RECOGNITION-BASED DIAGNOSTIC REASONING

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ABSTRACT

Expertise in fault diagnosis often depends on recognizing particular patterns in observed data corresponding to situations that have previously been 6een and correctly interpreted This approach can result in significant efficiencies by avoiding a costly and detailed analysis based on the causal relationships between faults and observable data Recognition-based reasoning requires highly focused search strategies in order to extract relevant information from a knowledge base that contains a large number of prototypes for various possible faults This paper describes a model of recognition-based reasoning and describes an implementation of the model. designed to solve diagnostic problems in pediatric cardiology The model first analyzes portions of the patient data to hypothesize a small set of potential diseases Knowledge specific to each selected hypothesis is then used to refine these initial choices The model has been validated against actual hospital cases and performs in a manner comparable to expert physicians

1. Introduction.

Expertise in many diagnostic problem domains depends on both an extensive, detailed knowledge base about potential faults and their manifestations and on control structures capable of selecting only those purtions of the knowledge base likely to be relevant to the current problem We have developed a model of diagnostic reasoning for such problems which performs at expert levels through the use of three components. An initial recognition component triggers a small set of working hypotheses about a fault Refinement rules, specific to each triggered hypothesis, work to accept, reject, or modify the hypothesis, or to suggest alternate hypotheses for consideration A causal model component generates precise expectations about each fault for use by the refinement rules Successful diagnosis results from good first hypotheses about possible defects, efficient mechanisms for refining these hypotheses, and accurate techniques for determining the plausibility of possible defects

In fault diagnosis, expertise often consists of the recognition of specific situations as instances of ones that have been seen and successfully corrected in the past [1,2,3]. Such recognition-driven expertise is baaed upon a large repertoire of domain specific knowledge in the form of rules that trigger models of possible faults. These models in turn contain expectations for testing whether a proposed model fits the conditions of an observed situation. The complexity and level of detail of this knowledge leads to serious problems with the computational complexity of search processes. Our model focuses computational resources so that this detailed. prototypical knowledge can be utilized in an efficient manner

Not all fault based task environments lend themselves to this treatment, of course. The complexity of possible defects may be such that predefined fault models cannot be constructed In such cases, more complex causal reasoning is often required

2. Problem Domain.

The above approach to diagnostic problem solving has been demonstrated in a program we call GALEN GALEN diagnoses cases of congenital heart disease in children In this domain, GALEN is presented with some observable data about a circulatory system that contains one or more faults or *defects.* All possible defects that can occur within the circulatory system are assumed to be known in advance, as are the immediate effects they have on the rest of the system Some sets of defects uniquely correspond to named *diseases* It is GALENs task to identify what disease is present (and therefore what defects are present) given this patient data. The disease whose expectations best match the observed data is selected as the final diagnosis

3. Knowledge base.

The knowledge base in GALEN consists primarily of an organized collection of *hypotheses* The hypotheses specify potential defects, expectations associated with these defects, and rules for refining the hypotheses or suggesting other possible hypotheses

3.1. Hypotheses.

Each hypothesis has two main parts The first part is a model of the hypothesized phenomena themselves. In congenital heart disease, this is a partial model of a diseased circulatory system The models provide expectations about what should be observed if the hypothesis is true The second part is a group of production rules that describe how to investigate the truth or falsity of the hypothesis. These rules describe conditions under which a hypothesis should be accepted, rejected, or modified. They also describe when to temporarily abandon a hypothesis in favor of another

Three different forms of hypothesis have proven useful in diagnosing faults in the domain of congenital heart disease. *Category hypotheses* describe groups of diseases that have some important features in common. For example, the category hypothesis 'diseases_with_increased _PBF' includes all diseases that involve increased pulmonary blood flow. The category hypothesis 'cyanotic_heart_disease' includes hypotheses that could explain why the patient is cyanotic. The

models of a category hypothesis describe important expectations that are common to all its member hypotheses. The production rules in a category hypothesis deal primarily with choosing the most appropriate member hypotheses for the given situation.

Disease hypotheses describe individual diseases. Disease hypotheses typically contain several models that represent the state of the circulatory system under several variants of the disease. These variants describe differences in severity (e.g mild aortic stenosis, moderate aortic stenosis) or the presence of significant co-occurring defects (e.g. aortic stenosis with mitral insufficiency). The production rules in a disease hypothesis act to select an appropriate variant model of the disease, in much the same way that the rules in a category hypothesis act to select an appropriate disease.

Pathophysiological hypotheses describe individual faults within the circulatory system. They contain a sin gle model of the portion of the system that contains the fault. Pathophysiological hypotheses contain rules that trigger diseases (or categories of diseases) which result from the faults they describe

3.2. Hierarchical structure.

An important reason for the success of our model is the use of a hierarchically organized knowledge base which allows the program to consider possible diagnoses at appropriate levels of abstraction These levels range from very general (categories of diseases), to mid-range (specific diseases), to very specific (defects within diseases)

Describing hypotheses at different levels of abstraction is important in achieving efficiency during diagnosis Category hypotheses are included because some data cues suggest broad classes of hypotheses without necessarily suggesting any specific disease within the class Further specification of the exact defect can then be postponed until analysis of additional data have been completed, rather than hypothesizing a disease that might have to be rejected later. We can also prune away large areas of the problem space with a minimum of effort if we can reject an entire category of hypotheses, instead of having to reject hypotheses one at a time At the other end of the spectrum, pathophysiological hypotheses are included primarily as a way to organize patient data into meaningful chunks We can use a pathophysiological hypothesis as a short form for the complex of data that led to its proposal

The set of all hypotheses known to GALEN forms a search graph, in which the graph's vertices arc the hypotheses themselves and the graph's edges are production rules. A given hypothesis is "connected" to another one if the first hypothesis has a rule whose consequent specifies an operation on the second Triggering a hypothesis activates its production rules, which in effect starts a search of the graph at the corresponding point.

4. Control.

GALEN contains two major procedures for recognizing defects: *propose* and *review.* Together, these two procedures implement a highly focused search strategy for moving through the hierarchy of hypotheses. GALEN operates primarily by alternating cycles of propose and review: propose to suggest an initial place to begin searching the hypothesis graph, and review to continue the search until more data is needed.

4.1. Propose.

The propose procedure allows initial attention to be focused into the graph of hypotheses at some appropriate point. This is done by applying rules which look for specific features of the incoming data and *trigger* one or more hypotheses if they are successful When a hypothesis is triggered, a specialized set of rules within the hypothesis are applied which specify the most appropriate model for the hypothesis based on an examination of the data and of other hypotheses For example, the disease hypothesis 'tetralogy_of_allot' will choose a model reflecting a 'severe' form of the disease if the patient is less than two months old:

```
(if
  (data
    (description
      (age (lt 2))))
  then
  (specify severe_tetralogy))
```

These rules may also trigger additional related hypotheses; the hypothesis 'increased_PBF' also triggers the category hypothesis 'diseases_with_increased_PBF' and the pathophysiological hypothesis 'left_to_right_flow':

```
(if
  (hypotheses
    (increased_PBF))
  then
  (trigger left_to_right_flow
       diseases_with_increased_PBF))
```

The level of abstraction of triggered hypotheses may vary due to differences in the usefulness and specificity of data cues. For example, clubbing of the fingertips can indicate cyanosis, which would result in 'cyanotic_heart_disease' being triggered This is a category hypothesis that represents some six possible diseases; the cyanosis data cue alone is not powerful enough to select among them. An early diastolic murmur heard near the pulmonary valve, however, would result in the more specific hypothesis 'insufficient_pulmonary_valve' being directly triggered:

```
(if
  (diastolic
    (time early)
    (heard_best pulmonary_area))
  then
  (trigger insufficient_pulmonary_valve))
```

4.2. Review.

Once the propose procedure has identified plausible places to begin searching the graph of hypotheses, the review procedure can carry out a search from there. The review procedure compares a hypothesis' models against accumulated data, then applies rules within the hypothesis that examine the results of the comparison. These rules typically look for violated expectations in the patient data and specify new models of the original hypothesis that do not give rise to the same violations. (The rules can also accept or reject current hypotheses or trigger new ones). For example, if the disease hypothesis 'tetralogy-oLiallot' detected that its model 'severe__tetraiogy' expected increased vascularity on X-ray but none was observed, a new model, 'mild_tetralogy' would be specified as an alternative:

```
(unobtained
  (X-ray (vascularity)))
then (specify rnild_tetralogy))
```

Under some conditions, a hypothesis may have its models compared against other hypothesis' models in addition to being compared against data The decision to make this extra comparison is made by rules in the affected hypotheses themselves. Such additional comparisons can be useful if there is overwhelming evidence in favor of one or more hypotheses, or if there is some important pathophysiological hypothesis that should be taken into account whenever a new hypothesis is triggered

5. Expectations

A third procedure is also critical to Galen's performance. The *modeler* uses two types of causal rules to precompile models of the circulatory system that are associated with hypotheses in the knowledge base One type of rule determines the hemodynamic results of a defect or abnormality in the circulatory system Another type of rule determines the observable results of a defect or abnormality For example, a leaky valve can cause changes in flow and pressure in its vicinity hemodynamic results) as well as an audible murmur observable results).

The modeler starts with a model of the normal circulatory system and adds a set of defects corresponding to those present in a specific disease. Next, all relevant rules are applied to the defects, yielding a circulatory system containing new hemodynamic and observable abnormalities. Rules are then applied in turn to the resulting abnormalities, until no new rules are applicable. The result is a model of how a real circulatory system would appear in the specific disease.

6. Performance.

GALEN's knowledge base contains information sufficient to diagnose 70 congenital cardiac diseases and disease variants. It is able to respond to cases covering approximately 95% of the diseases found in the flies of the Pediatric Cardiology Clinic at the University of Minnesota heart hospital. In a typical run, GALEN inspects 30-30 pieces of patient data, distributed in the categories of history, physical examinatioa X-ray. and EIKG. Hypotheses are proposed as each item of data is examined. In such a run, 10-20 specific disease models are usually considered. Following the last item of data, a priority rating of these models is established, based upon the degree of fit between GALEN's expectations for each disease and the actual patient data values. Results of validation studies carried out using patient cases from the hospital files Indicate that: 1) GALEN is able to reach the same diagnosis as medical staff on the most common forms of congenital heart disease, 2) GALEN is also able to reach a correct diagnosis on selected cases of congenital heart disease that were initially misdiagnosed by medical staff, and 3) the reasoning steps employed by the program and the expert diagnosticians are similar [4,5].

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