

## **A REVIEW OF KNOWLEDGE BASED SYSTEMS IN MEDICAL DIAGNOSIS**

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Knowledge based system incorporates the expert knowledge that has been coded into facts, rules, heuristics and procedures. The power of expert system resides in the specific, high-quality knowledge about task domain. Knowledge is stored in a knowledge base where it is possible to add new knowledge or refine the existing knowledge without recompiling the control and inference programs. Sophisticated expert systems can be enhanced with additions to the knowledge base or to the set of rules. Medical diagnosis is one of the first knowledge based area to which Expert System technology was applied. The primary objective of the paper is to survey the research work accomplished in the field of medical sector.

**Keywords :** Knowledge, Knowledge Based Systems, Rule Based System, Expert Systems, Inference Engine, Knowledge Engineer.

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### **1. INTRODUCTION**

A knowledge is an information that can be used in decision making process. A knowledge base uses knowledge representation formalism to capture the subject matter experts knowledge and codifying it according to the formalism which is called the knowledge engineering. A knowledge based system uses a knowledge base containing accumulated experience and a set of rules for applying the knowledge to a particular situation. Knowledge based system /expert system have been developed in the last decade for many different applications by adopting artificial intelligence techniques. The knowledge based system in medical diagnosis was started in 70's and its usage actually started in 80's. In the last decade it is observed that the use of Knowledge Based System is embedded with other technologies.

#### **1.1. Statement of the Problem**

To undertake a brief survey of the research work done in Knowledge Based System /Rule Based Expert Systems in the field of medical diagnosis.

#### **1.2. Purpose of the Study**

The goal of the study is to conduct a brief survey of the research work in the area of knowledge based system in medical diagnosis based in the past two decades. This study has been undertaken keeping in the mind the research work to be undertaken by the authors of this paper.

### **1.3 Significance of the Study**

It is difficult to develop a system which is capable of duplicating the diagnosis process of a medical expert which depends on the tacit knowledge and explicit knowledge. An attempt is made to develop a computer logic as decision support system or second opinion for medical diagnosis. For this purpose the researcher has undertaken the survey about the research work done in the area of Medical Diagnosis over past decades. This will help the researcher to create a new prototype of an expert system for medical diagnosis.

## **2. EXPERT SYSTEM**

An expert system also known as a knowledge based system, is a computer program that contains the knowledge and analytical skills of one or more human experts in a specific problem domain. Recent advances in the field of artificial intelligence have led to the emergence of expert systems, computational tools designed to capture and make available the knowledge of experts in a field. The goal of the designer of the expert system is to capture the knowledge of a human expert relative to some specific domain and code this in a computer in such a way that the knowledge of the expert is available to a less experienced user. The various definitions of expert system are available in the literature. from a general point of view.

### **2.1 Definitions**

The British Computer Society's Committee of the specialist group on Expert Systems has framed the following definition [1] : "The embodiment within a computer of a knowledge based component from an expert skill in such a form that the machine can offer intelligent advice or take an intelligent decision about a processing function. A desirable additional characteristic which many would regard fundamental, is the

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capability of the system on demand to justify its own line of reasoning in a manner directly intelligible to the inquirer. The style adopted to attain these characteristics is rule-based programming.”

A precise and general definition of an Expert System is [2] : An expert system is a computer program that simulates the judgement and behaviour of a human or an organization that has expert knowledge and experience in a particular field.” Typically, such a system contains a knowledge base containing accumulated experience and a set of rules for applying the knowledge base to each particular situation that is described to the program. Sophisticated expert systems can be enhanced with additions to the knowledge base or to the set of rules.

Expert system seeks and utilizes relevant information from their human users and from available knowledge bases in order to make recommendations. With the expert system, the user can interact with a computer to solve a certain problem. This can occur because the expert system can store heuristic knowledge. The area of human intellectual endeavor to be captured in an expert system is called the *task domain*. *Task* refers to some goal-oriented, problem-solving activity. *Domain* refers to the area within which the task is being performed. Typical tasks are diagnosis, planning, scheduling, configuration and design.

### 2.1 Basic Components of Expert System Shell

An expert system tool, or shell, is a software development environment containing the basic components of expert systems. Associated with a shell is a prescribed method for building applications by configuring and instantiating these components. Some of the generic components of a shell are shown in Figure 1.1 and described below. The core components of expert systems are the knowledge base and the reasoning engine.

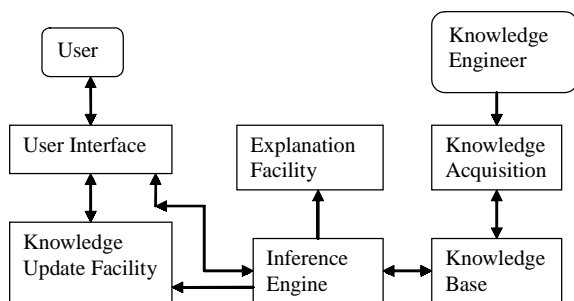


Figure 1: Architecture of Expert System

**Knowledge Base:** It is a warehouse of the domain specific knowledge captured from the human expert via the knowledge acquisition module. To represent the knowledge production rules, frames, logic, semantic net etc. is used. An Expert System tool provides one or more knowledge representation schemes for expressing knowledge about the

application domain. The knowledge base of expert system contain both factual and heuristic knowledge. Factual knowledge is that knowledge of the task domain that is widely shared, typically found in textbooks or journals. Heuristic knowledge is the less rigorous, more experiential, more judgmental knowledge of performance. In contrast to factual knowledge, heuristic knowledge is rarely discussed, and is largely individualistic. It is the knowledge of good practice, good judgment, and plausible reasoning in the field. It is the knowledge that underlies the “art of good guessing.”

**Inference Engine:** Inference Engine is used to perform the task of matching antecedents from the responses given by the users and firing rules. The major task of inference engine is to trace its way through a forest of rules to arrive at a conclusion. Here two approaches are used i.e. Forward chaining and Backward chaining. The inference mechanism can range from simple *modus ponens* backward chaining of IF-THEN rules to case-based reasoning.

**Knowledge Acquisition :** It is a subsystem which help experts to build knowledge bases. For knowledge acquisition, techniques used are protocol analysis, interviews, observation etc. Collecting knowledge needed to solve problems and build the knowledge base continues to be the biggest bottleneck in building expert systems.

**Explanation Facility:** A subsystem that explains the system’s actions. The explanation can range from how the final or intermediate solutions were arrived at to justifying the need for additional data. Here user would like to ask the basic questions why and how and serves as a tutor in sharing the system’s knowledge with the user.

**User interface:** It is a means of communication with the user. It provides facilities such as menus, graphical interface etc. to make the dialog user friendly. Responsibility of user interface is to convert the rules from its internal representation (which user may not understand) to the user understandable form.

To build the expert system personnel involved in expert system development are user, knowledge engineer, domain expert and system maintenance personnel. Knowledge engineer is involved in the development of the inference engine , structure of the knowledge base and user interface. Domain expert transfers the entire knowledge about the domain to the system.

## 3. RESEARCH WORK

### 3.1. To Build Medical Knowledge in Knowledge Based System

In healthcare sector computers are used to keep patient details, details of diseases, treatment given, rooms allotted, pathology report and billing. Computers are also used to keep details of hospital such as registration, appointment

scheduling, day care units, operation theatres, laboratories, radiology, pharmacy, inventory management, hospital resources, revenues/expenditures etc. Using Artificial Intelligence (AI) techniques, computers are able to give diagnosis of a specific disease called as medical expert systems. However the practical benefits of such automated reasoning systems have fallen short to give independent expert advice about the particular disease.

The failure of the efforts to create automated medical diagnosis has led to a reassessment and reclassification of research aim. The key objective has been transformed into the creation of knowledge based systems. The purpose of the knowledge based system is to act as a decision support system or as a second opinion for the doctors in critical cases. Knowledge based system is developed to incorporate medical knowledge and reasoning strategies into the automation of medical diagnosis. With the help of new approaches in AI which have recently emerged, may overcome some of the limitations inherent in earlier attempts to automate medical diagnosis system. It is possible to prepare knowledge based system for medical diagnosis to assist the junior doctors or doctors who are practising at remote places. The medical knowledge of specialized doctor is required for the development of an expert system.

### 3.2 A literature Study–Application of Knowledge Based Systems in Medical Diagnosis :

The past study shows that over the past two decades lot of work has been carried out in the development of Knowledge Based/ Rule Based Expert System in medical diagnosis in many countries. In order to know the previous research done in this direction, the authors examined several studies dedicated to this topic or to broader topics including references to knowledge based system. A literature survey was carried out to know about the Knowledge Based Systems in medical diagnosis since 1970.

It is observed that the research work in the knowledge based system can be categorized into the following types :

- Rule Based reasoning and its application in medical diagnosis.
- Case based reasoning in medical diagnosis.
- Use of hybrid model by combining rule based reasoning and case based reasoning.
- Fuzzy logic with rule based reasoning.
- The relationship between inferencing and the other technologies of hypertext and text retrieval.
- FEL-EXPERT is a family of diagnostic rule-based shells using a Prospector-like model for uncertainty handling.

In the subsequent part the authors present the study conducted in the field of medical diagnosis since 1974.

**1. INTERNIST–I (1974) [5] :** INTERNIST is one of the first clinical decision support systems developed by Pople and Myers at the University of Pittsburgh in 1974 for the diagnosis of complex diagnosis problems in general internal medicine. INTERNIST-I is a rule based expert system which uses patient observations to deduce a list of compatible state of disease based on a tree structure database that links diseases with symptoms. By the early 80s, it was recognized the most valuable product of the system which was used as the basis for successor systems including CADUCEUS and Quick Medical Reference (QMR), a commercialized diagnosis Decision Support System for internists.

**2. MYCIN [6] :** MYCIN was the first well known rule-based medical expert system developed by Shortliffe at Stanford University to help doctors, not expert in antimicrobial drugs, prescribe such drugs for blood infections. It was designed to aid physicians in the diagnosis and treatment of meningitis and bacteremia infections. MYCIN was the first large expert system to perform at the level of a human expert and to provide users with an explanation of its reasoning. Clinical knowledge in MYCIN is represented as a set of IF-THEN rules with certainty factors attached to the diagnosis. It was a goal –directed system , using a basic backward chaining reasoning strategy. MYCIN operated using a fairly simple inference engine, and a knowledge base of 600 rules. It would query the physician running the program via a long series of simple yes/no or textual questions. At the end, it provided a list of possible culprit bacteria ranked from high to low based on the probability of each diagnosis. Its confidence in each diagnosis probability, the reasoning behind each diagnosis (that is, MYCIN would also list the questions and rules which led it to rank a diagnosis a particular way), and its recommended course of drug treatment make it a successful expert system.

MYCIN has three sub-systems: Consultation system, Explanation System, Rule Acquisition system. In the 1980s, expert system “shells” were introduced, known as E-MYCIN and supported the development of expert systems in a wide variety of application areas.

**3. CADUCEUS [7] :** CADUCEUS is a medical expert system finished in the mid-1980s by Harry Pople building on Pople’s years of interviews with Dr. Jack Mayers, one of the top internal medicine diagnosticians and a professor at the University of Pittsburgh. Their motivation was an intent to improve on MYCIN which focused on blood-borne infectious bacteria to focus on more comprehensive issues than a narrow field like blood poisoning. Instead of embracing all internal medicine, CADUCEUS eventually could diagnose 1000 diseases. While CADUCEUS worked using an inference engine similar to MYCIN’s, it made a number of changes like incorporating abductive reasoning to deal

with the additional complexity of internal disease. There can be a number of simultaneous diseases, and data is generally flawed and scarce.

**4. QMR–Quick Medical Reference** [8] : QMR is developed out of INTERNIST-I, which is an in-depth information resource that helps physicians to diagnose adult diseases. It provides electronic access to more than 750 diseases representing the vast majority of the disorders seen by internists in daily practice as well a compendium of less common diseases. QMR uses more than 5,000 clinical findings to describe the features of diseases in the QMR knowledge base. Findings include medical history, symptoms, physical signs, and laboratory test results. QMR findings represent abnormal conditions, e.g., “Abdomen Pain Severe” or “Blood Hepatitis B Virus By Polymerase Chain Reaction.” Every disease profile included in the QMR knowledge base is the result of an extensive review of the primary medical literature. Consultation with experts is used to resolve any inconsistencies or deficiencies found in published reports. QMR is used in hospital and office practice.

**5. PUFF–Pulmonary Function System** [9]: PUFF expert system diagnoses the results of pulmonary function tests. PUFF was initially developed on the SUMEX computer, using EMYSCIN and was later rewritten in a production version to run on the hospital’s own minicomputer. Its task is to interpret set of pulmonary function (PF) test results, like volume of the lungs, the ability of the patient to move air into and out of the lungs, and the ability of the lungs to get oxygen into the blood and carbon dioxide out, then it produces a set of interpretation statements and a diagnosis for the patient. Its’ Purpose is to generate reports from a set of interpretation statements for pulmonary function diagnosis and saving the staff a great deal of tedious work. The staff themselves would not be displaced by this tool because their expertise still would be necessary to verify PUFF’s output, to handle unexpected complex cases, and to correct interpretations that they felt were inaccurate prior to printing the final report for physician signature and entry into the patient record. Approximately 85% of the reports generated are accepted without modifications. PUFF’s performance is good enough that it is used daily in clinical service.

**6. ATHENA** [10] : ATHENA is developed by Stanford Medical Informatics, VA Palo Alto Health Care System, and Stanford Center for Primary Care and Outcomes Research.

The ATHENA Decision Support System (DSS) implements guidelines for hypertension.

ATHENA DSS encourages blood pressure control and recommends guideline-concordant choice of drug therapy in relation to comorbid diseases. ATHENA DSS has an easily modifiable knowledge base that specifies eligibility criteria, risk stratification, blood pressure targets, relevant

comorbid diseases, guideline-recommended drug classes for patients with comorbid disease, preferred drugs within each drug class, and clinical messages. Because evidence for best management of hypertension evolves continually, ATHENA DSS is designed to allow clinical experts to customize the knowledge base to incorporate new evidence or to reflect local interpretations of guideline ambiguities. Together with its database mediator, Athenaem, ATHENA DSS has physical and logical data independence from the legacy Computerized Patient Record System (CPRS) supplying the patient data, so it can be integrated into a variety of electronic medical record systems.

To assess the quality of ATHENA DSS, researchers monitored real-time clinician feedback during point-of-care use of the system. Comments ( $n = 835$ ) were submitted by 44 of the 91 (48.4 percent) study clinicians (median 8.5 comments/clinician). Twenty-three (2.8 percent) comments identified important, rarely occurring problems. Timely analysis of such feedback revealed omissions of medications, diagnoses, and adverse drug reactions due to rare events in data extraction and conversion from the electronic health record. Analysis of clinician-user feedback facilitated rapid detection and correction of such errors. Based on this experience, new technologies for improving patient safety should include mechanisms for post-fielding QA testing.

**7. CEMS** [11] : CEMS is the mental health decision support system. It is developed by the Institute of Living (the Mental Health Network of Hartford Hospital), Hartford, Connecticut, USA. CEMS supports input clinical functions like evaluation, diagnosis, treatment, outcome assessment by allowing queries (e.g., of treatment protocols) and by monitoring data in the central medical database for adherence to more than 100 practice guidelines. Alert messages are sent to clinicians when a practice does not follow the guideline, and a response is generated by changing the not-standard practice e.g., change in diagnosis or ordering a required laboratory test or completing an on-line form to document the rationale for the deviation from the standard. The central database contains all diagnoses, the diagnostic criteria selected to support each diagnosis, all laboratory orders and results, all pharmacy orders (current and historical), and selected demographic and payment data.

**8. ERA–Early Referrals Application** [12] : ERA is a web-based decision support and cancer referrals system developed by Advanced Computation Laboratory, Cancer Research UK, London in collaboration with Infer Med Ltd., London. ERA is an interactive decision support tool to support General Practitioners in identifying those patients with suspected cancer that should be referred under the 2-week standard outlined by the Department of Health’s “Referral Guidelines for Suspected Cancer”. The web pages are clear and concise as the user must read and comprehend their content within the environment of the consultation. It



also ensures that minimal time is spent interacting with ERA rather than the patient.

**9. GIDEON–Global Infectious Disease and Epidemiology Network [13]** : GIDEON is developed by the gideononline.com for diagnosis of infectious diseases, tropical diseases, epidemiology, microbiology and antimicrobial chemotherapy. The system generates a Bayesian ranked differential diagnosis based on signs, symptoms, laboratory tests, country of origin, and incubation period and can be used for diagnostic support and simulation of all infectious diseases in all countries. A trial is done on 495 patients, the correct diagnosis was included in the differential diagnosis list in 94.7% of cases (sensitivity) and displayed as the first disease in the list in 75% (specificity). The data in GIDEON are derived from all peer-reviewed journals in the fields of infectious diseases, pediatrics, internal medicine, tropical medicine, travel medicine, antimicrobial pharmacology, and clinical microbiology; a monthly electronic literature search based on all relevant terms in GIDEON (e.g., diseases, drugs, etc.) all available health ministry reports (both printed and electronic); standard texts; and abstracts of major meetings.”

The database incorporates 327 diseases, 205 countries, 806 bacterial taxa and 185 antibacterial agents.

**10. PERFEX–Knowledge Based Interpretation of Myocardial SPECT Imagery [10]** : Myocardial perfusion imaging represents the most widespread procedure for assessing infarction and/or ischemia. The PERFEX project has as overall objective of developing a clinically useful, computer-based methodology to aid in the diagnosis of heart disease. The methodology combines well established mathematical methods, visualization techniques, and artificial intelligence approaches for representing medical knowledge and integrating visual, numeric, textual, and temporal information. This project has the following specific aims:

- i) Automatic Determination of the orientation of the left ventricular myocardium; This is being presented as a separate sub-project, called DISHA.
- ii) Extension and enhancement of a knowledge base for interpreting myocardial perfusion imagery and other relevant information.
- iii) Prediction of resting perfusion from resting thickening distributions through connectionist methods .
- iv) Integration of connectionist and symbolic methods;
- v) Implementation and automation of the methodology into a fully integrated system.

But interpreting the image information in combination with related clinical data remains a difficult and ill-defined problem.

**11. Iliad [12]** : It is the expert system for internal medical diagnosis at the University of Utah School of Medicine’s Dept. of Medical Informatics, has been under development for several years. Iliad uses Bayesian reasoning to calculate the posterior probabilities of various diagnoses under consideration, given the findings present in a case. Iliad was developed primarily for diagnosis in Internal Medicine, now covers about 1500 diagnoses in this domain, based on several thousand findings. The Iliad shell has also been used to develop knowledge bases for diagnosis in other domains. Iliad was developed initially for the Apple Mac; a version for the PC-AT running windows has also been released. Current use: Iliad is used as a teaching tool for medical students. Particular cases can be simulated through’ this program and the students have to diagnose the case. This helps the students to sharpen their skill in differential diagnosis. Iliad can prove useful to a health care provider as a personal consultant and can suggest relevant diagnoses, give advice regarding cost-effective workup strategies, and explain relationships of findings to diseases.

**12. Isabel [14]** : Isabel is a web-based diagnosis decision support system created in 2001 by physicians to offer diagnosis decision support at the point of care. Isabel has been extensively validated and been shown to enhance clinician’s cognitive skills and to improve patient safety and the quality of patient care. Isabel covers all ages (neonates to geriatrics) and all major specialties and sub-specialties in Internal Medicine, Surgery, Gynecology & Obstetrics, Pediatrics, Geriatrics, Oncology, Toxicology and Bioterrorism. Isabel is fast and easy to use & gives the clinician an instant list of likely diagnoses for a given set of clinical features like symptoms, signs, results of tests and investigations etc. Following on from history taking and clinical examination, Isabel assists the provider by reconciling patient data sets with data sets as described in established medical literature (textbooks and journals). Isabel has been interfaced with electronic medical record systems [EMR]. It helps to answer clinical questions with up to date knowledge from textbooks and journals containing 11000 diagnosis and 4000 drugs and heuristics.

**13. LISA [15]** : LISA is a clinical information and decision support system for collaborative care in childhood acute lymphoblastic leukaemia. A centralised Oracle database holds all patient information (drug schedules, blood and toxicity results, doses prescribed etc.) and accessible by health professionals from different sectors in different locations. Acute Lymphoblastic Leukaemia (ALL) is the most common paediatric malignancy. A web-based decision support module, implemented using PROforma guideline development technology, designed to provide advice about dose adjustments in the treatment of acute childhood lymphoblastic leukaemia. LISA is primarily concerned with providing support in the maintenance period during which drug dose decisions have to be made weekly.

**14. Expert System for the Diagnosis of neonatal jaundice for use by medical field personnel [16] :** The main objective of this project is to develop an expert system module that uses the clinical decision criteria of experts in the neonatology field to advice paramedical and semi-skilled personnel who require guidance to diagnose the etiology of neonatal jaundice. The project was developed with a commercially available software package EXSYS (Multilogic Inc.). A rule-based system is used to build the decision tree that would aid the paramedical personnel to arrive at a appropriate etiology for hyperbilirubinemia in a neonate. The project is field tested, where the residents are asked to come up with the correct diagnosis given the clinical tests.

**Note:** The disadvantage of this model is that it becomes obsolete in a few years time unless it is constantly being updated by developers.

**15. MEDUSA [17] :** MEDUSA is a fuzzy expert system for medical diagnosis of acute abdominal pain. Even today, the diagnosis of acute abdominal pain represents a serious clinical problem. The medical knowledge in this field is characterized by uncertainty, imprecision and vagueness. This situation lends itself especially to be solved by the application of fuzzy logic. A fuzzy logic-based expert system for diagnostic decision support is presented in MEDUSA. The representation and application of uncertain and imprecise knowledge is realized by fuzzy sets and fuzzy relations. The hybrid concept of the system enables the integration of rule-based, heuristic and case-based reasoning on the basis of imprecise information. The central idea of the integration is to use case-based reasoning for the management of special cases, and rule-based reasoning for the representation of normal cases. The heuristic principle is ideally suited for making uncertain, hypothetical inferences on the basis of fuzzy data and fuzzy relations.

#### 4. FUTURE RESEARCH ACTIVITY

The evolving paradigm shift resulting from IT, social and technological changes has created a need for developing an innovative knowledge based healthcare system which can effectively meet global healthcare system demands. It has been observed that since expert's knowledge is not stored in repository, so when expert become retired, his tacit knowledge cannot be utilized for future period for the treatment of the diabetic patients. The researcher has prepared a knowledge based system for the diagnosis of type of diabetes and its complications depending on symptoms, physical signs and investigation tests. The researcher has prepared a paper "A Knowledge Based System in Healthcare with Special Reference to Diabetes" which is going to be published in "**International Journal of Intelligent Information Processing**" June 2009 issue of the journal. The system will act as a decision support system or second

opinion for physicians in critical cases. This system will guide to the junior doctors or doctors who are practising at remote places for the treatment of a diabetic patient for acute/ chronic complications. The system will also act as a tutor for a layman. The researcher is trying to implement the Expert System in an Expert System Shell such as CLIPS or JESS.

#### 5. CONCLUSION

The views expressed above are statements which convey that medical knowledge for diagnosis purpose can be expressed in form of the rules in the knowledge Based System. The authors of this paper conclude that the knowledge based systems have been extensively used in the medical diagnosis. Medical expert systems will begin to appear, and researchers in medical artificial intelligence continue to make a progress in key areas such as knowledge acquisition, model-based reasoning and system integration for clinical environments.

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