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XBONE: A Hybrid Expert System 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.

Keywords: hybrid expert systems, decision support systems, knowledge representation.

1. Introduction

Medical diagnosis is a very active field as far as introduction of artificial intelligence methods is concerned. Expert systems (ESs) are used to perform diagnoses based on patient data, as they can represent the way experts reason. Diagnosis of bone diseases is greatly facilitated by the use of nuclear medicine methods, more specifically of scintigraphic images (or scintigrams or scans), and a number of relevant expert systems have been developed [1, 7]. These systems, however, are based on a single representation scheme. Expert systems technology is moving towards hybrid representations, that is integrations of more than one representation scheme. A promising integration is that of a symbolic representation, e.g. frames or rules, with a connectionist one, i.e. various artificial neural networks (ANNs), [5, 6, 2].

In this paper, we present a hybrid medical expert system, called XBONE, which uses a hybrid representation formalism integrating rules and an 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 scintigraphic images acquired that depict the concentration of an administered radio-pharmaceutical (99m Tc-MDP) on patient's osseous tissue. Patient data are related to called *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 a two-fold procedure. An initial diagnosis, called *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 a *late diagnosis* based on the NMI data. This later diagnosis may or may not coincide with the initial.

The second type of diagnostic knowledge, *heuristic diagnostic knowledge*, represents experience accumulated through years and concerns the way an expert uses the patient data to make diagnoses. We acquired heuristic knowledge by interviewing experts in the field 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 in a scintigram, which is based on qualitative features, such as whether the concentration is uniform or not etc, and quantitative features, such as whether the concentration is normal, slightly increased etc. Each pattern gives an indication of one of the following involved bone disease categories: metastases, hyperplasia of spinal cord, traumas, orthopeadic abnormalities, arthrites, metabolic diseases, spinal cord diseases, Paget disease, benign tumors and malignant tumors.

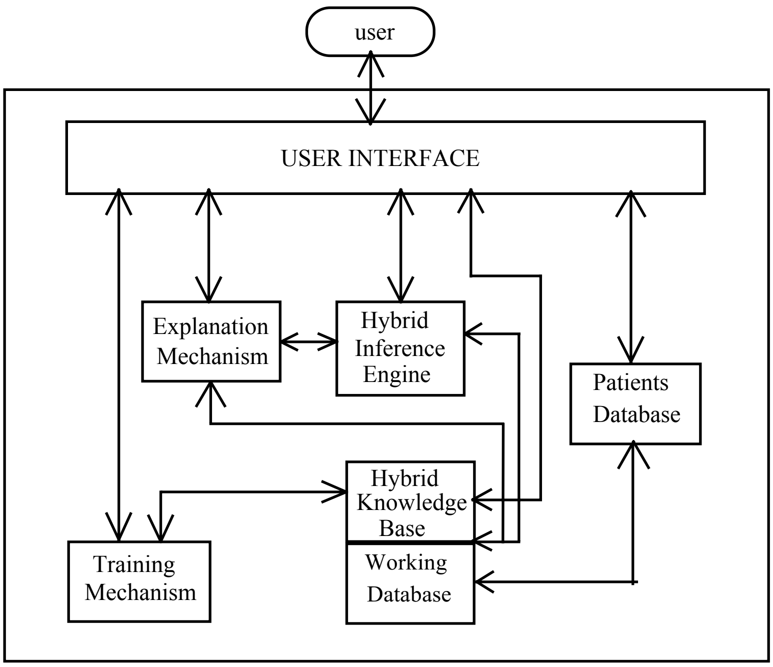

Fig.1 The Architecture of XBONE

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 automaticaly transferred to the system [4]. *Hybrid Knowledge Base* (HKB) contains the domain and the heuristic diagnostic knowledge, represented via a hybrid representation formalism. *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. Demographic patient data are transferred to WDB from PDB. *Hybrid Inference Engine* (HIE) realizes the procedural knowledge and uses the available heuristic knowledge in HRB to draw conclusions and make diagnoses. *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 as far as user interaction with the system is concerned.

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*, corresponding to the weight of the input of the adaline unit. Each significance factor represents the significance of the corresponding condition in drawing the conclusion of the rule. Moreover, each rule itself is assigned a number sf_0, called the *bias factor*, corresponding to the weight of the bias input ($C_0 = 1$) of the unit.

Each input takes a value from the following set of discrete values:

$$C_i = \begin{cases} 1 & \text{if condition is true} \\ 0 & \text{if condition is false} \\ 0.5 & \text{if value is unknown} \end{cases}$$

This gives the opportunity to easily distinguish between the falsity and the absence of a condition, in contrast to conventional rule-based systems.

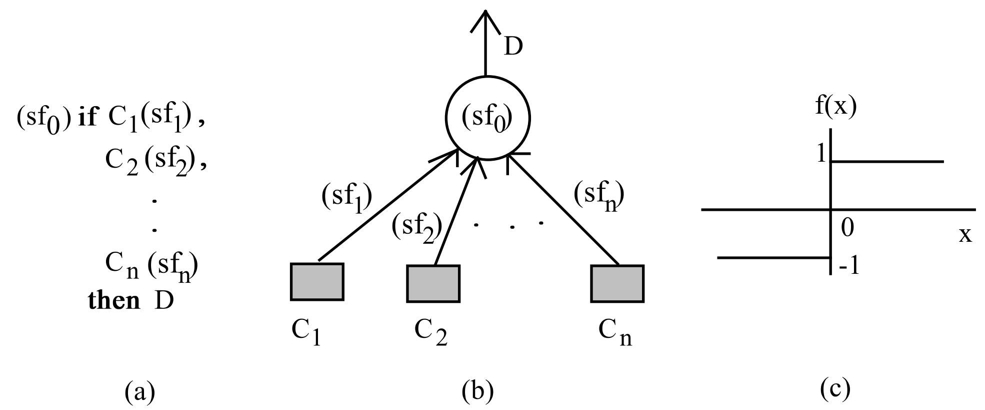

Fig.2 (a) A neurule (b) Corresponding adaline unit (c) Activation function

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. [3]):

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

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[1]:

<rule> ::= [(<bias-factor>)] **if** <conditions> **then** <conclusions>
<conditions> ::= <condition> {**,** <condition>}
<conclusions> ::= <conclusion> {**,** <conclusion>}
<condition> ::= <object> <l-operator> <value> [(<significance-factor>)]
<conclusion> ::= <object> <r-operator> <value>.

<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 operator (e.g. $<, =, >$), whereas <r-operator> can be only "is". <value> denotes a value of the <object>, numeric or symbolic. Finally, <bias-factor> and <significance-factor> are real numbers. As it is clear, significance factors and the bias factor are optional in a rule. Thus, neurules (with factors) and conventional production 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

HRB consists of *Domain Knowledge Base* (DKB) and *Hybrid Rule Base* (HRB). DKB contains domain knowledge, represented via variable declarations. HRB consists of *Early Diagnosis Rule Base* (EDRB) and *Late Diagnosis Rule Base* (LDRB), which contain knowledge concerning the early and the late diagnosis respectively, represented via rules. 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 next section).

R1:
if sex **is** man **,**
age >= 20 **,**
age < 35
then patient_class **is** man_20_35

R2:
(-8) **if** pain **is** continuous (5) **,**
patient_class **is** man_20_35 (2.5) **,**
fever **is** medium (2) **,**
fever **is** high (2)
then disease_type **is** inflammation

Fig.3 A conventional rule and a neurule.

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. Each rule is optionally given initial values to its significance factors and bias factor, when introduced in HRB. Training of the neurules takes place in a period prior to the initial use of the system and every time the system is updated. The training sets are

[1]A BNF notation is used here, where '[]' denotes optional occurrence and '{}' zero, one or more occurrences of the enclosed expression. Also, '<>' denotes a nonterminal symbol. All other symbols are terminal.

extracted from known (old) patient cases and/or the diagnostic tree. The standard least mean square (LMS) learning algorithm (see [3]) is employed to calculate the values of the factors.

However, there are cases where TM fails to find converging factors: the training set of a rule corresponds to a non-separable function (see [3]). This is an inherent weakness of the Adaline model. Then, conventional rules should be employed.

4.4 The Hybrid Inference Process

An inference process is peformed in two stages, the *early diagnosis stage* and the *late diagnosis stage*. During the early diagnosis stage EDRB is activated, the system asks questions about patient's clinical data and produces a first diagnosis. During the late diagnosis stage, LDRB is activated. Activation of LDRB 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. There are two stacks used, a *goal stack*, where the *current goal* (conclusion/condition) to be matched is always on its top, and a *rule stack*, where the *current rule* under evaluation is always on its top. A rule succeeds if it *evaluates* to 'true', that is all of its conditions evaluate to 'true', in the case of a conventional rule, or its output is computed to be '1', via formula (1), after evaluation of its conditions, in the case of a neurule.

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. [7]), image processing techniques are not very reliable and are not preferred (e.g. [1]). On the other hand, this makes participation of the user-physician more active.

Knowledge is represented via a formalism integrating conventional rules and neurules. This results in better representation, since one can represent more complex relations between conditions, and facilitates knowledge acquisition. All that an expert has to do is to determine the symptoms involved in diagnosing various diseases and the training sets.

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.

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