Short Report

An expert system for the diagnosis of epilepsy: Results of a clinical trial

SANJEEV V. THOMAS, JAYALEKSHMI R. KURUP, ABRAHAM KURUVILLA, B. NARAYANAN NAIR, K. L. THOMAS, P. SANKARA SARMA

ABSTRACT

Background. Artificial intelligence is an area where computer systems are used to solve real-life problems that require expert human intelligence. Expert systems serve as an effective alternative to supplement the dearth of human experts in a narrow domain of applications. We developed an expert system named SEIZ using DIAGNOS (an expert system shell for diagnostic applications) for the diagnosis and management of epilepsy.

Methods. A clinical trial was done to test the reliability of SEIZ. The clinical and demographic data from the medical records of 50 patients with epilepsy who attended an epilepsy clinic were provided to the expert system. The system-generated diagnosis was compared with the clinical diagnosis.

Results. The seizure types and epileptic syndromes for the 50 patients included generalized tonic–clonic seizure (14), absence (4), complex partial seizure (18), simple partial seizure (4), juvenile myoclonic epilepsy (5) and other epileptic syndromes (3). There were two cases of hysterical conversion reaction. There was concordance in the diagnosis between the expert system and clinician in 47 cases (94%). The overall sensitivity was 94% and the specificity was 100% for absence, generalized tonic–clonic seizures, simple partial seizures and juvenile myoclonic epilepsy; 94% for complex partial seizures and 98% for hysterical conversion reaction.

Conclusion. This expert system could generate reliable diagnoses for patients with epilepsy. Such a system may be useful for a doctor in a remote or peripheral area where an expert on epilepsy is not available.

Natl Med J India 2001;14:274–6

INTRODUCTION

Computers are increasingly being used in clinical settings to handle administration, accounts and clinical data. With the availability of more powerful computers, it has now become possible to use advanced computing in the management of complex problems in patient care. Artificial intelligence (AI) is the study of how to make computers do things, which, at the moment, people do better. The different areas where AI has been tried include natural language processing, computer vision, robotics, pattern recognition, expert systems, etc. An expert system (ES) is a computer programme that relies on a body of knowledge to carry out a complex task usually performed only by a human expert. An ES successfully deals with problems for which clear algorithmic solutions do not exist. It embodies a knowledge base (KB) derived from an expert and a method of applying it to solve problems. Performance at the level of a human expert implies the ability to explain the line of reasoning used to arrive at a solution. An ES brings to the user the expertise of a specialist or a team of specialists in the respective field. Such systems have been successfully tried in several aspects of clinical diagnosis and treatment. A system that can identify epileptiform discharges from continuous electroencephalographic data has recently been developed. An ES that assists in the histopathological diagnosis of breast cancer has been designed. We examined the scope of a dedicated ES called SEIZ in the diagnosis and management of epilepsy. SEIZ was developed jointly by the Electronics Research and Development Centre and Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram.

METHODS

SEIZ was developed using DIAGNOS—an ES shell for diagnostic applications. Since this is a shell for diagnostic applications, the KB of DIAGNOS is structured as complaints, hypotheses and parameters. These are frames which hold rules and relevant information of the KB in different slots. The principal power of an ES is derived from the knowledge the system embodies rather than from search algorithms and specific reasoning methods. The KB consists of about 4000 rules split into different pieces of knowledge, based on the International League Against Epilepsy (ILAE) classifications of seizures and epileptic syndromes. Hypotheses are possible reasons for the set of complaints suggested by the domain experts. Parameters are data required for the confirmation or rejection of hypotheses. Experts' knowledge about the domain is represented using rules, facts, relations and procedures. Wherever possible, heuristics or 'rules of thumb' can be incorporated into the KB to reduce search space. Uncertainty in the domain is handled by assigning a weightage to different hypotheses and is controlled through different values of parameters, which contribute shares of weightage to hypotheses. A hypothesis is either confirmed or rejected depending upon whether the accrued weightage of the hypothesis exceeds an upper threshold or falls below a lower threshold value. The experts, depending upon their expertise in the domain, determine these values.

SEIZ works on a logic similar to that applied by a clinician in everyday practice (Fig. 1). The system examines the presenting symptom on the basis of the KB. It further enquires about a number of other clinical features (parameters), the various options of which have different weightage for different diagnoses (hypotheses), e.g. the parameter 'aura' carries a certain positive weightage for complex partial seizure and a negative weightage for typical absence. Instructions are also incorporated to examine or skip certain parameters based on the available data. After
Fig 1. Algorithm comparing the diagnostic process followed by a clinician and an expert system

On a structured proforma, we extracted from the medical records the clinical data of 50 consecutive patients with epilepsy. The mean age was 21.3 years; 28 were men and 22 women. These patients had been evaluated by different clinicians to arrive at the diagnosis. Fourteen patients had generalized tonic-clonic seizures (GTCS), 4 had absence, 18 had complex partial seizures with or without generalized seizures (CPS±GS), 4 had simple partial seizures with or without generalized seizures (SPS±GS). Two patients had hysterical conversion reaction (HCR), 5 had juvenile myoclonic epilepsy (JME), 2 had late-onset seizures and 1 had infantile spasm. The ES examined their clinical data independently and generated its own diagnosis. We compared the diagnosis made by the clinician with that generated by the ES.

Statistical methods were applied to measure the sensitivity and specificity of the diagnosis. The sensitivity of the ES was estimated in two parts. Sensitivity A was the percentage of each ES diagnosis that concurred with that of the clinician. It also included those cases where the system gave an additional diagnosis. Sensitivity B was the percentage of each ES diagnosis that concurred absolutely with that of the clinician, and there was no other diagnosis given by the system (Table I).

RESULTS
In 47 of 50 cases (94%), the ES concurred with the clinical diagnosis. In 6 of the 47 cases (5 cases of CPS±GS and 1 case of SPS±GS), the ES suggested one more diagnostic possibility in the order of priority. The terminology recommended in the ILAE classification of seizures and epileptic syndromes was followed in the ES.

In 3 of the 50 cases (6%) there was disagreement between the ES and the clinician regarding the diagnosis.

The ES correctly diagnosed all 14 cases of GTCS, 4 cases of absence, 2 cases of HCR and 5 cases of JME. There was agreement in the diagnosis between the ES and the clinician in all 18 cases of CPS±GS. However, the ES gave an additional diagnosis of HCR in 5 of them. The ES concurred with the clinician in all 4 cases of SPS±GS but gave a second diagnosis of HCR in 1 of them.

There was disagreement in the diagnosis of 3 cases, 2 of late-onset seizure and 1 of infantile spasm. The ES diagnosed them as CPS±GS and HCR, respectively. Late-onset seizure is not an ILAE-approved seizure terminology. The ES was programmed to follow only the ILAE terminology. Hence, this cannot be considered as a failure of the ES. However, the ES failed to correctly diagnose 1 case of infantile spasm.
TABLE I. Comparison of the sensitivity and specificity of ES to clinical diagnosis

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Clinician confirmed (other)</th>
<th>Sensitivity A</th>
<th>Specificity B</th>
<th>Sensitivity A</th>
<th>Specificity B</th>
</tr>
</thead>
<tbody>
<tr>
<td>Absence</td>
<td>4</td>
<td>100</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Generalized tonic–clonic seizures</td>
<td>14</td>
<td>100</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Complex partial seizures</td>
<td>18</td>
<td>72</td>
<td>94*</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>with or without generalized seizures</td>
<td>4 (1)</td>
<td>100</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Simple partial seizures</td>
<td>4</td>
<td>75</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>with or without generalized seizures</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hysterical conversion reaction</td>
<td>2</td>
<td>100</td>
<td>100</td>
<td>98†</td>
<td>100</td>
</tr>
<tr>
<td>Juvenile myoclonic epilepsy</td>
<td>5</td>
<td>100</td>
<td>100</td>
<td>100</td>
<td>100</td>
</tr>
<tr>
<td>Others</td>
<td>3†</td>
<td>94</td>
<td>82</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>50</td>
<td>47 (6)</td>
<td>94</td>
<td>82</td>
<td></td>
</tr>
</tbody>
</table>

* Expert system had made a diagnosis of complex partial seizures in 2 cases of late-onset seizure. † Expert system had made a wrong diagnosis of hysterical conversion reaction in 1 case of infantile spasm. ‡ Late-onset seizures 2, infantile spasm 1.

Overall, the ES had a sensitivity A of 94% (47 cases in which the diagnosis made by the ES concurred with that of the clinician). This includes 6 cases in which the ES gave a second diagnosis. The sensitivity B of the ES was 82% (41 cases where the ES gave one diagnosis which was the same as that of the clinician). Similarly, sensitivities A and B were calculated separately for each specific diagnosis (Table I). The specificity for individual diagnoses ranged from 94% to 100%.

DISCUSSION

An ES is an intelligent computer program that uses knowledge and inference procedures to solve difficult problems that require human expertise. Epilepsy is a common condition in which the application of an ES is convenient. Epilepsy has a sound KB, and the clinician weighs many factors such as the type of seizure, electroencephalographic findings, other imaging findings, clinical background, etc. to arrive at a diagnosis. Recently, an ES has been used to distinguish temporal lobe epilepsy from frontal lobe epilepsy. Smeets et al. developed an ES to recommend the appropriate antiepileptic drug for the treatment of patients with epilepsy. We attempted to apply ES in the diagnosis and management of epilepsy and have shown that it is possible to apply ES for the complex logic and reasoning required to diagnose epilepsy.

The clinical material tested in this study covers a wide spectrum of commonly encountered seizures, epileptic syndromes and pseudoseizures (HCR). The diagnoses in these patients were ascertained by a set of doctors other than those involved in the development of the ES. The high degree of agreement between the clinician and the ES is an indicator of its reliability. In this study the ES agreed with the clinical diagnosis in 94% of cases. The ES successfully diagnosed all cases of HCR as well. There was disagreement in 3 cases (6%). There were certain deficiencies in the clinical data as these were extracted retrospectively from the case records. In some instances, the clinician probably had not written down all the relevant findings that enabled him to diagnose the condition. The ES was constrained to formulate a diagnosis from the limited data that were made available to it and one such instance was infantile spasm. A prospective study alone can overcome this handicap. Among the 47 cases where the ES and clinician agreed broadly, there were 6 cases in which the ES suggested another diagnostic possibility rather than confirming one diagnosis. This is advantageous as it broadens the differential diagnosis and there is room for further investigation. The results of this preliminary study are encouraging. Further clinical trials with a larger cohort drawn from multiple centres would be required to validate the sensitivity and specificity of the ES.

There is some debate regarding the utility of an ES and other AI tools in clinical practice. However, there is wide scope for ES in the management of epilepsy and other disorders. An ES has the advantage that its KB can be updated and improved by pooling the expertise of many professionals in the field. It is possible to validate the reliability of the ES in actual clinical settings by appropriate clinical trials such as this. Once the psychological barrier to the use of ES is broken, it would find wider acceptance among clinicians. In developing countries such as India, patients often have to travel long distances to be seen by experts who work in major cities. In such situations, an ES can be of assistance to doctors working in peripheral centers and remote areas.

REFERENCES