MODELLING THE DIAGNOSTIC PROCESS

by

G. Anthony Gorry

No. 370-69

February 20, 1969

The author is indebted to Dr. G. Octo Barnett of the Massachusetts General Hospital for his advice and comments.

I. Introduction

In this paper, the process of diagnosis will be considered as the problem-solving activity directed toward the classification of a patient for the purpose of relating experience with past patients to him and assessing the therapeutic and prognostic implications of his condition. Because the ability to solve diagnostic problems is crucial to the delivery of appropriate medical care, the development of the requisite skills in young physicians is particularly important. The present process of medical education does not provide the student with a logical structure within which he can develop his abilities. Generally the student learns about diagnosis as a derivative of his learning of disease patterns. The approach is sometimes ill-structured and erratic. This situation arises out of the absence of a formalism for diagnosis in medical education. That such a formalism does not exist may result from a belief that the diagnostic processes are so highly individualistic and obscure as to elude precise specification.

Certainly it is true that when one considers the process of diagnosis as performed by physicians, one is faced with important complexities. A given doctor may employ highly individualistic methods in his approach to a diagnostic problem. Secondly, even the physician himself may have difficulty in isolating the fundamental determinants of his diagnosis. In some cases, he cannot verbalize his methods. Thus it has been observed:

> The clever diagnostician remembers many little relationships because of some common denominator which may be rather obscure on superficial examination of the actual logic used at the bedside. This is the experience of which physicians are proud. This is what appears to be artistic ability in the practice of medicine and why it may seem to many physicians that they are gifted in making correct decisions without definable data and why they profess that rare diagnoses can be correctly made when no statistician would dare stick his neck out on the basis of the data presented [1].

In considering ways to improve the development of diagnostic skill in medical students, it seems natural to question this view. Is diagnosis an entirely individualistic process, and if not, to what extent can general strategies be isolated and described? If such general strategies can be outlined, the educational experience of the medical student would be improved, because diagnosis could be taught directly. The common indirect method of teaching by example places a heavy burden on the student, because he is forced to infer the elements of diagnosis employed by his teachers. The pivotal role played by diagnosis in the delivery of appropriate medical care makes even marginal improvements in its teaching very valuable. Thus there is considerable motivation for the investigation of the diagnostic process in an attempt to describe strategies and procedures which can be taught explicitly.

The focus of this investigation should be the formulation of a model of diagnosis. The formal definition of what constitutes a model of either the behavior or structure of a given system is not an easy task. Informal

definitions are more readily obtained. Here the choice is ease of exposition over rigor. An attempt will be made to outline in general terms those properties which a model of the diagnostic process should have. The potential value and limitations of such models also will be discussed. Finally, a specific model of diagnosis will be reviewed. The key issue, however, is the usefulness of modelling activity in improving the teaching of diagnosis rather than a consideration of the details of any particular model.

II. Modelling and Systems

To the extent that two systems resemble one another from some point of view, one serves as a model of the other. As there is an enormous diversity of systems, so there is an equal diversity of models. Some models represent systems pictorially or visually while others employ one set of properties to represent another (as the flow of water through a pipe can be taken as a model of the "flow" of electricity in a wire) [18]. The use of systems of equations and mathematical relations results in a third kind of model, an abstract model of a system. It is this latter type of model which will be of principal interest here.

The motivation for a model may come from several sources. A model may isolate and illuminate certain relationships or properties of the modelled system and hence promote an improved understanding of that system. Also, manipulation of the model may be easier than experimentation with the modelled system. Either or both can be a reason for constructing a model of a system.

The concept of a model is intimately related to that of a system. Generally, the two aspects of a system with which modelling is concerned are its structure and its behavior. The structure of a system is the totality of the interrelationships among its elements. The behavior of a system is composed of the interactions between the system and its environment. In various contexts, we may wish to model either the structure of a system, the behavior of a system or both. The focus of this paper is on abstract models of the behavior of the "diagnostic system". More general discussions of system modelling are available in a variety of references (e.g., 2, 3, 4, 5, 18).

As suggested above, the extent to which one system (in this case an abstract system) can serve as a model of another system (the cognitive system of diagnosis) depends on the use to which the model is to be put. In order to make this point more specific, we want to consider the modelling of the behavior of a system in more detail.

In Figure 1, schematics of a system and model of the system are presented. Because we are interested in the behavior of the system, no details of system structure are shown. Hence for the system in question, we have the "black box" representation with inputs from and outputs to the environment. The same representation for the modelling system is used with the important addition that there is an intermediate stage between the inputs and the modelling system and one between the modelling system and the output. Formally these intermediate stages represent transformations, and they are required because the inputs and outputs of the modelling systems are representations of the inputs and outputs of the modelled system. For example, an abstract model of response to visual stimuli might employ as inputs mathematical representations of the light patterns sensed by the eye.

The test of the validity of the model is the determination of whether for each set of relevant inputs to the modelled system, the representation of these inputs produce outputs of the modelling system which are representations of the outputs of the modelled system. Notice that implicit in this notion of the validity of a model is a level of resolution. A system may be a valid model of another system when the inputs and outputs are viewed at one level of resolution, but not when viewed at a greater level of

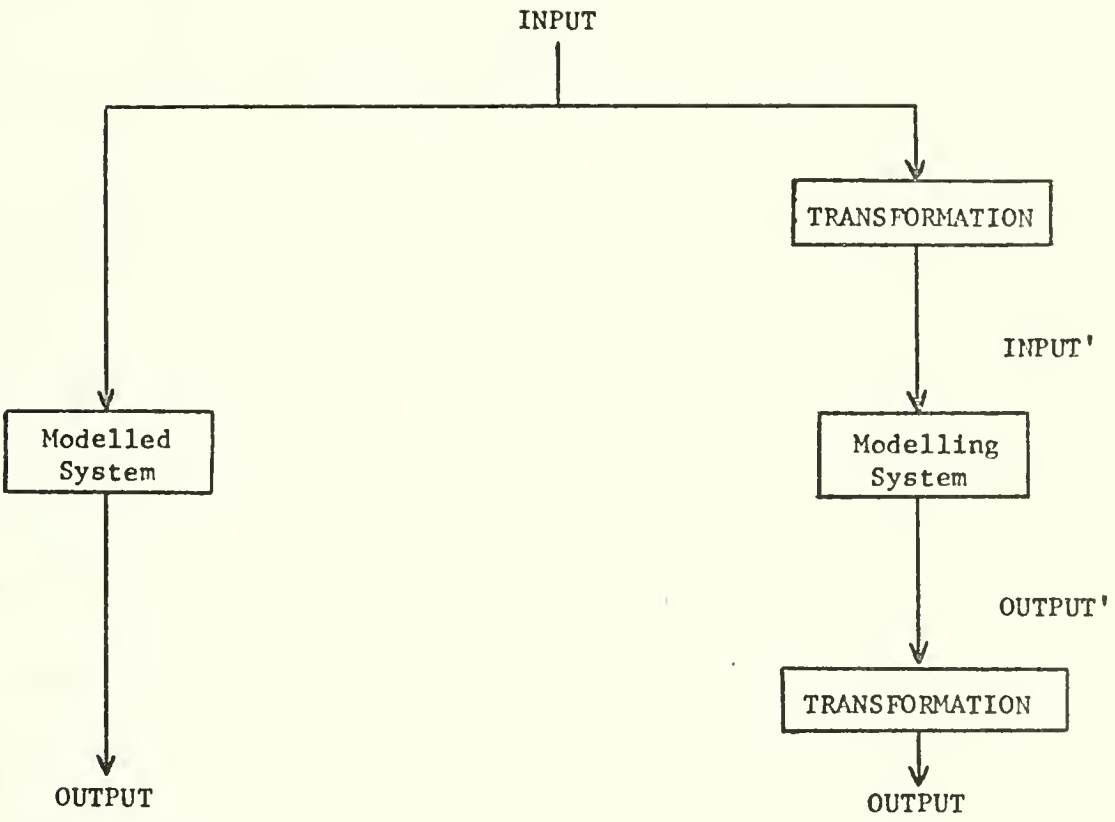


Figure 1: System and Model

resolution. Thus a model of a lung might be valid for the gross transport phenomena, but be quite inappropriate at the microscopic level. Because the level of resolution is chosen most often to suit the purposes for which a model is required, the validity of using one system as a model of another is dependent on the use to which the model is to be put.

A system can be a valid model of the behavior of another system, but be entirely inappropriate as a model of structure. For example, a set of equations may be a very good model of the input-output characteristics of a piece of electrical equipment without revealing anything of themselves about the structure of the device. Similarly, an analogue model of a human heart might be valid at a macroscopic level of resolution without accounting for microscopic structure or behavior.

Considerations such as these are important for several reasons. First, although use has been made in medicine of mathematical models of physiological systems such as the heart, the use of such models in studying diagnostic problem-solving has been virtually nonexistent. Part of the difficulty here may be that the tradition of formal modelling is not strong in the general medical community. This difficulty is compounded by the failure to appreciate the relevance of the resolution level to the modelling activity. Many medical people seem to feel that a model of diagnosis must be complete, and hence so complex as to be infeasible. A second difficulty is that the potential advantages of modelling as an activity (in addition to the value of the resultant model) are not generally recognized. The extensive success of modelling in other areas of science does not appear to have had significant impact on the main body of the medical community.

Because we are concerned with the use of abstract models of the behavior of doctors in solving diagnostic problems, we want to indicate some of the potential benefits of such formal descriptions of diagnostic information processing.

First the formulation of an abstract model (for example, a mathematical model of diagnosis) requires many of the assumptions about the process be made explicit.[3] When the sources of ideas for the model are observation and introspection, the formal nature of the model requires that these activities be undertaken with particular care. In the process of refining the ideas for the model in order to make them sufficiently precise, new insights into the nature of the information processing function may be gained.

One of the difficulties in constructing a mathematical model is that it can become very complex. This potential complexity, however, can serve in a positive way as an important constraint on the modelling activity. In order to keep the model tractable, we may be forced to dispense with much detail. In some cases, the reduction in detail enhances the value of the model by revealing general properties which are otherwise obscured. Caution must be taken, however, to avoid oversimplifications which lead to generalizations which are not correct. If the modelled system is in fact very complex, it may be necessary to elaborate a simple first model to account for more detail. Finding the appropriate level of complexity in the model can require considerable work and skill.

---

[3]Of course, implicit assumptions will remain, and some can be quite subtle. The degree to which assumptions are made explicit, however, is generally high.

Formal models are more easily tested than informal (e.g., verbal) ones. Hence their inadequacies are more apt to be exposed, prompting a re-design. The relative lack of ambiguity in a formal model may encourage people other than the designers to test it. Their involvement in this activity may generate new ideas which further improve the model. Finally, the structured model is more easily taught. An unambiguous model, stated formally, can be studied and analyzed by a student, and it will provide a good framework within which he can organize his understanding of the modelled system.

For these reasons, then, a formal model of diagnosis is highly desirable for use in teaching medical students. The major question remaining is whether such a model can be formulated. The answer to this question can be framed only within the context in which a model of diagnosis is viewed. For example, if we required that the model explain all aspects of diagnosis, encompassing every subtlety of the process, then there is little hope of success. Fortunately we need not place such stringent requirements on the model, particularly in view of its intended use in teaching.

The medical student is in real need of basic strategies and principles of diagnosis. A model of diagnosis would have considerable value if it were able to elucidate these matters -- even though it does little to account for idiosyncratic aspects of the process. Again, the level of resolution is crucial in assessing validity. Because the emphasis in teaching the student should be on general procedures, the level of resolution in the model should be such as to eliminate much detail. The important point is that the model is not intended as a final statement on diagnosis to be learned by the student, but rather as a conceptual framework within which he can organize and enlarge his understanding of the actual process.

This latter goal seems much more attainable. Formal models of cognitive information processing activities in other areas [6, 7, 8] have proved quite successful in stimulating interest in and study of these activities. Work done on formalizing diagnostic procedures [9, 10, 11] has shown sufficient promise to merit expanded effort. In the remainder of this paper, we will outline a view of diagnosis which lends itself to formalization. In fact, it serves as the basis for an interesting, interactive computer program for diagnosis [10]. This discussion, however, will emphasize only the general features of diagnosis from this point of view. There undoubtedly exist other formalizations of this view which are valid models of diagnosis and differ significantly from that discussed in the references cited above. The principal purpose here is not to argue for one formalization, but instead to encourage new contributions to the area.

III. Considerations for a Model of Diagnosis

Consider an admittedly simplified description of a diagnostic problem for a clinician. In general, the clinician may draw on two sources of information in developing a diagnosis: the presenting signs and symptoms of the patient, and his medical knowledge of the relevant diseases. He must then use this information in arriving at certain decisions: what additional information about the condition of the patient should be collected; what disease states are most probable; what are the potential consequences of the particular clinical situation. A source of complexity in diagnosis is that these decisions are not independent, and the physician must consider their interrelationships.

The physician maintains his medical knowledge largely in terms of abstractions. That is, his concept of a particular disease is really a generalization of his past experience with patients he has read about or seen himself. As noted by Lusted, these generalizations are:

> ...representations of more or less loose association of similar patients about which some statements may be made regarding common mechanisms or causes of illness or regarding therapeutic choice, response, and prognosis [12].

The diagnostic process employed by a physician, then, can be viewed as an attempt to establish the similarity of the presenting signs and symptoms of the patient to a particular disease prototype. The criterion of similarity employed is a fundamental factor in diagnosis.

In general, the clinician does not have sufficient information about the patient initially to assess the similarity of the patient's sign and symptom pattern to various disease prototypes with sufficient certainty to make a diagnosis. Hence an important aspect of diagnosis is the selection of certain questions, tests, etc., which yield additional information about the disease state of the patient.

Therefore it appears that there are three major components of the cognitive system for diagnosis employed by a physician. The first is his general medical experience. The second is the process whereby he assesses the similarity of a partial pattern of signs and symptoms to those of relevant disease prototypes. We will call this process the <u>inference function</u> of diagnosis. The third component is the process by which the

clinician selects potentially useful questions, laboratory procedures, etc., to obtain more information on which to base a diagnosis. For convenience, we will use the term test to denote any such question, laboratory procedure, etc., which can yield information relevant to the diagnosis, and will call the process involved the test selection function of diagnosis.

In order to construct a model of this view of diagnosis, then, we must describe each of the three components: medical experience, the inference function, and the test selection function, as well as the manner in which they interact. As noted above, modelling of this type requires a clear view of the use to which the model is to be put and the level of resolution required. Because we have limited requirements on the explanatory power of a diagnostic model, we can work at a relatively low level of resolution. Also we will be concerned with a model of the behavior of the diagnostic system employed by physicians. No attempt will be made to model its structure. We will be concerned with descriptions of the information processing aspects of the components mentioned above, ignoring the questions of the structure and operation of the brain.

In Figure 2, a schematic representation of a diagnostic model is presented. The three major components of the system are shown as elements of the model. To make the model useful, we must describe the behavior of each of these elements. Before attempting to formulate these descriptions, we want to point out an important aspect of our view of diagnosis implicit in the figure. This is the sequential nature of the problem-solving activity in question.

In considering a particular test, the physician should weigh the expected value of possible test results against the expected cost of the test. Because tests can be costly (in terms of patient discomfort, time of skilled persons, money, etc.) the number of diagnostic tests should be kept to a minimum. On the other hand, the physician seeks to minimize the consequences of possible misdiagnoses. In general, the probability of such an error is reduced as more information about the patient is obtained through the use of diagnostic tests. Hence the physician may wish to perform many tests to reduce his uncertainty about the condition of the patient. Because these are contradictory objectives, he must strike a balance between the two. Because he can re-evaluate the various test alternatives after each test result is obtained, he can solve this problem in a sequential manner. This is the reason for the relation between the inference and test selection functions shown in Figure 2. Using his current view of the problem in conjunction with his medical experience, he can sequentially develop a testing strategy which he expects to yield significant information. The sequential mode of solving this problem permits him to modify the strategy in the light of new (and perhaps unexpected) test results.

Now we turn to a consideration of the three elements of the model of diagnosis. Descriptions of these elements are provided in detail elsewhere [11, 13], but they reflect only one point of view. The emphasis here will be only general considerations which it is believed are relevant to any formalization of the behavior of these elements.

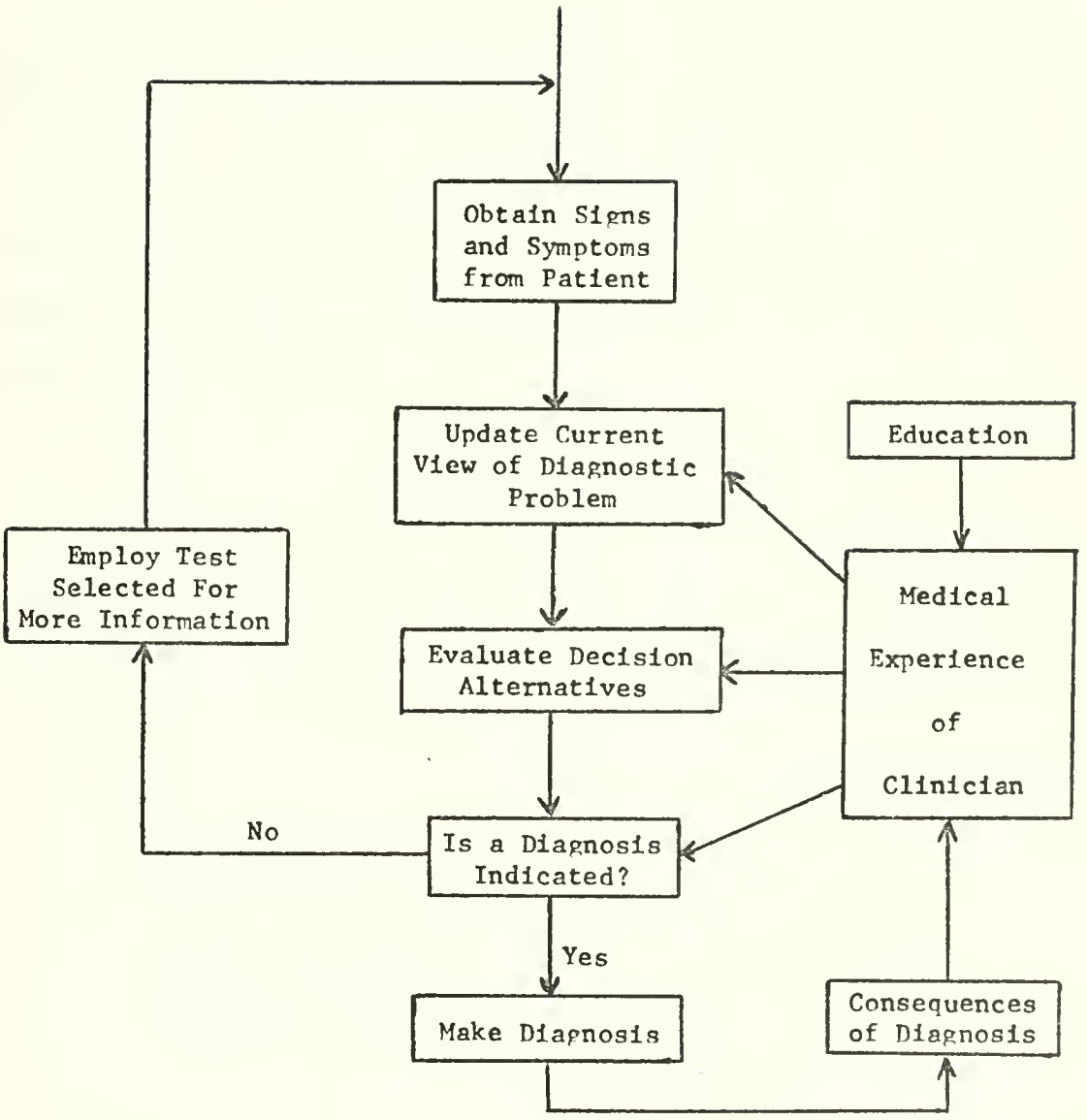


Figure 2: Schematic Model of Diagnostic Problem-Solving

In considering medical experience, we will not be concerned with the manner in which this experience is obtained by the physician, but rather with the way in which it is used in the inference and test selection functions. The schematic of the model, however, does indicate the two principal sources of medical experience, education (such as reviewing medical literature or attending medical school) and the evaluation of experience with patients.

The way in which the medical experience of the physician is modelled depends on the use to which that experience is to be put in the inference and test selection functions. Basically, however, we can begin with some observations on the physician's conception of disease.

It has been argued that:

> Physicians think of diseases in a dynamic context as a sequence of changes occuring in certain organs, and they remember the signs and symptoms because of their understanding of the mechanisms which bring about these indices of the hidden disease process. They do not usually memorize long lists of signs or symptoms any more than we remember stories by memorizing a list of nouns, verbs, adverbs, etc. [1].

If we attempt to base our model of medical experience on this view, we will have serious difficulty in formalizing the process. This is not because the view is incorrect, but rather because it implies thought processes of which we understand very little. If we are willing to treat the activities involved in structuring and accessing medical experience as subsumed in an element of our model, however, the problem is much less severe. Thus we assume that the element associated with medical experience works in a manner

(currently unknown to us) such that it can produce a pattern of signs and symptoms corresponding to a given disease whenever this pattern is required by either the inference or the test selection functions. No attempt is made to say how this element is organized so as to meet this demand. We will, however, suggest some of the properties of the sign and symptom patterns produced.

These patterns represent abstractions of disease manifestations. They are generalizations based on experience, and it is not expected that all (or even most) of the patients with a given disease will present signs and symptoms which exactly match the pattern. One way to represent this variability is through the use of probabilities. Thus in addition to the prototypical pattern for a given disease, the element of medical experience can assess how likely a specific sign-symptom pattern for a patient with the disease is to deviate from the prototype. When we consider the inference function, we will see further need for this probabilistic association between signs and symptoms and diseases in the medical experience.

In addition to these associations, the relative costs of various diagnostic tests and those of possible misdiagnosis should be accessible from medical experience. These costs are crucial in the determination of a good diagnostic strategy. Again these are important inputs to the other elements of the model.

The problem to which the inference function is addressed is the interpretation of evidence from a real patient in terms of the abstractions of disease produced by medical experience. The problem of attaching a

diagnostic label to a patient is equivalent to establishing the similarity of his presenting signs and symptoms to these prototypical patterns [14]. Because it is clear that few patients will exactly match a specific pattern developed by medical experience, it seems appropriate to base the measurement of similarity on probabilistic considerations. This has been done successfully in some studies of computer-aided diagnosis [9, 10, 11].

Specifically, we suggest the use of Bayes rule as the basis for the inference function. Such a use of Bayes rule is discussed in detail elsewhere [11, 13]. Basically its use is in obtaining conditional probability assessments for disease given a partial pattern of signs and symptoms from probabilistic representations of prototype patterns and the likelihoods of the various diseases in question.

It is precisely this ability to rationally adjust prior opinions in keeping with current evidence which needs to be developed in new doctors. When one observes them attempting to develop a diagnosis, one is struck by the extent to which their ability to reconstruct sign-symptom patterns for disease entities outstrips their ability to discriminate the manifestations of one disease from those of another. Part of this is because diagnosis can be a difficult intellectual exercise regardless of the approach employed. The larger part, however, seems attributable to a lack of an orderly procedure for establishing the similarity of the pattern of presenting signs and symptoms of the patient to those patterns associated with particular diseases. The development of these skills might well be accelerated through the use of Bayes rule in teaching.

The use of Bayes rule as a model of the inference function of diagnosis exposes the inherently probabilistic nature of the process. Indeed, one can argue that the essence of diagnostic inference is the development of subjective probabilities of the diseases in question from the evidence at hand and previous medical experience; and hence Bayes rule is a natural vehicle to use in training students.

The problem to be solved by the test selection function of diagnosis is the following. Given a current view[4] of the problem developed from partial evidence about the patient and past medical experience, decide whether to make a final diagnosis (and perhaps initiate treatment), or to seek additional information. In the event the latter alternative is chosen, determine the means by which this information is to be sought. A number of factors weigh upon this decision, but basically they can be collected as follows. If the physician chooses to make a diagnosis, he assumes the risk will be determined by the probabilities of various misdiagnoses and the seriousness of the errors in question. On the average, the information he would obtain from further testing will reduce the expected risk of misdiagnosis. The cost of the test employed, however, in terms of patient discomfort, money, etc., must be considered. Only if the expected reduction in the risk of misdiagnosis provided by the test results justifies the cost of the test can the use of the test be part of a good diagnostic strategy.

---

[4]If the inference function is modelled using Bayes rule, the current view can be identified with the latest assessment of the probabilities of the diseases in question.

Here the interaction between the inference function and the test selection function is apparent. The assessment of the risk of misdiagnosis depends in large part on the probabilities of the diseases in question as developed by the inference function. The inference function is affected in turn by the tests employed. Good tests will improve the quality of inference.

Again there are many ways to formalize a description of this activity. The important point is that some attempt to make the procedures explicit should be made, even at the expense of detail. This holds for each of the other two elements of the model as well. The considerations suggested here for a model of diagnosis are not exhaustive. Certainly attempts to formalize such a model will yield new insights into its requirements. It is believed, however, that certain of the considerations raised here will be basic to any good model. These are: 1) the sequential nature of the problem-solving activity; 2) the probabilistic basis for inference; and, 3) the comparison of the expected cost of a test with the expected reduction in the risk of misdiagnosis resulting from the use of the test.

IV. A Model of Diagnosis

A formal model of diagnosis has been developed and serves as a basis for an interesting interactive computer program. Both the program and its performance have been discussed elsewhere [9, 10, 11, 13]. Here the basic components of the underlying model will be reviewed. The schematic in Figure 2 will be used as the basis for the discussion. One part of that

schematic is not represented in the model, namely the process by which the physician perceives diagnostic clues. This, of course, is an extremely important aspect of diagnostic problem-solving. It is with good reason that so much effort is spent in medical schools training students in the techniques and skills of information gathering. Hopefully the model has didactic value in spite of this incompleteness.

Through training and experience, a physician develops his understanding of disease mechanisms. He learns of sign and symptom patterns as manifestations of various disease processes. When faced with a diagnostic problem, however, he must solve the problem of associating sign and symptom patterns with disease mechanisms. The process by which he constructs this latter association constitutes the inference function of diagnosis. Simply stated, the need for inference arises because he knows sign and symptom patterns given a particular disease, but the problem is to assess the likelihood of the disease given one such pattern.

The inference function employed in the diagnostic program mentioned above is based on Bayes rule. The assumption is that medical experience is used to estimate the a priori probabilities of the diseases in question and the conditional probabilities of observed signs or symptoms given these

diseases. Note that the latter probabilities are subjective estimates of the likelihood of signs or symptoms in the event that a certain disease mechanism is present. In view of his training, the physician's conception of disease lends itself to this type of association.

The Inference Function and Bayes Rule

Bayes rule is employed as follows: Given a set of diagnostic possibilities $D = \{D_1, D_2, \ldots D_n\}$ with a priori likelihoods $P(D_i/E)$ "the probability of disease i, given experience to date", where E denotes experience to date, assume that sign (or symptom) S_j is observed. The new assessment of the likelihoods, $P(D_i/S_j, E)$ is given by

$$P(D_i/E') = P(D_i/S_j, E) = \frac{P(S_j/D_i,E)P(D_i/E)}{\sum_i P(S_j/D_i,E)P(D_i/E)}$$

where $P(D_i/E')$ is used to indicate that our new estimate of the probability is conditioned on increased experience, namely S_j and E. The next time we use Bayes rule (say when we observe S_k), $P(D_i/E')$ will be used in place of $P(D_i/E)$. In this way, the significance of any diagnostic evidence is interpreted in terms of the latest opinion about the probabilities of the various diseases in question.

Suppose a physician is attempting to diagnose a patient with oliguric acute renal failure. Assume that his experience to date has convinced him that the condition of the patient can be attributed to one of five causes. These causes and the subjective probability assigned to them by the physician

are: 1) obstruction, 0.40; 2) acute tubular necrosis, 0.25; 3) functional acute renal failure, 0.15; 4) acute glomerulonephritis, 0.15; and 5) cortical necrosis, 0.05.

Assume the immediate problem is to interpret the diagnostic significance of a urine volume of 50 - 400 cc. per day. To make explicit use of Bayes rule, the physician proceeds as follows. He estimates the conditional probabilities of this urine volume given each of the diseases in question. His estimates of this probability given each disease are: 1) obstruction, 0.20; 2) acute tubular necrosis, 0.90; 3) functional acute renal failure, 0.10; 4) acute glomerulonephritis, 0.50; and 5) cortical necrosis, 0.05. Because he now has both $P(D_j/E)$ for each disease in question and $P(S_i/D_j, E)$ for S_i = "urine volume between 50 and 400 cc. per day", he can apply Bayes rule to obtain an updated view of the problem. The application of Bayes rule yield the values of $P(D_j/S_i, E)$ which for this example are: 1) obstruction, 0.20; 2) acute tubular necrosis, 0.56; 3) functional acute renal failure, 0.04; 4) acute glomerulonephritis, 0.20; and 5) cortical necrosis, essentially zero.[1]

Of course, one would expect that as the student gains experience both in the estimation of conditional probabilities and the use of Bayes rule, the need to employ the technique explicitly will diminish. That is, he will

---

[1]Specifically, for acute tubular necrosis we have $P(D_2,E)P(S/D_2/E) = 0.25 \cdot 0.90 = 0.225$ and $P(S) = \sum_i P(D_i)P(S/D_i) = 0.395$. Therefore:

$$P(D_2/E') = \frac{0.225}{0.395} = 0.56.$$

associate with a disease those sign and symptom patterns which have the properties: 1) that the likelihood of the pattern is high given the disease is present, and 2) the conditional probability of the disease is high given the pattern. Then when he observes such a pattern, he can perform the inference function without resorting to the detailed steps required to employ Bayes rule. Note, however, that when the pattern of the presenting signs and symptoms does not match one of these learned patterns, he can employ the formal analysis he has learned to assess the significance of the various pattern differences.

While the study of Bayes rules may prove useful to physicians developing their diagnostic skills, it provides some insight into one aspect of the process only. Thus, while Bayes rule is a useful model of the inference function, more is required in order to model the test selection function. This problem was faced in the design of the diagnostic program. Because in many cases the initial signs and symptoms are not extensive enough to permit a diagnosis to be made, a method of seeking further information was devised. The emphasis was placed on the development of precise procedures, suitable for incorporation into a program. As with the inference function, an attempt was made to define the test selection function with sufficient generality to be useful in a number of ways.

The decision problem to which the test selection function is addressed can be represented schematically as a decision tree as in Figure 3. The topmost node in the tree, node A, denotes the position of the physician to the diagnostic problem. He must choose among the alternatives denoted

{0, 1, ..., K} where 0 denotes his making a diagnosis, and {1, ..., K} each denote his choosing a corresponding one of the K tests available. In Figure 2, each decision alternative is represented by a branch emanating from node A. Given that he selects a decision alternative J in {1, ..., K}, he arrives at a node like B. The path away from B is determined by the test outcome (which the physician cannot control). Thus he arrives at node A', at which he must again choose among alternatives, but now he haa more information, namely the test results from test J.

For the program, the problem of choosing a diagnostic strategy involves the analysis of such a tree. That is, the problem is to select a decision alternative at A. The program evaluates each alternative in turn. Basically the analysis considers the possible outcomes of an alternative in turn. For each test result, the inference function is used to simulate the observation of that result. Thus a new probability distribution is obtained on the assumption that the result will be obtained. A value is assigned to this distribution which reflects the potential risk of misdiagnosis and the likelihood that the test will in fact yield this result if it is performed. An overall value for the testing alternative is built up from the values for specific results. When each testing alternative and the diagnosis alternative have been ranked, the alternative with highest value is chosen. If it is a test, the results obtained are used in the inference function in Bayes rule. Then the test selection decision is faced again.

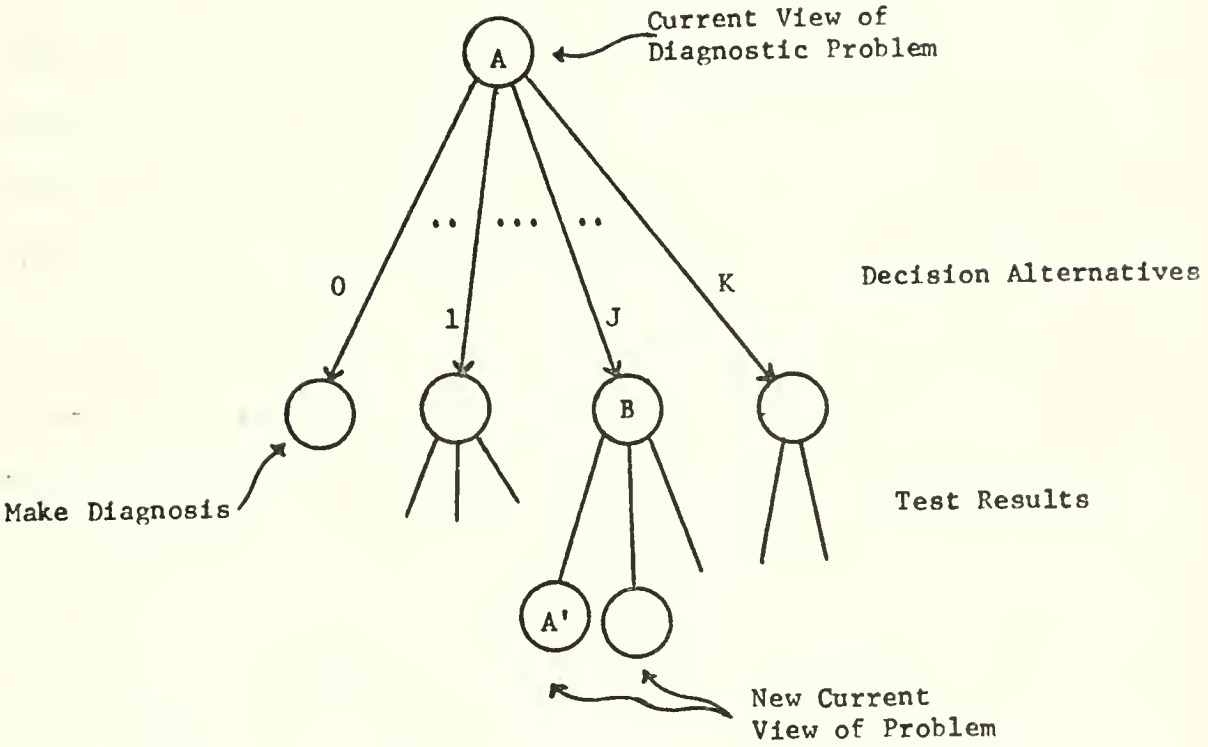
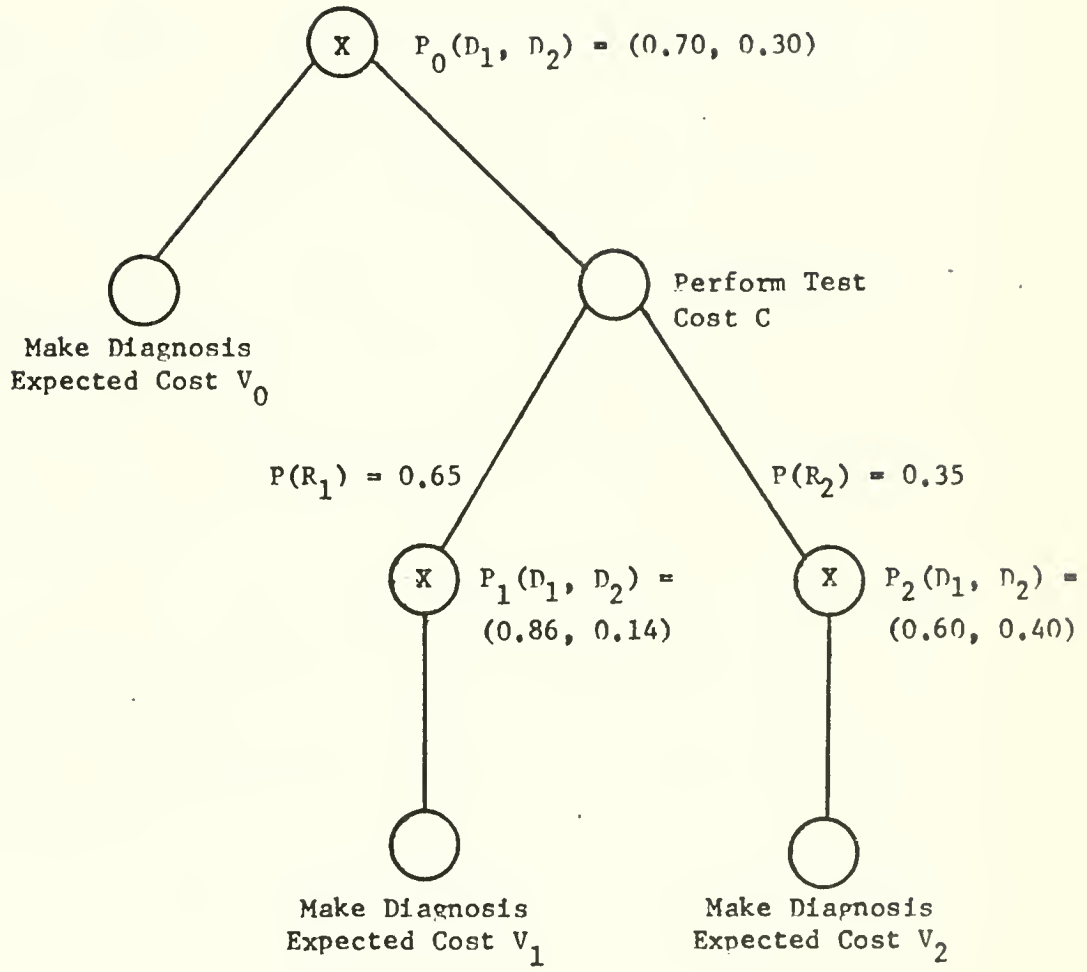


Figure 3: Decision Tree for Test Selection

Consider a very simple case of such a decision problem. Assume that the problem is to decide whether to make a diagnosis now, or to run a test (cost C) which has two mutually exclusive outcomes (R_1, R_2), and after obtaining the result, make the diagnosis. Suppose there are only two diseases in question, D_1 and D_2 with a priori probabilities 0.70 and 0.30, respectively. The relevant probabilities and the appropriate decision tree are shown in Figure 4.

In the decision tree there is one branch corresponding to the "diagnosis now" alternative and one corresponding to the use of the test. Assume that the physician assigns a value V_0 to the risk of misdiagnosis. This assessment will reflect both the probabilities of the two diseases as well as the seriousness of misdiagnosing one as the other. Thus, V_0 is the value assigned to the "diagnosis now" alternative.

The branch corresponding to the use of the test leads to a node from which there is a branch for each test result. Each "results branch" leads to a node when a new probability distribution is calculated. The new distribution is obtained from Bayes rule and the observation of the new result. At either of these nodes, the physician will have to make a diagnosis, and so he assigns values V_1 and V_2 to the nodes respectively, reflecting the risk of misdiagnosis at each. Note that in general $V_0 \neq V_1 \neq V_2$ because each is based on a different probability distribution.



Expected Cost of "Diagnosis Now" = V_0

Expected Cost of "Test then Diagnosis" = $C + P(R_1)V_1 + P(R_2)V_2$

		$P(R_1 \mid D_j)$	
A Priori		R_1	R_2
0.70	D_1	0.80	0.20
0.30	D_2	0.30	0.70

Figure 4: Sample Test Selection Problem

If the physician chooses to perform the test, he expects to obtain result R_1 with probability 0.65 and result R_2 with probability 0.35.[2] Therefore the expected risk of a diagnosis <u>after</u> performing the test is $0.65V_1 + 0.35V_2$. The total expected cost of the "test then diagnosis" strategy is $C + 0.65V_1 + 0.35V_2$. Hence if $C + 0.65V_1 + 0.35V_2 < V_0$, he will choose to test before making a diagnosis. Another way to view this result is if $C < V_0 - 0.65V_1 - 0.35V_2$, the cost of the test is less than the expected reduction in the risk of misdiagnosis resulting from its use, and so it should be performed.

Although this example is a simple one, it does demonstrate the basic decision tree analysis employed in the model. More detailed discussions of this analysis, including some comments on the specification of the V_i are available in [11] and [13].

---

[2]Note that $P(R_1) = P(R_1/D_1)\ P(D_1) + P(R_1/D_2)\ P(D_2) = 0.80 \cdot 0.70 + 0.30 \cdot 0.30 = 0.65$ and similarly for $P(R_2) = 0.35$.

V. Conclusions

In the above discussion, we have tried to indicate a point of view which can be taken with regard to the modelling of the diagnostic process. The underlying motivation for the discussion was the perceived lack of a formalism within which a student would study diagnosis and upon which he could build his skills. That sufficient knowledge of the process exists to alleviate this need seems clear. Introspection and observation have produced several discussions of diagnostic problem-solving [16, 17]. Such work can be a good basis for the development of a model. It seems, however, that certain problems have retarded this activity among those who teach medical students.

The first problem is the concept of a model is not well understood. In our discussion of models of cognitive function, we emphasized: 1) the level of resolution employed, 2) the importance of the purpose to which the model is to be put in determining its validity. The dependence of the first upon the second was also noted. These considerations have importance in teaching diagnosis for several reasons.

First, a basic model of the diagnostic process may be valid without accounting for the detailed processes employed by individual doctors. If the model elucidates general principles of diagnosis, then it has validity (and value) in teaching. Second, such a model need not account for the structure of the cognitive system involved in diagnosis. Third, certain complex aspects of the process may be treated as elements of the system, with only statements of their input-output behavior included in the model.

Finally, certain simplifications of the interaction of the system with its environment may be made. Thus, for example, we have ignored the question of how a doctor perceives diagnostic clues, although this perception is an important factor in the process.

With these considerations in mind, we outlined some of the issues which seem relevant to a model of diagnostic problem-solving. Evidence exists that a formal model can be constructed within this restricted context [11, 13] which is sufficiently interesting to merit further study and elaboration.

Such work is important because of the potential advantages it holds for improving the educational experience of students. Even in its current simple form, the model outlined in Section IV can be useful as a focus for discussions of diagnosis. Undoubtedly, it could be significantly expanded in ways which would increase its didactive value.

The development of that model (or any other such model) will require the efforts of physicians experienced in diagnosis. To a certain extent, they should consider concepts developed in other fields including cybernetics, cognitive simulation, probability theory, statistical decision theory, utility theory and computer science. All these fields have something which is relevant to the understanding, teaching and improvement of diagnosis. Perhaps an even better source of ideas about improving all aspects of diagnosis will be the physicians themselves, once they begin to wrestle with the problems of modelling their activities.

REFERENCES

[1] Mason, E. E., and Bulgren, W. G., Computer Applications in Medicine, Chapter III, Charles Thomas (Publisher), 1964, p. 39.

[2] Klír, J., and Valach, M., Cybernetic Modelling, Van Nostrand, Princeton, New Jersey, 1967.

[3] Ashley, W. R., General System Theory and the Problem of the Black Box, R. Oldenbourg Verlag, Munich, 1961.

[4] Shannon, C. E., and McCarthy, J., Automata Studies, Princeton University Press, Princeton, New Jersey, 1956.

[5] Bush, R. R., and Mosteller, F., Stochastic Models for Learning, John Wiley, New York, 1955.

[6] Reitman, W. R., "Information Processing Models in Psychology", Science, Vol. 144, 1964, No. 3623, pp. 1192-1198.

[7] Simon, H. A., and Newell, A., "Computer Simulation in Human Thinking and Problem Solving", Computers and Automation, Vol. 10, 1961, No. 4, pp. 18-26.

[8] Feigenbaum, E. A., and Feldman, J., Computers and Thought, McGraw-Hill, New York, 1963.

[9] Gorry, G. A., "Strategies for Computer-Aided Diagnosis", Mathematical Biosciences 2, 1968, pp. 293-318.

[10] Gorry, G. A., and Barnett, G. O., "Sequential Diagnosis by Computer", Journal of the American Medical Association, Vol. 205, September 16, 1968, pp. 849-854.

[11] Gorry, G. A., and Barnett, G. O., "Experience with a Model of Sequential Diagnosis", Computers and Biomedical Research, Vol. 1, No. 5, May 1968, pp. 490-507.

[12] Ledley, R. S., and Lusted, L. B., "Reasoning Foundations of Medical Diagnosis", Science, 130, July 3, 1959, pp. 9-22.

[13] Gorry, G. A., A System for Computer-Aided Diagnosis, Technical Report MAC-TR(44), Project MAC, M.I.T., September, 1967.

REFERENCES (Continued)

[14] Manning, R. T., and Watson, L., "Signs, Symptoms, and Symptomatics", Journal of the American Medical Association, December 12, 1968, Vol. 198, pp. 1180-1184.

[15] Platt, J. R., "Strong Inference", Science, October 16, 1964, Vol. 146, No. 3642, pp. 347-353.

[16] Levinson, D., "Teaching the Diagnostic Process", Journal of Medical Education, September, 1968, Vol. 43, pp. 961-968.

[17] Feinstein, A. R., Clinical Judgment, Williams and Wilkens, Baltimore, 1967.

[18] Churchman, C. W., et al., Introduction to Operations Research, John Wiley and Sons, New York, 1957.

Date Due