Computer-assisted Diagnosis of Pediatric Rheumatic Diseases

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ABSTRACT. Objective. AI/RHEUM is a multimedia expert system developed originally to assist in the diagnosis of rheumatic diseases in adults. In the present study we evaluated the usefulness of a modified version of this diagnostic decision support system in diagnosing childhood rheumatic diseases.

Methodology. AI/RHEUM was modified by the addition of 5 new diseases to the knowledge base of the system. Criteria tables for each of the diseases included in the knowledge base were modified to suit the needs of children. The modified system was tested on 94 consecutive children seen in a pediatric rheumatology clinic.

Results. AI/RHEUM made the correct diagnosis in 92% of the cases when the diagnosis was available in the knowledge base of the system. It was also shown to be effective in the education of pediatric trainees through its multimedia features.

Conclusions. AI/RHEUM is an expert system that may be helpful to the nonspecialist as a diagnostic decision support system and as an educational tool. Pediatrics 1998;102(4). URL: http://www.pediatrics.org/cgi/content/full/102/4/e48; computer-assisted diagnosis, multimedia, rheumatic diseases, expert system.

Computers have come to play a major part in patient care activities, including literature searches, patient monitoring, and drug prescriptions. Imaging techniques such as magnetic resonance imaging and nuclear medicine would be impossible without computer technology. All of these areas are grouped under computer-based clinical decision support systems by Habbema, who recognized four levels of medical decision-making: 1) collecting background information on patients, both general and specific; 2) assessing diagnostic and therapeutic methods that include algorithms; 3) developing a global clinical policy, such as protocols and guidelines, which are becoming increasingly important in this era of cost-containment; and 4) developing an individualized clinical strategy that includes medical expert systems.

The term expert system refers to a computer program that simulates the diagnostic reasoning of an experienced clinician in specific areas of medicine such as rheumatology. Expert systems belong to the general area of artificial intelligence in which symbolic reasoning methods are used. An expert system has two components—a knowledge base and an inference mechanism. The knowledge base can include information about specific diseases and an inference mechanism designed to work with the knowledge base and with patient data to reach a conclusion such as a diagnosis or treatment plan.

There are complex software packages that can build general expert systems, called expert system shells. CTX, written at the National Library of Medicine (Bethesda, MD), is one such shell. AI/RHEUM, a knowledge-based medical consultant system built using the CTX shell, is a multimedia expert system originally developed by Kingsland et al$ in the 1980s to assist general physicians with the diagnosis of rheumatic diseases in adults. We now report on a modification of this system to address childhood rheumatic diseases.

MATERIALS AND METHODS

AI/RHEUM runs on IBM-compatible personal computers and UNIX systems. The knowledge base includes diagnostic criteria tables for 54 common rheumatic diseases (Table 1). The criteria tables are designed using the most important clinical and laboratory features of each of the diseases. These tables were originally developed at the University of Missouri, Columbia, MO, and reviewed by a panel of rheumatologists. More recently, H. James Williams, MD, at the University of Utah has modified some of these tables. One of the authors (B.H.A.) modified the criteria tables to suit the needs of children, prepared a set of screening criteria to enter patient data into the system, and also added 5 more diseases to the knowledge base.

AI/RHEUM's reasoning uses the criteria table paradigm with 1 criteria table for each of the diseases covered. Criteria tables (Fig 1) consist of the following elements: major criteria, minor criteria, required criteria, and exclusions. The decision elements may be symptoms, signs, or laboratory data from the patient data file. The decision element may also be an intermediate hypothesis derived from a combination of findings. An example of an intermediate hypothesis is when the data entry includes pain in the joints and limitation of range of movement and the program generates arthritis as the decision element.

The expert system matches the individual patient findings with the criteria tables and reaches diagnostic conclusions at three different levels of certainty: definite, probable, and possible. An example of a patient with definite diagnosis of Wegener's granulomatosis is given in Fig 2. When a patient has vasculitis of small vessels, midline granuloma, and antineutrophil cytoplasmic antibody the diagnosis is definite. If the same patient has small vessel vasculitis and antineutrophil cytoplasmic antibody, the diagnosis of Wegener's granulomatosis will be probable. Whenever the diagnosis is suggested at the possible level, the system recommends that you keep the diagnosis in the differential list and look for further clues to exclude or include it.

Patient findings are entered on data forms online for each patient. The data form consists of multiple screens to enter clinical
findings and laboratory data. The system can capture newly added criteria tables.  
Wegener’s granulomatosis  
Vasculitis, nonspecific  
Tuberculous arthritis  
Trochanteric bursitis  
Takayasu’s arteritis  
Systemic lupus erythematosus  
Sjogren’s syndrome, primary  
Rotator cuff tendinitis  
Fibrosis  
Giant cell arteritis  
Gonococcal arthritis  
Gout  
Henoch-Schonlein purpura  
History of systemic lupus erythematosus  
Hypermobile joint syndrome*  
Hypersensitivity vasculitis  
Hypertrophic osteoarthropathy, primary and secondary*  
Intervertebral disc herniation  
Juvenile rheumatoid arthritis, pauciarticular, type 1*  
Juvenile rheumatoid arthritis, pauciarticular, type 2*  
Juvenile rheumatoid arthritis, polyarticular, RF negative*  
Juvenile rheumatoid arthritis, polyarticular, RF positive  
Juvenile rheumatoid arthritis, systemic onset (Still’s disease)  
Kawasaki disease  
Lateral epicondylitis (tennis elbow)  
Lyme disease  
Mixed connective tissue disease  
Nonspecific back pain  
Nonspecific joint pain  
Polyarteritis nodosa  
Polymyalgia rheumatica  
Polyarthritis  
Primary Raynaud’s  
Psoriatic arthritis  
Reiter’s syndrome or reactive arthritis  
Relapsing polychondritis  
Rheumatic fever  
Rheumatoid arthritis  
Rotator cuff tear  
Rotator cuff tendinitis  
Sjogren’s syndrome, primary  
Spinal stenosis  
Systemic lupus erythematosus  
Systemic sclerosis  
Takayasu’s arteritis  
Trochanteric bursitis  
Tuberculous arthritis  
Vasculitis, nonspecific  
Wegener’s granulomatosis  

* Newly added criteria tables.
recognized that criteria tables were not available for 5 diseases and refused to come to a conclusion. In the other 13 patients, the system offered some other diagnosis. Seven of them were appropriate. For example, panniculitis and retroperitoneal fibrosis are not included in the system; however, the system suggested nonspecific vasculitis as a possibility. In 6 other patients, the alternatives suggested by the sys-

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**Criteria Table for: Juvenile RA, systemic-onset (Still's Disease)**

Criteria for Juvenile RA, Systemic-onset (Still's Disease) (3-21-95)

**Major Criteria**

1. Patient's age at the onset of symptoms < 16 years
2. Daily temperature intermittent
3. RA rash
4. Arthritis of more than 6 weeks

**Minor Criteria**

1. Joint pain (Arthralgia) or generalized aches 
2. Joint stiffness
3. Repeatedly normal
4. Splenomegaly
5. Pericarditis by echocardiogram or EKG
6. Pericarditis by examination
7. Myocarditis
8. Pneumonia
9. Malignancy
10. WBC > 20,000/mm
11. Platelet count > 600,000/mm

**CLINICAL COMBINATIONS OF FINDINGS**

<table>
<thead>
<tr>
<th>Definite</th>
<th>Probable</th>
<th>Possible</th>
</tr>
</thead>
<tbody>
<tr>
<td>4 Majors</td>
<td>Majors 1, 2, 3</td>
<td>Major 1</td>
</tr>
<tr>
<td>2 minors</td>
<td>1 other major</td>
<td>3 minors</td>
</tr>
</tbody>
</table>

**REQUARED FINDINGS**

| None | None | None |

**EXCLUSIONARY FINDINGS**

| Infections and malignancy | Infections and malignancy |

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**Summary of Findings for Demo**

**CASE NUMBER:** Demo 16  
**DATE OF VISIT:** 8/13/97  
**CASE NUMBER:** Demo 16  
**VISIT NUMBER:** 1

1. Fever  
2. Rash  
3. Vasculitis by biopsy  
4. Westergren ESR (normal < 20 mm female, < 30 mm male) 55  
5. I would like to continue data entry.  
6. Patient in male  
7. Patient age at this workup 11  
8. Patient age at onset of symptoms 11  
9. Joint or joint related complaint  
10. Systemic illness with articular and/or constitutional features (jaw, rash, panniculitis, etc.)  
11. Joint swelling (less than 4 joints)  
12. Chronic arthritis, longer than six weeks duration  
13. Fatigue  
14. Malaise  
15. Symmetrical distribution of synovial swelling  
16. Skin findings  
17. Edema, nose and throat findings  
18. Pulmonary findings  
19. Renal findings, including biopsy  
20. Vasculitic rash  
21. Vasculitic skin findings  
22. Vasculitic rash  
23. Acute sinovitis  
24. Chest X-ray: normal, non-infectious, non-malignant nodular or cavitary infiltrates  
25. Necrotizing granulomata on respiratory tract bx  
26. History of hematuria  
27. CBC, Erythrocyte sedation rate or Congestive study  
28. Urinalysis or 24 hour urine collection  
29. Rheumatologic serology (ANA, RF, ANCA, etc.)  
30. Hematocrit (normal 40-50%) 21  
31. WBC (normal) 12000, < 12,000/mm

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Fig 1. Example of a criteria table.

Fig 2. Summary of patient data from AI/RHEUM.
tem were not appropriate. Three of these children had leukemia, 1 had reflex sympathetic dystrophy syndrome, 1 had lymphangioma of the synovium, and 1 had lymphedema.

**DISCUSSION**

Computer-assisted decision making is currently confined to specific areas of patient management such as routine follow-up and drug monitoring. It has not played a major part in clinical diagnosis despite the availability of some excellent programs. The CTX expert system can be adapted to many areas of medicine for developing a consultation and an educational program for nonspecialists in that field. It is ideal for performing triage functions for primary care physicians and emergency room physicians. The multimedia capacity of the program allows for interactive learning and immediate feedback.

AI/RHEUM developed originally to assist in the diagnosis of rheumatic diseases in adults has been tested extensively. In the initial study, the system was tested in 384 carefully selected adult cases with an accuracy of 94%. Another study, from Keio University in Japan, involved 59 patients with various connective tissue diseases. The diagnosis made by AI/RHEUM was in full or partial agreement with those of the Japanese rheumatologists in 54 cases (92%). The criteria table for mixed connective tissue disease was found to have a sensitivity of 90% and specificity of 96%.

Moens and Van der Korst tested the performance of AI/RHEUM in 570 different cases from a Dutch outpatient rheumatology clinic and compared it with predictions of diagnostic outcome made by rheumatologists. In this study, physicians made 839 diagnoses. Of these, 694 were definite and 145 were possible. Of these, the original model ranked 735 (88%) and the modified model 727 (87%) among the 5 most common diagnoses. Sensitivity was 62% but the specificity was 97% to 98%.

The modified AI/RHEUM system when tested in children was shown to make the diagnosis correctly in 92% of patients when the diagnosis was available in the knowledge base. Obviously the system cannot make those diagnoses for which there are no criteria tables (Table 3) in the system and should refuse to make a diagnosis for these conditions. This was what we found for 5 conditions. In 13 patients, the program offered a different diagnosis because some of the signs and symptoms are common to many rheumatic diseases.

Two diseases account for 6 of the 18 incorrect conclusions—Lyme disease accounting for 2 of 5 among the group of diseases for which criteria tables are available and leukemia and malignancy account-
ing for 5 of the 13 among the diseases without criteria tables. We propose to review the criteria table for Lyme disease and add a criteria table for leukemia. We also plan to perform sensitivity and specificity analysis on all the criteria tables.

More than the consultative role, this expert system provides a strong educational component. All through the case-entry mode, are prompts requesting written and visual information pertaining to the clinical features of the disease in question. This aspect was not tested in formal way. However, trainees in pediatrics who used the system found it to be educational. Some of them said that AI/RHEUM helped them consider a diagnosis they would not have considered otherwise.

The system is simple to use and capable of per-
forming some functions of an expert in pediatric rheumatology, but it needs to be tested at other pediatric centers. The educational component needs formal testing. The sensitivity of the system in diagnosing conditions listed in the knowledge base needs to be tested.

Finally, the legal ramifications of an expert system for use by generalists is an uncharted territory. Although an expert system such AI/RHEUM can be made available through the Internet, the legal and financial issues have to be resolved before such expert systems are available more widely.

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REFERENCES
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