DOMAIN SPECIFICITY AND KNOWLEDGE UTILIZATION IN DIAGNOSTIC EXPLANATON

Vimla L. Patel
José F. Arocha
Guy J. Groen

McGill University

ABSTRACT

This paper examines the performance of cardiologists, psychiatrists, and surgeons in diagnostic explanations of cases within and outside their domain. The protocols were analyzed by techniques of transforming a propositional representation into a semantic network. Some graph-theoretic criteria for analyzing semantic networks are used for precision of analysis. The results show that the subjects interpret cases in terms of the familiar component of the problem, using specific domain knowledge. This is related to forward directed reasoning. Unfamiliar or uncertain components of the disorder are either ignored or explained using backward reasoning strategies. A tendency to move from a forward driven strategy to a backward driven strategy and viceversa is also seen in some protocols. This sequence is repeated a number of times to form a chain consisting of forward/backward reasoning sequences. This has implications for how subsequent patient information is processed in order to make decisions for treatment and management.

INTRODUCTION

Recent research on medical problem solving has shown that expert physicians solve clinical cases in a way that reflect their familiarization with specific disease types. Studies by Joseph and Patel (1986) and Patel, Arocha and Groen (1986), suggest that when experts have extensive knowledge of the disease classification in a clinical problem they are trying to solve, they work inductively, reaching a diagnostic hypothesis directly from the clinical findings. This has been documented even in situations where the physicians are forced to make tentative hypotheses due to insufficient information to diagnose the case correctly. Joseph and Patel showed that when physicians process a case serially, by reading a segment at a time, experts differ from subexperts (physicians working on a problem that falls outside their area of specialization) in the amount of information needed to reach the accurate diagnosis. That is, the experts correctly diagnose the problem early in the case. Once the experts reach the diagnosis, they interpret further findings as supporting evidence for their first hypothesis, as opposed to subexperts who produce subsequent diagnoses as they process more of the clinical information, generating multiple new hypotheses along the way.

The tendency of physicians to interpret clinical cases in terms of their specialization has also been shown by Patel, Arocha, and Groen (1986). When presented with an endocrinology case, cardiologists interpret some of the information in the text as support for cardiac involvement, even though this constituted a secondary manifestation of the primary disease of hypothyroidism. This situation is more evident when the case is at a high level of difficulty i.e., when the pattern of findings for the case overlap with that of other diseases. Precisely how this occurs was not addressed by the authors.

In a paper by Patel and Groen (1986) it was shown that in order to reach a correct diagnosis cardiologists have to make use, at least partially, of all the components in the case. However, those cardiologists who solve the case of bacterial endocarditis (see Table 1) in a more complete way, made use of the rules linking puncture wounds to intravenous drug use to bacterial infection and the rule linking early diastolic murmur to aortic valve insufficiency. Although most cardiologists recognized the infection component of the disease, all of them focused predominantly on the cardiac aspect of the problem.

The facility with which experts interpret clinical cases within their clinical specialty suggest that a fast process of pattern recognition may be taking place. This has been more clearly documented in domains involving a strong perceptual component, such as radiology (Lesgold, Robinson, Feltovich, Glaser, & Klopfer, in press). Research suggests that forward reasoning may be the counterpart of pattern recognition in more perceptually oriented domains. The issue of perception becomes a pattern of comprehension in a verbally rich domain such as medicine. The research described in Patel and Groen has pointed out that expert cardiologists process cardiology cases in a forward fashion by encoding cases in terms of hypotheses that lead to the diagnoses. Other research comparing the performance of cardiologists and endocrinologists in explanation tasks within and outside their domain specialties suggest that when a case is in the physician's own specialty and is typical of the domain, physicians use forward directed reasoning. Contrarily, if the case is outside the physicians specialty, or is at a high level of difficulty, physicians employ backward directed reasoning.

TABLE 1
ACUTE BACTERIAL ENDOCARDITIS TEXT

THIS 27-YEAR OLD UNEMPLOYED MALE WAS ADMITTED TO THE EMERGENCY ROOM WITH THE COMPLAINT OF SHAKING CHILLS AND FEVER OF FOUR DAYS DURATION. HE TOOK HIS OWN TEMPERATURE AND WAS RECORDED AT 40°C ON THE MORNING OF ADMISSION. THE FEVER AND CHILLS WERE ACCOMPANIED BY SWEATING AND A FEELING OF PROSTRATION. HE ALSO COMPLAINED OF SHORTNESS OF BREATH WHEN HE TRIED TO CLIMB THE TWO FLIGHTS OF STAIRS IN HIS APARTMENT. FUNCTIONAL ENQUIRY REVEALED A TRANSIENT LOSS OF VISION IN HIS RIGHT EYE WHICH LASTED APPROXIMATELY 45" ON THE DAY BEFORE HIS ADMISSION TO THE EMERGENCY WARD.

PHYSICAL EXAMINATION REVEALED A TOXIC LOOKING YOUNG MAN WHO WAS HAVING A RIGOR. HIS TEMPERATURE WAS 41°C, PULSE 120, BP 110/40. MUCUS MEMBRANES WERE PINK. EXAMINATION OF HIS LIMBS SHOWED PUNCTURE WOUNDS IN HIS LEFT ANTECUBITAL FOSSA. THE PATIENT VOLUNTEERED THAT HE HAD BEEN BITTEN BY A CAT AT A FRIEND'S HOUSE ABOUT A WEEK BEFORE ADMISSION. THERE WERE NO OTHER SKIN FINDINGS. EXAMINATION OF THE CARDIOVASCULAR SYSTEM SHOWED NO JUGULAR VENOUS DISTENSION, PULSE WAS 120 PER MINUTE, REGULAR, EQUAL AN DISYNCRONOUS. THE PULSE WAS ALSO NOTED TO BE COLLAPSING. THE APEX BEAT WAS NOT DISPLACED. AUSCULTATION OF HIS HEART REVEALED A 2/6 EARLY DIASTOLIC MURMUR IN THE AORTIC AREA AND FUNDOSCOPY REVEALED A FLAME-SHAPED HEMORRHAGE IN THE LEFT EYE. THERE WAS NO SPLENOMEGALY. URINALYSIS SHOWED NUMEROUS RED CELLS BUT THERE WERE NO RED CELL CASTS.

This paper focuses on the domain-specific knowledge of psychiatrists, cardiologists and surgeons which lead them to emphasize certain components of the case with which the physicians are more familiar. The problem text to be presented describes a subject with a diagnosis of acute bacterial endocarditis. The case has five main components which taken together would lead to an accurate diagnosis. These are as follows: First, the patient is admitted with signs of infection represented by fever, chills, and rigors; more specifically, the signs are an indication of bacteremia or bacterial infection. Second, he shows signs of aortic valve insufficiency indicated by low diastolic pressure and normal systolic pressure, and an early diastolic murmur. Third, the patient presents embolic phenomena suggested by a previous episode of transient blindness, hemorrhage in one eye, and the presence of red cells in his urine. The fourth component is suggestive of an acute process and it is indicated by the short duration of the illness and by the normal size of both the spleen and the heart. This is important because a failure to identify the problem as acute may change the treatment and endanger the patient's life. The fifth component, of social rather than biomedical nature, is suggested by the presence of puncture wounds on the patient's left arm, which together with the fact that he is a young, unemployed male may suggest that the patient is an intravenous drug user, which in turn, may be suggestive of bacterial endocarditis as this disease is common among intravenous drug users.

It is hypothesized that experts focus on their own domain of expertise in diagnostic reasoning because the clinical rules in these domains for solving specific problems are easily accessible. Thus expert psychiatrists explaining the clinical problem would tend to focus on the social and the "illness" aspect of the patient rather than on the biomedical explanation. A surgeon under the same condition would be expected to emphasize the infective process of the disease. A cardiologist, in contrast, would focus on the cardiac problem leading to endocarditis. This paper reports the findings from these predictions using specific and precise methods of analyses.

EXPERIMENTAL METHOD

Rationale:

The empirical paradigm used in the study involves a diagnostic explanation task in which the subject is asked to explain the underlying pathophysiology of the patient's condition. This probe has proved to be useful for assessing differences in knowledge that may underlie the problem-solving process (Patel & Groen, 1986).

We have found that physicians respond to the probe by explaining the patient's symptoms in terms of a diagnosis. The diagnosis is requested after the explanation task, giving the subjects the opportunity to provide a diagnosis during the explanation, such that the resulting protocols may reflect elements of the solution process.

It has been argued elsewhere (Groen & Patel, in press) that in highly verbal knowledge domains, problem solution requires methodologies different from those traditionally used in the literature. The attempt to combine comprehension and problem solving has led some researchers (Kintsch & Greeno, 1985; Patel & Groen, 1986) to utilize a combination of propositional analysis and production rule systems. Another way of representing the same information is through structured representations known as semantic networks, which can be rendered more precise by introducing a few concepts from graph theory (Sowa, 1984; Groen & Patel, 1987). Formally a graph is defined as a non-empty set of nodes and a set of arc leading from a node N to a node N'. A graph is connected if there exists a path, directed or undirected between any two nodes, where a directed path has every node as a source of an arrow connecting to its immediate successor. If the graph is not connected then it breaks down into disjoint components.

In terms of these notions, a semantic network is formed by nodes and connecting paths. Nodes may represent either clinical findings or hypotheses and the paths represent directed connections between nodes. Forward reasoning corresponds to an oriented path from a fact to a hypothesis. Backward reasoning corresponds to a directed path from a hypothesis to a fact. Thus forward directed rules are identified whenever the physicians attempt to generate a hypothesis from the findings in the case. Backward directed rules are identified when a physician first formulates a hypothesis and then attempts to put the data in. The reader is referred to Groen & Patel (in press) for details of the application of graph theory to semantic network.

Description of Method:

The clinical problem selected was the case of ACUTE BACTERIAL ENDOCARDITIS, previously described. It has five sub-components, infectious process, aortic insufficiency, embolic phenonema, acuteness of disease and intravenous drug use.

The subjects included cardiologists, psychiatrists and surgeons, six at each level of specialty.

The general procedure involved presentation of the clinical case, obtaining a free-recall protocol and a pathophysiological explanation protocol, and finally asking for a diagnosis.

RESULTS AND DISCUSSION

Overall summary of results in terms of number of subjects in each specialty that focus on particular components of the disease is provided on Table 2. Here focus refers to providing an explanation for the component. Most of the subjects focus on the infection component of the disease with a strong tendency for the surgeons to providea detailed explanation for the infective process. The cardiologists have easily accessible rules for the cardiac problem, thus they focus on the aortic insufficiency aspect of the problem. In contrast, more psychiatrists than cardiologists or surgeons interpret the problem in terms of social phenomena of intravenous drug use. Surgeons identified and explained the infectious process and the acuteness of the patient's condition more than any other aspect of the disease.

The details of the results are explained with one example from each of the generated explanation protocols from a cardiologist, surgeon and a psychiatrist. Table 3 gives the explanation

protocol of a psychiatrist and Figure 1 presents the semantic network or structural representation generated from the protocol.

This physician organized his protocol around the likely drug injection by the patient which was signaled in the text by the puncture wounds on the patient's arm. The psychiatrist seems to interpret the problems in terms familiar to his specialization, that is, as a drug toxicity problem. He interprets fever as a reaction to the drugs, not to the use of a contaminated needle, i.e., infective component of the disease. The subject does not mention information about the patient's aortic insufficiency or embolic phenomenon. The flame-shaped hemorrhage is also explained from the injected drug rather than from an infectious process.

TABLE 2
NUMBER OF SUBJECTS IN EACH SPECIALTY THAT EXPLAIN THE SPECIFIC COMPONENT OF THE DISEASE

SPECIALTY	NUMBER OF SUBJECTS	INTRAVENOUS DRUG USE	INFECTION	ACUTENESS	AORTIC INSUFFICIENCY	EMBOLI
CARDIOLOGISTS	6	2	4	1	6	5
PSYCHIATRISTS	6	5	5	2	2	2
SURGEONS	6	6	6	5	2	3

TABLE 3
PATHOPHYSIOLOGY PROTOCOL BY PSYCHIATRIST # 2

THE PATIENT HAS BEEN REACTING TO STRESS LIKELY BY HIS INJECTING A DRUG (OR DRUGS) WHICH HAS RESULTED IN TACHYCARDIA, A FALL IN BLOOD PRESSURE AND ELEVATED TEMPERATURE. HE IS IN OR NEAR SHOCK.

THE FLAME-SHAPED HEMORRHAGE MAY REPRESENT A SEQUELA OF AN UPSURGE IN BLOOD PRESSURE POSSIBLY AS A RESULT OF HIS INJECTION OF DRUGS.

The whole structural representation is tied to the hypothesis of drug injection. This hypothesis is causally linked to the two other hypotheses, reaction to to stress and upsurge in blood pressure and to the findings in the case i.e, tachycardia, fall in blood pressure, high temperature, toxicity and the eye hemorrhage. It is important to note that the critical information for the accurate diagnosis of bacterial endocarditis regarding the involvement of heart was completely ignored.

Figure 2 shows the four major components of the structural representation of the explanation protocol. None of the four components separately show any signs of pure forward or backward reasoning. The first component, reaction to some "stress leads to injection of drugs", has both the nodes inferred. The injection of drug is believed to be the patient's reaction to some stress and it is the drug that causes toxic problems. This component explains the injection of drugs. The second component of the network explains the diagnosis of the shock state of the patient resulting in three clinical findings and inference on the findings. The third component explains the diagnosis of the toxic state of the patient induced by the drug. The final component explains the flame-shaped hemorrhage as a result of increase in blood pressure caused by the drugs. It should be noted that everything is explained in terms of the drug induced toxicity and stress. It is the injection of drug aspect of the network that is the overlapping factor in explaining the clinical cues. Table 4 gives the pathophysiological protocol of a surgeon.

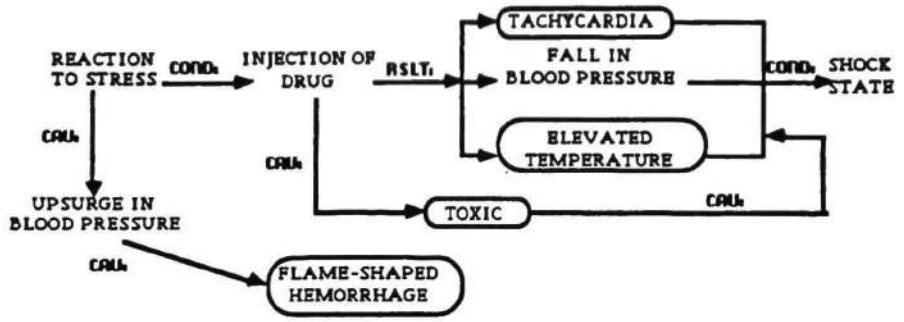

FIGURE 1
STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY PSYCHIATRIST # 2

KEYS FOR ALL FIGURES:

TEXT CUES

COND: conditional links
CAU: causal links
RSLT: resultive links
LOC: locative links
EQUIV: equivalence links

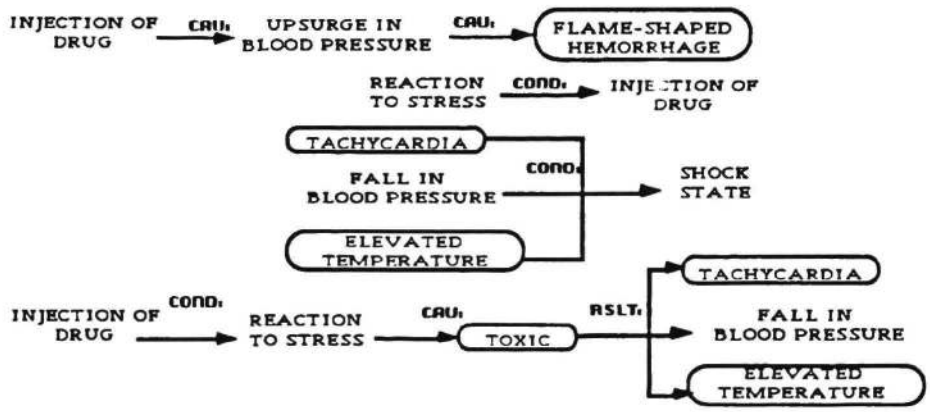

FIGURE 2
COMPONENTS OF STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY PSYCHIATRIST # 2

TABLE 4
PATHOPHYSIOLOGY PROTOCOL BY SURGEON # 3

ACUTE FEBRILE ILLNESS OF SOME SEVERITY, APPARENT BACTEREMIA OR VIREMIA. BACTEREMIA SEEMS MORE LIKELY BECAUSE OF THE HIGH FEVER, CHILLS, TOXICITY, AND IMPLICATION OF THE HEART I.E., POSSIBILITY OF ACUTE BACTERIAL ENDOCARDITIS, MULTIPLE ORGAN INVOLVEMENT, E.G., EYES, KIDNEYS - POSSIBLE SEPTIC EMBOLI. APPROPRIATE INVESTIGATIONS WOULD DEFINE THE ETIOLOGY. LIKELY SECONDARY TO INJECTING HIMSELF WITH DIRTY NEEDLES.

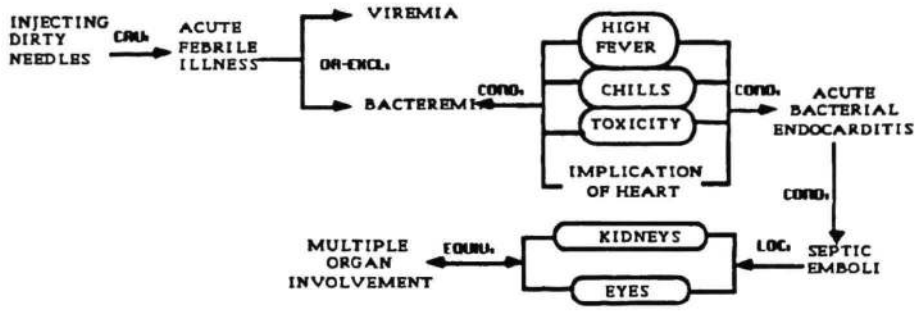

FIGURE 3
STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY SURGEON # 3

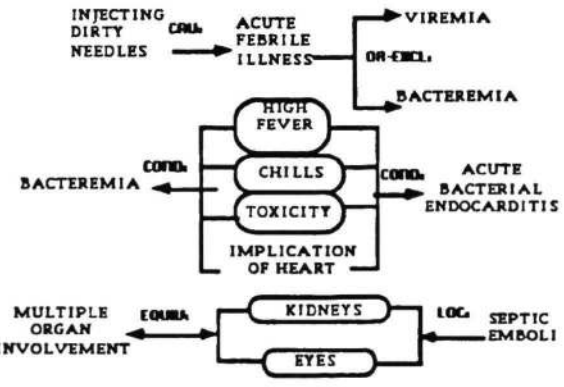

FIGURE 4
COMPONENTS OF STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY SURGEON # 3

Figure 3 shows the structural representation of the case by a surgeon. He identifies the problem as acute febrile illness of either viral or bacterial origin. The surgeon decides that the patient has bacteremia because the patient has fever, chills, toxicity and has implications of heart involvement. His differential diagnosis is acute bacterial endocarditis. Given endocarditis, the most likely source of infection is from a dirty needle. The above description can be decomposed into three component of the representational network (Figure 4). The first component deals with acute fever due to dirty needles leading to bacteremia. The second component also deals with explaining bacteria from the given clinical cues and make the secondary diagnosis of acute bacterial endocarditis. The third component deals with septic embolism. Each of the three components deal

with acute infection or sepsis, a condition that surgeons have to be very familiar with in their practice.

Figure 5 gives a structural representation of the protocol generated by a cardiologist and Figure 6 provides the four major components of this representation.The major diagnosis is of endocarditis affecting the heart valve. The mixture of forward and backward reasoning towards a diagnostic hypothesis of endocarditis affecting aortic valve as seen in component one. It should be emphasized that except for noting that infection exists, it is not explained in any details. The second component also deals with aortic valve involvement and is again a mixture of forward and backward reasoning. The use of intravenous injection by the patient is not mentioned. The third component uses a pure forward reasoning to explain the aortic valve involvement from two given textual cues. The final component also deals with the cardiac problem although it does not have any relationship to the overall coherence. All the components of the network deal with cardiac problem with little focus on the infective process.

The results show that a psychiatric emphasizes the drug toxicity and stress reaction part of the problem, a surgeon emphasizes the acute infective nature of the problem, and a cardiologist focuses on the heart valves and heart murmur that relate to the disease. The subjects make extensive use of their domain knowledge to represent the clinical problem. A mixture of forward and backward reasoning is seen in all the cases. A more forward directed reasoning strategy is used in a familiar situation and a more backward reasoning strategy is used in an unfamiliar situation.

TABLE 5
PATHOPHYSIOLOGY PROTOCOL BY CARDIOLOGIST #7

THE DIAGNOSIS APPEARS TO BE ENDOCARDITIS, THE SITE IS THE AORTIC VALVE. IT IS AORTIC RATHER THAN PULMONARY BECAUSE 1) LOW DIASTOLIC PRESSURE, 2) EVIDENCE OF EMBOLI IN EYE AND URINE, RBC CASTS, 3) HYPERDYNAMIC SYSTEMIC CIRCULATION. EXAMPLE, THE INTENSITY OF FIRST HEART SOUND. IT SHOULD BE DEMONSTRATED WITH PREMATURE CLOSING OF ATRIAL VALVE. DYSPNEA MAY POINT TO EARLY HEART FAILURE, A VERY OMINOUS SIGN WHICH MAY DEMAND EARLY SURGICALINTERVENTION.

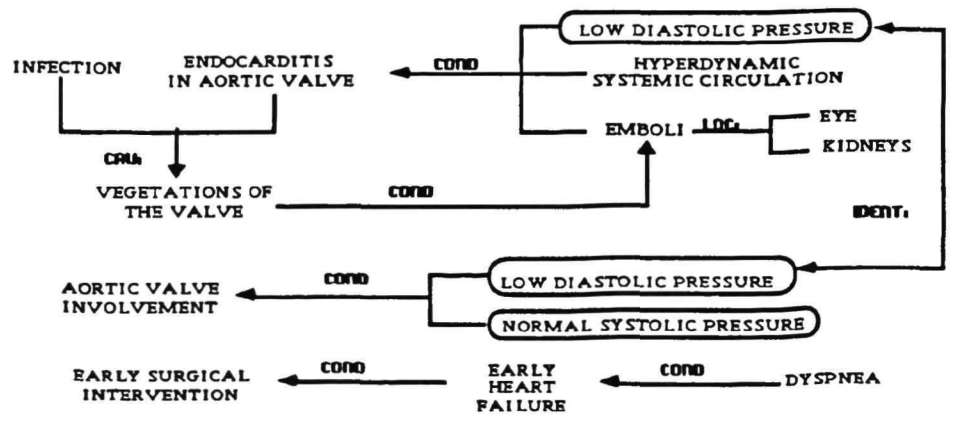

FIGURE 5
STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY CARDIOLOGIST 7

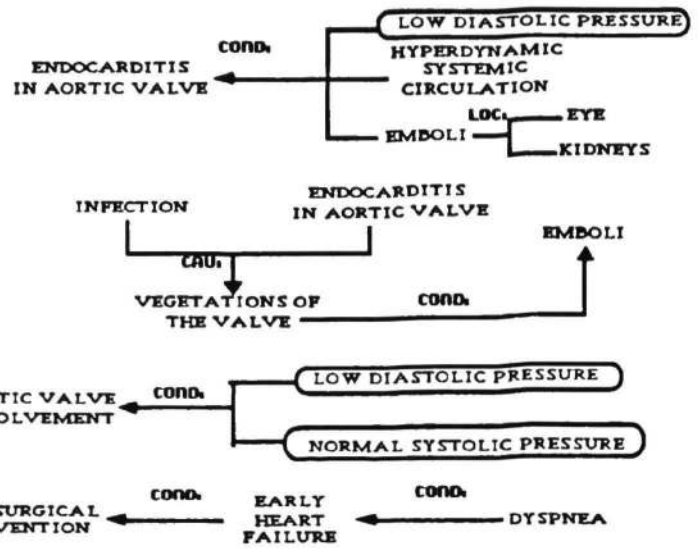

FIGURE 6
COMPONENTS OF STRUCTURAL REPRESENTATION OF ACUTE BACTERIAL ENDOCARDITIS PROBLEM BY CARDIOLOGIST # 7

ACKNOWLEDGEMENTS

This work was supported by a grant from the Josiah Macy Jr. Foundation (B8520002) and the National Science Engineering Research Council of Canada (A2598) to Vimla Patel. We would like to acknowledge the support of all the medical specialists who took part in our study and to Susan Young for typing the manuscript.

REFERENCES

Groen, G.J. and Patel, V.L. (in press) The relationship between comprehension and reasoning in medical expertise. In M. Chi, R. Glaser & M. Farr (Eds.) *The Nature of Expertise.*

Joseph, G.-M. and Patel, V.L. (1986) Specificity of expertise in medical reasoning. In *Proceedings of the Eighth Annual Conference of the Cognitive Science Society* (pp.331-343). Hillsdale, N.J.: Lawrence Erlbaum.

Kintsch, W. and Greeno, J.G. (1985) Understanding and solving word arithmetic problems. *Psychological Review, 92*: 109-129.

Lesgold, A., Robinson, H., Feltovich, P., Glaser, R. and Klopfer, D. (in press) Expertise in complex skills: Diagnosing x-ray pictures. In M.Chi, R. Glaser, & M. Farr (Eds.) *The Nature of Expertise.*

Patel, V.L. and Groen, G.J. (1986) Knowledge based solution strategies in medical reasoning. *Cognitive Science, 10*, 91-116.

Patel, V.L., Arocha, J.F. and Groen, G.J. (1986) Strategy selection and degree of expertise in medical reasoning. In *Proceedings of the Eighth Annual Conference of the Cognitive Science Society* (pp.780-791). Hillsdale, N.J.: Lawrence Erlbaum.

Sowa, J.F. (1984) *Conceptual structures..* Reading, MA.: Addison Wesley.