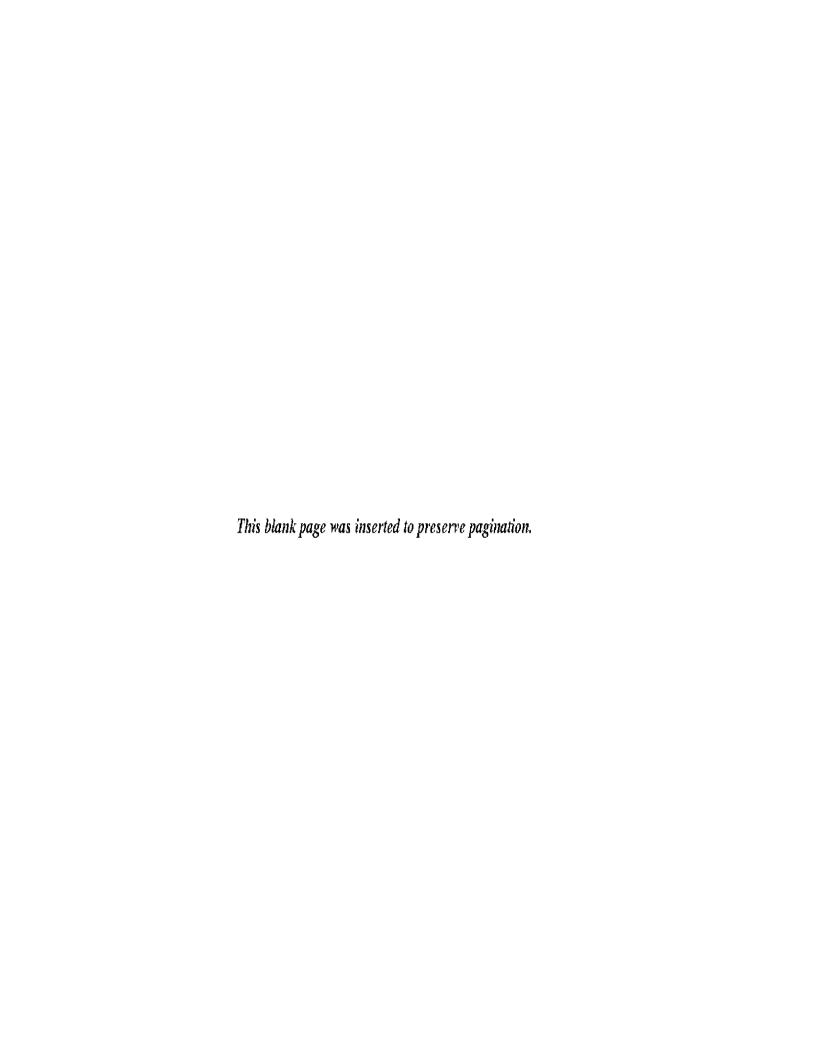
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STRATEGY SELECTION IN MEDICAL DIAGNOSIS

Peter B. Miller

September 1975



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Strategy Selection in Medical Diagnosis

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PROJECT MAC

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STRATEGY SELECTION IN MEDICAL DIAGNOSIS

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ABSTRACT

The recorded, verbal problem-solving behavior of doctors performing the diagnostic task of taking a present illness was analyzed in this research. The goal of the analysis was to discover what data-acquisition strategies were used by the doctors to accomplish the task. A model called the strategy frame model was dreated to describe the strategies that were found and to provide a mechanism for the selection of a strategy. In this model strategy selection is determined by the problem space of the doctor—his internal diagnostic configuration. A scheme for classifying strategies as confirmation, elimination, discrimination or exploration was also developed.

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THESIS SUPERVISOR: Professor G.A. Gorry
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CHAPTER 1

INTRODUCTION

"We're experts. We ask the right questions."
----- Henry Block (of H. & R. Block)

To begin, I ask that the reader imagine the following scene: A middle-aged woman enters a doctor's office at a large metropolitan hospital. She tells the doctor that she has felt nauseous and has been vomiting. She also tells him that she has had abdominal pain and has had to urinate quite often during that time. The following dialogue ensues:

- D: How long have you had the nausea and vomiting?
- P: For about three weeks.
- D: Did the nausea and vomiting start before the increased urination?
- P: They happened about the same time.
- D: Did you have a burning sensation when you urinated?
- P: Yes I did. Almost all the time.
- D: Have you lost any weight during the past month?
- P: Yes, I've lost about ten pounds.
- D: Do you remember having any fever or chills?
- P: I've been having some bad chills?
- D: Did your urine appear dark or bloody?
- P: No.
- D: Do you have any pain in your side, in the flank area?
- P: Yes, on the left side.

While the dialogue above is hypothetical, it is realistic enough to give a flavor of the kind of interchange that actually takes place in most instances when a doctor first encounters a patient. Doctors call this initial interview with a patient taking the present illness. This activity, the initial stage of data acquisition in the process of

formulating a diagnosis, is one in which virtually all doctors participate every day. By the end of the interview a doctor will have gathered enough information to guide him in making the necessary decisions in the management of the petient including further diagnostic procedures and initial therapeutic decisions. These decisions will be based, in large measure, on the diagnostic hypotheses formed in the process of taking the present illness.

The motivation for the research I undertook was the fundamental question that arises when one examines the process of taking the present illness - What is the origin and research for each of the specific questions asked by the doctor?

1.1 Methodologu

The method I chose to attack the fundamental question is to model the process of taking the present illness. In the design of models of the clinical decision-making process, two distinct approaches have been used. The first, the <u>normative</u> approach, emphasizes the development of models that are <u>prescriptive</u>. The decisions made by a normative model are said (under certain assumptions) to be <u>optimal</u> decisions. If this is true, it is claimed that decisions <u>ought</u> to be made this way, disregarding the way that doctors make the same decisions. While in most cases no real claim of optimality can be made, the normative approach has had some success in certain limited areas <Gorry 73>.

The second approach, the development of a <u>descriptive</u> model, is the one I chose to attack the fundamental question. A model based on this approach seeks to describe the actual decision-making process of clinicians performing a problem-solving task such as diagnosis. The basis for the model I have developed is the recorded and transcribed verbal behavior of doctors taking a present illness called a <u>protocol</u>. In the analysis of a protocol, the verbal behavior of the doctor is seen as a record of the sequence of steps taken by the doctor in solving a diagnostic problem.

Protocol analysis has been used by different researchers in various problem domains. Newell and Simon Here among the first to apply this technique to aid in the understanding of human problem solving <Newell and Simon 72>. In the area of medical diagnosis, Kleinmuntz analyzed the protocols of neurologists diagnosing eight areas of neurological disease and compared the performance Hithin different levels of clinical experience <Kleinmuntz 68>. Dombal examined the differential diagnosis of abdominal pain again using clinicians Hith varying degrees of expertise <Dombal 73>. Recently, Rubin used protocol analysis applied to a case of presenting hematuria as a basis for a model of hypothesis formation and verification <Rubin 74>. A comprehensive survey of protocol analysis and other "process tracing" methodology has been compiled by Schulman and Elstein <Schulman and Elstein 74>.

1.2 Why Protocol Analysis?

An as yet unrealized goal of research into the diagnostic process is a comprehensive theory of how doctors obtain, assimilate and evaluate medical data — what Gorry has termed the process of clinical cognition:

"The major reason that cognitive psychology has made relatively little progress with respect to understanding behaviors as complex as that involved in clinical decision-making is because there was a serious shortage of ways to describe the more procedural aspects of that behavior."

Thus there is a crucial link between our understanding of clinical behavior and our ability to describe it. And further, it is my belief that our ability to replicate behavior in the form of competent programs that embody the level of expertise found in highly-trained clinicians is predicated on our understanding of how doctors perform clinical tasks such as diagnosis in their day-to-day practice. I believe that protocol analysis is the best tool available for constructing descriptive models of clinical behavior as well as yielding a data base upon which to test theories of clinical cognition <Gorry 74>.

An alternative to protocol analysis in deriving descriptions of a doctor's problem-solving is introspection. In this approach, a doctor is asked to think about and report on how he solved a diagnostic problem. While this can also be a useful tool, there are some serious drawbacks that forced me to reject it as an experimental approach. The major problem with introspection is validation. There is no way to confirm that the problem

uas actually solved by the doctor in the way that he described. In contrast, a protocol reveals each of the separate steps taken by the doctor in his problem-solving process. Because introspection can yield useful insights, in the experiment I performed the doctors were encouraged to report their current thinking about the problem in addition to asking for data.

1.3 Goals of the Protocol Analysis

The original question - the origin and reason for each question asked by the doctor - I felt was to broad and general to have any hope of my giving a complete or definitive answer. Instead, I have focused on one specific issue raised by this question - the data-acquisition strategies used by the doctors. In particular, the goals I set for my analysis were the following:

- 1. Determining what strategies are used by doctors in the gathering of data for the purpose of diagnosis.
- 2. Developing a model to describe these strategies.
- Including in this model a mechanism to describe the selection of a particular strategy.

1.4 A Description of the Experiment

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1.4.1 Experimental Design and Constraints

With the aid of Dr. Jerome Kassirer, I selected a case history from the patient records of the Tufts-New England Medical Center. Each of the six doctors who participated in the experiment was given the following instructions:

- You will be presented with a case, initially starting with the age, sex and chief complaints of the patient.
- 2. You will take the present illness of this patient with the following conditions:
 - A) Dr. Kassirer will not directly simulate the patient in giving answers to your questions. Instead, he will answer any question you ask about the patient from the point of view of a medical person who knows as much as can be known about the medical history of the patient.
 - B) The questions you ask should be as specific as possible, asking for specific facts. General questions such as "What complaints did the patient have in the past?" will not be answered.
 - C) When you ask a question you should provide a reason for asking it.
 - D) You are to tell what you learned from the answer to the question. You should also report any hypotheses you are considering.
 - E) When you feel satisfied that you have reached a final diagnosis or feel you have gone as far as you can, you can stop asking

questions and summarize the case.

The entire dialogue was recorded and transcribed. Of the six doctors who participated in the experiment, four were renal Fellows at the NEMC, one was a gastroenterologist and one a cardiologist also from the NEMC.

1.4.2 Criteria for Case Selection

A number of considerations were discussed in the selection of a case to be used. An important fact not directly connected to the experiment itself was that the case be centered on a renal problem. This was deemed necessary because Dr. Kassirer, who would be answering the questions in the experiment is a renal specialist and indicated that he felt most comfortable in this area.

The criteria that did directly bear on the experiment were:

- 1. The case chosen should be one that a doctor might see in the everyday course of his clinical practice in the hospital. The case should not be a "trick" case involving a very obscure disease.
- 2. The case should be normal in the sense that the clinical presentation and history should be both suggestive and consistent with the final diagnosis. No effort was made, however, to find a "classic" case for a particular disease. Both aspects were important because the experiment was designed to capture data about "standard" presentations rather than being a test of diagnostic skill.
- 3. The case should be rich in history with enough data available about the medical history to provoke consideration of a number of possible diagnostic options. It was felt that the case should contain

an element of chronic disease as well as an acute presentation in that we wished to see if different approaches were used for the two different categories or what effect one had on the other.

1.4.3 The Case

The case that was finally chosen was one of a 57 year old woman who presents at the hospital with the chief complaints of nausea, vomiting, abdominal pain and frequency of urination. The patient had a history of a previous hospitalization for kidney stone removal and a hospitalization for urinary tract infection. At the actual time the patient came to the NEMC the doctors felt that the patient's findings were due to a combined etiology. The diagnosis made then and listed on the discharge summary was:

- 1. Acute pyelonephritis (APN)
- 2. Chronic renal disease (CRD)
 either a) Chronic pyelonephritis (CPN)
 or b) Phenacetin nephritis
 or both together.
- 3. Chronic renal failure (CRF)
- 4. Metabolic acidosis, secondary to CRF.
- 5. Anemia secondary to CRF and folic acid deficiency.

Each doctor was told at the start that the patient was being discharged from the hospital after a three week hospitalization.

1.5 Preview

In Chapter 2 an analysis of a protocol is presented and discussed. In Chapter 3 a model for the description and selection of data-acquisition strategies called the <u>strategy frame model</u> is presented. In the last chapter a classification system for strategies is presented.

CHAPTER 2

THE ANALYSIS OF PRESENT ILLNESS PROTOCOLS

A present illness protocol is a record of the problem-solving behavior of a doctor performing diagnosis. The analysis of each protocol was directed towards uncovering the strategies used by a doctor in performing this task. The kernel of the analysis was the assignment of a set of goals and methods to each question. The strategies used by the doctor were then specified through the goal-structure for each question and the relationships among the goals for different questions.

The major part of this chapter consists of the analysis of one of the protocols. The analysis consists of two components — a formal and an informal one. The formal component specifies (among other things) the goals and methods for each question and the relationship of the goals to the current strategy of the doctor. The informal component is a commentary that seeks to explain in greater detail the medical facts that the doctor used. It attempts to provide a reader whose background in medicine is limited insight into the interpretation made by the doctor of the data that was presented.

In analyzing the protocols it was felt that it was important to try to maintain as wide a perspective as possible. By this I mean that the broad outlines and overall patterns of questioning were kept in mind as well as the specific details of each question. One interesting (but not unexpected) discovery was the tendency of the doctors to digress from a

principal line of questioning in order to obtain data that they thought might be significant at a later stage in the interview and then to resume the principal line again. While examples of classical problem-solving techniques such as recursion, depth-first and breadth-first search and back-up were found, most of the strategies were not "pure" but pragmatic. There was considerable jumping around among different areas of concern, multiple focusing and conditional (priority) interupting. Redundant questions were asked. (One doctor asked the exact same question at three different times in the interview.) Extra or "unnecessary" questions were also asked. One hypothesis that was considered was that some of the questions were asked simply to give the doctor time to think of a better one.

2.1 Formal Annotation Scheme

The following is a description of the components of the formal annotation scheme:

- A. Question A verbatum reproduction of the question asked by the doctor.
- B. Data Requested The specific datum the doctor wanted.
- C. Goals What the doctor hoped to accomplish by obtaining

the requested data. In making the decision about assigning goals many factors were weighed; the reason the doctor supplied, the current context of questioning and the opinion of Dr. Kassirer as to the possible interpretation of the data.

Structural abstraction was the guiding principle in formulating the statement of the goal. By this I mean that wherever possible the goal that is stated is formulated in terms of the structural relationship between the data and a specific knowledge "chunk" describing a disease, clinical state, etc Included in the goal statement in these cases is the instantiation of the abstract version with the specific entities under consideration filled in. If a goal is a subgoal of a higher-level strategy, the goal structure is also given.

Two types of goals where assigned to each question, a primary goal and (where applicable) a set of secondary goals.

1) Primary goal - The assignment of the primary goal represents an estimate of the principal purpose for asking the question through an evaluation of the most important medical significance of the data sought in the context in which the question was asked. If there where clearly two or more equally significant implications that could be drawn from the data, this was represented as a multiple primary goal.

- 2) Secondary goals Many of the questions asked by the doctors where noncommittal. For example, most of the doctors asked "What were the values of the renal function tests?" even when they strongly suspected that the values would be elevated. Even though they were directed to be as specific as possible in their questions, they tended to ask questions that were very broad in terms of the range of answers that could be given. It was felt that the doctor had thought about the range of possible payoffs that could be obtained from a question. It is, of course, impossible to be certain a postiori whether they did or not. Upon retrospective examination, many insisted they did have these things in mind. It was felt in annotating the questions that these possible payoffs should be included in the form of secondary goals.
- C. Methods Associated with each goal is the method used to obtain the goal. The methods can be <u>direct</u> or <u>indirect</u>. In a direct method the finding of interest is asked for. In an indirect method the premise of a rule that associates the finding of interest with the some other evidence is asked for.

- D. Expectation If the doctor had some expectation of uhat answer he would get to a question, it was noted. These expectations are classified as strong, moderate, weak or uncommitted.
- E. Answer The data supplied by Dr. Kassirer in response to the question.
- F. Result To what extent was the primary goal satisfied.

 To what extent were the secondary goals satisfied.
- Possibilities list (PLIS) The PLIS (a list of ough to being thateleanh whom book hypotheses) is a representation of the doctor's thinking Tarritron - Life on italiana di rigliaba di Labori. about the present illness of the patient after hearing on the college of the college of the college of the answer to his question. It is by no means the complete pacos y wheathy he m representation but reflects the most significant part in terms of the final diagnostic conclusions Above PERO Series Amerik the doctor makes. The possibilities list is divided into fier off. . As each ocamin. six parts: CONFIRMED, SATISFIED, LIKELY, POSSIBLE, UNLIKELY reference and border does the colored and RULED-DUT. The first hypothesis on the LIKELY list is 自然的一点,我在你们一点了你去的一定<mark>数数保护数据的一点数,给</mark>第二个人的办法 called the principal disease hypothesis (PDH).
- H. <u>Doctor's Commentary</u> Any reasons or explanation supplied by the doctor.

2.2 Analysis of Protocol 1

Many of the discoveries made about strategy are best presented in the context of an analyzed protocol. The protocol that is presented here was chosen because the doctor exhibits a wide range of strategies and touches most of the important issues that the particular case that was used raises. The subject is a Fellow in the Renal Department of the New England Medical Center.

Initial Presentation - This is a 57 year old lady who is being discharged from the hospital after a three week hospitalization. Just before she came into the hospital her complaints were nausea, vomiting, abdominal pain and frequency of wrination.

Initial Possibilities List -

Confirmed: None

Satisfied: None

Likely: Acute pyelonephritis (APN) = PDH

Acute lower urinary tract infection (ALUTI)

Possible: Chronic renal disease (CRD)

Chronic renal failure (CRF)

Acute GI disease Chronic GI disease

Unlikelu: None

Ruled-out: None

Initial Commentary — The initial presentation is a set of findings that, even before interpretation, has a fairly complex structure. The symptoms of nausea and vomiting are specifically GI but commonly occur in many forms of renal disease. Abdominal pain is too general a finding to refer to any particular organ system while frequency of unination is a very specific unological symptom. Acute lower uninary tract infection is a reasonable initial hypothesis; it is activated by the uninary frequency. In addition, women are twice as likely to get one as men. ALUTI by itself is not sufficient, however, to explain all the symptoms known thus far. An acute uninary tract infection (AUTI) can start in the lower uninary tract (bladder) and retrograde up the uninary tract to infect the kidney and produce acute pyelonephritis (The inflamatory reaction and interstitial

lesions of the kidney due to infection). APN is normally used to refer to the infection of the entire urinary tract including the kidney and bladder.

Because APN includes the findings of ALUTI as a principal-part (a principal-part of a disease is a subset of related findings of the disease that is viewed as a distinct clinical unit), it is a better hypothesis and could explain all of the know symptoms if the following are true:

- 1. The abdominal pain is abdominal flank pain
- 2. The onset of the symptoms was sudden.
- 3. The duration of the symptoms had been fairly short.
- 4. There were systemic manifestations of infection.

Another factor that the doctor must take into consideration is the length of the hospitalization. Normally, uncomplicated acute pyelonephritis is treated with antibiotics and clears up within a week. The patient was hospitalized for a period of three weeks, however. This would make the doctor suspect that either the illness was very severe, resistant to antibiotic therapy or that the patient had more problems, specifically, some chronic condition. Thus, it is reasonable for the doctor to hypothesize as possible a chronic renal disease that is being complicated by an acute urinary tract infection. It is a fact that many chronic renal diseases make a patient more susceptible to urinary tract infection. In addition, certain chronic renal diseases can lead to chronic

renal failure for which nausea and vomiting are key symptoms. At this point the doctor can not rule out the possibility that he is not looking at a renal problem but a GI problem or a combined renal and GI problem.

Question 1 - How long has she had the nauses and vomiting?

Data Requested - Duration of the nauses and vomiting.

Goals

Primary - Discriminate Acute vs. Chronic illness subsoal to Explore time-pattern of the illness.

Secondary - 1. Confirm symptom time-duration component of APN prototype symptom time-pattern of APN prototype symptom to Confirm APN prototype symposi to Confirm APN subgoal to Confirm APN.

2. Explore the need for immediate treatment of a clinical condition that can be caused by a reported symptom: severe, short-term vomiting can cause dehydration, alkalosis.

Methods - P: Indirect: The duration of the nausea & vomiting Is-Suggestive-Evidence-For the duration of the illness. S1: Direct.

S2: Indirect: The duration of the nausea & vomiting Is-A-Suggestive-Measure-Of the likelihood of dehydration and/or alkalosis.

Expectation - Moderate : Less than 2 weeks.
Strong : Less than 3 weeks.

Answer - She had the nauses and vomiting for three weaks before coming into the hospital.

Result - P: Partially satisfied \$1: Satisfied \$2: Partially satisfied

PLIS - Unchanged

Doctor's Commentary - The reason for the first question is to get a time

course as far as the patient's illness is concerned.

Commentary - The strategy chosen by the doctor reflects his decision to first focus on the acute vs. chronic characterization of the illness. As a method for achieving the goal he uses a heuristic (indirect method) which says that the time duration of the symptoms is a good indication of the "acuteness" of the illness. The sumptomatic duration (of the nausea and vomiting) is just consistent with an acute process and is suggestive of an underlying chronic process: for this reason the primary goal is only partially satisfied. As a part of the strategical decision, the doctor chose to ask about the duration of the nausea and vowiting rather than the abdominal pain or the frequency. This can be explained by the following argument: of the three reported sumptoms (nausea and vomiting get linked together) the patient would have most likely remembered when the nausea and vomiting began since this is a very distressing condition. Another important reason for choosing this symptom is that by itself, nausea and vomiting is a serious condition if it has occurred for an extended period of time. This relates to the secondary goal of exploring the need to treat such clinical conditions as dehydration (with resulting loss of renal function or even damage to the kidney), acute weight loss or metabolic alkalosis that can result from an extended period of nausea and vomiting.

There can be no doubt that the doctor was also thinking of whether the symptomatic duration is consistent with his principal disease hypothesis.

This is reflected in the assignment of the first secondary goal. This goal

is a subgoal of a higher-level strategy that the doctor has invoked called case-building. This strategy is invoked when the doctor wishes to confirm a hypothesis he views as likely. (See Section 4.1.2.1 for a detailed description of case-building and its variants.) The particular variant of case-building the doctor is using requires determining if the prototype of the principal disease hypothesis matches the patient's findings. (A disease prototype consists of the signs and symptoms that a doctor would expect a patient to have if the patient had the disease.) This is accomplished by setting up as subgoals the confirmation of each component of the disease prototype. For the hypothesis of APN the goal-tree is the following:

(GOAL (CONFIRM APN)
(TO (CONFIRM APN) (CASE-BUILD APN)
(TO (CASE-BUILD APN) (CONFIRM (PROTOTYPE APN))
(TO (CONFIRM TIME-PATTERN

(TO (CONFIRM TIME-PATTERN)

(AND (CONFIRM (BURATION (< (3 WEEKS)))))

(CONFIRM (ONSET SUCCEN))))
(CONFIRM SYSTEMIC-INFECTION)
(CONFIRM KIDNEY-INFLAMATION))))

Question 2 - Did the nausea and vomiting begin before the frequency or did the nausea and vomiting occur after the frequency had occurred for several days?

<u>Data Requested</u> - Sequence and relationship of symptom development.

Goals
Primary - Discriminate GI etiology of symptoms from renal etiology subgoal to Explore organ-system of disease origin.

- Secondary 1. Confirm symptom onset-pattern of APN prototype subgoal to Confirm principal-part: symptom time-pattern of APN subgoal to Confirm prototype APN subgoal to Case-build APN subgoal to Confirm APN.
- Methods P: Indirect: Characterize the time sequence of symptom development.
 S1: Direct.
- Expectation Strong: The nausea and vomiting and the frequency occured within a few days of each other.

 Moderate: The frequency preceded the nausea and vomiting by a few days or both symptoms occurred together.
- Answer The occurrence was pretty much simultaneous.
- Result P: Partially satisfied
 S1: Satisfied
- PLIS Unlikely: Acute GI disease Chronic GI disease

Doctor's Commentary - The reason for that question is to try to determine whether the nauses and vomiting is the primary problem, trying to see which came first and which is the secondary sequence.

Commentary - This question is very complex in terms of the kind of information that could have been obtained. The doctor might have been given many different answers, each one of which would have a different interpretation. The doctor's primary concern here, however, is to determine the ethology of a complex of symptoms from different organ systems. The first step towards achieving this goal is to determine which organ system is responsible. In order to put this into perspective consider the range of interpretations if the answer had been that the

nausea and vomiting had preceded the frequency by many days.

- 1. An independent for unrelated 51 presion and unotogical problem developed at about the same time.
- 2. The origin was the GI system (an infection)
- 3. There was a chronic renal disease which developed into chronic renal failure and preceded the urinary tract infection.
- 4. The universe tract infection developed by the descending (blood-borne) routs.

The hypothesis of independent problems that develop in two different organ systems at about the same time is not considered very likely by most doctors. Doctors use the principle of barsimony in considering explanations for findings. This principle says that the simplest explanation should be considered before a more complex explanation.

Clearly, a set of independent (unrelated) problems is not the simplest explanation. The doctor did not expect to hear that the names and vomiting had been a chronic condition for several months (or years). If this had been the case he probably would have opted for the GI system as the cause of both symptoms. The fact that the onset of all the symptoms was simultaneous is consistent with his principal disease hypothesis but does not completely rule-out a GI etiology.

Question 3 - Along with the frequency was there any burning on urination, any dysuria?

Data Requested - Presence of dysuria observed by the patient.

Goals

Primary - Confirm principal-part: Bladder irritation of APN prototype subgoal to Confirm prototype ARN.

Methods - P: Direct: Dusuria Is-Prima-Facie-Evidence-For Bladder irritation.

Expectation - Strong : Dysuria present.

Answer - Yes, she did complain of some burning on urination.

Result - P: Satisfied

PLIS - Confirmed: Bladder irritation (of APN)

Commentary - The prototype for bladder irritation/inflamation specifies specifies frequency or urgency and dysurfa. Since the frequency is siready known, the doctor asks for what now can be considered prima facile evidence for a bladder irritation, the presence of dysuria. The doctor is now clearly focusing on the principal disease hypothesis and is attempting to confirm it by confirming each principal-part.

Question 4 - Had she gained or lost weight during that 3 week interval? Data Requested - Amount of weight-loss or weight-gain.

Goals

Primary - P1: Explore the severity of the illness. P2: Confirm a previously reported finding: three week period of nausea and vomiting.

Secondary - 1. Eliminate Acute GI problem.

Methods - P1: Indirect: Weight-lass Is-A-Suggestive-Measure-Of Severity. P2: Indirect: Assess a finding that will validate and/or support a previously reported finding: Weight-loss due to nausea and vomiting. S1: Indirect: No weight-lose Is-Negative-Evidence-For

Acute GI problem.

Expectation - Moderate: Height-loss 5 - 15 lbs.

Strong: Any weight-loss.

Heak: Stight weight-gain.

Answer - She lost 18 lbs. of weight in the two weeks before admission.

Result - P1: Partially satisfied.
P2: Satisfied.

S1: Partially satisfied.

PLIS - Unchanged.

Page 1

Doctor's Commentary - That is to assess, if you will the severity of the illness. If she had gained several pounds during that time you would be much less likely to believe the information about the nausea and vowiting. Whereas with the 18 ib. weight-loss, again from the patient's history, that makes that more believable.

Commentary - There are two points of interest about this question. The first is that it is the first example in the protocol so far of a multiple primary goal. The second but related point is that it seems as if the focus has immediately shifted away from case-building for the principal disease hypothesis. It is, in fact, not so much a shift away than a broadening of the focus to include another espect of the patient's condition that is significant by itself and also provides some evidence for the sufficiency of the principal disease hypothesis law an explanation for the findings). In developing questions to ask, the doctor always has a set of goals that he would like to satisfy. Among them are:

- 1. Arrive at a satisfactory diagnosis
- 2. Assess the need for immediate treatment
- 3. Gather sufficient information to begin formulating a therapeutic plan.

- 4. Develop a prognosis for the patient.
- 5. Determine what further information needs to be gathered as part of the management plan for the patient.

In the early stages of the interview, when most of these goals have not yet been satisfied, a good strategy is try to develop questions that might satisfy as many of these goals as possible.

In the present case, while severity is not specifically a part of the prototype for APN, the doctor has a fairly good idea of the range that can be expected. Clearly, a weight-loss of 30 lbs or more would make him suspect a serious gastrointestinal problem. It should be noted that, in general, the amount of weight-loss is a good indicator of how acutely ill the patient is.

The question also serves as a check on the degree of nausea and vomiting. The doctor could have asked another set of questions in order to characterize the degree of nausea and vomiting. The information about weight-loss, however, is sufficient to indicate a level that is consistent with the reported time duration and also with the principal disease hypothesis.

Question 5 - Was she febrile or having chilly sensations during the three week interval between the time of the onset of her symptoms and the time or presentation to the hospital?

Data Requested - Presence of fever or chills.

Goals

one of some contraction of the contraction of the

Primary - Confirm principal-part: Systemic evidence of active infection of APN prototype subgoal to Confirm prototype APN.

Methods - P: Indirect: Fever or chills 1s-Strong-Supporting-Evidence-For Active infection.

Expectation - Moderate: Fever or chills present.

Answer - She said she had chills but her temperature wasn't taken.

Result - P: Satisfied

PLIS - Satisfied: Active infection (of APN).

Doctor's Commentary - I am trying to establish whether or not this is an infectious disease.

Commentary - In this question the doctor has returned to confirming the APN prototype as his primary goal. An essential feature of APN is that it produces systemic findings associated with an active infection such as fever or chills. This is normally not the case for an acute uninary tract infection that is restricted to the lower uninary tract (cystitis). The fever associated with APN can be quite high (181 - 183) and can be a serious condition in an older person if it has persisted for any length of time. It is quite possible there is an ordering consideration in confirming each principal-part of the disease prototype: the more serious and potentially health- (or life-) threatening symptoms are asked about first. This would tend to show that even in diagonatic games the fundamental concern of the doctor is focused on the well-being of the patient. This kind of subtle (but demonstrable) strategical choice will probably have to be embeded into a present lilness program for it to be

Question 6 - Along with the dysuria and the frequency were there any episodes of gross hematuria where she passed blood or dark urine?

Data Requested - Presence of gross hematuria (observable by the patient).

Goals

Primary - P: Eliminate a complication of the PDH (= APN): Hemorrhagic custitis.

Secondary - S1: Explore diseases that are complicated by the PDH:
Obstruction, renal tumor, renal calculi.

Methods - P: Direct: Absence of gross hematuria Is-Prima-Facie-Evidence-Against Hamorrhagic custitis. S1: Indirect: Gross hematuris Is-Suggestive-Evidence-For Obstruction, renal calculi.

Expectation - Moderate: No gross hematuria.

Answer - No.

Result - P: Satisfied
S1: Partially satisfied.

PLIS - Ruled-out: Hemorrhagic custitis.

Commentary - There are a number of possible interpretations of this question. While the focus is still on the PDH it is not completely clear whether the primary goal is part of case-building or elimination of a complication. The reason that elimination of hemorrhagic cystitis was chosen was that the doctor later told me that is what he had in mind. The absence of gross hematuria could be a component in the doctor's prototype for APN, however. As far as the secondary goal is concerned, urinary tract infection is a common complication of diseases such as urinary tract

obstruction, renal tumor or renal calculi. While there has been no direct evidence of these conditions, the finding of abdominal pain is suggestive and the presence of gross hematuria would certainly have activated these hypotheses.

Question 7 - Were there any episodes of pussy unine or foul-smelling unine or change in the odor of her unine?

Data Requested - Presence of pyuria or foul-smelling urine.

Goals

Primary - Confirm principal-part: Kidney/biadder inflamation of APN prototype <u>subgoal</u> to Confirm prototype APN.

Methods - P: Indirect: Pyuria or foul-smelling urine Is-Supporting-Evidence-For Kidney/bladder inflamation.

Expectation - Moderate: Pyuria or foul-emelling urine present.

Answer - No.

Result - P: Not Satisfied.

PLIS - Unchanged.

Commentary - Pyuria is a key sign in the diagnosis of a uninery tract infection. The fact that the patient did not report observing either pyuria or its side effects (the foul-smelling unine) is not disturbing, however, as it can be easily overlooked by a patient. The doctor will ask about the pyuria again when he starts asking about laboratory findings. If at that time no pyuria is reported, the diagnosis of APN might be in some doubt.

Question 8 - Was there any history of pain in one side or the other, flank pain specifically?

Data Requested - Presence of flank pain.

Goals

Primary - Confirm principal-part: Kidney inflamation of APN prototype subgoal to Confirm prototype APN.

<u>Methods</u> - P: Indirect: Flank pain Is-Supporting-Evidence-For Kidney inflamation.

Expectation - Strong: Flank pain present.

Answer - She had had abdominal flank pain in the past on the left side.

Result - P: Satisfied.

PLIS - Satisfied: Acute pyelonephritis.

Doctor's Commentary - Well, I am not sure how this is to be played but what I'd be homing in on now is the situation of the newsea, vomiting.

frequency, dysuria and flank pain as somebody with acute pyelonephritis.

Commentary - Flank pain (either unilateral or bilateral) is another key symptom in the diagnosis of acute pyelonephritis. At this point in the protocol the doctor has finishing confirming the component of the acute pyelonephritis prototype that refers to symptomatic history. It is clear that while he feels satisfied that she has APN, no evidence strong enough to confirm that diagnosis has been presented yet. This evidence will be gathered when the doctor reaches the part of the present illness concerning physical examination findings and laboratory results. It is then that signs such as costovertebral-angle tenderness, pyuria, white blood-cell casts and bacteriuria will be asked about.

Question 9 - 0K., He've got nausea, vomiting, 18 pound Height loss, frequency, dysuria and at least left-sided flank pain. And now I'll begin to question specifically for pyelonephylitis, which is what I'd be aiming for at the present time. Now, I'd like to know whether before this 3 week episode, before this acute illness whether in the past she has ever had any episodes of a similar nature?

Data Requested - History of similar past episodes.

Goals
Primary - Confirm principal-event: Acute flare-up(s) of urinary tract infection of CPN developmental scenario subgoal to Confirm scenario CPN subgoal to Case-build CPN subgoal to Confirm CPN.

Methods - P: Direct.

Expectation - Weak: History of similar past episodes present.

Answer - Yes, she has.

Result - P: Satisfied.

PLIS - Likely: Chronic pyelonephritis (CPN) (with scute flare-ups)
Possible: Other chronic interstitial nephritis (CIN)
Unrelated episodes of AUTI
Acute flare-ups of chronic bacteriuria
Chronic renal failure (CRF)

Commentary - This question indicates that a new phase of the protocol has been reached - the patient's past medical history. The findings that the doctor has heard so far, while fairly convincing evidence for an acute uninary tract infection and acute pyelonephritis, are also suggestive of an underlying chronic condition. The length of the hospitalization, the duration of the symptoms and the severity of the neusea and vomiting (as indicated by the 10 pound weight loss) are all clues in that direction. It is only at this point, however, that he is willing to specify more

precisely what chronic renal diseases might be present and commit himself to pursuing one of them - chronic puelonephritis. The strategy he chooses is another variant of case-building. The variant he used before was applicable to acute diseases and only involved confirming the disease prototupe. In case-building for a chronic disease another component must also be confirmed - the developmental scenario (See Section 4.1.2.1). One common scenario for the development of CPN specifies that the patient experiences a number of episodes of acute uninary tract infection over a fairly long time period. This is generally not due to reinfection by new organisms but due rather to acute flare-ups of a long-standing, chronic infection. It is entirely possible (and also quite common) for the patient to experience both unrelated episodes of acute infection or acute flare-ups (of chronic bacteriuria) without having CPN. It is also possible for the patient to have another chronic intenstitial disease that simply predisposes him to urinary tract infections. CPN is one of the most difficult diseases to diagnosis based on a patient's current symptoms and requires knowing facts about the patient's medical history. This consideration leads to the next question.

Question 18 - Does she know that? Can she tell me whether she's had one. tho, three? Has she been hospitalized here before or elsewhere?

Data Requested - History of previous hospitalizations.

Goals
Primary - Confirm a body of reliable information on the patient's past medical history is available.

Secondary - 1. Confirm similar past episodes were serious enough to require hospitalization.

Methods - P: Indirect: Past hospitalizations Is-Evidence-For Availability of reliable patient information (hospital records). S1: Indirect: Past hospitalizations Is-Suggestive-Evidence-For Past episodes being serious.

Expectation - Moderate: At least one past hospitalization.

Answer - She has in fact been hospitalized before. In fact her first hospitalization was for a kidney stone.

Result - P: Partially satisfied.
S1: Partially satisfied.

PLIS - Likely: CPN (with acute flare-upe).

Possible: Kidney-stone (past) Caused-by UTI

Kidney-stone (recurrence) (# AUTI)

Other CIN

CRF Cause-by CPN or other CIN

Unlikely: Unrelated episodes of AUTI

Commentary - The doctor learned two important facts with this question. The first is that she indeed was hospitalized a number of times. The second piece of data, that the first hospitalized a number of times. The second piece of data, that the first hospitalized in number of a kidney stone was free information. This was an error in the experimental procedure. (The primary opinion of the doctor in asking the question is to see if reliable information about the patient's past medical history is available.) The answer he obtained, however, while consistent with a chronic uninary tract infection suggests a few help hypotheses. Certain kinds of kidney stones (called infection stones) are formed around a nucleus of bacteria associated with a uninary tract infection. Kidney stones in general, however, also predispose the patient to having further uninary tract infections. These infections in turn can cause the formation

of new kidney stones. The doctor's new hypothesis is that the patient may now be having a recurrence of this cycle. The linked hypothesis of recurrence of kidney stone and acute urinary tract infection could explain all of the known findings.

Question 11 - Do we know which side that was?

Data Requested - Lateralization of past kidney stone.

Goals
Primary - Confirm/Eliminate scenario-prerequisite: Consistent
lateralization of previous kidney stone and current abdominal
pain in hypothesis: Kidney stone (recurrence) (+ AUTI)
subgoal to Confirm scenario Kidney stone (recurrence)
(+ AUTI) subgoal to Confirm/Eliminate Kidney stone
(recurrence) (+ AUTI).

Methods - P: Direct: Expert witness.

Expectation - Moderate: Left side.

Answer - She had had a kidney stone 17 years before. We don't know anything about that. We don't know which side that was.

Result - P: Not satisfied.

PLIS - Unlikely: Kidney stone (recurrence) (+ AUTI)

Commentary - The fervor with which the new hypothesis was originally considered has been considerably dampened by the new information. Seventeen years is too long a period of time for the cycle to be happening again with any reasonable degree of probability. The lack of any information makes it very difficult to pursue even if the time period had been shorter. The alternative hypothesis of CPN is strengthened, however in an indirect way. The doctor has now established a possible



precipitating event in the developmental scenario. CPN requires that there be a long-standing, chronic infection of the kidney(s). It is quite possible that the stone could have been associated with a uninary tract infection (possibly as the result of the clinical procedure to diagnosis and remove the stone) and that the whole process began 17 years ago.

Question 12 - And that stone wasn't analyzed?

Data Requested - Results of stone analysis,

Goals
Primary - P1: Confirm precipitating-event: initial UTI in CPN developmental scenario <u>subgoal</u> to Confirm scenario CPN.

P2: Confirm Kidney stone caused-by UTI.

Methods - P1: Indirect: Past kidney stone was an infection stone Is-Strong-Supporting-Evidence-For initial UTI preceding kidney stone.
P2: Direct: Expert witness.

Expectation - Moderate: Infection stone.

Answer - We have no history of stone analysis. She had also been hospitalized 8 months earlier.

Result - P: Not satisfied.

PLIS - Unchanged.

Commentary - The doctor would like to pin down when the chronic urinary tract infection (of CPN) started. If the kidney stone is of the type that results from a preceding infection, he would assume that the patient probably had bacteriuria for a period longer than 17 years and the likelihood of CPN would be slightly higher. Some free information was

given to the doctor by telling him that she had a episode similar to the current episode for which she was hospitalized & months prior to the current admission.

Clearly, there are other ways to find out about a possible infection associated with the kidney stone. I hypothesize, however, that the reason this particular question was asked is because it is closely related to the previous question in that a feature of the same finding (or event in this case) is being asked about. Among certain doctors there is a proclivity to derive as much information as possible from (and about) the finding or event that is currently under consideration rather than "change the subject." (See Section 3.4 for a discussion of diagnostic style.)

Clerical bridge- D: At our hospital?

K: No, at another hospital.

D: And are we able to get those records?

Commentary - The exchange above need not be analyzed by the formal means I have been using. Its purpose is simply to establish the framework for the next set of substantive questions the doctor will ask. For that reason I

K: Yes, they are available. I'm sorry
I just found which side. It was the
left side.

D: That was 17 years ago?

K: Yes, that was 17 years ago.

D: 8 months ago she was hospitalized for a similar episode?

K: That's correct

D: And do we know whether she had renal function tests, urine cultures and urinalysis done at that time?

K: Some information is available on that.

call it a clerical bridge. It also gives the doctor a chance to think things over before committing himself to a new line of questioning. The doctor does learn that the stone 17 years ago was on the left side. This strengthens his belief that the chronic urinary tract infection goes back at least that far.

Question 13 - What is the urine-culture report?

<u>Data Requested</u> - Has the culture positive? Type of bacteria.

Goals
Primary - P1: Confirm principal-event: Acute flare-up of uninary tract infection of CPN developmental scenario <u>subgoal</u> to Confirm scenario CPN.
P2: Confirm principal-part: Chronic bacteriuria of CPN prototype <u>subgoal</u> to Confirm prototype CPN subgoal to Case-build CPN subgoal to Confirm

Methods - P1: Indirect: Postive urine culture Is-Supporting-Evidence-For Acute flare-up of UTI. P2: Direct.

Expectation - Strong: Positive urine culture.

 $\underline{\text{Answer}}$ - I don't have any unine culture report except that the unine culture grew out e-coli.

Result - P1: Partially satisfied.
P2: Partially satisfied.

CPN.

PLIS - Unchanged.

Doctor's Commentary - The reason I am asking for that specifically (although in most patients with a renal problem I'd be interested in the overall renal function), first is again I'm aiming for a presumptive or sort of speculative diagnosis of ichronic! pyelonephritis and I would like to find things that are most strongly suggestive for that or make that hypothesis enough.

Commentary - The doctor has returned to the line of questioning that was started with Question 9. In order to confirm the chronic UTI associated with CPN it is necessary to ascertain if the cultures for past episodes grew significant numbers of becteria and that the type of becteria reported in each episode was the same. At this point the dector does not yet know the kind of bacteria that grew out in the wrine culture done for the present episode so this point cannot be checked. This is not the crucial aspect of the diagnosis, however. It is quite pessible that the bacteriumia of 8 months ago was cured (The cultures became sterile.), but because of underlying CPN she was predisposed to reinfection by a new strain of bacteria. The significant aspect is the episode of AUTI itself. Note that the goal is partially satisfied since the culture report did not state whether the colony count was significant.

Question 14 - Do we have a uninalysis? At the same time I am looking to find out whether or not there was any pour a or white cells.

Data Requested - Presence of pyunia.

Goals

Primary - P1: Confirm principal-event: Acute flare-up of unimary tract infection of GPN developmental scenario.

P2: Confirm principal-pert: chronic bacteriums of CPN prototupe.

Methods - P1: Indirect: Pyuria Is-Supporting-Evidence-For Acute flare-up (episode) of Uff. P2: Indirect: Pyuria Is-Supporting-Evidence-For Chronic bacteriuria

Expectation - Strong: Pyuria present.

Answer - Yes, there were many white cells and red cells present in the urine.

Result - P: Satisfied

PLIS - Satisfied: Episode AUTI (8 months PTA)

Doctor's Commentary - I know she's had an episode of at least bacteriuria, that she's got both a positive unine culture and white cells and that she's got a uninary tract infection which presumptly was the cause of her episode 8 months ago and which may be a similar episode going on right now. I need to know two other pieces of information from 8 months ago and I need to know what her renal function was 8 months ago and I need to know what her kidney x-ray, looke like, whether she has one kidney, two kidneys whether one kidney is working the other is not, what the kidneys look like and whether they are scarred by infectious disease on not.

a kaa waa Mii ahii ah waxa

<u>Commentary</u> - Sufficient evidence (for this doctor) for a previous episode THE CARLO CARREST OF THE STATE OF THE PARTY of UTI (i.e. bacteriuria) consists of a positive urine culture and pyuria. the boundaries of shirts of thing of This would satisfy a goal of confirming a past episode of acute urinary and education and the sections tract infection regardless of any possible relationship to her current symptoms. The doctor's hypothesis, however, is that the two episodes are MARKER BERTHAM both acute flare-ups due to the same (chronic) infection. (Orea box alternatively, CPN predisposed hereto reinfection possibly with a different organism.) It is interesting to note that the doctor seems either not to JUNEVEN LE COMP DO MARTINED : hear or chooses to ignore the fact that hematurie was found 8 months ago (This can only be inferred from the fact that he makes no comment on it.). There are two possible interpretations of this. The first is that some degree of hematuria is consistent with AUTI. The other explanation is that doctors tend not to notice certain reported findings unless they have a place to "hook" the finding onto (i.e. some hypothesia that the finding is significant in).

Question 15 - What was her renal function?

Data Requested - Reported value of the renal function tests - creatinine clearance, BUN or NPN 8 months PTA.

Goals
Primary - P1: Confirm principal-event: Development of renal insufficiency (CRF) of CPN developmental ecenario subgoal to Confirm scenario CPN.
P2: Confirm principal-part: Decreased renal function of CRF prototype subgoal to Confirm prototype CRF subgoal to Case-build CRF subgoal to Confirm CRF.

Secondary - 1. Explore the severity of the illness.

2. Explore the stage of development of a chronic disease: CPN.

Methods - Pl: Indirect: Renal function test decreased
Is-Supporting-Evidence-For renal insufficiency.
P2: Direct.
S1: Indirect: Renal function test Is-A-Suggestive-Measure-Of the severity of the illness.
S2: Indirect: Degree of decrease of renal function indicated by a renal function test Is-A-Suggestive-Measure-Of the stage of development of CPN.

Expectation - Strong: Renal function tests indicate normal renal function or some degree of renal insufficiency.

Answer - The NPN was 51.

Result - P: Partially satisfied. S1: Partially satisfied. S2: Partially satisfied.

PLIS - Unchanged

Doctor's Commentary - That's an old record. What I learned from that is that her renal function was close to normal or only slightly impaired. The normal NPN goes up to somewhere around 45. And that was when she was acutely ill.

Commentary - A chronic, degenerative renal disease such as CPN can eventually result in the loss of functioning renal mass causing the loss of normal renal function. The final stage in the developmental scenario of CPN is advanced renal failure. Of course this process can take many years. It is also true that even with the loss of one kidney, and the partial degeneration of the remaining one, renal function can remain at about normal levels. The doctor has really has heard no definitive evidence as to what stage of development the disease might have reached in this patient. Therefore, it is important to differentiate between the doctor's goal and his expectation. If he had found out that the degree of loss of renal function was indicative of advanced renal failure, his primary goal would have been more fully satisfied. The answer he does hear, however, is not unexpected and is consistent with all the other evidence.

The measurement of renal function that he is given (the NPN) is outdated and is normally not used anymore. That is what he meant by it being an "old record." The only other disturbing factor is that an acute episode superimposed on a chronic condition tends to amplify the loss of renal function. Thus the relatively low value of the NPN would seem to indicate that the kidneys were still functioning about normal and that the chronic condition had not progressed to the point of significant loss of renal function. While 17 years is not that unusual in the progression of CPN to end stages (in fact it might have been much longer than than this period) a shorter development period is more common.

Question 16 - And the IVP?

Data Requested - Reported results of the introvenous pyelogram, a kidney x-nay...

Goals Primary - Confirm principal-part: Scarred kidneys of CPN prototupe <u>subgoal</u> to Confirm prototupe CPN <u>subgoal</u> to Case-build CPN <u>subgoal</u> to Confirm CPN.

Secondary - 1. Explore permanent structural damage to kidneys subgoal to Explore status of primary organ system under consideration subgoal to Explore level of organ/organ sustem function.

2. Explore a predisposition to PN: uninary tract obstruction.

3. Eliminate Ridhey stone (recurrence).

Methods - P: Direct.

S1: Direct.

S2: Direct.

S3: Direct.

Expectation - Moderate: Scarred kidneys, contracted kidneys.

Answer - The IVP was not satisfactory due to poor infiltration of the dye. It was repeated and there was only minimal appearance, mostly in the left urinary tract. No signs of stone or other abnormalities.

Result - P: Not satisfied.

S1: Partially satisfied.

S2: Satisfied. S3: Satisfied.

PLIS - Unlikely: Uninary tract obstruction (8 months PTA). Ruled-out: Kidney stone (recurrence).

Commentary - The Introvenous puelogram can provide crucial data in the attempt to characterize the status of the kidneus. Among the things that can be determined (if the TVP procedure is successful) are:

- 1. The size and configuration of the kidneys.
- 2. An estimate of the level of renal function.
- 3. The presence of gross abnormalities or stones.

4. Whether there is obstruction or not.

The doctor asked the question in a non-committal style. He did not ask for any specific feature of the IVP. From his previous comments, however, we know that he is primarily interested in whether the kidneys are scarred as the result of chronic infection. But clearly, since he can explore the overall status of the kidneys, he doesn't went to restrict the question to just that. The IVP report does not answer the question of whether her kidneys are small and scarred. It is suggestive of same degree of loss of renal function (the minimal appearance of the due) and strangly suggestive that the patient wasn't obstructed (at least totally questructed) at the time the IVP was performed.

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Question 17 - Do we know if there were two kidneys?

Data Requested - Did both kidneys appear on the IVP.

Goals
Primary - P1: Explore prognosis relative to the POH,
P2: Explore the gross configuration of the kidneys
subgoal to Explore status of the primary organ system
under consideration subgoal to Explore lavel of
organ/organ system function for decree of organ system
impairment).

Methods - P1: Indirect: CPN + missing kidney Is-Exidence-Formation appropriate to the state of t

Expectation - Moderate: Both kidneys present.

Answer - I can't tell you that. It's not known, and

Result - Pl: Not setting the down to set in section and the set in the section of the section of

PLIS - Unchanged.

Doctor's Commentary - OK, that's the information I've been looking for from 8 months ago, now to get back to the present episode. I really don't think I have any other questions as far as that acute episode. I would like to know some general information. He know that she's acutely III, with an illness which is quite similar to that which she had 8 months ago and we've got some nice information that 17 years ago she had a stone. He know she's got nawses, vomiting, frequency, dysuria and abdominal pain.

Commentary — There has been no evidence to suggest that the patient is missing a kidney. In terms of possible therapy for CPN (which the doctor is considering as he goes along), the absence of one kidney would be very serious. Nephrectomy (surgical removal of a kidney) is often required if serious hypertension has developed in association with CPN. If the patient has only one kidney, transplantation might be the only option. Because of the age of the patient this option would not be viable. There has been some suggestive evidence that the disease might be unilateral. If this is the case and both kidneys are present with the undiseased kidney functioning normally, the prognosis would be much better.

Question 18 - Is she a diabetic?

Data Requested - Blood sugar, history of diabetes.

<u>Goals</u>

Primary - Explore a predisposition to CPN: Diabetes subgoal to Case-build CPN.

Methods - P: Direct.

Expectation - Uncommitted.

Answer - She's not a known diabetic. The blood sugar was normal.

Result - P: Satisfied.

PLIS - Ruled-out: Diabetes.

Doctor's Commentary - What I would like to know is whether there is anything that would predispose this individual to having pyelonephritis. That was what I was looking for before, that she might be obstructed on one kidney or the other 8 months ago. I would like to know whether she is a diabetic, since they are supposedly more susceptible to infection because of the disease or instrumentation and whether she takes phenacetin for headaches or not.

Commentary - An component of the variant of case-building that the doctor is using is determining if there are any significant factors that would predispose the patient to getting the disease. Most doctors use a weight-of-evidence procedure in evaluating a hypothesis <Shortliffe 74>. This involves adding up factors that support a hypothesis and subtracting those that are against it (Factors that are neutral don't count.).

Predispositions are "indirect" evidence for a disease or other clinical condition. They cannot be used to support a hypothesis unless "direct" evidence for a hypothesis (e.g. the characteristic findings of a disease) are also present. It should be noted how quickly (i.e. how few questions were needed) the doctor characterized the previous episode and its relationship to the current episode and has moved on.

Question 19 - Does she take phenacetin? Does she take aspirin containing compounds?

Data Requested - History of medication for headaches, specially compounds containing phenacetin or aspirin.

Goals

Primary - Explore a predisposition to CPN: Phenacetin and/or analgesic abuse.

Methods - P: Direct.

Expectation - Uncommitted.

Answer - The patient had taken 6 empirin tablets a day for 15 years.

Result - P: Satisfied.

PLIS - Possible: Phenacetin (analgesic) nephritis

Papillary necrosis
Urinary tract obstruction
Gram-negative (septic) shock

Doctor's Commentary - I'm sure it took a lot longer to dig that information out than simply asking that time around. There is an increased incidence of infectious disease of the kidney particularly papillary necrosis in patients who take increased amounts of phenacetin or phenacetin-containing compounds. So that would make me concerned first of all not only that she's got a urinary tract infection, which I'd be quite sure of by this time even before I'd done a physical examination or laboratory data but make me concerned that she might have papillary necrosis and might be sloughing a papilla and blocking file. Obstructing one kidney or the other. And I'd be more concerned about this patient than another patient with it. I wouldn't ask any other questions at this time and I'd begin the physical examination.

Commentary - This finding is one of the key findings in the diagnosis of this patient. (The final diagnosis was APN superimposed on chronic renal disease, either phenacetin nephritis, chronic pyelonephritis or both and chronic renal failure.) Before discussing the interpretation of this finding, consider the different methods that were used by the other doctors in obtaining it:

- 1. Through a review of systems in the section about history of medications (Protocols 4 and 5).
- 2. Through a review of systems (focused on renal disease & hypertension)

asking about history of headaches (Protocol 6).

(It should be pointed out that the four doctors who uncovered the finding of phenacetin abuse had established CPN early-on in the interview as the most likely hypothesis for the underlying chronic renal disease.) The more common route for uncovering abuse of a drug is probably not the one used by this doctor. Questions concerning the use of medications are normally asked as part of a systems review. It is asked about here because determining predispositions is part of case-building, the strategy the doctor is using to confirm his principal hypothesis. The reason the doctor says that it probably took longer to get that information is that patients are sometimes very reluctant to divulge any history of self medication.

The element of phenacatin abuse significantly alters the doctor's hypothesis structure. While both CPN and phenacetin nephritis are forms of chronic interstitial nephritis and both can occur in the same patient at the same time, the developmental history is non clouded. A complex relationship can exist among chronic pyelonephritis, phenacetin nephritis, urinary tract infection and papillary necrosis. The degenerative effects of both chronic diseases are cummulative. It may be impossible even with a histological examination of the lesions in the kidney to differentiate between a single or combined etiology. While both disease predispose the patient to now episodes of acute urinary tract infection, it is phenacetin nephritis that is indicated in the development of renal papillary necrosis. For this reason the doctor works under the assumption that phenacetin

nephritis is present because of the seriousness of papillary necrosis. He is primarily concerned with the possibility that some obstruction has occurred as a result of sloughing a necrotized papilla. Obstruction is a very serious condition in a patient with an acute urinary tract infection. This concern is demonstrated in a partial way by his decision to immediately go to the physical examination.

Question 28 - What is her blood pressure? Pulse? Temperature?

Data Requested - Vital signs on admitting physical exam.

Goals
Primary - Confirm/Eliminate a complication of APN (UT[):
Gram-negative (septic) shock subgoal to Explore the need for immediate treatment subgoal to Explore the severity of the illness (how acutely III is the patient).

Secondary - 1. Confirm principal-part: Systemic eyidence of active infection: temperature > 181. of APN prototype <u>subgoal</u> to Confirm prototype APN <u>subgoal</u> to Case-build APN.

Methods - P: Indirect: Extreme hypotension + fever/chills
Is-Strong-Supporting-Evidence-For Gram-negative
shock.
S1: Direct.

Expectation - Weak: Evidence of shock.

Answer - Her blood pressure is 148/88, lying, 138/88 siting. Pulse 88. Temperature 98.4.

Result - P: Satisfied. S1: Not satisfied.

PLIS - Ruled-out: Gram-negative shock.

Doctor's Commentary - I am going to ask a sort of directed physical examination related to the problem at issue end skip things which would need to be done such as a neurological examination. I upuld like to know what her blood pressure is. If she were hypotensive on admission to the

floor with pyelonephritis, gram-negative shock happens and is much worse and needs to be treated much more rapidly than somebody who simply has an acute pyelonephritis which is often treated as an outpatient..... (after answer) OK, then at least although the temperature is helpful there are some patients with gram-negative shock (which she doesn't have) who can be afebrile.

Commentary - In terms of strategy selection, the basis for for choosing this question is the high priority of determining the need for immediate treatment of a potentially fatal condition. The possibility of obstruction secondary to papillary necrosis superimposed on an acute urinary tract infection is what originally activated the possibility of septic shock. While the doctor is aware that this is a diagnostic game, it is almost impossible for him to forget that he is a doctor first and remove himself from his normal daily concerns and routines. Even though an elevated temperature is part of the APN prototype and confirming it part of case—building for APN it is clearly a secondary goal.

Question 21 - Does she have CVA pain posterially or can you feel large polycystic kidneys?

Data Requested - Presence of CVA pain or palpable kidneys.

Goals

Primary - Confirm principal-part: Kidney inflamation of APN prototype subgoal to Confirm prototype APN.

Secondary - 1. Explore a predisposition to PN: polycystic kidneys.

Method - P: Indirect: CVA pain or tenderness Is-Evidence-For Kidney inflamation of APN.

S1: Indirect: Kidneys not parpable Is-Evidence-Against Polycystic kidneys

Expectation - Strong: CVA pain or tenderness.

Moderate: Kidneys not palpable.

Answer - The kidneys were not palpable. She had left CVA tenderness.

Result - P: Satisfied. S1: Satisfied.

PLIS - Unlikely: Polycystic kidneys.

Doctor's Commentary - The CVA pain would simply be evidence of pyelonephritis. Polycystic kidney disease (which is not suspected by the history) I must say would simply be another thing that can predispose to urinary tract infection..... (after answer) Which first of all fits with her history and fits interestingly enough with that stone 17 years ago on the left side.

Commentary - The doctor is continuing the case-building for acute pyelonephritis that started with Question 1. As the protocol moves from symptoms and history to physical exam and finelly to laboratory data, the evidence available is more objective (in the sense of providing increasingly more reliable tests of a hypothesis). Referring to the Prototype of APN in Appendix 1, we can see that it is divided into the three areas mentioned. Confirming the prototype requires confirming each section. For APN, the physical exam section specifies that CVA pain or tenderness should be present. The doctor's comment that left CVA tenderness "fite" with the stone 17 years ago indicates that he may feel that the pyelonephritis may be unilateral (or at least more severe on the left side).

The second part of the question, about the possible presence of large polycystic kidneys, is very interesting. The hypothesis that Dr. Kassirer and I worked out is that it is easier for the doctor to visualize

himself performing the physical examination that asking questions second hand. Thus, when he sees himself examining the flank area for CVA pain or tenderness, he would almost automatically palpate for large kidneys. It is like a reflex: while I'm there I might as well find out about it since it costs very little. As the doctor indicates there has been nothing in the history to indicate the presence of polycystic kidneys.

Question 22 - Does she have superpublic tenderness?

Data Requested - Presence of tenderness over the bladder.

Primary - Confirm principal-part: Bladder irritation of APN prototype subgoal to Confirm prototype APN.

Method - P: Indirect: Superpublic tenderness Is-Evidence-For Bladder Irritation/inflamation.

Expectation - Moderate: Superpublic tenderness present.

Answer - It's not mentioned.

Result - P: Not satisfied.

PLIS - Unchanged.

Commentary - Superpublic tenderness is optional. If present, it is simply supportive evidence. If absent, it weighs little against the hypothesis. The frequency and dysuris are sufficient to confirm the bladder irritation. Again it's simply a case where the data is very inexpensive to obtain and the doctor would himself have checked for it himself while conducting the physical exam.

Question 23 - Now as far as specific GI findings, does she have bowel sounds, rebound tenderness or not?

Data Requested - Presence of normal bonel sounds, absence of rebound tenderness.

Goals
Primary - Eliminate acute GI disease.

Method - P: Indirect: Normal bowel sounds + soft abdomen
Is-Strong-Supporting-Evidence-Against Acute GI disease.

Expectation - Moderate: Normal GI findings.

Answer - The abdomen was soft with normal bowel sounds.

Result - P: Satisfied.

PLIS - Ruled-out: Acute GI disease

Doctor's Commentary - We sort of neglected the nausea and vomiting....lafter answer? We sort have gone through this as nausea vomiting frequency and dysuria but if you go back to that nausea, vomiting and weight loss you'd obviously have to keep at least on one track of your mind that rather this being a kidney problem that this is some acute abdominal problem. With soft abdomen, normal bowel sounds that goes to the bottom of the list, if you will and we can continue down the stream that we have been working on. So she has left CVA tenderness, no palpable kidneys, no comment on superpublic tenderness, soft abdomen and normal bowel sounds.

Commentary — As the doctor has indicated he has pretty much gone along with his principal hypothesis and has ignored the possibility of acute GI disease. (He has thought this unlikely all along though.) The best opportunity to rule-out acute GI disease is in the physical examination. He is possibly thinking about such diseases as appendicities or peritonitie. A soft abdomen (absence of rebound tenderness) and normal bouel sounds rule these out as active possibilities.

It is interesting to note the effect of a doctor's specialty in

strategy selection. The protocol obtained from a gastroenterologist is almost reversed in the order of which hypothesis gats tested first. The GI specialist first explored and then eliminated the possibility of chronic/acute GI disease before going on to renal disease.

Question 24 - What was her hematocrit?

Data Requested - Value of hematologic test: hematocrit.

Goals
Primary - Confirm/Eliminate anemia subgoal to Confirm/Eliminate principal-event: Development of advanced CRF of CPN developmental scenario subgoal to Explore developmental stage CPN subgoal to Confirm scenario CPN.

Secondary - 1. Explore the severity of the Illness.

Methods - P: Direct: Hematocrit low + hemoglobin low Is-Prims-Facia-Eyldence-For anamia. S1: Indirect: Degree of anamia Is-An-Objective-Measure-Of the severity of the illness.

Expectation - Moderate: Hematocrit near normal (37 - 47)

Answer - Her hematocrit was 20%. Hemoglobin 6.8 grams.

Result - P: Satisfied. S1: Satisfied.

PLIS - Confirmed: Anemia

Doctor's Commentary - I would go to laboratory information now. I would like to know what her hematocrit, white count and differential are. I would like to know what her hematocrit simply as general information to see if her hematocrit was 20, I'd be concerned that she has been in renal failure (although we don't know that yet) for a longer period of time or something else is going on which I've totally neglected so far...(after enemes) That's interesting. Then in addition to this question of infectious acute pyelonephritis there is more going on than we are aware of. There are two gensibilities. One, she's got a separate reason for being anemic; we're going to have to run down the evaluation of her anemia, or secondly the anemia is part of her

renal disease and if it is it's likely to be due to chronic renal failure which is odd because we are told that she had relatively normal renal function or only slightly impaired renal function & months earlier. I don't have any answer for that yet.

Commentary - The doctor has in some sense opened up Pandora's Box with this question. Anemia is a clinical condition with many different etiologies: blood loss, excessive hemolysis (destruction of redublood cells) or impaired production. The anemia of CRF is associated with impaired production but there are many other causes of impared production (folic acid deficiency, splenic disorders, etc.). The doctor is facing the following dilemma: he would expect to see some degree of impairment of renal function: that would be consistent with either phenacetin nephritis or CPN. It is the degree of renal failure implied by the anemia as compared with the measure of renal function 8 months PTA that is disturbing. At this point in the protocol the doctor has started the last Branch Carlotte Charles phase of questioning - the laboratory findings. He does not want to throw out the hypotheses he has been working on and try something new as yet. · 真然 "自己" 安安村 [) 完全稿 (· · · · · · · ·) [] (· · · · · For this reason he puts aside the finding (does not pursue it using a differential diagnostic strategu) and continues down the infection/chronic ina gran ing Santanan ing Palakan Baring ang manggan ba renal disease line.

Question 25 - White count and differential?

Data Requested - White blood cell count, differential breakdown of white cell population.

Goals
Primary - Confirm Leukocytosis + Left-shifted differential

subgoal to Confirm principal-part: Systemic evidence of active infection of APN prototype subgoal to Confirm prototype APN subgoal to Case-build APN subgoal to Confirm APN.

Methods - P: Direct.

Expectation - Strong: Elevated white count (leukocytosis) and differential shifted-to-left.

Answer - 9858, 63% polys, 30% lymphocytes, 3% monos, 1% eosinophils.

Result - P: Partially satisfied.

PLIS - Unchanged.

Doctor's Commentary - Her white count I'd be expecting to see elevated with a shift to the left, because everything that I've heard so far leads me to suspect there is an acute infectious process... (after answer) That is not as striking as I would have expected, but doesn't eliminate the possibility.

Commentary — The white count and differential are two key signs in the confirmation of an active bacterial infection. The normal white count is 5 — 10 thousand. With an infection the expectation would be that it should be above 10 thousand. But as the doctor has said a white count of 9850 while not supportive of the hypothesis, doesn't eliminate it. Similarly for the differential. The expectation is that the percentage of polys should be larger; but the figure given is within the appropriate consistent range. The laboratory-data section of the APN prototype is now being confirmed.

Question 26 - The next thing I would like to know is what her urinalysis looks like

Data Requested - Results of urinalysis.

Goals

Primary - Confirm pyuria <u>subgoal</u> to Confirm principal-parts
Kidney-inflamation of APN prototype <u>subgoal</u> to
Confirm prototype APN.

Secondary - 1. Confirm urinalysis (excluding pyuria) is
Consistent with APN subgost to Case-build
APN.
2. Confirm/Ellminate (late) CRF subgost to Explore
Developmental stage CPN/Phenacetin nephritis.

Methods - P: Direct.
S1: Direct.
S2: Indirect: Low specific gravity + renal failure casts
Is-Evidence-For (fate) CRF.

Expectation - Strong: Pyuria present.

Moderate: UBC casts present.

Weak: Renal fallure casts present.

Answer - Specific gravity 1.818, no sugar, 1+ protein, acid reaction, no red cells, filled with white cells.

Result - P: Satisfied.

51: Partially Satisfied.

S2: Partially Satisfied.

PLIS - Unchanged.

Doctor's Commentary - I am again looking at this specifically as an infectious disease of the kidney and trying to find things for it or against it. I would expect to see again, pyuria..... [after answer] That specific gravity fits with either chronic renal failure or normal renal function depending on whether she is dry or not. I know she can acidify her unine which doesn't surprise me very much. I know she specifically bit of protein in her unine which fits with some element of inflamatory renal disease and that her sediment is loaded with white sells which is again most consistent with acute infectious uninary tract infection or acute pyelonephritis if she has flank pain as well.

Commentary - The uninalysis is one of the three important sources of data when renal disease is the central focus. (the other two are the kidney x-ray and renal function tests). In some sense the spirit of the experiment

was violated by allowing the doctor to ask for the whole urinalysis rather than asking about each of its constituent findings. It was very difficult to stop this, however, as this is the normal form for inquiring and reporting the results. (Remember that Dr. Kassirer who was answering the questions is also a renal specialist.) Regardless of this, however, it is clear what the doctor was expecting to hear and how it fit into his strategical plan. His primary concern was to confirm the component of the urinalysis that is evidence of acute infections, white cells or pyuria. If there had been no puuria reported, the diagnosis of acute infection would have been in doubt. As far as the secondary goals are concerned, it is important that the other components of the uninalusis are consistent with the diagnosis of acute infection. (Consistent in this sense means that while they may not support the diagnosis, neither do they argue against it.) An optional component of this variant of case-building is to ask about the most common consistent findings relative to the principal hupothesis.

Question 27 - I would like to know if organisms were seen unspun?

Data Requested - Presence of bacteria in unspun unine sediment.

Expectation - Moderate: Bacteria present.

Goals

Primary - Confirm principal-part: Evidence of active UT infection of APN prototype <u>subgoal</u> to Confirm prototype APN.

Methods - P: Direct: Bacteria in unine sediment le Rnima-Facie-Evidence-For Active UTI.

Answer - They were not seen. There were no casts.

Result: P: Not satisfied.

PLIS - Unchanged.

Commentary — If the uninary tract infection is still active normally the stained sediment will reveal numerous bacteria. Therefore, the answer is slightly disturbing to the doctor. The confirmation can be made on a quantitative unine culture (and was in this case), so the hypothesis need not be rejected on the basis of this finding. The finding is also suggestive (though not very strongly) of obstruction, especially if the infection is localized to one kidney.

Question 28 - The next thing I would like to know would be the level of her renal function.

Data Requested - Values of renal function tests.

Goals

Primary - Confirm/Eliminate (late) CRF subgoal to Explore developmental stage CPN/Phenacetin nephritis.

Methods - P: Direct: Elevated renal function test Is-Prima-Facie-Evidence-For Renal failure.

Expectation - Strong: Slightly impared renal function.

Answer - BUN 96. Creatinine 9.4

Result - P: Partially satisfied.

PLIS - Satisfied : CRF (late)

Possible: "Severe bilateral pyelonephritis" (a severe form

Doctor's Commentary - That's enough to stop and make a tentative diagnosis

and then begin to look for some other things. She's got renal failure. It's acute in terms of months, that is from 8 months ago till the present time, she's had a very rapid deterioration in her renal function. And we know she's got an infectious process going on in her kidneys or at least everything is consistent with that. That degree of rapidity of renal failure over 8 months is much more likely that I would expect to see in somebody with chronic pyelonephritis and would make me suspect something superimposed, either severe bilateral pyelonephritis and some preexisting disease. That could happen. Or obstruction. Again, we've got that history of phenacetin we've been told about and papillary necrosis.

Commentary - Again, some degree of impairment of renal function would be consistent with the hypothesis of underlying chronic renal disease. What the doctor must now explain is the degree of decline in renal function over such a short time. The actual fact is that the measurement from 8 months ago was not accurate and her renal function had been much lower at that time. What is concerning him now (and concerned him for the remaining part of the protocol) is the possibility of obstruction resulting from papillary necrosis secondary to phenacetin nephritis. If a papilla had disloged and had obstructed one or the other kidney, this could explain what seems to be a very acute drop in renal function.

The analysis of the remaining portion of the protocol will not be included here for two reasons. The most important is that as an answer to his next question concerning kidney size by IVP, he was given inaccurate data. (As this was the first protocol, we had not discovered that the x-ray report was in error). The second reason is that by this time the reader should have had sufficient exposure to the annotation techniques

that were applied to the protocols.

In the next chapter I present a model to describe the strategies used in this and the other protocols.

CHAPTER 3

THE STRATEGY FRAME MODEL

Most theories of problem-solving incorporate the concept of a strategy. By a strategy what is meant is a plan that specifies a sequence of steps that will (hopefully) result in achieving a desired goal. The sequence of steps that form a plan is normally arrived at through a decomposition of the original problem into a set of sub-problems that are considered easier in some sense to solve. The relationship between the sub-problems and the original problem can be formally represented by an AND/OR goal-tree.

There are two distinct but closely related planning activities involved in taking a present illness: data acquisition and diagnosis. One plan is needed that specifies what to do with each piece of data once it has been obtained and another is need that specifies what data to look for next. Diagnostic strategy is defined here as the set of goals and methods that guide the evaluation and interpretation of findings, the formation and testing of hypotheses and the handling of competing hypotheses and discrepant information. Deta-gathering (or data-acquisition) strategies determine the content, form and sequence of the questions that are asked. Since the focus of study in my research has been describing data-gathering strategy, whenever strategy is mentioned it is meant to refer to this strategy.

The model I developed to describe strategies and strategy selection I

call the <u>strategy frame model</u> (SFM). The essence of the model can be stated as follows:

A strategy frame is a data-structure for describing a strategy. A particular strategy frame is intially suggested to the doctor by some feature of his internal diagnostic configuration (IDC). The IBC is the doctor's problem space - his internal representation of the external environment for the task of diagnosis. A strategy frame contains a set of conditions of the IDC under which the strategy is potentially applicable. If these conditions are met, the associated strategy is selected.

The concepts underlying this model have been influenced by the work of many people; Minsky's <u>frame theory</u> for the representation of knowledge contributed significantly to the theoretical basis. <Minksy 74>. The application of frame theory to the representation of medical knowledge by Pauker and Sussman (later refined by Schwartz and Gorry) was a major influence. <Pauker 75>. Newell and Simon's work on the representation of problem spaces and Newell's MERLIN program also played a significant role <Newell 74>.

3.1 Strategy Selection via Strategy Frames

What is required to apply frame theory to strategy selection is a representation of the doctor's problem space (which I have called his internal diagnostic configuration) such that classes of configurations appropriate to the selection of a particular strategy can be identified.

In the strategy frame model, the IDC is represented by a set of <u>descriptors</u> divided into two basic components. The first is a component I call the <u>patient model</u> and the second is the <u>current status</u> of the doctor in the diagnostic task. The patient model is a representation of the doctor's <u>diagnostic thinking</u> about the present illness of the patient <Silverman 74>. It consists of such things as a list of classified and ordered hypotheses. A typical disease hypothesis on this list is classified by features such as being acute or chronic, single or multiple etiology and episodic or non-episodic. The list is ordered by likelihood. The features of reported findings are also included in this component. Included in the current status component are such things as the phase of the interview and the strategy being used.

The stereotyped objects to be "recognized" by examining the IDC are those configurations that are associated with the selection of the strategies that have been identified through the protocol analysis. Each of these configurations forms the set of conditions of a strategy frame. For example, associated with a strategy of confirmation is a configuration that characteristically has a single hypothesis classified as LIKELY, while elimination is associated with three or more LIKELY hypotheses. Chronic diseases are associated with a variant of case-building that requires confirming the disease's developmental scenario.

The methodology described above for strategy selection is related to the selection of methods based on problem-space configurations found in the work of Newell and Simon, Heuitt's stereotypes < Heuitt 75>, Schank's

scripts <Schank 75>, and Malhotra's ruse/strategem frames <Malhotra 75>.

3.2 Constituents of Strategy Frames

A strategy frame consists of two basic components. The first is a prototype which is a description of the class of configurations that is suited to the use of the strategy. The second component is the strategy — a plan detailing a next step or sequence of steps in the present illness process if the prototype is successfully matched. The second component can be optional.

The prototype consists of a set of terminals as in most frame structures. Each terminal refers to a specific feature of the internal diagnostic configuration such as the number of active hypotheses, the presence of a causal link between two active hypotheses or if an immediately life-threatening symptom has been reported. The terminal specifies a condition that must be met by the feature refered to. If this condition is met the terminal has been matched successfully. Associated with each terminal is a score that indicates the relative importance of the feature to the selection of the strategy. A terminal can also consist of a logical construction formed from AND, OR or NOT operators applied to set of features. Another strategy frame can also serve as a component of a terminal.

CASE-BUILD-1 is a confirmation strategy that is applicable under the following conditions:

- 1. The principal disease hypethesis (PDH) is an acute, single-etiology disease.
- 2. There are no other likely hypotheses.
- 3. Absolute score of PDH > .8
 Relative score of PDH > 1.8
- 4. No CAUSE or COMPLICATION links into the PDH.

Programme to the state of the con-

```
(PROTOTYPE CASE-BUILD-1
 (TERMINAL NUM-HYPS (TRIGGER)
    ((TOTLIK 1) (SCORE 6))
  (TERMINAL HYP-CLASIF
    ((AND (ACP ACUTE)
          INETTOC SINGLEDY
     (SCORE 5)))
 (TERMINAL HYP-SCORE
    ((AND (ABSCORE ( > .8))
(RELSCORE (> 1.0)))
 (TERMINAL HYP-STRUCTURE
    ((AND (NOT (LINK-TYPE PDH CAUSE-OF IN))
(NOT (LINK-TYPE PDH COMPLICATION-OF IN)))
     (SCORE 3)))
(FAILURE-LINKS
  (IF (TOTLIK 2) (ACTIVATE DISCRIMINATE-1))
  (IF (TOTLIK (> 2)) (ACTIVATE ELIMINATION-1))
  (IF (ACP ACUTE-STAGED) (ACTIVATE CASE-BUILD-2))
  (IF (ACP CHRONIC) (ACTIVATE CASE-BUILD-3))
  (IF (NETIOL HULTIPLE) (ACTIVATE CAUSAL-EXCLUSION)))
(STRATEGY
 ((GOAL (CONFIRM (PROTOTYPE PDH)))
  (SUGGESTED-METHODS
      (IF (PHASE LABORATORY) (TRY DIRECT-CONFIRMATION))
      (ELSE (ASSESS (PRINCIPAL-PARTS (PROTOTYPE PDH)))))))
```

Figure 3.1 - Strategy frame for CASE-BUILD-1

The strategy component specifies the plan in the form of an AND/OR goal-tree. Associated with each node of the goal-tree is a (possibly empty) set of suggested methods. These methods can either be direct or indirect. A direct method represents the conversion of the goal (through syntactic means) into the form of a question. An indirect method can either be the premise of an IF-THEN type rule or a reference to another strategy frame. In the situation where there are no suggested methods associated with a goal, the (bound) goal will index a library of methods in order to select a method appropriate to the finding or disease hypothesis under consideration.

Figure 3.1 shows an example of a strategy frame for a variant of the confirmation strategy called case-building. Appendix 2 contains a listing of variables and the feature of the IDC to which each refers.

3.3 Organization and Operation of the Strategy Frame System

A strategy frame can be activated by the appearance of a particular feature in the IDC through the terminals of the frame that serve as triggers. A strategy frame can be considered as a candidate only if it has been activated. The frame system is arranged in such a way, however, that the majority of the strategy frames can only be activated through the suggested methods of the goal-tree of a successfully-metched, higher-level strategy. The suggested methods associated with each goal act as guides in filling out the details of the tree. These detail links can bind together

large numbers of strategy frames into complete sub-systems.

If the set of suggested methods contains references to other strategy frames, each of these may be activated and a match attempted. If such a subframe is also successfully matched, the associated strategy replaces the original method in the goal-tree. Global considerations are taken into account by imposing conditional tests on the selection of a member from the set of suggested methods at each node of the goal-tree. In this way overall control of the process of strategy selection can be maintained while still allowing more local details of the configuration to determine lower-level strategy. This process continues until all the terminal nodes of the goal-tree are associated with either a single direct or indirect method. The process of transfering global information can be seen in the sample strategy frame where the phase of the interview is taken into consideration.

Another form of organization is imposed through failure links. Each strategy frame maintains a list of alternative frames to activate if the terminal-matching procedure is not successful. (The terminal-matching procedure is not successful if a feature mentioned in the premise of a failure-link rule does not exactly match the condition imposed in the terminal it is a part of.) Each frame mentioned as an alternative is either an unrestricted alternative or is associated with a specific reason for failure. It is important to note here that failure in this context does not mean the failure of a strategy to be diagnostically productive after it has been tried, but that it fails to meet selection conditions

before it is applied.

The process of constructing a strategy from the strategy frames should be viewed as a form of <u>progressive refinement</u>. Some features of the IDC are more important in the selection of strategy than others. The sequence of choices made in the process of constructing a complete goal-tree, from the highest-level node to the leaf nodes, reflects this implicit <u>hierarchy</u> of features.

3.3.1 Strategy Binding and Special Strategies

Strategy binding is the process of replacing the abstract structural elements (such as the principal-part of a disease prototype) mentioned in the goal-tree with the specific referents of a disease frame (e.g. Bladder irritation is a principal-part of the APN prototype.). As an example, consider the class of diseases that have as a principal-part the reduction in function of an organ or organ system. Examples of this kind of disease include cirrhosis of the liver, Kimmelstein-Hilson disease and myxedema. In applying a strategy of confirmation to each of these diseases, a disease prototype match must be made. A part of this matching procedure specifies determining the level of organ function. This would be represented by a goal such as:

(GOAL (CONFIRM (PRINCIPAL-PART (PROTOTYPE DISEASE-NAME) (DECREASED-FUNCTION ORGAN-SYSTEM)))) Included in the set of suggested methods associated with this goal would be the following method:

(HETHOD INDIRECT (ASSESS (QUANTITATIVE-HEASURE ORGAN-SYSTEM-FUNCTION)))

Binding the general strategy to each specific disease entity results in the following bound strategies:

```
((GDAL
   (CONFIRM
     (PRINCIPAL-PART (PROJUTYPE CIRRHOSIS)
(DECREASED-FUNCTION LIVER) 177
 (METHOD INDIRECT
   (ASSESS
     (AND (SCPT (> 35 UNITS))
(SCOT (> 48 UNITS))))
                sign of the liver of the first of the second of the liver
( (GOAL
   (CONFIRM
     (PRINCIPAL-PART (PROTOTYPE KINNELSTEIN-HILSON)
 (DECREASED-FUNCTION KIDNEY))))
(HETHOD TNOTRECT
   (ASSESS
       (AND ICREATININE (> 1.0))
           (BUN (5. 58)))))) nspri no napri ne
( (GOAL
                              positiva 🕍 🗱 📆 🖟 🖟 🖟 🖟 🖟 💮
    (PRINCIPAL-PART (PROTOTYPE MYXEDEMA)
       (DECHERSED-FUNCTION THYROTO) )7)
  (METHOD INDIRECT
                        MARKET AND THE AND THE PARTY OF THE
   (ASSESS
      (AND (PBI (< 3.5))
```

This binding procedure results in the construction of a strategy that is tailored for the specific disease entity under consideration.

It can be argued that experienced clinicians within a specialty tend

to see manu cases that are very similar both in mode of presentation and in final diagnosis. A Fellow at the Renal Clinic at the NEMC estimated that one-third of all new cases refered to them have unexplained hypertension as the presenting complaint. Hypertension is a disease that is closely related to a strategy called causal exclusion (see Section 4.3). It is thus reasonable to assume that repeated binding of a strategy with respect to a specific disease or hypothesis structure tends to associate the bound strategu with that disease. A strategu that is retained and associated with a disease is called a special strategy. The strategy frame model is flexible enough to allow special strategies. The existence of a special (pre-bound) strategy associated with one of the currently active hypotheses can be made a feature of the IDC. A strategy frame is then added which essentially acts like a "buck-passer" to lit notes the presence of a special strategy as part of its prototype. When this frame is activated and successfully matched, the suggested methods will point to the special strategy contained in the disease frame (which can then be selected as the current strategu).

3.3.2 Is It a Realistic Model?

The use of frame theory to describe strategies in some sense does violence to the original conceptions of Minsky. While there is not direct evidence for frame theory as an explanatory mechanism (i.e. a theory of diagnostic problem-solving), it is possible to argue that it is not an

unreasonable explanation:

- 1. Through repeated experience in the process of taking a present illness doctors develop strategies to deal with each possible configuration of the IDC.
- 2. At each level of expentise, within a particular specialty the underlying knowledge base of medical facts possessed by each doctor is, to a close approximation, the same.
- 3. Knowledge of strategy is an integral part of the dector's knowledge base.
- 4. It is unreasonable to assume that doctors associate with each individual disease and with each possible grouping of diseases a separate set of strategies.
- 5. Therefore, it seems likely that to a significant degree what determines the selection of a strategy are the features shared by sets of diseases and findings and the classes of structural relationships that can exist among diseases.

By features of diseases I mean such things as the acute vs. chronic classification. Structural relationships are CAUSE, COMPLICATION, etc. If indeed doctors do key their strategy selection in part on these features and relationships (and the protocol analysis provides some confirmation that they do) then the strategy frame model is not unrealistic.

3.4 Diagnostic Style and the Strategy Frame Model

If it is indeed true that (at least four of) the doctors who were subjects have approximately the same underlying (medical) knowledge base

(which includes the knowledge of how to take a present illness), how can the differences in behavior found in the protocols be accounted for and incorporated in the SFM? One obvious answer is that even though they had heard the same information, they had different diagnostic configurations (e.g. different hypotheses). If we assume, however, that they had the same configurations this variation in behavior is disturbing. A subset of this variation can be easily accounted for by the SFM as it stands. Variations can be produced by assuming that the choice of a node from the set of nodes at a particular level within the goal-tree is a a free choice. One kind of behavioral variation that this can produce is a permutation in the question order within a set of closely related questions. This can be seen, for example. In a strategu of confirmation that calls for matching the principal-parts of a disease prototype. The selection of which principalpart to inquire about first can be made a free choice (in manucircumstances). Free choice could result in significantly altering the diagnostic configuration (and the resulting behavior) if a crucial fact that is at variance with expectations is uncovered at an earlier stage.

The choice of completely different strategies (under the assumption of identical diagnostic configurations) must still be accounted for within the SFM. I use the term <u>diagnostic style</u> to refer to the variations in behavior among doctors resulting from the selection of different strategies. It is useful to get a feeling for what this kind of variation is like. Some doctors seem to be more aggressive in pursuing a hypotheses; they ask more questions directly related to its final confirmation at

earlier stages in the interview. In contrast to the "aggressive" style there is a more "cautious" style. A doctor having this style tends to explore many different areas before focusing in on his principal disease hypothesis or he might intermix the confirmational questions with explorational questions. This kind of behavior can be explained in a number of ways. There can be a high cost associated with missing important information and thus some doctors are careful not to be led down a "garden path." Alternatively, the doctor may not feel comfortable with a high level of complexity in the hypothesis structure or with a certain area of disease and might try to eliminate unlikely possibilities before attempting confirmation. Doctors are also sware of the fact that a patient may have multiple, unrelated problems while only presenting with findings of one of them. Many disease processes such as cancer or pre-azotemic renal failure can be "silent" in early stages.

Variations can be seen in how doctors choose and pursue a finding.

Some doctors choose to exhaustively characterize a finding already obtained before seeking any new findings. Other doctors try to expand the total picture they have of a patient before going after specific finding characterizations. Occasionally, a doctor will reverse the normal sequence of current symptoms - past related medical history and immediately ask about the history.

The descriptions I have presented are imprecise and impressionistic; they were intended to give a reader a broad overview of the range of variation that is observed. Any model that claims to be able to reproduce

the behavior to doctors in taking the present illness must include the factor of diagnostic style. The SFM provides a framework for including diagnostic style. It can be included by weighting the scoring of the strategy frames.

Associated with the matching of each terminal of strategy frame is a score. This score is a measure of the importance of the feature in the selection of the particular strategy. The total score is obtained by adding the scores of each terminal. This score represents the weight-of-evidence in favor of selecting the strategy. By assigning a weighting system to the elements of the diagnostic configuration variations in the selection of strategy can be produced. (Note: The weights are assigned to the variables of the IDC not to the features of the case that caused assignments to the variables.)

When a terminal of a strategy frame is matched to a element of the IDC, the matching score is now calculated by multiplying the element's weight with the terminal's raw score. (Of course the weighting system must use normalized weights.) The entire frame is then scored by adding up the individual weighted scores for each terminal. This will reorder the scores of each strategy frame in a different way.

It is not obvious how this would work, so consider the effect of a particular weighting scheme. If we want to produce an aggressive style we would assign heavy weights to each element of the IDC that involves the presence of a LIKELY hypothesis. By doing this we tend to swamp the contributions by the other elements of the configuration. On the other

hand if we want to produce a more cautious style we would weigh these same elements very little. To produce a style that tends to characterize a finding once obtained, we heavily weight the IBC element that specifies the presence of a differential characterization network for a finding. While this description does not tell the whole story of how style is incorporated into the SFM, it shows that the model is flexible enough to include it in a rather simple way.

Style is perhaps the least understood facet of human problem-solving behavior and specifically, the problem-solving behavior of doctors.

Dombal, who examined the protocols of doctors taking a case of abdominal pain was forced to conclude that (probably because of the effects of style) that there was no such thing as the "diagnostic process." <Dombal 73> In other problem domains it may have as much effect in determining time-to-solution as intelligence and domain-specific knowledge.

CLASSIFICATION OF STRATEGIES

In this chapter a scheme is presented for classifying the data-gathering strategies from the protocols. For a long time one type of strategy has dominated the thinking of the medical profession - the differential diagnosis of a symptom (such as abdominal pain). The codification of this approach in a book such as French's <u>Index of Differential Diagnosis</u> was considered an important step forward in the systematic organization of diagnostic procedures. Of course, many doctors now accept that diagnosis requires a larger repertoire of strategies.

The classification scheme presented below was developed by assessing the intended effect of each question. It was clear that the answers to certain questions (or groups of questions) would have the effect of establishing a hypothesis if the answers met the expectations of the doctor. On the other hand, there were questions that were aimed at removing a hypothesis from contention or deciding which of two different but closely related hypotheses was better. Finally, there was a class of questions whose intended effect was to develop a new hypothesis or sharpen an existing one. The names I have assigned to each of these different types of goals are Confirmation, Elimination, Discrimination and Exploration. A strategy is assigned to one of these four categories based on the intended effect of its top-level goal. The protocol analysis revealed that within each of these categories there was a wide range of

variation; there are different types of confirmation, elimination, discrimination and exploration strategies. The classification of strategies within a category is made on the basis of the method used. In a direct method, the data sought is the same as (or equivalent to) the entity to which the goal is being applied. In an <u>indirect</u> method, the data sought is derived from the premise of a rule that associates the data with the entity to which the goal is applied.

A scheme for classifying strategies says nothing about the conditions for the selection of a particular strategy. In addition to its value as an observation of underlying structure in the experimental data, however, it can give us a way of viewing strategies in terms that can aid in our understanding of the clinical decision-making process. If each question asked by the doctors represented a different tupe of strategy our ability to describe and understand the doctor's problem-solving behavior would be very limited. It is the existence of similarities among the goals of the questions that makes a classification scheme for strategies possible.

4.1 Confirmation Strategies

Confirmation strategies are strategies to establish or validate a hypothesis. They are characterized by questions that seek evidence that will support the hypothesis to be confirmed. Further, the type of evidence that is sought is positive evidence. This means findings that are characteristic of the disease, clinical state, etc. under consideration.

This is in contrast to supporting a hypothesis by evidence that that tends to weigh against competing hypotheses.

Evidence can be classified by its relationship to a hypothesis.

Sufficient (or prima facie) evidence is sufficient to confirm a hypothesis.

For example, dysuria is sufficient evidence for bladder irritation.

Necessary evidence consists of findings that must be present or the hypothesis is rejected. Supporting evidence consists of findings that add weight in support of a hypothesis. Consistent evidence is similar to the legal concept of circumstantial evidence; it can not be used to support a hypothesis unless there is direct supporting evidence. Negative evidence consists of findings that weigh against or are inconsistent with a hypothesis.

4.1.1 Direct Confirmation

Direct confirmation strategies are confirmation strategies that use direct methods. They can be further subdivided into expert witness and prima facie confirmation. Expert witnesses are those doctors who have observed the patient at some time in the past and have evaluated their medical status. In addition, the doctor has reason to trust their conclusions. An example of this strategy is the following, the doctor wants to confirm a history of previous uninary tract infections:

D: Did they (the doctors at the hospital) arrive at a conclusion from the IVP.

K: She was said to have a bilateral staph. pyelonephritis.

As our protocols demonstrated direct confirmation using the expert witness method will be quickly abandoned once the witness has proven to be unreliable. In general, however, doctors must rely on the previous hospital records or reports from other doctors to a certain extent.

Prima facie direct confirmation is normally restricted to physical examination or laboratory findings but can be used in other phases as the example of the dysuria demonstrates. This strategy can best be understood by observing that asking for prima facie evidence is equivalent to asking if the patient has the condition being tested for. Asking if the creatinine value is elevated is equivalent to asking if the patient has renal failure.

4.1.2 Indirect Confirmation

Indirect confirmation was the most common strategy found in the protocols. A single application of this strategy generated from one to as many as fifteen questions. Indirect confirmation strategies are based on the use of finding-disease association rules. The general form of these rules is:

evidence-for <collection of findings> -----> <clinical condition>

In the simplest case for example, fever is evidence for an active infection. For anything other than very simple conditions the collection of findings is a complex, highly-structured set consisting of many elements. If a sufficient collection of positive findings is found, the clinical condition is considered confirmed.

4.1.2.1 Case-Building - An Indirect Confirmation Strategy

Consider the task of a district attorney prosecuting a case against a defendant. In order to be successful he must show a number of things:

- 1. A crime has been committed.
- 2. The defendant had the opportunity to commit the crime.
- 3. The defendant had the necessary "tools" available to him.
- 4. The defendant had a motive for committing the crime.

In addition, he must demonstrate a prima facie case. (A case built entirely on circumstantial evidence will be thrown out by the judge.) A case of homicide is a good example. To show that a homicide was committed a body must be found. To show that the defendant had opportunity there must be evidence (witnesses, credit—card trails, etc) that the defendant was near the victim at the time of the murder. If the victim was shot, the prosecutor must produce a gun and show the defendant had possession of it at the time of the murder. To show motive the DA must prove that the

defendant had a reason to kill, either gain or animosity, for example. Of course if enough credible witnesses saw the defendant pull the trigger, the other elements might not be necessary (Although metive is important in determining which degree of homicide the defendant will be prosecuted for.).

Case-building in medical diagnosis is similar to this legal example. (Of course the doctor is not bound by the rules of evidence.) The concept of opportunity is found, for example, in the consideration of a patient who seems to have a disease that has a clear demographic distribution. The past presence of a patient in a place where he would have been exposed to a certain kind of infectious disease would normally be confirmed if the doctor is considering as a diagnosis an infection that is localized to that part of the world. The analogu of "tools" can be seen in diagnosing alcoholic cirrhosis of the liver. The doctor must demonstrate that a sufficient amount of alcohol was available and consumed by the patient. The concept corresponding to motive in medical diagnosis is predisposition. A person with diabetes mellitus is predisposed to having urinary tract infections; he has a "motive" for getting the disease.

This analogy (like most analogies) can be overdone. The point I would like to make is that in attempting to confirm a diagnostic hypothesis a great deal of evidence of different types must be gathered. Case-building is not a single strategy but a collection of strategies. The variant of case-building that the doctor uses depends on the nature of the disease hypothesis to which it is applied. The three major variants that I have

identified correspond to the classification of a disease as acute, singlephase, acute, multiple-phase or chronic.

Case-Building-1 - Acute, Single-phase Diseases

An acute, single-phase disease is one that develops over a short time period and has only one phase of development. Examples are influenza, acute myocardial infarction (heart attack) and APN. Case-building applied to this kind of disease is characterized by an attempt to confirm the disease prototype. It is difficult to give an exact definition of what a prototype is. The working definition that most of the doctors seemed to use is that it is the collection of signs and symptome that they would expect the patient to have if the patient had the disease. It includes all the findings that are necessary evidence. For example, the prototype for influenza is:

(PROTOTYPE INFLUENZA (SYMPTOMATIC-HISTORY-REVEALS (ONSET (CONSTELLATION (FEVER AND CHILLS AND MALAISE)) (BETLEEN: (18 MIN) (2 HR))) AND MUSCLE-ACHES AND COUGH AND NASAL-STUFFINESS AND (OCCASIONALLY PROSTRATION AND NAUSEA AND CORYZA)) (PHYSICAL-EXAMINATION-REVEALS MILD-PHARYNGEAL-INJECTION AND FLUSHED-FACE AND CONJUCTIVAL-REDNESS) (LABORATORY-DATA-REVEAL LEUKOPENIA))

The prototype for APN can be found in Appendix 1.

A variant of this form of case-building is used for acute, multipleetiology diseases when the etiology is a factor in determining a treatment, acute pericarditis, for example. This variant of case-building will typically invoke a sub-strategy to determine the etiology.

Case-Building-2 - Acute, Multiple-phase Diseases

The diseases that fall under this classification are short-term with two or more distinct phases of development. Examples are acute glomerulonephritis and acute tubular necrosis (acute renal failure). This strategy is characterized by confirming a <u>set</u> of prototypes, each representing a phase of the disease. The prototype-set also includes the time-relationships among the phases. For example, the set of prototypes for acute tubular necrosis:

(PROTOTYPE-SET ACUTE-TUBULAR-NECROSIS

((PROTOTYPE GLICURIC-PHASE

(OLIGURIA AND

RED-CELLS AND

GRANULAR-CASTS ... ETC.))

(PROTOTYPE DIURETIC-PHASE

(POLYURIA ... ETC.))

(TIME-BETHEEN (OLIGURIC-PHASE DIURETIC-PHASE)

(BETHEEN (2 DAYS) (6 NEEKS))))

In the case of a multiple-ethology disease such as acute renal failure, where the ethology is a factor in determining the treatment, a sub-strategy may be invoked to determine the cause.

Case-Building-3 - Chronic Diseases

Chronic diseases are long-term diseases that may or may not have acute episodes. In many chronic diseases the presentation may not have a characteristic prototype depending on the stage of development. In order to characterize chronic diseases a developmental scenario is needed. This scenario is similar to the connected phases associated with acute, multiple-phase diseases. It consists of a sequence of events and their associated time relationships. Occasionally, the sequence of events is very clear such as the three stages in the development of suphilis or a history of rheumatic fever in rheumatic heart disease. In many cases, however, there is no single sequence of events that can characterize the development of a chronic disease. When this is the case a set of alternative scenarios is encased together and called the story of the development of the disease. Included in this story are the common features of the different scenarios. For each scenario there can be findings that are independent of the stage of development. A tupical pattern of questioning in the protocols was for the doctor to tru and establish the initial point of the story. In many diseases the initial event is the common locus for all the scenarios. Even when this is not the case, the initial event can be a good guide to the selection of the appropriate scenario.

This variant of case-building does not have as rigid a sequence of actions as do the other variants. There are, however, definite components of the strategy that are normally attempted (regardless of the order in

uhich each is tried). They are:

- 1. Confirm the initial event (including prenequisites). The initial event is normally associated with the cause of the disease. In this situation it is called the precipitating event.
- 2. Confirm the developmental scenario by confirming each principal event. This pen determine the stage of development.
- 3. Confirm the time-independent findings.
- 4. If the disease has a typical acute presentation, confirm the associated prototype.
- 5. Confirm any prediapositions to the precipitating event.

An example of this strategy is in the protocol enalyzed in Chapter 2. Here the doctor wished to confirm chronic pyelonephritie. The precipitating event was an initial uninary tract infection in association with a kidney stone 17 years earlier. The scenario that the doctor chose to match specified chronic bacteriums with scyte flare-upe of uninary tract infection. After establishing this a the doctor established a predisposition to CPN, phenacetin abuse.

4.2 Elimination Strategies

Elimination strategies are invoked when the doctor wishes to remove a hypothesis from active contention. This normally means moving the hypothesis to the RULED-OUT section of the PLIS. (Maying a hypothesis from

LIKELY to UNLIKELY will also be considered an application of an elimination strategy. This is because in the phases preceding the physical exam and lab data it is normally not possible to RULE-GUT or CONFIRM a hypothesis.) The most important element of the IDC in determining whether elimination will be selected is the total number of LIKELY and POSSIBLE hypotheses. The larger this number, the greater the likelihood that elimination will be selected. This, however, is not the only criterion for the selection of elimination. The number of separate areas covered by the set of hypotheses is another crucial element. As shown in the analyzed protocol, the presence of independent GI hypotheses on the POSSIBLE list was a factor in the doctor's selection of an strategy at Question 4. (The secondary goal was to eliminate GI as a separate, independent problem.).

Many factors go into the decision as to which hypothesis to eliminate after elimination has been selected. The following factors weigh in the favor of a hypothesis being selected for elimination:

- 1. The position on the PLIS. The lower down on the list, the more likely the hypothesis is to be selected for cutting.
- 2. The increase in the compactness of the PLIS to be gained by cutting a hypothesis. Hypotheses that will remove whole areas from consideration are prefered.
- 3. The relationship of the cut findings of a hypothesis to the current focus of questioning. (Cut findings are those findings that are considered to be necessary evidence for a hypothesis or have a very high frequency of occurrence in the disease.) Hypotheses whose cut findings are closely related to the group of findings under consideration are prefered choices for eliminating.

If the current focus of questioning is urinalysis findings, for example, a hypothesis that can be eliminated by asking about the presence of red-cell casts would be preferred.

- 4. The potential decrease in the PLIS density: if a whole set of hypotheses on the hypothesis list connected through relational links can be removed by cutting a hypothesis. Cutting a hypothesis that has many CAUSE or COMPLICATION links on the PLIS is generally prefered.
- 5. The prognosis of a disease hypothesis. The protocol analysis revealed that the more serious the disease was in terms of difficulty of treatment or having a poor prognosis the more likely the dactor was to try and eliminate it.
- 6. The number of findings left unexplained by a hypothesis. A hypothesis that leaves many of the findings unexplained is a prefered choice for elimination.

Elimination strategies are characterized by questions about the absence of findings that are either necessary evidence for a disease or are so often found if a disease is present that their absence weighs very heavily against a disease hypothesis. What distinguishes an elimination strategy from a discrimination strategy is that a finding sought that is evidence against the disease hypothesis to be eliminated is not necessarily evidence that supports any other hypothesis on the list.

The goal of elimination must also be distinguished from the possible results of exploration. An exploration strategy may have the result of eliminating areas of consideration, but since the hypotheses that are eliminated were never on the PLIS to begin with, it can not be interpreted as an elimination strategy.

4.2.1 Direct Elimination

Direct elimination is similar to direct confirmation. In direct confirmation the findings that are sought are those that are prima facie evidence in support of a hypothesis. In direct elimination the findings sought are prima facie evidence against the hypothesis. Asking about these findings is thus equivalent to asking if the patient does <u>not</u> have the condition. Consider the following example taken from Protocol 2:

- D: First I'm going to ask some questions about the character of her uninary stream because I'm thinking in terms of infection in the lower uninary tract. Did the patient notice any blood in her unine?
- K: No. she didn't.
- D: That she didn't have gross hematuria makes me turn away from one possibility that she might have past a stone in association with infection. She might have had a hemorrhagic cystitis and that makes it unlikely.

Another example from the same protocol:

- D: The one other thing I might be interested in this lady with what you told me is the fact that she continued to have white cells in her urine. Atthough it's completely consistent with chronic pyelonephritis I would be interested in getting a TB culture on her just because of her history a long period of time ago. I would expect them to be negative.
- K: TB cultures were performed and were negative.

A special case of direct elimination occurs when the finding (if positive) is sufficient evidence for the hypothesis and if negative is

sufficient evidence against the hypothesis. In the annotation of the protocols I noted this type of strategy with a top-level goal of Confirm/Eliminate. There are many examples. Asking if due could be seen in the uneters: if yes, no total uneteral obstruction, if no, obstruction is present. Another example is the unine culture as evidence of active uninary tract infection.

4.2.2 Indirect Elimination

Indirect elimination is a class of strategies that have structural similarities to indirect confirmation strategies such as case-building. It is tempting to call these strategies negative case-building. These strategies were not as commonly seen in the protocols simply because in the case we used, they probably were not needed. Indirect elimination is characterized by the following set of subgoals:

- 1. Confirm symptoms inconsistent with the hypothesis.
- 2. Confirm the absence of a precipitating event, sticlogical agent, and predisposition
- 3. Confirm the absence of principal events in the developmental acepanio.
- 4. Confirm physical exam findings inconsistent with hypothesis.
- 5. Rule-out (eliminate) the hypothesis using Laboratory data.

Consider an example from protocol 4. The doctor is trying to eliminate chronic abdominal problems:

- D: Has this lady ever had any abdominal complaints which required surgery in the past?
- K: She had a cholecustectomy and eppendectomy seventeen years earlier.
- D: Was this the first time she ever had abdominal problems.
- K: Yes
- D: She'd never had any trouble as a child where she missed school because of abdominal complaints.
- K: Not that we're aware of.
- D: Or had irregular bouel movements—again I'm looking. the goal here ... is this a chronic pattern of a person who has had abdominal complaints all their life and had an appendent on and choiceystectomy done on the basis of just chronic complaining. ... Util indeed at the time of her choiceystectomy she have galistones?
- K: We don't have that information.

As the reader has probably noticed, the interpretation of this set of questions is subtle. It could be argued that the doctor was trying to confirm a developmental scenario for chronic abdominal problems. The reason Dr. Kassirer and I chose to interpret this as indirect elimination was the absence of any links made by the doctor to her current symptoms and the remarks made by the doctor after the protocol had been taken in which he said that it is very common to see a pattern of chronic complaints and procedures performed without any underlying chronic problems being found.

4.2.2.1 Causal Exclusion - An Indirect Elimination Strategu

An example of indirect elimination is a strategy called causal exclusion. When a patient presents with findings that suggest a multiple-etiology syndrome, clinical or physiological state such as nephrotic syndrome, renal failure, sodium retention or hypertension, a set of the possible causes may be placed on the PLIS. Depending on the place in the protocol where the resulting condition is hypothesized, this list can be very long or quite short. Beeson and McDermitt, for example, list 44 different etiologies for chronic renal failure «Beeson and McDermitt 71». A diagnosis must include the underlying causal mechanism. As has been stated previously this is important for a number of reasons. Among them is to separate out treatable causes from untreatable causes. The mode of treatment may also depend on the etiology.

Causal exclusion is a strategy that is invoked in order to eliminate from consideration a subset of the possible causes of the resulting state. This strategy is characterized by seeking findings that are consistent with resulting clinical state but inconsistent with the cause (or set of causes) to be eliminated. An example of causal exclusion can be found in Protocol 2.

D: ...Did she have any history of high blood pressure? The reason I'm asking that question is that in association with certain kinds of renal insufficiency hypertension is a very common feature.

K: No, she didn't.

D: The answer to that question leads me away from something like chronic glomerulonephritis as causing her renal insufficiency. It's consistent with chronic pyelo. You could have hypertension or no hypertension.

4.3 Discrimination Strategies

Discrimination could be viewed as a form of elimination in that the goal is to eliminate one or another of two competing hypotheses. Looked at in the other direction, elimination could be seen as a form of discrimination, where the two hypotheses to be discriminated are "Has-X" and "Does-not-have-X." Discrimination strategies, in fact, do have a unique characterization that sets them aside from elimination strategies:

1. They are applied to pairs of hypotheses. 2. In terms of the information sought, any evidence that supports one of hypotheses is also evidence against the other hypothesis. Medical books are not very precise in making distinctions between elimination and discrimination. Doctors use the term differential diagnosis to refer to the combined use of elimination and discrimination strategies to determine which of two or more diseases with similar symptoms the patient has.

It might be argued that elimination strategies tend to indirectly support the hypotheses that remain under consideration. If we eliminate an alternative hypothesis from a list that is known to be an exhaustive enumeration of all possible diagnostic hypotheses we in fact do tend to add weight to (at least) a subset of the hypotheses that remain. Of course,

this situation may not occur with with any frequency in real diagnostic situations. In the same light, by eliminating all other options but one, under the same conditions, we could conclude that we have confirmed the remaining hypothesis. These techniques are clever and no doubt are used by most physicians some of the time. In the protocol analysis, however, I did not observe these techniques being used. What was evident, however, was that there were two different kinds of questions associated with the application of a discrimination strategy: 1. Asking as a noncommittal question for the presence or absence of a finding or for the value of some measure (such as the hematocrit). 2. Asking a set of questions to characterize a finding, normally a symptom.

4.3.1 Direct Discrimination

Direct discrimination is a strategy that has one form. The finding that is being asked about, if positive is considered to be prima facial evidence in support of one hypothesis while the negative finding is necessary evidence for the other hypothesis. An example that was found in nearly every protocol was to discriminate an acute from a chronic problem. Five out of the six doctors asked for the duration of the nausea and vomiting. Nausea and vomiting of long duration is prima facial evidence for a chronic problem, while nausea and vomiting of short duration is necessary evidence for an acute problem. Due to the nature of clinical medicine there are not many examples where such a clear cut discrimination is

possible.

4.3.2 Indirect Discrimination

Most discrimination strategies that were observed fall into the category of indirect discrimination. Indirect discrimination is characterized by questions derived from rules that associate the positive finding as supporting evidence for one hypothesis and negative evidence for the competing hypothesis. In Protocol 4 an example of the first form of discrimination strategy can be found (+ or - finding). In this example the doctor is trying to discriminate a bowel problem from a kidney problem:

- D: I'm thinking more of a kidney thing than a bouel thing and I would ask her did she have any change in bouel movements associated with this (the abdominal pain)? Is this a possibility.
- K: We don't have any information about it.
- D: There is no diarrhea that we're aware of?
- K: We don't have any information about that.

Just prior to this set of questions the doctor used the second form of discrimination (symptom characterization):

- D: I'm feeling ...it would be more likely a kidney problem or a bladder problem I'd be concerned with. I would ask about the the abdominal pain...dld the patient describe it or where it where it is located?
- K: The location was said to be in the area of the left flank.

Later on in the protocol the doctor again returned to the characterization of the abdominal pain in order to discriminate the flank pain associated with obstruction or renal calculi from the flank pain of (uncomplicated) puelonephritis:

- D: Did the pain in the flank radiate at all... did it move?
- K: It was said to radiate around to between the left flank and the left kidney area.
- D: Did the pain at all radiate down into the grain?
- K: She had also superpubic pain and tenderness at times.

The final example of differential symptom characterization comes from Protocol 2. In this example the doctor is trying to discriminate uremia from other causes of nausea and vomiting (specifically, GI causes).

- D: Was she nauseated for the whole period of time? Again I'm trying to characterize this nausea that she had. And the vomiting: was it associated with food, was it spentaneous. First of all, was she continually nauseated?
- K: She first became nauseated toward the beginning of the illness and later on began to vomit.
- D: And was the vomiting associated with eating or did she wake up in the morning with nausea and vomiting?
- K: I don't have information about that but I can tell you she did lose weight during that time.
- D: I'm still thinking renal disease and I don't know why at this point...But I was thinking with that question in terms of uremia.

4.4 Exploration Strategies

The discovery of a class of strategies whose top-level goals could neither be classified as confirmation nor as a form of elimination or discrimination was unexpected (even though it is not difficult to imagine strategies that are not hypothesis driven.). The top-level goal of diagnosis specifies developing a diagnostic hypothesis and then confirming it. If anyone has even had to repair a car or a radio there are many times when the presenting "sumptoms" suggest nothing more specific than "the electrical system" or "the power supply" and even these non-specific hypotheses might have a low certainty. The strategies that car mechanics, radio and TV repairmen use have a certain similarity to the strategies that the doctors used. In the presence of non-specific clues these diagnosticians can (and do) use variations of what could be called checkout lists. A check-out list is simply a pre-compiled set of questions or conditions to be tested. The nature of these lists is such that in almost all cases they will turn up something that will generate a very specific hypothesis. The diagnostician then can either complete the check-out list or immediately turn to the hypothesis (or hypotheses), that has been generated. These lists can be quite complex: they can contain many branch points and levels of detail. In medicine, the comprehensive check-out list is called a review of sustems.

The protocol analysis revealed three different intended effects whose top-level goals I have classified as exploration. They are:

- 1. Developing a hypothesis in an area where there are no hypotheses at present.
- 2. Sharpening a hypothesis by making it more
- 3. Checking for additional problems in the presence of a hypothesis structure that is already sufficient to explain the known findings.

The use of the word "area" in 1. Is meant to be very general. It can refer to an area of internal medicine such as valvular heart disease or carcinomas of the pancreas. It can also refer to an aspect of the relational structure of a disease such as complications of urinary tract obstruction or causes of renal failure. It can also refer to aspects of the patient's condition that cut across all clinical categories such as the severity of the illness.

sharpening a hypothesis. It is not uncommon for a doctor to start with a broad hypothesis such as chronic renal disease or acute abdominal problem and try to sharpen it resulting in, for example, chronic pyelonephritis in the former case and acute pancreatitis in the latter.

An important consideration in making a final diagnosis is that it should be "complete" in the sense of not missing any secondary, subsidiary or complementary problems of the patient. Even though these might not be the major problems (or at least the most sevious sheet that the patient is manifesting, any management decisions about the patient should only be made with as complete a diagnostic picture as possible. In the case that we

used all of the doctors uncovered the secondary anemia and five out of six, the metabolic acidosis.

4.4.1 Direct Exploration

Direct exploration can easily be confused with direct confirmation or direct elimination. As in these two strategies it is characterized by asking for prima facie evidence – thus it is equivalent to asking if a condition is present. It can be differentiated from these two strategies by two conditions: 1, The hypothesis being tested is not on the PLIS and/or 2. There has been no evidence presented as yet that the condition is present. Direct exploration can result in the simultaneous activation and either the confirmation or elimination of a hypothesis. This strategy was used by one doctor and resulted in uncovering and confirming the patient's metabolic acidosis. From Protocol 2:

- D: Now, in terms of a lady who is presenting with unemia with what I think are unemic symptoms in association with an acute bacterial infection superimposed on chronic pyelo. I'd be concerned with her electrolyte status at the time of admission as well.
- K: You want to know her Na. K. CI and CO2?
- D: Yes
- K: 140, 3.7, 109 and 12.
- D: Given that information which makes me think she's got a metabolic acidosis with an increased anion gap, I'd like to know her pH just to make certain that's what is going on.

K: Her pH was 7.18 , PC02 15.

Occasionally, direct exploration can use the expert witness method. From Protocol 6:

- D: The next question would be had she any history of any kidney disease in the past. Specifically has she ever been told that she had a kidney disease as a youngster or in the course of pregnancies or anything of that sort?
- K: She did have a history of difficulties with her kidneys in the past.

4.4.2 Indirect Exploration

Indirect exploration is characterized by questions derivedfrom rules that associate the finding sought to the <u>activation</u> of a disease hypothesis. This strategy can encompass a single question or whole groups of questions. The following example from Protocol 3 can be considered as a single question. The doctor has invoked exploration in order to develop a hypothesis in the area of chronic renal disease:

- D: ... Now she literally had nothing else going on in the intervening time, is that right?
- K: The history that she gave said that she had no serious illnesses during that time.

- D: No urinary symptoms, no abdeminal pain, no nausea, no vomiting?
- K: That's correct.
- D: No bouts of unexplained fever?

K: No unexplained fever.

One area that concerned the doctors was the severity of her illness.

Question 4 of the analyzed protocol is an example of indirect exploration focused on the question of severity. In the next example (from Protocol 6) the issue of severity in a previous illness is explored:

- D: Was she hospitalized on that occasion?
- K: Yes she was. Why did you want to know that?
- D: Because I would assume to some extent the severity of her symptoms might be reflected in whether or not she was hospitalized. It would give me an indication of how seriously lil she was at the time.

The next example shows how indirect exploration can be used to develop a hypothesis of early renal failure. Again from Protocol 6:

- D: Was there any modification of her diet?
- K: Not that we're aware of.
- D: Was there any reduction in the amount of protein in her diet?
- K: We're not aware of any change in diet.

If a doctor had found renal insufficiency in the past, one therapy plan might include a reduction in the amount of protein the patient could incorporate in her diet.

The final example of a single-question, indirect exploration strategy shows how it can be used to check for secondary or associated conditions (in this case hypertension of renal origin). Again from Protocol 6:

- D: Also one question I should ask is do you have any information about her blood pressure. Is her blood pressure elevated during that hospitalization or has it ever been?
- K: I'm sorry but I don't have that information.
- D: I raise that question now because of its association with chronic renal disease. My computer tells me that whenever you get chronic renal disease you need to know about the blood pressure.

4.4.2.1 The Review of Systems

The review of systems is the best example I found of an extended, indirect exploration strategy. Many doctors consider asking this collection of questions to be a routine procedure to be performed regardless of any diagnostic hypotheses. Indeed, the second halves of Protocols 4 and 6 were extensive and nearly complete reviews of systems. The normal review of systems will cover such areas as the head, ears, eyes, nose and throat (HEENT), the skin and muscular-ekeletonal system, the cardiovascular, genitourinary, gastrointestinal and neurological systems. Most doctors will include a history of medications and previous illnesses (although these are usually considered part of the medical history).

As an example of this strategy I will focus on a protocol that, so far, has not been used as an example - Protocol 5. This protocol was by far the most difficult one to analyze. Most of the protocol was a review of systems, but one that seemed to be tailored to the case in question. By using this strategy the doctor very effectively uncovered the phenacetin

abuse and eliminated the possible abdominal etiology. The reason for the difficulty in analysis was that while each question could be viewed as having a confirmation or elimination goal there seemed to be no clear hypothesis structure to which to apply these goals. For this reason both Dr. Kassirer and I decided that each question was really meant to develop hypotheses rather than confirm or eliminate them even though in the process of developing a diagnostic hypothesis, the doctor does eliminate certain areas of consideration.

- D: I think I'd like to go to sort of a systematic review of her health to see if we can pick up any ancillary information. Had she ever been told abouthad she ever had any problems with her skin? Rashes or allergies?
- K: No.
- D: Had she been subject to headache?
- D: She had a long history of headaches.
- K: Was she treated? Did she treat herself?
- D: Yes, she has taken some medication for some time.
- D: Do we know the nature of this medication?
- K: She took Empirin tablets.
- D: How manu did she take.
- K: As far as we can tell she took around 6 tablets a day.
- D: Over what period of time.
- K: For about 15 years.
- D: What I'm concerned about now is whether she has

had an abuse of this compound.

After the history of headaches has been established the possibility of drug abuse arising from self medication immediately follows. Chronic headaches are associated with hypertension, thus the next series of questions:

- D: Did she have any problem with her blood pressure?
- K: She never was told she had high blood pressure.
- D: Had it been examined?
- K: It had been examined on numerous occasions some time ago.
- D: How about her vision?
- K: No problems with her vision.
- D: Has she been subject to seizures?
- K: No.
- D: How about depressions? Psychiatric....?
- K: No.
- D: Had she any problems with her breathing? Any shortness of breath, cough?
- K: No.
- D: Hemoptysis, chest pain?
- K: She mentioned that she has had a chronic dry hacking cough without sputum production.
- D: Obviously the most cogent thing we've picked up in this review is the very heavy abuse of Empirin which I think could well be related to the problem she had with kidney stone; could well in fact have been a sloughed papilla from papillary necrosis. The episode she's having now could well represent episodes of renal infection or papillary necrosis

related to phenacetin abuse or analgesic abuse.

The doctor is being a little modest at this point. The hypothesis he states is clearly his PDH. The remaining questions can either be viewed either as exploration or a weak form of elimination. It was difficult to decide because the doctor's PLIS now contains a very sharp hypothesis.

- D: Had she had any problems with cardiac disease or angina?
- K: She has no cardiac symptoms.
- D: Ankle suelling?
- K: No.
- D: Aside from this weight loss that she experienced with this present illness, had she had any change in her weight over the past year?
- K: No.

The focus is now in the area of GI problems.

- D: Any problems with her appetite or any difficulty swallowing?
- K: No.
- D: Any previous episodes that we know about of gastrointestinal upset, any ulcers?
- K: No.
- D: The cholecystectomy was performed because of what...jaundice? Pain?
- K: I don't have any information about that.
- D: Bouel habits?
- K: She has a rectocele. She has had a history

of severe consitipation more sever since March.

- D: Has she taking any medication?
- K: No she was not.
- D: Had she noticed any change in the color of her stool?
- K: No.
- D: I want to get a feel for her menstrual history пом...onset of menses was...?
- K: Normal age.
- D: Does she have any children?
- K: She has one child who is 35 and well.
- D: Did she have any difficulty with that pregnancy?
- K: Not that we're aware of.
- D: And was it delivered normally?
- K: I don't have any information.
- D: But she didn't have any incision or cesarean section?
- K: No.
- Di Did she have any problem with her joints? Any swelling? Pain like arthritis?
- K: No.
- D: Well again, I think the main thing we've learned from that brief review of systems is that she's had this large lingestion of Empirin.

In Chapter 3 I discussed diagnostic style. Hithout any considerations of style the preceding dialogue could not be explained in a satisfactory

way. For another doctor, the hypothesis of phenacetin nephritis might have immediately invoked case-building. In this example, however, the doctor continues the review of systems to its conclusion.

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Appendix 1 - A Frame for Acute Puelogephritis

```
(FRAME ACUTE-PYELONEPHRITIS
   (CLASSIFICATION-OF *
      ACUTE EPISODIC SINGLE-ETIOLOGY INTERSTITIAL RENAL-DISEASE)
   (ULTIMATE-ETIOLOGY-OF *
       BACTERIAL-INFECTION-OF-URINARY-TRACT)
   (ULTIMATE-SEQUEL-OF *
       NONE)
   (STORY-OF *
    (ALMOST-ALMAYS
      (ASCENDING-PATHUAY-ROUTE SCENARIO
         (BEGINS-WITH (EPISODE-OF LOWER-URINARY-TRACT-INFECTION))
        ((EPISODE-OF LOWER-URINARY-IRACT-INFECTION) PROCRESSES-TO (EPISODE-OF KIDNEY-INFECTION) ((EPISODE-OF KIDNEY-INFECTION) CAUSES (EPISODE-OF *))
         (TIME-PATTERN
               ((INTERVAL-BETHEEN
                  (EPISODE-OF LOWER-URINARY-TRACT-INFECTION)
                  (EPISODE-OF KIONEY-INFECTION))
                (BETHEEN IN DAYS) (7 DAYS)))
                ((INTERVAL-BETHEEN
                  (EPISODE-OF KIDNEY-INFECTION)
                  (EPISODE-OF *))
                 (BETHEEN (8 DAYS) (4 DAYS))))
         (COTEMPORANEOUS (EPISODE-OF LOUER-URINARY-TRACT-INFECTION)
                          (EPISODE-OF KIDNEY-INFECTION)
                          (EPISODE-OF *))))
    (OCCASIONALLY
        (DESCENDING-PATHHAY-ROUTE SCENARIO
          (BEGINS-WITH (EPISODE-OF BLOOD-BORNE-INFECTION))
          ((EPISODE-OF BLOOD-BORNE-INFECTION) PROGRESSES-TO
                         (EPISODE-OF KIDNEY-INFECTION))
          (OCCASIONALLY ((EPISODE-OF KIDNEY-INFECTION) PROGRESSES-TO
                      (EPISODE-OF LOHER-URINARY-TRACT-INFECTION) ).
          ((EPISODE-OF KIDNEY-INFECTION) CAUSES (EPISODE-OF *))
          (TIME-PATTERN
            ((INTERVAL-BETHEEN
              (EPISODE-OF BLOOD-BORNE-INFECTION)
              (EPISODE-OF KIDNEY-INFECTION))
             (BETWEEN (0 DAYS) (5 DAYS)))
            ((INTERVAL-BETHEEN
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(EPISODE-OF KIDNEY-INFECTION)
           (EPISODE-OF LOWER-URINARY-TRACT-INFECTION))
          (BETHEEN (0 DAYS) (3 DAYS)))
         ((INTERVAL-BETHEEN
           (EPISODE-OF KIDNEY-INFECTION)
           (EPISODE-OF *))
          (BETWEEN (0 DAYS) (3 DAYS))))
       (COTEMPORANEOUS (EPISODE-OF BLOOD-BORNE-INFECTION)
                        (EPISODE-OF KIDNEY-INFECTION)
                        (EPISODE-DF *))))
  (COMMON-FEATURES
    SELF-LIMITED-WITH-TREATMENT
    SYMPTOMS-SELF-LIMITED
     (TIME-DURATION (EPISODE-OF *) (BETHEEN (2 DAYS) (14 DAYS)))
     (TYPICAL-TIME-DURATION (EPISODE-DF *) (5 DAYS)))
  (SEQUEL AE
     (OCCASIONALLY (CHRONIC-BACTERIURIA OR
                     WEAKNESS))))
(PROTOTYPE-OF *
     (PATIENT-DESCRIPTION-REVEALS (SEX FEMALE))
     (SYMPTOMATIC-HISTORY-REVEALS
          ((ONSET-OF
             (CONSTELLATION (HIGH FEVER OR SHAKING CHILLS)
                              (ACHING FLANK-PAIN OR SEVERE FLANK-PAIN
                              OR ACHING CVA-PAIN OR SEVERE CVA-PAIN))
                                 (BETHEEN (3 HOURS) (2 DAYS)))
           AND (SYMPTOMS-OF BLADDER-IRRITATION)))
      (PHYSICAL-EXAMINATION-REVEALS CVA-TENDERNESS)
      (LABORATORY-TESTS-REVEAL (PYURIA AND
                                POSITIVE GRAM-NEGATIVE URINE-CULTURE AND
                                HHITE-BLOOD-CELL-CASTS AND
                                 (MILD LEUKOCYTOSIS OR MODERATE
                                 LEUKOCYTOSIS)
                                AND DIFFERENTIAL SHIFTED-TO-LEFT)))
(SUFFICIENT-EVIDENCE-FOR (EPISODE-OF *)
   (FEVER AND PYURIA AND POSITIVE URINE-CULTURE))
(ASSOCIATED-CONSISTENT-FINDINGS-IN *
    (SYMPTOMATIC-HISTORY-REVEALS
        ((USUALLY (MALAISE OR WEAKNESS))
                    AND
         (OCCASIONALLY (ABDOMINAL-PAIN OR
                         BACKACHE OR
                         LUMBAR-PAIN OR
                          (NAUSEA AND YOMITING) OR
```

```
FOUL-SMELLING-URINE OR
                           DIARRHEA DR
                           CONSTIPATION OR
                            GROSS-HEMATURIA DR
                           CLOUDY-URINE))
                      AND
          (RARELY OLIGURIAL)
    (HISTORY-OF-PREVIOUS-ILLNESSES-REVEALS
         (OCCASTONALLY (HISTORY-OF
                           CONE FOR MORE FOCUMPRENCES-OF
                              ((EPISODE-OF URINARY-TRACT-INFECTION)
                               (EPISODE-OF *)))))))
    (PHYSICAL-EXAMINATION-REVEALS
          ( (USUALLY (NORMAL BLODD-PRESSURE AND
                     (NORMAL SKIN-TURGOR OR SLIGHTLY-DECREASED SKIN-TURGOR) AND
                     NO EDEMA))
                       AND
           (OCCASIONALLY ABDOMINAL-TENDERNESS)))
    (LABORATORY-TESTS-REVEAL
          ( (USUALLY (LIGHT-PROTEINURIA AND
                     NORMAL BUN AND
                     NORMAL CREATININE AND NORMAL HENATOCKIT AND
                     NORMAL HEMOGLOBIN AND
                     NEGATIVE BLOOD-CULTURE))
           (OCCASIONALLY (ENLARGED-KIDNEY OR HEMATURIA))))
(ASSOCIATED-DISEASES-OR-STATES-OF *
    (COMPLICATION-OF URINARY-TRACT-OBSTRUCTION)
    (COMPLICATION-OF CHRONIC-RENAL-DISEASE)
    (COMPLICATION-OF PHENACETIN-NEPHRITIS)
    (COMPLICATION-OF PAPILLARY-NECROSIS)
(COMPLICATION-OF PERINEPHRIC-ASSCESS)
(COMPLICATION-OF RENAL-ABSCESS)
    (COMPLICATION-OF RENAL-CALCULI)
    (COMPLICATED-BY BACTEREMIC-SHOCK)
(PREDISPOSITIONS-TO *
```

DIABETES-MELLITUS
PHENACETIN-ABUSE
CHRONIC-BACTERIURIA
POTASSIUM-DEPLETION
URINARY-TRACT-DESTRUCTION

```
PREGNANCY
       SICKLE-CELL-TRAIT)
(* PRESENTS-AS
  ((USUALLY ((SYMPTOMS-OF BLADDER-IRRITATION) AND
           SYSTEMIC-MANIFESTATIONS-OF-INFECTION) JOR
  (OCCASIONALLY (FEVER OR CHILLS) AND HALAISE!) OR
  (OCCASIONALLY (LUMBAR-PAIN OR BACKACHET)))
(TREATMENT-FOR * PARENTERAL-ANTIBIOTIC)
(HEURISTIC-RULES-FOR *
 (IF (CHRONIC-RENAL-DISEASE OR URINARY-TRACT-DBSTRUCTION)
       (USUALLY (HISTORY-OF (ONE-OR-MORE-OCCURRENCES-OF
     ((EPISODE-OF *) OR (EPISODE-OF URINARY-TRACT-INFECTION))))))
 (IF (ACUTE HYPOTENSION AND (FEVER OR CHILLS))
     (CONSIDER BACTEREMIC-SHOCK))
 (IF (HYPERTENSION OR EDEMA)
     (CONSIDER ((RENAL-FAILURE AND CHRONIC-RENAL-DISEASE) OR
                 ACUTE-GLOMERULONEPHRITIS OR
                 CARDIOVASCULAR-DISEASE)))
 (IF ((ABDOMINAL-PAIN OR (NAUSEA AND VOMITING))
                    and
      (NO PYURIA AND NEGATIVE URINE-CULTURE))
     ((RULE-OUT (EPISODE-OF *)) AND (CONSIDER (APPENDICITIS
                     OR CHOELECYSTITIS OR PANCREATITIS))))
 (IF (HIGH BUN OR HIGH CREATININE)
     (CONSIDER (RENAL-FAILURE OR CHRONIC-RENAL-DISEASE)))
(IF (FEVER AND LEUKOCYTOSIS AND (FLANK-PAIN OR CVA-PAIN)
           AND NO PYURIA)
     (CONSIDER RENAL-ABSCESS))
 (IF (FLANK-PAIN RADIATES-TO (UPPER ABDOMEN OR BACK))
     (CONSIDER PERINEPHRIC-ABSCESS))
 (IF SMALL-KIDNEY (CONSIDER CHRONIC-RENAL-DISEASE))
 (IF SCARRED-KIDNEY (CONSIDER CHRONIC-PYELONEPHRITIS))
 (IF (LOW HEMATOCRIT OR LOW HEMOGLOBIN)
     (CONSIDER (RENAL-FAILURE AND CHRONIC-RENAL-DISEASE)))
 (IF ((HIGH BUN OR HIGH CREATININE) AND (DECREASED SKIN-TURGOR))
     (CONSIDER PRE-RENAL-AZOTEMIA))
 (IF ANURIA
     (CONSIDER ACUTE-RENAL-FAILURE OR URINARY-TRACT-OBSTRUCTION))
 (IF OLIGURIA
     (CONSIDER URINARY-TRACT-OBSTRUCTION))
 (IF (URETERAL-PAIN OR (FLANK-PAIN RADIATES-TO GROIN))
```

URINARY-TRACT-ANATOMICAL-ABNORMALITIES

RENAL-CALCUL1

URINARY-TRACT-INSTRUMENTATION

The state of the second of the second second

Appendix 2 - A Set of Descriptors for an IDC

Presented below is a set of descriptors that can be used as a basis for the internal diagnostic configuration of a present illness program. In constructing this set I have attempted to include every consideration that could be inferred from the protocol analysis.

The Patient Model Component

Global Vaciables

The information in this subcomponent is not specific to any one hypothesis or subset of hypotheses, but is instead a collection of descriptors about the general medical status of the patient.

Immediate Treatment Considerations

Need-for-Immediate-Treatment (IMTREAT) - The doctor's primary concern is with the well-being of the patient. This must be reflected in a present illness program. This is the key variable in determining if the choice of focus will be on an emergency situation. The possible values for this variable are UNKNOWN, CONFIRMED, (STRONG, WEAK) -EVIDENCE-(FOR, AGAINST).

Reason-for-Immediate-Treatment (RMTREAT) - The evidence or finding that suggests the need for immediate treatment. This can be a specific disease hypothesis or finding. Examples: GI-Bleeding (FINDING MELENA),

Dehydration (FINDING SKIN-TURGOR-DECREASED), Shock (FINDING SEVERE-HYPOTENSION).

Nature-of-Treatment (NMTREAT) - How the condition should (or could) be treated. Once there is strong evidence for the need for immediate

treatment, a set of possible therapies is made the value of this variable. Example: IV fluids for dehydration.

Caveats-Against-Treatment (CMTREAT) - If there are any caveats such as contra-indications or possible complications in the consideration of a treatment assigned to MTTREAT, they are made the value of this variable. Clearly, the focus of the strategy will be to inquire about these if the need for immediate treatment has been established.

<u>Caveats-Explored (XMTREAT)</u> - A flag to specify if the caveats have been explored.

Prognosis-if-Treatment-Given (PMTREAT) - Future questioning will be affected by the prognosis and expected results of therapy for the condition needing immediate treatment as this is a factor in determining a total management plan for the patient. Values for this variable are POOR, GOOD or UNKNOWN.

Information Sources

Sources-of-information-Available (ISOURCE) - The choice of strategy is influenced by what sources of information about the patient are available. Sources can range from the patient himself, to the patient's friends and relatives, the LMD, a small regional hospital or a large teaching hospital. This variable is a list of all known sources of information that are available for this particular patient. If a doctor needs an expert opinion about the patient's past medical history he might ask if she has ever been hospitalized.

Credability-of-Information-Sources (CRED) - Associated with each source of

information is a measure of its credability. This can be known a priori by the doctor or it can be computed based on the data that is reported from the source. If clearly contradictory data is reported, the source's credability may be in doubt. In one protocol, after the doctor found out that the source of the information he wanted was a certain hospital with a questionable reputation, he tended to discount much of the hospital record reported.

Prefered-Information-Sources (PSOURCE) - A list of the information sources in order of preference. Expert witnesses such as recognized specialists or consultants will have a higher preference than LMDs. This list can not be computed directly from the credabilities, as some them might not be known. Thus, this list can consist of an ordered list of unordered sublists.

Findings Subcomponent

The findings subcomponent consists of information about the features of the findings that have been reported. These features are independent of interpretation with respect to a disease hypothesis; the primary concern is with the findings as objects of consideration by themselves.

Anu-Finding-Life-Threatening (FLIFE) - This flag is turned-on if a reported finding is potentially life-threatening. (The variable RMTREAT can be set to this variable.) A condition such as a high serum-cholesterol level is life-threatening over a long period of time while bleeding is immediately life-threatening. For this reason the variable values are NOW, SHORT-TERM and LONG-TERM.

Differential-Net-for-Any-Reported-Finding (FNET) - Many symptoms are

associated with a differential discrimination net based on their dimensions of characterization. The nodes in such a net point to different disease hypotheses. A common discrimination strategy used by doctors is to run down one of these nets for a particular finding. For example, symmetric periorbial edema is suggestive of nephrotic syndrome while asymmetric leg edema is suggestive of cellulitis. This flag specifies if any of the reported findings has such an associated net.

Majority-System-Association-of-Findings (MSYS) - Most findings can be associated with a particular organ system of the body. For example, nausea, vomiting, melena and diarrhea are associated with the GI system, while dyspnea and rales are associated with the respiratory system. Some findings, such as weakness and fever, have no such association. This flag indicates if the majority of the findings are specific to any particular organ system. It can play a role in the selection of an exploration strategy in the absence of any LIKELY hypotheses.

Specificity-of-Two-or-More-Findings-Identical (SPECS) - Certain findings are almost always associated with certain diseases or disease classes. For example, equeezing chest pain is elmost always associated with heart disease. This association will, of course, be refelected in the hypotheses structure. This flag specifies if two or more findings have the identical association.

Individual Finding Descriptors

Finding-Classification (FCLASIF) - Findings are classified as symptoms, historical events, physical-exam or faboratory data.

Source-of-Finding (FSOURCE) - Patient, LMD, hospital record, etc.

Finding-Credability (FCRED)

Seriousness-of-Finding (FSERI)

Organ-System-Association-of-Finding (FSYST)

Differential-Net-for-Finding (FDNET)

Specific-Disease-Associations (FSPEC)

The Hupotheses Sub-component

Hupothesis List Descriptors

The first set of descriptors characterizes the hypothesis list in terms of density, specificity and compactness. The underlying nosology is a hierarchical KIND-tree such as used by Pople Pople 75>.

Measures of Hupothesis List Densitu

Total-Active-Hupotheses (TOTHYP), Total-Likely-Hupotheses (TOTLIK), Total-Possible-Hupotheses (TOTPOS), Total-Unlikely-Hupotheses (TOTUNL) - The density of the hypothesis list (the number of hypotheses in active consideration) plays an important role in strategy selection. A low density implies the use of confirmation strategies, a high density, elimination and a zero density, exploration.

Measures of Hupothesis List Specificity

Highest-Classification-Level-of-(LIKELY, POSSIBLE)-Hupotheses (HCLSL, HCLSP), Louest-Classification-Level-of-(LIKELY, POSSIBLE)-Hupotheses

(LCLSL, LCLSP) - The specificity of a hupothesis is the number of separate diseases to which it can refer. For example, a hupothesis of regional enteritis can refer to Chron's disease or regional ileitis, in a KIND-

tree, the <u>classification level</u> is the height (number of levels) of the node from the fringe of the tree. This measure tends to indicate the number of different discriminations that would have to be made to arrive at a more specific disease diagnosis. The spread between the highest and lowest levels is a factor in deciding between discrimination and elimination strategies. For example, if the spread is large as in a hypothesis list containing heart disease, liver disease and acute glomerulonephritis, an elimination strategy aimed at heart or liver disease might be appropriate. Whereas, if the spread is small such as with renal disease, heart disease and liver disease, discrimination might be indicated.

Number-of-Nodes-Covered-by-(LIKELY, POSSIBLE)-Hypotheses (LNODES, PNODES) —
The total number of nodes covered by the hypotheses can be used in deciding between discrimination and elimination. A small covering fits better with discrimination strategies, while a large covering is better suited to elimination. The covering is an indicator of the total range of diagnostic options that must ultimately be considered. In selecting a hypothesis to eliminate, the larger the covering the better the hypothesis is as a choice to cut.

Measures of Hypothesis List Compactness

Classification-Level-of-First-Common-Ancestor-Node-for-ILIKELY, POSSIBLE) - Hypotheses (CLANL, CLANP) - The compactness of the hypotheses list refers to the range of different areas covered by the hypotheses. This is reflected by how high up the disease classification tree one must go to find a common ancestor. Elimination strategies are suggested by high

values and discrimination strategies by low-values.

Hupotheses Structure Descriptors

Possibilities-List (*PLIS*) - This variable specifies the active hypotheses: they are ordered by absolute score and classified by relative score:

((CONFIRMED-List) (SATISFIED-List) (LIKELY-List) (POSSIBLE-List) (UNLIKELY-List) (RULED-OUT-List))

The first entry on the LIKELY-List is called the Principal Disease
Hypothesis (PDH).

Hupotheses-Structure-Graph (#HYGRAPH#) - A graph of the relational structure of the hypothesis list. Links include CAUSE, COMPLICATION, and PRINCIPAL-PART.

LIKELY-or-POSSIBLE-Hypothesis-COMPLICATION-OF-or-CAUSED-BY-SATISFIED-or-CONFIRMED-Hypothesis (LPCSC) - Certain patterns of the hypotheses structure graph are preclassified because of their relative importance in strategy selection. In this particular pattern, one hypothesis is a consequence of another hypothesis that is believed true. The effect on strategy selection might be to force a confirmation strategy aimed at the cause or complication that will seek explanations for inconsistent data (if any) or attempt to match uncommon scenarios (if necessary) in order to satisfy the confirmation goal.

SATISFIED-or-CONFIRMED-Hypothesis-COMPLICATION-OF-or-CAUSED-BY-LIKELY-or-POSSIBLE-Hypothesis (SCCLP) - This is the reverse of the previous

situation. Urinary tract infection is a hypothesis that is easily confirmed. It is, however, very commonly a complication of another clinical condition such as obstruction. A doctor might hypothesize the generating condition even if there is no direct evidence for it as yet. He might then try to confirm or eliminate this new hypothesis. The effect on strategy selection might be to force the focus to be shifted to this new hypothesis regardless of its order on the *PLIS*. (See Question 11 in Chapter 2.)

Combined-Hypothesis (COMB) - A combined hypothesis is two or more <u>unrelated</u> diseases hypothesized together as a diagnosis. The effect on strategy selection might be to favor exploration strategies in order to develop alternative hypotheses (If it is the only LIKELY or POSSIBLE hypothesis).

Individual Hupothesis Descriptors

System-Classification (SYSTEM) - The systems considered are GI,
Cardiovascular, GU, Respiratory, Neurological, Hepatic, Hematopoietic,
Endocrine, and Bone-and-Joint. Any specific subsystem (such as the thyroid of the Endocrine system) is also noted. If multiple organ systems are involved the value would be MULTIPLE followed by a list of the systems.

For example, for Wilson's disease the value would be (MULTIPLE (NEUROLOGICAL BRAIN) (HEPATIC LIVER)).

Disease-Clinical-or-Physiological-State (DSCS) - Each hypothesis is tagged with its basic clinical classification. These are disease, clinical-state or physiological-state. Chronic renal failure is a clinical-state while sodium retention is a physiological-state.

Acute-Staged-Chronic (ACP) - A hypothesis is classified as being either acute (single-phase), acute-staged (multiple-phase) or chronic. Examples: acute - APN, acute-staged - AGN, chronic - CPN.

Episodic-Non-Episodic (EPSID) - This variable specifies if the hypothesis is episodic or not. Focal GN and malaria are episodic diseases.

Single/Multiple-Etiology (NETIOL) - Classifies the hypothesis as either single or multiple etiology. Examples: Single-Etiology - Rubella, Multiple-Etiology - Acute pancreatitis.

<u>Treatable-Not-Treatable (TREAT)</u> - If there is a known treatment for the hypothesized disease.

Etiology-Implicated-In-Treatment (ETREAT) - If the etiology is a factor in determining the nature of the treatment for multiple-etiology conditions.

<u>Differential-Relatives-on-Hypothesis-List (DIFFR)</u> - Indicates if any diseases that can be eliminated through differential diagnosis of a key symptom (already reported) are present on the hypothesis list.

Absolute-Score (ABSCORE) - A score reflecting the evaluation of the hypothesis frame. Normally, a weight-of-evidence measure.

Relative-Score (RELSCORE) - A score used to classify the hypothesis as CONFIRMED, LIKELY, etc.

Hupothesis-Summary (*HYSUM*) - The state of a hypothesis with respect to the reported findings. The summary consists of two parts, the scenario summary (for a chronic disease) and the prototype summary (for all acute and most chronic diseases). For acute-staged diseases, the prototype summary would be a set of prototype summaries. The structure of this

variable is the following:

```
(HYPOTHESIS-SUMMARY

(SCENARIO-SUMMARY

(PREREQUISITES-VERIFIED)

(EVENTS-REPORTED-IN-AGREEMENT)

(EVENTS-NEEDED-BUT-NOT-YET-KNOWN)

(EVENTS-KNOWN-BUT-NOT-YET-MEEDED)

(EVENTS-TESTED-BUT-REPORTED-UNKNOWN))

(PROTOTYPE-SUMMARY

(FINDINGS-REPORTED-IN-AGREEMENT)

(FINDINGS-REPORTED-IN-DISAGREEMENT)

(FINDINGS-NEEDED-BUT-NOT-YET-KNOWN)

(FINDINGS-KNOWN-BUT-NOT-NEEDED)

(FINDINGS-TESTED-BUT-REPORTED-UNKNOWN))))
```

If a reported finding (event) is not part of the prototype (scenario) but is considered consistent with the hypothesis it is classified as known but not needed. It it is inconsistent with the hypothesis it is classified as known but in disagreement.

<u>Direct-Confirmation (DIRECT)</u> - If there is a finding that can be used to directly confirm the hypothesis (prima facile evidence). For example, a positive unine culture directly confirms a uninary tract infection.

<u>Direct-Elimination (ELIMD)</u> - If there is a finding that can directly eliminate (i.e. rule-out) a hypothesis (necessary evidence).

ELIMD-DIRECT (CUTCON) - If the same finding can be used to both directly confirm or directly eliminate the hypothesis.

Special-Strategy (SPSTRAT) - If there is a special strategy associated with the hypothesis.

The Current Status Component

The current status component is a description of how the diagnoser

views himself in carrying out a specific present illness. This component plays the role of defining what is normally considered the state of a process.

Process State Descriptors

<u>Current-Present-Illness-Phase (PHASE)</u> - The section of the present illness currently being performed. The sections are:

- 1. Symptom discovery and characterization.
- 2. Past medical history.
- 3. Social and family history (optional).
- 4. Physical examination.
- 5. Standard laboratory tests.
- 6. Complex diagnostic procedures.

Review-of-Systems-Flag (RSYSF), Review-of-Systems-Pointer (RPOINT) - If a review of systems is currently being conducted (RSYSF). The section of the review corresponding to the current present Illness phase (RPOINT).

Strategy Bescriptors

Strategy-Frame-for-Current-Question (SFQ) - The instantiated prototype of the strategy frame from which the current question has been derived.

Current-Goal-Tree (*GOALTREE*) - A specification of the goal-tree resulting from the binding of the strategy components of the strategy frames. There are two varieties of root nodes: entry and continuation nodes. An entry node is the top node of the subtree generated by a strategy frame. The expansion of an entry node includes the name of the strategy frame, the top-level structural and bound goal and a flag specifying the AND/OR structure of its immediate descendants. A continuation node is a root node that is not an entry node. The expansion of a continuation node includes

the structural and bound goal and an AND/OR flag.

The expansion of a leaf node includes the structural goal, the bound goal, the method and the question. For example:

```
(L1 ((STRUCTURAL-GOAL
(CONFIRM (DECREASED-FUNCTION ORGAN-SYSTEM)))
(BOUND-GOAL
(CONFIRM (DECREASED-FUNCTION KIDNEY)))
(METHOD
(IF-THEN (CREATININE (> 1.2))
(DECREASED-FUNCTION KIDNEY)))
(QUESTION
(CREATININE VALUE?)))
```

Also included in the expansion of a leaf node is the expected answer (if any). This is to decide if the goal has been satisfied. Goals are marked as satisfied, not satisfied or partially satisfied.

Future-Question-List (*QUESL*) - The questions of the leaf nodes are arranged in a list corresponding to the phases of the present illness. Within each phase the questions are ordered by the left-to-right sequence of the original goal-tree. If the goal tree undergoes transformations, this variable is correspondingly updated.

Phase-Transition-On-Next-Question (PHTRAN) - If the next question to be asked represents a phase transition. Under certain circumstances this can cause the current strategy to be recomputed due to the desirability of not having to return to a previous phase.

Predicates on IDC Variables

To facilitate the matching of terminals of the strategy frames to the variables of the IDC a collection of predicates (in addition to the normal

logical operators) is required.

Hupothesis List Predicates

(LINKS <into out-of> Node-Set Num) - The value of this predicate is TRUE if the number of links into (out-of) the collection of nodes specified by Node-Set is equal to Num.

(LINK-TYPE Node-Set Type <into out-of>) - TRUE if any of the links into (out-of) the Node-Set are of the type specified.

(M*PLIS* List) - The general hypothesis-list matching predicate. List specifies a sample hypothesis list to be matched against the current *PLIS*. Example:

(M*PLIS*
((LIKELY NONE)
(POSSIBLE (< 2))))

This will match any hypothesis list with no LIKELY hypotheses and two or less POSSIBLE hypotheses.

(M*HYGRAPH* Graph) - A predicate for testing the hypotheses structure graph. For example:

(MxHYGRAPH*

(AND (COMPLICATION-OF (1 POSSIBLE) PDH)

(CAUSE-OF 1 (2 POSSIBLE))))

uill test if any POSSIBLE hypothesis is both a complication of the PDH and a cause of some other POSSIBLE hypothesis.

Goal-Tree Predicates

(NUMBER-OF <entry continuation leaf> <above below> Anchor Num) - This predicate is TRUE if the number of the type of node specified above or

below the Anchor node is equal to Num.

(M*GOALTREE* Tree) - The general goal-tree matching predicate. Tree is the pattern to be matched against *GOALTREE*.

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