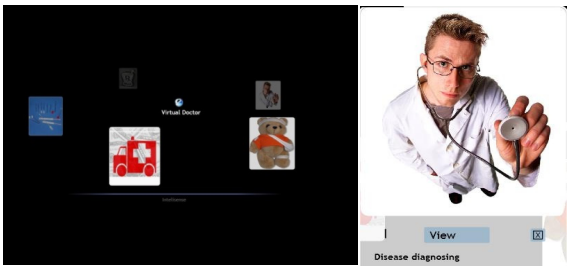

Fig. 1. (a) Virtual Doctor Main –with a 3D rotation menu for each of the sub-modules (b) Disease diagnosis sub system

Disease Diagnosis. Disease diagnosing sub-system prompts the physician to gather set of preliminary medical investigation related information, such as body temperature, blood pressure from patient. Based on this information, the system will intelligently prompt more related questions, in order to acquire detailed information of the disease. After analyzing the given information, the system derives conclusions accordingly. i.e. (1) the system may either report the decision about the disease and prescribe medication (i.e. when the disease is diagnosed with higher certainty) or (2) when clinical symptoms are not sufficient to come up with a diagnosis decision, the system recommends appropriate laboratory tests in order to help further diagnosis of the disease with more accuracy. Besides, the system stores all the data related to the patient for future references.

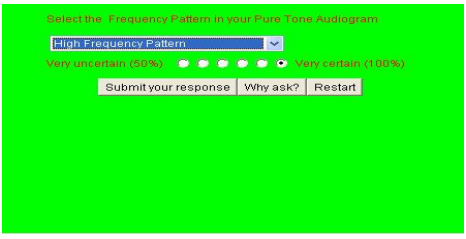

Fig. 2. Analyzing the objective evidence

Prescribing. During the disease diagnosis process, if the disease is accurately identified with higher level of certainty, the disease information (ENT disease) will be

transferred to 'Prescribing' sub-system. Before prescribing medication, this sub-system would inquire about the patient's allergic conditions (to drugs or food), and patient's current medication details (such as whether the patient is currently being administered any other drugs, for some other disease). Based on this information the system would prescribe appropriate medication (i.e. system takes into account the interaction or contradiction between drugs). Moreover, this sub-system also stores the information for future reference.

Recommend Testing. When the clinical symptoms are not sufficiently enough to decide on the diagnosis with certainty, appropriate and comprehensive clinical tests and supporting tests will be recommended in order to decide on the diagnosis with higher level of certainty. Laboratory testing, X-ray or other relevant checkups will be recommended. If the patient provides details of the clinical and supporting tests, the test results will be entered into the system or scanned image will be uploaded into the system. Image processing techniques will identify patterns. Details of the clinical and supporting tests results will be used for detailed diagnosis with greater level of accuracy. In addition, the information gathered through this sub-system will also be transferred to database for future references. Table 1 provides a list of tests that can assist in ENT disease diagnosis. Table 2 provides details of the test results and possible diagnoses.

Table 1. Types of specific tests

Category	Test
Hearing Tests	Pure Tone Audiometry, Tympanometry, Speech & Voice Analysis
Nasal Tests	Finger-nose test, Standard Smell Test, Nasal endoscopy
Throat Tests	Throat Culture, Rapid Strep Test

ENT Specialist Referral. With the given symptoms and test results if the 'Virtual Doctor' expert system is unable to diagnose the ENT disease with certainty, the system will refer the patient to a real ENT expert or Otolaryngologist. The system will request for patient's current residence location and recommend an appropriate ENT expert (i.e. consultant), who is in the close proximity to the patient. Based on the seriousness of the illness this sub-system can advise the patients to get admitted to closer hospitals with facilities (i.e. equipment, ENT consultants) and facilitate appointments with ENT specialists.

Track Patient History. The system store all the details related to the patients, including symptoms, test results, diagnosis details, medicine prescribed, etc. The information gathered will be used for further analysis and will be used as an input for the self-learning learning system.

Table 2. Specific Tests for objective evidence

Test	Test Result	Possible Diseases
Pure Tone Audio-gram	High Frequency Pattern	Age Related Hearing Loss
		Vesticular Neuronitis
	Low Frequency Pattern	Meneire's Syndrome
	Part Frequency Pattern	Age Related Hearing Loss
	Unilateral Hearing Loss	Acoustic Neuroma
CT Scan	Tumor affecting the VC Nerve	Acoustic Neuroma
X Ray - Sinuses	Thickening of lining in Mucosa, Fluid in the Sinus	Sinusitis
	Thickening of lining in Mucosa, Fluid in the Sinus, Post Nasal Space	Epitasis
Blood Test (Full Blood Count)	Hemoglobin level outside the base lines. WBC > 12000	Infection
	Hemoglobin level outside the base lines. Platelet < 80000	Dengue

4 Virtual Doctor Design and Implementation

Virtual Doctor is intended to be used by physicians who are not advanced computer users. Therefore the user interfaces were designed to be more user-friendly. Following sections describe the details of implementation.

Knowledge Engineering for the E.N.T. Disease Diagnosis Expert System. Several different AI techniques were considered for the design of 'Virtual Doctor', including multi-agent systems and expert systems to solve the diagnosis problem. After discussing with knowledge engineer, AI expert, medical professionals and analyzing the existing literature, we decided to choose rule-based expert system, as it would best represent the diagnosis process of an ENT expert. Once the AI technique was chosen, discussions with a panel of ENT experts were carried out in order to gather relevant data. We gathered all necessary information, including a list of important ENT diseases, their symptoms, diagnosis procedures, treatment options, clinical and laboratory tests (see Figure 4). In addition, ENT related publications and books were analyzed as an additional source of information. Once the required knowledge was gathered, the details were organized and presented to ENT specialist for verification. Certain items were removed and few items were added based on the advice from ENT specialists. Once the items were finalized the acquired knowledge was encoded into rules in the knowledge base.

Table 3. Knowledge Acquisition – ENT disease and their symptomps

	Disease	Diagnosis investigation
Ear Diseases	Presbyacusis	1. Is the patient having difficulty in speech discrimination? 2. Is the patient having difficulty in phone conversation? 3. Has the patient had any trauma? 4. Has the patient got any infection? 5. Does the patient get fits? 6. Does the patient get Tinitus?
	Mener's Syndrome	1. Is the patient having hearing loss? 2. The hearing loss is sudden onset or Episodic? 3. Does the patient have associated Tinitus or Virtigo? 4. Is the patient having fever?
	Vestibular Neuronitis	1. Does the patient have hearing loss? 2. The hearing loss in patient in sudden onset or Severe? 3. Does the patient have Vertigo or tinnitus 4. Does the patient have fever?
	Acoustic Neuroma	1. Does the patient have Hearing loss? 2. Does the hearing loss gradually progressing form months to years? 3. Does the patient have initially Tinitus then Vertigo? 4. Does the patient has unilateral (one sided hearing) loss?
Nose Diseases	Allergic Rhinids/Catarrh	1. Does the patient have nasal discharge? 2. Is the nasal discharge clear or watery? 3. Does nasal discharge increases during morning or night? 4. Does the patient have sneezing? 5. Does the patient have itching in eyes, nose, or throat? 6. Does the patient have tearing of eyes? 7. Does the patient have fever?
	Sinusitis	1. Does the patient have nasal discharge? 2. Is the Nasal discharge thick or yellow discharge? 3. Does the patient have nasal block? 4. Is the nasal block is bilateral? 5. Does the patient have headache? 6. Does the patient have bleeding from nose?
	Nasal Discharge	1. Does the patient have discharge from the nose? 2. Is the nasal discharge is unilateral or bilateral? 3. Is the discharge from the nose is yellow or clear? 4. Is the discharge watery or thick? 5. Does the patient have Post Natal Drip (PND)? 6. Is the PND yellow or clear? 7. Does the patient have nasal block? 8. Is the nasal block unilateral or bilateral? 9. Is the nasal block episodic or persistent? 10. Does the patient have fever?

Table 3. (*Continued.*)

		11. Does the patient have headache? 12. Does the patient have sneezing? 13. Does the patient have itching in nose, eyes, and in palate or throat? 14. Does the patient have bleeding in nose or throat?
Nose Diseases	Foreign body in nose	1. Does the patient have nasal discharge? 2. Is the nasal discharge initially clear and then yellow?
	Bloody discharge or Epistaxis	1. Does the patient have bloody discharge or phlegmy discharge? 2. Did the patient have a trauma? 3. Does the patient have any infection? 4. Does the patient take any medication to thin the blood? 5. Does the patient involved in Drug abuse (Cocane)?
Throat Disease	Pharyngitis	1. Does the patient have sore throat? 2. Does the patient have fever? 3. Does the patient have hoarse voice? 4. How long the patient has hoarse voice is it a sudden onset (days) or long time (months to years)? 5. Does the patient have cough? 6. Does the patient have breathing difficulty? 7. Does the patient used to smoke or beetle chewing or alcohol use?
	Tonsulitis/ Oesophagitis	1. Does the patient have sore throat? 2. Does the patient have fever? 3. Does the patient have hoarse voice? 4. How long the patient has hoarse voice (is it a sudden onset (days) or long time (months to years))? 5. Does the patient have cough? 6. Does the patient have pain in swallowing or obstruction is swallowing?
	Laryngitis	1. Does the patient have Sore throat? 2. Does the patient have hoarse voice for a short duration? 3. Does the patient have fever?
	Cancer of Larynx/ Vocal code	1. Does the patient have fever? 2. Does the patient smoke? 3. Does the patient exposed to second hand smoking? 4. Does the patient have the habit of Betel Chewing?

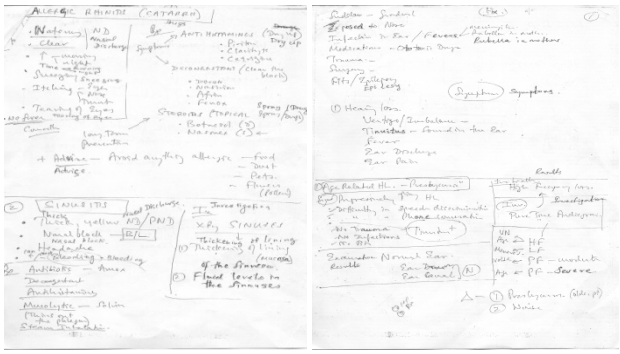

Fig. 3. Knowledge Acquisition

Knowledge Base Design. An expert system shell was used to create the knowledge base. Gathered information (such as symptoms, disease) was converted into a table format in order to implement the knowledge base (See Table 3). The rules were then written into expert system shell.

Inferencing. Once the knowledge is encoded into rules inferencing mechanism would diagnose the disease. Virtual Doctor – ENT diagnosis uses forward chaining mechanism to implement the inferencing logic using java classes. That java class logic takes user inputs (such as symptoms) through applets and subsequently the symptoms are matched with the knowledge base rules. Based on the match it will prompt further questions for clarification through applet and eventually diagnose the disease accordingly.

Reasoning with Uncertain Data. While the physician has to be certain about his/her assessment of patient's symptoms, the Virtual Doctor - ENT diagnosis system allows the user (i.e. physician) to provide confidence factor along with his/her response, which offers flexibility when he/she is not so sure about the symptoms (see Figure 3). Confidence measurements are usually called certainty factors or confidence factors (or CF). Similarly, based on the inputs' certainty, Virtual Doctor provides the diagnosis with a corresponding level of certainty or confidence.

Implementation and Integration. 'Virtual Doctor – ENT Diagnosis' comprises several sub-systems, which are built on top of an inference engine and knowledge base. All the sub-systems that are connected to the inference engine have the ability of communicating with each other using web services. The backend knowledge base (i.e. rule - based) together with the inference engine was implemented using java (e.g. forward chaining algorithms). The database server (i.e. Microsoft SQL Server) stores knowledgebase data and patient information. The front-end interfaces are built using Visual Studio & XAML, which provides a better look and feel for the user interfaces. Applets have been used to get the inference to the front end which is displayed with in XML web page. The XML web page uses Silver light to make it more attractive with

the animations. ASP.NET was used for the development of web application. Java scripts were used to capture user interaction with the systems and all the interactions are stored in the database. Moreover, C#.NET was used implement algorithms to update the knowledge base, with new acquired knowledge about disease or symptoms. This enables the self-learning capability of our system. The system as a whole combinedly provides assistance for physicians in ENT diseases diagnosis, prescribing, recommend clinical laboratory tests and refereeing patients to real ENT experts when necessary. To deploy the system one needs a web server (IIS) and an application server (java) to host the application, All the modules in this system use the common software and hardware interfaces as they have been connected together.

5 Evaluation and Validation

Evaluation of Virtual Doctor- ENT Diagnosis includes addressing issues related to accuracy and usefulness. The system we have developed is being evaluated by ENT experts and specialists. The aim of our system is to emulate ENT specialist. Thus, several reliability and validity checks are warranted. The process of evaluation is described below.

Initially a team of ENT specialist created a set of test cases based on real patients' histories and symptoms, who visited the hospital for ENT diseases diagnostic consultation. These test cases were chosen to be complex enough that physicians would be likely to request a diagnostic assistance from an ENT specialist. Ten ENT specialists were assigned 30 diagnosis cases (each three). The same test cases will be diagnosed by set of ten ENT specialists as well as by Virtual Doctor-ENT disease diagnosis system. This phenomenon is similar to Turing Tests which have been used in validating expert systems (such as MYCIN and ONCOCIN). Same test will be repeated with general physicians, so that we will be able to know how far Virtual Doctor-ENT disease diagnosis system can help physicians in diagnosing ENT diseases in the absence of ENT expert. In order to compare the real diagnosis with virtual doctor – ENT system, the physician and ENT specialist will be requested to produce a list of possible diagnoses, and rank the order of each diagnosis. Currently the system is being evaluated by ENT experts and general physicians. Once this process is completed Kappa scores(Cohen, 1960) will be calculated to evaluate the agreement beyond chance. Higher Kappa score will be perceived as greater accuracy of the system and thus confirming the validity of Virtual Doctor – ENT diagnosis system (i.e. Diagnosis similarity between real ENT specialist and Virtual Doctor Expert system). In addition, the tests will be repeated twice with another set of ENT specialist and physicians to ensure reliability of diagnosis.

Once the accuracy and reliability of the system is ensured, the usefulness aspect of 'Virtual Doctor' expert system will be validated based on the following key aspects: (1) Structure - How well the 'Virtual Doctor' ENT disease diagnosis expert system would fit into its environment(i.e. healthcare system), do doctors find it helpful? (2) Process – What effects does the 'Virtual Doctor' have disease diagnosis process, such as accuracy of decision(as discussed above) (3) Outcome – Are the effects on

healthcare process reflected on patient outcomes i.e. does the patient feel the same satisfaction that he/she would feel while diagnosed by an ENT expert? (how far it is similar to that of a real ENT expert) (Donabedian, 2005).

6 Discussion

The Virtual Doctor expert system was designed in a similar manner that an ENT consultant would approach an ENT disease diagnosis. The diagnosing processes includes steps such as initial gathering of subjective information(i.e. knowledge acquisition of symptoms related to ENT disease), generation of probable diagnosis list, gathering objective evidence(i.e. evidence from scan reports and laboratory tests), and hypothesis evaluation (i.e. finalize the disease based on subjective and objective information). Thus, Virtual Doctor ENT diagnosis system has important contributions for research and practice.

The major contribution of Virtual Doctor - ENT diagnosis system is to the healthcare domain. As the research on computerized diagnosis of ENT disease is limited, this study has an important contribution for ENT sub-specialty. Virtual Doctor - ENT diagnosis system can help general physicians in the diagnosis of ENT diseases in the absence of ENT expert. Thus, it can help in short listing the critical patients who can be referred to ENT specialists and reduce the backlog created due to busy schedules of limited accessibility of ENT experts. Hence our Virtual Doctor-ENT diagnosis can improve the efficiency, effectiveness of the healthcare system and improve the quality of patient care. With effective extensions, 'Virtual Doctor' can also be used in ENT education to train healthcare professionals and medical students to get a better understanding on ENT diseases and diagnosis processes. Besides this artifact (Virtual Doctor – ENT diagnosis system) has important implication for Design Science paradigm. This artifact is created to solve the problem faced by the healthcare community due lack of ENT experts. We demonstrate this artifact through experiment with real users of the system.

Despite the several contributions, Virtual Doctor has its limitations too. Even though the Virtual Doctor is designed mimic the diagnosis approach of an ENT specialist, certain aspects were not feasible to implement. First, complete expertise of an expert is difficult to extract and encode, because the knowledge of the specialist cannot be fully explained. Therefore, sometimes the required knowledge may not be available through the system. Second, ENT experts may sometimes use common sense, which cannot be programmed. Third, our system may also have problems in recognizing issues outside the knowledge domain. However, we overcome these limitations by referring the patient to consult real ENT specialists when the physician is unable diagnose the disease with the aid of 'virtual doctor' or by his/her medical knowledge. Fourth, the expert system have limited sensory experience compared to human ENT specialist, however our system try to overcome this issue to some extent by using image processing techniques that would process the webcam images of patients with abnormal conditions(e.g. swollen eye) and map it to the given patterns of normal and abnormal conditions. Besides, physicians can also record their observations manually, if the system is unable to recognize the patterns or image.

7 Conclusion

In this paper, we described the development of a rule based ENT diseases diagnosis expert system the 'Virtual Doctor', which can be used by physicians in their daily practice. The proposed system closely follows the steps followed by ENT specialist in diagnosing ENT diseases, therefore it is highly likely that this system would be accepted and adopted by physicians to assist them in the ENT disease diagnosis. However, extensive field trials need to be carried out in order to overcome the legal and ethical concerns, in order to incorporate such tools into healthcare systems.

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