Expert Systems in Medical Diagnosis

K.I. Nkuma-Udah[1], G.O. Onwodi[2], D. Njoku[3], G.I.N. Ndubuka[1] and F.B. Osang[2]

[1]Department of Biomedical Technology, Federal University of Technology, Owerri, Nigeria
[2]Department of Computer Science, National Open University of Nigeria, Lagos, Nigeria
[3]Department of Computer Science, Imo State Polytechnic, Umuagwo, Imo State, Nigeria

(Received August 22, 2013; Revised October 21, 2013; Accepted October 29, 2013)

Clinicians all over the world use several sources of data through a series of algorithms to make a diagnostic impression. This is aimed at arriving at an appropriate treatment decision. The cost of healthcare globally is high. So bringing intelligent healthcare informatics to bear on the dual problems of reducing healthcare costs and improving quality and outcomes is a challenge even in countries with a reasonably developed technology infrastructure. This research sets out to review expert systems in medical diagnosis, *ESMD*. ESMD is usually designed to enable the clinicians to identify diseases and describe methods of treatment to be carried out taking into account the user capability. Many formats use the C Language Integrated Production System (CLIPS) as the tool for use to design the ESMD. In the system, a number of patient cases is selected as prototypes and stored in a separate database. The knowledge is acquired from literature review and human experts of the specific domain and is used as a base for analysis, diagnosis and recommendations.

Key words: Developing, Affordability, Appropriateness, Expert System, Medical Diagnosis, Developing Countries, Artificial Intelligence

1. Introduction

Medical diagnosis is a complicated cognitive clinical process requiring high level of expertise. A clinician uses several sources of data through a series of algorithms to make a diagnostic impression. The whole aim of medical diagnosis is to arrive at an appropriate treatment decision, which in turn leads to a good prognosis for the particular ailment or disease. Therefore, any mis-diagnosis will lead to a wrong treatment and by extension an addition to the cost of medical or healthcare.

This research sets out to review expert system in medical diagnosis ESMD. The ESMD is usually designed to enable the clinicians to identify diseases and describe methods of treatment to be carried out taking into account the user capability. It uses inference rules and plays an important role that will provide certain methods of diagnosis for treatment.

TheCLanguage Integrated Production Syste m (CLIPS) is the tool mainly used to design the ESMD. In the system, a number of patient cases will be selected as prototypes and stored in a separate database. The knowledge is acquired from literature review and human experts of the specific domain and is used as a base for analysis, diagnosis and recommendations. Knowledge is represented via an integrated formalism that combines production rules and a neural network. This results in better representation, and facilitates knowledge acquisition and maintenance. Diagnosis is performed via the ES, based on patient data. The proposed system will be experimented on various scenarios in order to evaluate its performance.

2 Expert Systems and Artificial Intelligence

An *expert system* is a computer system that emulates

*Corresponding Author email: E-mail: drkinkumaudah@yahoo.com

ABC Publishers Inc

the decision making ability of a human expert [1]. By so doing, it acts in all respects like a human expert. It uses human knowledge to solve problems that would require human intelligence. The expert system represents an expertise as data or rule within the computer. These rules and data can be called upon when needed to solve problems. Expert System is itself a major aspect of *artificial intelligence,* which is the branch of computer science concerned with making computers behave like humans [2], i.e., concerned with automation of intelligent behavior.

2.1 Artificial Intelligence

Artificial intelligence (AI) is the intelligence of machines and the branch of computer science that aims to create it. In other words, AI is the branch of computer science concerned with making computers behave like humans [3][4][5]. The field of artificial intelligence attempts to understand intelligent entities. Thus, one reason to study it is to learn more about ourselves. But unlike philosophy and psychology, which are also concerned with intelligence, AI strives to *build* intelligent entities as well as understand them [6].

There are four ways in which AI can be defined. These are as a system that: thinks like humans; acts like humans; thinks rationally; and acts rationally. In thinking like humans, AI is "The exciting new effort to make computers think . . . *machines with minds,* in the full and literal sense" [7]. In acting like humans, AI is "The art of creating machines that perform functions that require intelligence when performed by people" [8]. Thinking rationally, AI is "The study of the computations that make it possible to perceive, reason, and act" [9]. Then acting rationally, AI is "A field of study that seeks to explain and emulate intelligent behavior in terms of computational processes" [10].

Common areas of applications of Artificial Intelligence include the following [11]: Computer Vision, Expert systems, Game Playing, Speech recognition, Natural Language Processing and Robotics. Computer vision is the artificial intelligence field that includes methods for acquirng, processing, analyzing, and understanding images and, in general, high-dimensional data from the real world in order to produce numerical or symbolic information, *e.g.*, in the forms of decisions.[12][13][14]. The world is composed of three-dimensional objects, but the inputs to the human eye and computers' TV cameras are two dimensional. Some useful programs can work solely in two dimensions, but full computer vision requires partial three-dimensional information that is not just a set of two-dimensional views. At present there are only limited ways of representing three-dimensional information directly, and they are not as good as what humans evidently use. [15]

Expert system as an application area of artificial intelligence is a computer system that emulates the decision-making ability of a human expert.[16] It is designed to solve complex problems by reasoning about knowledge, like an expert, and not by following the procedure of a developer as is the case in conventional programming [17]. Expert systems were one of the first applications to emerge from initial research in artificial intelligence, and the explanation of expert system reasoning was one of the first applications of natural language generation. Expert Systems attempt to capture the knowledge of a human expert and make it available through a computer program. There have been many successful and economically valuable applications of expert systems. Expert systems provide benefits such as reducing skill level needed to operate complex devices, diagnostic advice for device repair, interpretation of complex data, 'cloning' of scarce expertise, capturing knowledge of expert who is about to retire, combining knowledge of multiple experts and intelligent training [18].

Artificial intelligence game playing refers to techniques used in computer games to produce the illusion of intelligence in the behaviour of non-player character (NPCs). Emphasis of game playing in AI is on developing rational agents to match or exceed human performance.

In artificial intelligent, games are interactive computer program, made by creating human level artificially intelligent entities, eg. enemies, partners and support characters that act just like humans [19]. Games are good vehicles for research because they are well formalized, small, and self-contained. They are therefore easily programmed and can be good

models of competitive situations. So principles discovered in game-playing programs may be applicable to practical problems.

Speech recognition in artificial intelligence is the translation of spoken words into text [20]. In speech recognition, the user speaks to the computer through a microphone, which in turn, identifies the meaning of the words and sends it to natural language processing (NLP) device for further processing. Once recognized, the words can be used in a variety of applications like display, robotics, commands to computers, and dictation. Some speech recognition systems use "speaker independent speech recognition" while others use "training" where an individual speaker reads sections of text into the SR system [21]. These systems analyze the person's specific voice and use it to fine tune the recognition of that person's speech, resulting in more accurate transcription. Systems that do not use training are called "speaker independent" systems. Systems that use training are called "speaker dependent" systems. Speech recognition applications include voice user interfaces such as voice dialing, call routing, domestic appliance control, search, simple data entry, preparation of structured documents, and aircraft.

Natural Language Processing, NPL is the area of application of artificial intelligence focused on developing systems that allow computers to communicate with people using everyday language [22]. NPL does automated generation and understanding of natural human languages. In natural language generation system, it converts information from computer databases into normal-sounding human language, while natural language understanding system converts samples of human language into more formal representations that are easier for computer programs to manipulate [23].

Artificial Intelligence robot is a mechanical creature which can function autonomously. “Intelligent” here implies that the robot does not do things in a mindless, repetitive way; it is the opposite of the connotation from factory automation. The “mechanical creature” portion of the definition is an acknowledgment of the fact that our scientific technology usually use mechanical building blocks, not biological components. It also emphasizes that a robot is not the same as a computer. A robot may use a computer as a building block, equivalent to a nervous system or brain, but the robot is able to interact with its world: move around, change it, etc. A computer doesn’t move around under its own power. “Function autonomously” indicates that the robot can operate, self-contained, under all reasonable conditions without requiring recourse to a human operator. Autonomy means that a robot can adapt to changes in its environment (the lights get turned off) or itself (a part breaks) and continue to reach its goal [24].

Artificial Intelligence may be subdivided into two main branches or aspects. The first branch, *cognitive science*, has a strong affiliation with psychology. The goal is to construct programs for testing theories that describe and explain human intelligence. The second branch, *machine intelligence*, is more computer science oriented and studies how to make computers behave intelligently. Expert systems fall in the later branch [25].

2.2 Expert Systems

The idea of expert systems came from the fact that human experts are able to solve problems at a high level because they exploit knowledge about their area of expertise [26]. Therefore, the design of programs with *expert-based* problem solving capabilities that use specific knowledge about a specialty area in order to obtain competence comparable to that of a human expert became an innovative. The specific knowledge may be accumulated in form of a database obtained by interviewing one or more experts in the area in question.

Expert systems were the first successful and largest area of applications of Artificial Intelligence to real-world problems solving problems in medicine, chemistry, finance and even in space (examples, space shuttle, robots on other planets) [27]. It is recently widely applied in medical diagnosis and practice. The expert system seeks to exploit the specialised skills or information held by of a group of people on specific areas. It can be thought of as a computerized consulting service. It can also be called an information guidance system. Such sys-

tems are used for prospecting medical diagnosis or as educational aids. They are also used in engineering and manufacture in the control of robots where they inter-relate with vision systems.

An expert system is usually a computer program which performs complex data processing similar to evaluation made by a human expert. The program is able to draw conclusions and make decisions, based on knowledge, represented as a database, it has. An expert system doesn't have to be a replacement for a human expert. Such systems are often used as a support when a human cannot collect all vital information due to their amount or complexity. There are two reasons for this: one, there is a need for systems that work in real-time and perform their functions faster and better then a human is able to do. Two, computer programs are much more cheaper than human experts, especially in terms of maintenance, costs of education, salaries etc. If there is a way to duplicate a part of knowledge a human expert has, it is economical to do that using a computer program [28].

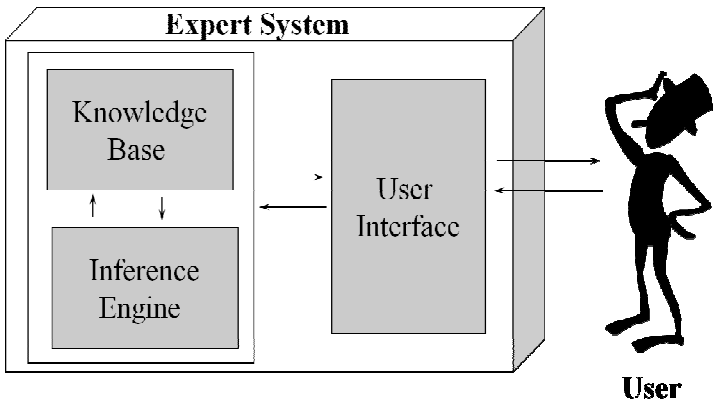

Figure 1: Expert System Architecture

Expert systems usually consists of two core parts in its kernel: a knowledge base and an inference engine [28]. A knowledge base represents a knowledge in certain domain and stores all the facts and rules about a particular problem in that domain. An inference engine is a set of algorithms, which perform judgment and reasoning. For a user to interact with the expert system, there has to be an additional part: the user interface, which is a means of communication between a user and the expert system problem-solving processes. Therefore, the entire expert system architecture in the most simplified version consists of three parts: a knowledge base, an inference engine and a user interface (figure 1).

3. Expert Systems and Medical Diagnosis

3.1 Medical Diagnosis and Care

Patients seek medical help for determination (or diagnosis) and treatment of various health problems [29]. Sometimes a combination of the patient's history and a clinical examination by a physician are enough to make diagnoses and decide whether medical treatment is needed, and what treatment should be given. However, often laboratory investigations or diagnostic imaging procedures are required to confirm a clinically suspected diagnosis or to obtain more accurate information. For example, malaria may be suspected by the presence of fever and by excluding other causes of fever on history and physical examination, but a firm diagnosis is made on microscopic examination of a blood slide.

Medical diagnosis or the actual process of making a diagnosis is a cognitive process. It is the pivotal cognitive activity of a practicing physician [30]. A physician uses several sources of data and puts the pieces of the puzzle together to make a diagnostic impression. The initial diagnostic impression can be a broad term describing a category of diseases instead of a specific disease or condition. After the initial diagnostic impression, the physician obtains follow up tests and procedures to get more data to support or reject the original diagnosis and attempts to narrow it down to a more specific level. Diagnostic procedures are the specific tools that the physicians use to narrow the diagnostic possibilities, which lead closer to correct diagnosis.

Correct diagnosis usually leads to appropriate treatment. With the high cost of health care, increased patient awareness, medico-legal and insurance pressures, every physician is expected to be responsibly accountable in patient care. Medical diagnosis must always be a logical, dependable and indeed cost effective system of medical care delivery. Investigations and treatment must be justifiable on the basis of the patients situational reality from the diagnosis rather than to compensate for the phy-

sicians diagnostic error or deficiencies. Every physician is therefore left to self audit for accountability of his/her actions or thoughts in the course of the medical care.

Generally four key factors influence clinical decision making following a medical diagnosis. These are quality, cost, ethics and legal concerns [31]. Indicators of the factor of quality are mainly the process and outcome of medical intervention. However, these depend on availability and accessibility of medical treatment and care. As a factor, patients and care payers are concerned about the costs of medical care, including the costs of diagnostic and laboratory testing, lengths of hospital stays, the costs of physician services, and the cost-effectiveness of various preventive diagnostic or treatment regimens.

Then, ethics on its own now play an important, although not dominant, role in decision making of primary medical care. In this regard, ethical considerations include what information to give patients, when to suggest alternate services because of a patient's inability to pay, and the appropriateness of various types and forms of treatment. On legal concerns, since 1980s the number of medical malpractice suits has increased with the amount of awards. This trend has so affected clinical decision making, that physicians now increasingly report that they practice 'defensive medicine', ordering tests and other procedures to protect themselves from liability rather than simply to provide the best care for their patients. This has tended to inflate medical care cost again.

3.2 Expert Systems in Medical Diagnosis

Since the whole aim of medical diagnosis is to arrive at an appropriate treatment decision, it will be worth the while for applications that improve medical diagnosis to be provided from time to time in order to also improve overall medical or health care quality. Besides, the cost of healthcare globally is high [32]. Therefore, bringing intelligent healthcare informatics to bear on the dual problems of reducing healthcare costs and improving quality and outcomes is a worthwhile venture, though a challenge even in countries with a reasonably developed technology infrastructure. This challenge is not insurmountable, though.

Moreover, medical diagnosis is a complicated process requiring high level of expertise [33]. So any attempt to develop an intelligent healthcare informatics or an expert system for medical diagnosis must be ready to confront the challenges and difficulties. These challenges include ensuring that the healthcare informatics is *appropriate* to the setting in which it's applied. That is why the design of the expert system for medical diagnosis here must be carefully implemented for the purposed.

3.2.1 CLIPS Expert System

One of the tools used to design the ESMD is the C Language Integrated Production System (CLIPS). CLIPS is an expert system tool originally developed by the Software Technology Branch (STB), NASA/Lyndon B. Johnson Space Center [34]. Since its first release in 1986, CLIPS has undergone continual refinement and improvement. It is designed to facilitate the development of software to model human knowledge or expertise [34][35][36]. CLIPS program is used widely by reason of the flexibility, portability, extensibility, capabilities and the low cost [37]. Knowledge can be represented in CLIPS in three ways: *rules*, which are primarily intended for heuristic knowledge based on experience; *deffunctions* and *generic functions*, which are primarily intended for procedural knowledge; and *object-oriented programming*, also primarily intended for procedural knowledge. CLIPS shall be used here in form of rules to design the ESMD.

The ESMD is designed to enable the clinicians to identify diseases and describe methods of treatment to be carried out taking into account the user capability. The expert system uses inference rules and plays an important role that will provide certain methods of diagnosis for treatment [38]. The proposed system is experimented on various scenarios in order to evaluate its performance

4.0 Research Methodology

Development methodologies of expert systems can be roughly classified into six categories [35]. These

are: Rule-based systems; Knowledge-based systems; Intelligent agent (IA); Database methodology; Inference engine; and System-user interaction. The expert system shell to be developed in this work shall consists of the user interface, the explanation system**,** the inference engine and the knowledge base editor. The structure of the expert system is as shown in Figure 2.

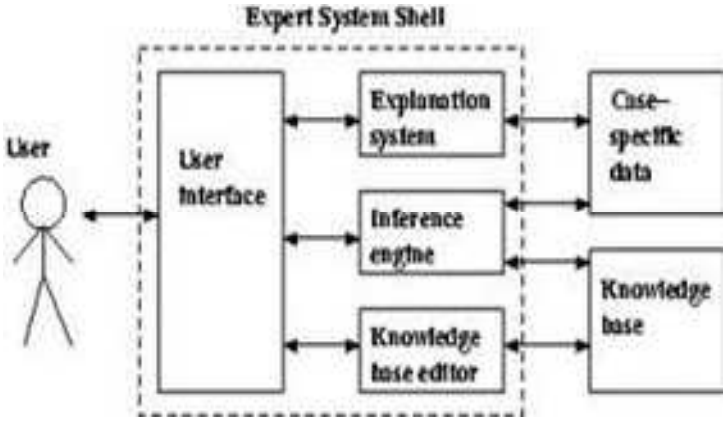

Figure 2: The Structure of the Expert System

Here, several human diseases shall be implemented by methodology of rule based systems. One of the well-known methods of representation of knowledge in the expert systems called the CLIPS shall be used. CLIPS (C Language Integrated Productive System) is a productive representation which keeps in memory a fact list, a rule list, and an agenda with activations of rules. Facts in CLIPS are simple expressions consisting of fields in parentheses [42]. Groups of facts in CLIPS, usually follow a fact-template, so that it will be easy to organize them and thus design simple rules that apply to them.

In the ESMD system designed using CLIPS, to accumulate factual knowledge, data is via a number of patient cases to be selected as prototypes and stored in a separate database. These data shall be collected concerning the association between sign and symptoms associated with patients [38]. Further knowledge shall be acquired from literature review and human experts of the specific domain and is used as a base for analysis, diagnosis and recommendations. Knowledge is represented via a production rule [39]. This facilitates knowledge acquisition and maintenance.

The sign, symptoms and test reports shall be the determining factor for diagnosis of a particular disease. These symptoms shall be organized in groups, which shall help in diagnosis. Each unit shall be provided with three groups of symptoms, say Key group (Kg), Sub group (Sg) and Unexpected (Ue). Kg represents a group of symptoms whose presence is necessary & sufficient to confirm the diseases where as the presence of Sg is not sufficient and it is a subset of Kg, while Ue is no match or symptom information is missing. Therefore, diagnosis is performed via the ESMD, based on patient data.

4. Conclusions

The CLIPS was used here as a case study for the ESMD. In the ESMD system using CLIPS, data from patient cases, literature review and specific domain human experts are used to accumulate factual knowledge. To facilitate diagnosis, the signs, symptoms and test reports, which shall be organized in groups, become the determining factor. The groups shall include Key group (Kg), a group of symptoms/ signs and test reports whose presence is necessary & sufficient to confirm the diseases; Sub group (Sg), a subset of Kg and a group of symptoms/signs and test reports whose presence is not sufficient to confirm the diseases; and Unexpected (Ue), a group in which there is no match or symptom/sign and test information are missing. Thus, diagnosis is eventually performed via the ESMD, based on patient data.

References

[1] Jackson & Peter, (1998) Introduction to Expert Systems, 3rd ed, Addison Wesley

[2] Harpreet K., et al., Artificial Intelligence: Bringng expert knowledge to computers, Discovery, 2012, 2(4), 4-7, http:// www.discovery.org.in/d.htm

[3] Bonn, Deutsche Gesellschaft fuer Luft- und Raumfahrt, 1986, p. 443-448

[4] Eric Tatro, Artificial Intelligence, Robotics, 2004, http://www.asimovlaws.com/articles/ archives/2004/07/why_we_need_fri_1.html

[5] Russell, Norvig P. Artificial Intelligence A Modern Approach, 2011

[6] Weyand J. IN: Yearbook 1986 II; DGLR, An-

nual Meeting, Munich, West Germany, Oct. 8-10, 1986, Reports, A87-48154 21-01

[7] Haugeland, J., editor (1985). Artificial Intelligence: The Very Idea. MIT Press, Cambridge, Massachusetts. In Russell, Norvig P. Artificial Intelligence A Modern Approach, 2011.

[8] Kurzweil, R. (1990). The Age of Intelligent Machines MIT Press, Cambridge, Massachusetts. In Russell, Norvig P. Artificial Intelligence A Modern Approach, 2011.

[9] (Winston, 1992) Winston, P. H. (1992). Artificial Intelligence. Addison-Wesley, Reading, Massachusetts, third edition. In Russell, Norvig P. Artificial Intelligence A Modern Approach, 2011.

[10] Schalkoff, R. I. (1990). Artificial Intelligence: An Engineering Approach. McGraw-Hill, New York. In Russell, Norvig P. Artificial Intelligence A Modern Approach, 2011.

[11]Nilsson N.J., 2010: The Quest for Artificial Intelligence, Cambridge University Press, UK

[12]Shapiro L.G. and Stockman G.C., 2001. Computer Vision. Prentice Hall. ISBN 0-13-030796-3

[13] Morris T., 2004. Computer Vision and Image Processing. Palgrave Macmillan. ISBN 0-333-99451-5.

[14]Jähne B. and Haußecker H., 2000. Computer Vision and Applications, A Guide for Students and Practitioners. Academic Press. ISBN 0-13-085198-1.

[15] Szeliski R., 2010, Computer Vision: Algorithms and Applications. Springer Inc

[16]Peter J., 1998, Introduction To Expert Systems (3 ed.), Addison Wesley, p. 2, ISBN 978-0-201-87686-4

[17] Barzilay R., McCullough D., Rambow O, DeCristofaro J., Korelsky T. and Lavoie B.: 1998 "A new approach to expert system explanations", CoGenTex, Inc

[18] Poole D., Mackworth A. and Goebel R., 1998: "Computational Intelligence: A Logical Approach", Oxford University Press, UK

[19] Luger G.F., 2002: Artificial Intelligence: Structures and Strategies for Complex Problem Solving. Addison-Wesley Publishing Company , USA

[20][20] Nilsson N.J, 1998: AI: A New Synthesis. Morgan Kaufmann Inc USA.

[21] Grant E. F. and Lardner R., 1952. "The Talk of the Town - It". The New Yorker.

[22] Mooney R.J., Artificial Intelligence: Natural Language Processing, University of Texas, Austin, USA accessed June 23, 2013.

[23]Dean T., 1994, Artificial Intelligence: Theory and Practice, Addison-Wesley Publishing Company , USA

[24] Murphy R.R. 2000. Introduction to AI Robotics, A Bradford Book The MIT Press Cambridge, Massachusetts, London, England.

[25] Helsgaun K. Ten Project Proposals in Artificial Intelligence. Roskilde University, Denmark. Assessed May 25, 2013

[26]Jayaraman V. and Srivastava R., 1996., Expert systems in production and operations management Current applications and future prospects, International Journal of Operations & Production Management, Vol. 16 No. 12, 1996, pp. 27-44. http://www.it.iitb.ac.in/~palwencha/ES/J_Papers/ES_APP_OPR.pdf

[27] Belavkin R.V, Introduction to Expert Systems , Lecture note 6, BIS4435, Middlesex University, London

[28] Melville S., Expert systems concept.., manuscript. AGH University of Science and Technology http://home.agh.edu.pl/~wojnicki/thesis/node6.html accessed date

[29] Carter J., Müller-Stöver I., Östensen H. and Claus Chr. Heuck C.C., 2005, Good Clinical Diagnostic Practice, WHO Regional Office for the Eastern Mediterranean, Cairo, Egypt

[30] Nkanginieme K.EO., 1997, Clinical diagnosis as a dynamic cognitive process: Application of Bloom's taxonomy for educational objectives in the from cognitive domain Med Educ Online [serial online] 1997;2:1. Available: URL http://www.utmb.edu/meo/.

[31] Leist JC. & Konen JC. Four factors of clinical decision making: a teaching model. Acad Med 1996; 71: 644-646.

[32]HealthCast: Global Best Practices in Bending the Cost Curve, 2012. www.pwc.com/btcc

[33] Hannan S.A.., Bhagile V.D., Manza R.R. and

R. J. Ramteke R.J., 2010. Development of an Expert System for Diagnosis and appropriate Medical Prescription of Heart Disease Using SVM and RBF, International Journal of Computer Science and Information Security, 8 (5)

[34]Giarranto J. C., 2007. CLIPS User Guide, Quicksilver Beta

[35] Naser S.S.A and Ola A.Z.A., 2008: An Expert System for Diagnosing Eye Diseases Using Clips, Journal of Theoretical and Applied Information Technology www.jatit.org/volumes/research-papers/Vol4No10/5Vol4No10.pdf

[36] Karagiannis S., Dounis A. I., Chalastras, T., Tiropanis P., and Papachristos D., 2007, Design of Expert System for Search Allergy and Selection of the Skin Tests using CLIPS, Journal of Information and Communication Engineering 3:6

[37] Giarranto J. C., 2007. CLIPS Reference Manual Volume I Basic Programming Guide.

[38] Patra P.S.K., Sahu D.P. and Mandal I., 2010, An Expert System for Diagnosis of Human Diseases, International Journal of Computer Applications Vol. 1 – No. 13

[39] Hatzilygeroudis I., Vassilakos P.J. and Tsakalidis A., 1994, An Intelligent Medical System for Diagnosis of Bone Diseases: 1st International Conference on Medical Physics and Biomedical Engineering (MPBE'94), Nicosia, Cyprus, May 1994, Vol. I, 148-152.

[40] Smitha V., 2010, An Expert System for Diabetes Diagnosis, an MPhil Thesis, Department of Computer Science, Christ University, Bangalore

[41] Eze A.O. and Opara F.K., 2012 An expert system (Autodoc) for diagnosing diseases and prescribing medication using visual basic.net, Information Technology Research Journal Vol .2(2)