Medical Informatics Europe '97
C. Pappas et al. (Eds.)
IOS Press, 1997

XBONE: A Hybrid Expert System for Supporting Diagnosis of Bone Diseases

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Abstract. In this paper, XBONE, a hybrid medical expert system that supports diagnosis of bone diseases is presented. Diagnosis is based on various patient data and is performed in two stages. In the early stage, diagnosis is based on demographic and clinical data of the patient, whereas in the late stage it is mainly based on nuclear medicine image data. Knowledge is represented via an integrated formalism that combines production rules and the Adaline artificial neural unit. Each condition of a rule is assigned a number, called its significance factor, representing its significance in drawing the conclusion of the rule. This results in better representation, reduction of the knowledge base size and gives the system learning capabilities.

1. Introduction

Expert systems are increasingly used to support medical diagnoses [1, 2, 3]. Diagnosis of bone diseases is greatly facilitated by the use of nuclear medicine methods, such as scintigrams (or scans), and a number of relevant expert systems have been developed [4, 5], which are based on a single representation scheme. Expert systems technology is moving towards hybrid representations [6, 7]. A promising integration is that of a symbolic representation, e.g. rules, with a connectionist one, i.e. various artificial neural networks.

In this paper, we present a hybrid medical expert system, called XBONE, which uses a hybrid representation formalism integrating rules and the adaline artificial neural unit. In section 2 the medical knowledge involved is presented. In section 3 the architecture of the system is discussed. Section 4 deals with the hybrid knowledge representation formalism, and finally Section 5 concludes.

2. Medical Knowledge

2.1 Patient Data

Patient data can be distinguished in three types: demograhic, clinical and nuclear medicine image (NMI) data. *Demographic data* concerns information such as patient's age, sex etc. *Clinical data* is further distinguished in physical findings and laboratory results. *Physical* *findings* are those detected by a physical examination of the patient, like the existence and the kind of a pain, called clinical *symptoms* as well. *Laboratory results* are those detected via laboratory tests, e.g. blood tests. Finally, *NMI data* is that extracted from scintigrams that depict the concentration of an administered radio-pharmaceutical (99m Tc-MDP) on patient's osseous tissue. Patient data are related to *domain knowledge*.

2.2 Diagnostic knowledge

Diagnostic knowledge concerns the way a diagnosis is performed. It is distinguished in two types. The first type, *procedural diagnostic knowledge*, reflects the diagnostic procedure. Diagnosis of bone diseases is considered a two-fold procedure. An initial diagnosis, called the *early diagnosis*, is made based on the demographic and clinical data of the patient. This is then used either to specify the kind of the scan needed (simple or 3-phase) or to be compared with the *late diagnosis*, which is based on the NMI data.

The second type of diagnostic knowledge, *heuristic diagnostic knowledge*, concerns experience and represents the way an expert uses patient data to make diagnoses. We acquired heuristic knowledge from an expert and constructed a *diagnostic tree* based on criteria such as the sex and the age of the patient, the existence and the acuteness of symptoms (e.g. pain, fever) etc., as far as non NMI data is concerned. As to the NMI data, criteria are related to the recognition of the *characteristic pattern* of the radio-pharmaceutical concentration, which is based on qualitative features (such as the uniformity of the concentration) and quantitative features (such as the extent of the concentration). Each pattern gives an indication for one of the following bone disease categories: metastases, hyperplasia of spinal cord, traumas, orthopedic abnormalities, arthrites, metabolic diseases, spinal cord diseases, Paget disease, benign tumors and malignant tumors.

3. System Architecture

The architecture of the system is illustrated in Fig.1. It consists of six main modules. *Patients database* (PDB) contains the demographic data and the scintigrams of the patients. Scintigrams are acquired by a γ-camera and then automatically transferred to the system [8]. *Hybrid knowledge base* (HKB) contains the domain and the heuristic diagnostic knowledge, represented as (neu)rules. *Working database* (WDB) contains the case-specific data, that is the (initial) patient data, partial conclusions and answers given by the user, represented as facts. *Hybrid inference engine* (HIE) realizes the diagnostic procedure and uses the available knowledge in HKB to draw conclusions. *Explanation mechanism* (EM) creates explanations when asked to do so. *Training mechanism* (TM) is used for rule training. Finally, *user interface* (UI) performs a number of functions related to user interaction with the system.

4. Knowledge Representation in XBONE

4.1 The Hybrid Formalism

We introduce *neurules* alongside conventional rules. Each neurule is considered as an adaline unit (Fig.2a,b). The inputs C_i, i=1,...,n of the unit are the conditions of the rule. Each condition C_i is assigned a number sf_i, called a *significance factor*, that represents the significance of the corresponding condition in drawing the conclusion.

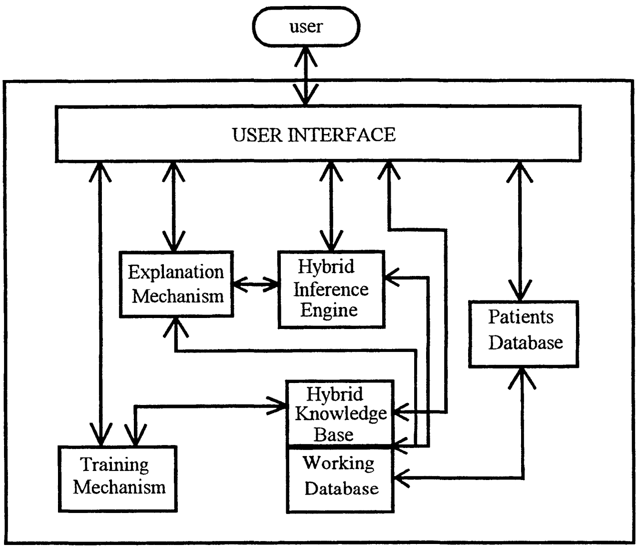

Fig.1 The Architecture of XBONE

Moreover, each rule itself is assigned a number sf_0, called the *bias factor*. Each input takes a value from the following set of discrete values: '1' if condition is *true*, '0' if it is *false* and '0.5' if its value is *unknown*. This gives the opportunity to easily distinguish between the falsity and the absence of a condition, in contrast to conventional rules. The output D, which represents the *conclusion* (decision) of the rule, is calculated as the weighted sum of the inputs filtered by a threshold function(see e.g. [9]):

$$D = f(\mathbf{a}), \quad \mathbf{a} = sf_0 + \sum_{i=1}^{n} sf_i \; C_i \tag{1}$$

where **a** is the *activation value* and *f(x)* the *activation (threshold) function* (Fig.2c). Hence, the output can be one of '-1' and '1', representing failure and success of the rule respectively.

The general syntax of a rule is the following:

<rule> ::= [(<bias-factor>)] **if** <conditions> **then** <conclusions>
<conditions> ::= <condition> {, <condition>}
<conclusions> ::= <conclusion> {, <conclusion>}

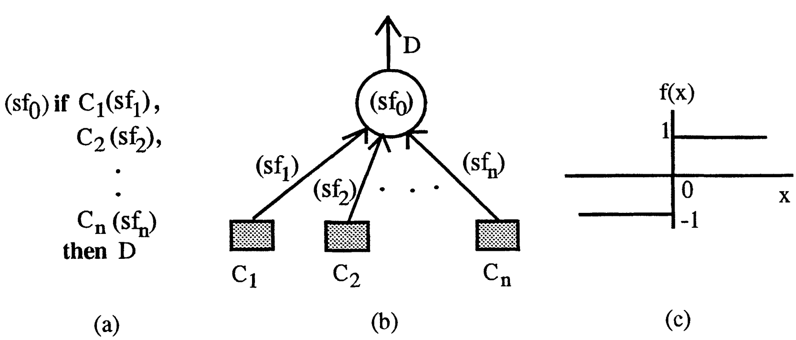

Fig.2 (a) A neurule (b) Corresponding adaline unit (c) Activation function

<condition> ::= <object> <l-operator> <value> [(<significance-factor>)]
<conclusion> ::= <object> <r-operator> <value>.
('[]' denotes optional occurrence, '{}' denotes zero, one or more occurrences of the enclosed expression and '<>' denotes a nonterminal symbol.)

<object> acts as a variable and represents a concept in the domain, e.g. "sex", "pain" etc. <l-operator> can be a symbolic (e.g. is, isnot) or a numeric (e.g. <, =, >) operator, whereas <r-operator> can be only "is". <value> denotes a value of <object>, numeric or symbolic. Finally, <bias-factor> and <significance-factor> are real numbers. Significance factors and the bias factor are optional in a rule. Thus, neurules (with factors) and conventional rules (without factors) may coexist in the knowledge base. (The terminal symbol "," in the case of a conventional rule denotes a conjunction).

The formalism also supports *variable declarations* that have the following syntax:

<variable-declaration> ::= <variable> **:** <multiplicity> **:** <value-domain>

and declare the types and the value domains of the variables.

<multiplicity> can be either "s" or "m" and denotes whether a variable is *single-valued* or *multi-valued* respectively. <value-domain> declares the possible values or the numeric type of a variable. Examples: "fever**:s:** (high, medium, low)", "symptom **:m:**(pain, fever)" and "age**:s:integer**".

Finally, the formalism supports *facts*. A fact has the same format as a conclusion of a rule. Facts represent either initial conditions or conclusions and are stored in WDB.

4.2 Hybrid Knowledge Base

HKB consists of the *domain knowledge base* (DKB) and the *hybrid rule base* (HRB). DKB contains domain knowledge, as variable declarations. HRB consists of two parts which contain knowledge concerning the early and the late diagnosis respectively. HRB may contain both conventional rules and neurules (Fig.3). Conventional rules are typically used to represent conclusions produced in a unique and exact way, in contrast to neurules, or conclusions that cannot be represented by a neurule (see Section 4.3). Thus, a neurule is actually a merger of more than one conventional rule. This greatly reduces the size of HRB.

4.3 Training neurules

The factors assigned to neurules are determined by TM. Each neurule is individually trained. To this end, a number of training patterns, called a *training set*, are supplied to TM for each rule. Training of the neurules takes place prior to the initial use of the system and every time the system is updated. The training sets are extracted from known (old) patient cases and/or the diagnostic tree. The standard least mean square (LMS) learning algorithm (see [9]) is used to determine the values of the factors.

However, there are cases where TM fails to find converging factors (case of non-separable functions, see [9]). This is an inherent weakness of the Adaline model. Then, conventional rules should be employed.

R1:
if sex **is** man ,
  age > 20 ,
  age < 36
then patient_class **is** man_21_35

R2:
(-8) **if** pain **is** continuous (5) ,
  patient_class **isnot** man_36_55 (2.5) ,
  fever **is** medium (2) ,
  fever **is** high (2)
then disease_type **is** inflammation

Fig.3 A conventional rule and a neurule

4.4 Inference Process

An inference process is performed in two stages, the *early diagnosis stage* and the *late diagnosis stage*. During the early diagnosis stage the first part of HRB is activated, the system asks questions about patient's clinical data and produces a first diagnosis. During the late diagnosis stage, the second part of HRB is activated. Activation of this part requires that a series of scintigrams of the patient be automatically loaded. Afterwards, the system asks questions about NMI data. Finally, it suggests a diagnosis that may or may not coincide with the first one. It is then up to the user-physician to decide on the final diagnosis. The inference mechanism is based on a backward chaining strategy.

5. Conclusions

In this paper, a hybrid medical expert system that supports diagnosis of bone diseases via scintigrams is presented. NMI data are extracted by the user-physician. Although there are systems using computer-based methods for NMI data extraction (e.g. [5]), image processing techniques are not very reliable and are not preferred (e.g. [4]). On the other hand, this makes participation of the user-physician more active.

Knowledge is represented via a formalism integrating rules and the adaline neural unit, to combine modularity and naturalness of rules with the representation and learning capabilities of neural units. This results in better representation, since one can easily represent imprecise relations, significantly reduces the size of the knowledge base, since each neurule is a merger of more than one conventional rule, and gives the system learning capabilities, since rules can be automatically updated.

A weak point of neurules is their inability to represent non-separable training patterns. To overcome this, a more complex (two layer) neural network is required. This, however, may make representation more complex, less comprehensible and modularity may be lost.

References

[1] B.N. Prasad, S.M. Finkelstein and M.I. Hertz, An expert system for diagnosis and therapy in lung transplantation, *Computers in Biology and Medicine* **26**(6) (1996) 477-488.

[2] S.D. Likothanassis, P. Adamidis and C. Giogios, Use of neural networks in medical expert systems, *Medical Informatics* **20**(4) (1995) 349-357.

[3] M. Fathi-Torbaghan and D. Meyer, MEDUSA: a fuzzy expert system for medical diagnosis of acute abdominal pain, *Methods of Information in Medicine*, **33**(5) (1994) 522-529.

[4] S.V. Ellam and M.N. Maisen, A Knowledge-based System to Assist in the Diagnosis of Thyroid Disease from a Radioisotope Scan, in Pretschner D.P. and Urrutia B. (Eds), Knowledge-based systems to aid medical image analysis, vol.1, Commission of the European Community, 1990.

[5] J.H.C. Reiber, G. Bloom, S.S. Gerbrands, E. Backer, H.J. van de Herik, A.E..M Reijs, I. van der Feltz and P. Fioretti, An Expert System Approach for the Objective Interpretation of Thallium-201 Scintigrams, in the same as [4].

[6] C.A. Huges, E.E. Gose and D.L. Roseman, Overcoming deficiencies of the rule-based medical expert system, *Computer Methods and Programs in Biomedicine* **32**(1) (1990) 63-71.

[7] B. D. Leao, E. B. Reategui, A. Guazzelli and E.A. Mendonca, Hybrid systems: a promising solution for better decision support tools, *Proceedings of the 8th World Conference on Medical Informatics* (1995) 823-827.

[8] I. Hatzilygeroudis, P.J. Vassilakos and A. Tsakalidis, An Intelligent Medical System for Diagnosis of Bone Diseases, *Proceedings of MPBE'94* (1994) 148-152.

[9] S.I. Gallant, Neural Network Learning and Expert Systems, MIT Press, 1993.