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The Diagnostic Process

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It is with deliberate intention that this paper, the first of three about computers, deals with the logic of medicine, in particular the diagnostic process, and not with the practical problem of how computers work. This is done to emphasise a most important concept: that until we have formulated a theoretical structure, a logic or a calculus of medicine, which is something medicine has never had, it is not possible to transfer all the activities of clinical medicine as we know them, diagnosis, prognosis, or decisions on treatment, to a computer. In other words our fundamental problem is that of formulation. The earliest attempt to apply any general formal structure to the problem of clinical medicine is that of Ledley and Lusted (1959). (For recent references *see* Lusted, 1968; Card and Good, 1970.)

A successful formulation would mean that the private world of the physician could be replaced by a public world of science, and in this public world we must inevitably use mathematical reasoning. First we have to define our objective. What is our objective in medicine?

Traditionally, the ultimate object of medicine is to improve the quality of life and its quantity in as many people as possible. The quality of a state of health might be called its 'value', but technically the word 'value' has long been appropriated by the mathematicians and we are forced to use the less happy word 'utility', which we take over from the economists. Our aim is therefore to maximise utility, but since our actions have only a certain probability of success, we must technically talk of 'expected utility'. We might, therefore, define our objective in medicine as: the maximisation of expected utility. If this is our objective, then the kind of mathematics we shall need is that concerned with probabilities and decisions, that is, a theory of rationality (Wald, 1950; Card and Good, 1970). According to this theory, the central activity of the doctor is the taking of decisions, and diagnosis, in the words of Lord Cohen, is only a tentative guide to action.

In this search for a theoretical structure for clinical medicine, and for diagnosis in particular, it is difficult to imitate a process of whose nature we are largely ignorant. We therefore have to start from certain axioms.

Our theoretical system might rest on the basis that the sick patient can be abstracted into a set of data. Each symptom, each physical sign, each laboratory or radiological examination that we can elicit can be regarded as an atomic piece of evidence. We have felt the need for a word to describe any phenomenon arising from a disease or constituting evidence of it and, for this purpose, we have resuscitated the word 'indicant'. A sick patient can then be considered as an ordered set of indicants. This is, of course, a tremendous assumption. In eliciting indicants, we have to include the concept of error, an imperfection that is acceptable provided we can measure its extent (for bibliography, *see* Nacke and Wagner, 1964). If we are now to allocate a set of indicants to a disease, that is, to make a diagnosis, we must define a disease. One way is to think of this concept geometrically.

If we regard the patient as an ordered set of indicants we can think of these as co-ordinates in some space; the patient can then be represented by a point in this space. Other patients with the same disease will be represented as points somewhere near him so that a cluster is formed characteristic of that particular disease. The problem of defining a disease then becomes a problem of defining a cluster that is separable from other clusters. This problem is part of the general problem of defining a class and is encountered in a number of disciplines, for example the definition of a species in biology, or the definition of a period in archaeology. Though the general problem cannot be said to be solved, there are a number of clustering techniques of some service (Sokal and Sneath, 1963; Hayhoe *et al.*, 1964), and perhaps we can say that at least such techniques can give us some confirmation of our clinical suspicions, for example, that Crohn's disease of the large bowel and ulcerative colitis are separate diseases (Hywel Jones *et al.*, unpublished data). Diagnosis now becomes the allocation of a set of indicants to a disease class.

One possible diagnostic method uses Bayesian probability. Let us imagine a handful of cards are found on a card table in a club, and in this club only two card games are played, bridge and poker. Given such a sample of cards, and knowing the constitution of the deck of cards used in bridge and the deck used in poker, it is possible to calculate the probability that this sample of cards is obtained from either deck. We also need to know the frequency with which these two card games are played at this particular club, and with this information we can now calculate the probability of which game was played at this table. In this analogy, the deck of cards represents the complete set of symptoms, signs, and laboratory findings in a particular disease—the set of indicants—while the cards on the table represent those indicants of which the particular patient complained. The frequency with which the two card games are played represents the frequency of encountering these two diseases, say, in an outpatient clinic. In an informal way this is all that is meant by Bayes' Theorem.

Such methods, which make certain simplifying assumptions, allow us to calculate the diagnosis if we know the initial probabilities of the diseases in question, and the probability of every indicant given a particular disease. Variants of this method have been used in a number of simple diagnostic problems, and the calculated diagnoses have been shown to compare in accuracy with the diagnoses as made independently by consultants skilled in that particular field (for bibliography, *see* Lusted, 1968). The allocation of a complete set of indicants to a disease class might be called the Mark 1 diagnostic model.

A little reflection will show us that this model is uneconomic. No doctor ever collects a whole set of all possible characters from the patients before making a diagnosis; he always acts sequentially. The presenting symptom suggests a set of possible diseases with some mental estimate of their probabilities. The doctor then asks the question most likely to alter this set of probabilities. From the patient's answer he then selects a further question, or makes a physical examination. Using his experience in this way, he aims to thread his way through what may be called a diagnostic search tree. Such a tree has two kinds of nodes, one of which the doctor selects, which we call a facet, and this comprises a set of indicants. The other kind of node is an indicant and is elicited by some test procedure (Fig. 1). During this diagnostic process the set of probabilities of the diseases are constantly altering, so that eventually the probability of one disease approaches unity, and the diagnosis is said to be reached. The doctor aims to do this by the fewest possible steps.

Movement through the diagnostic search tree by the expected fewest possible steps normally depends on the experience of the doctor in selecting the series of facets and carrying out the actual tests. With certain reservations, the optimal selection can now be carried out automatically (Good, 1968). In a given 'disease space', that is, a model dealing with a limited number of diseases, if the initial probabilities of the diseases are known and also the likelihoods, that is, the probabilities of the indicants given the diseases, then the selection of the facet of 'greatest expected informativeness' can be calculated. Very informally, we might call this the next best bet. Technically, the program does this by selecting the facet of greatest expected informativeness and the doctor then carries out the actual examination or test.

Such a model we might call Mark 2, and, although it is clearly an improvement on Mark 1, it is still quite unrealistic, since in eliciting evidence it makes no distinction between asking a patient's age and doing an exploratory laparotomy. It is here we have to introduce the concept of values, or the

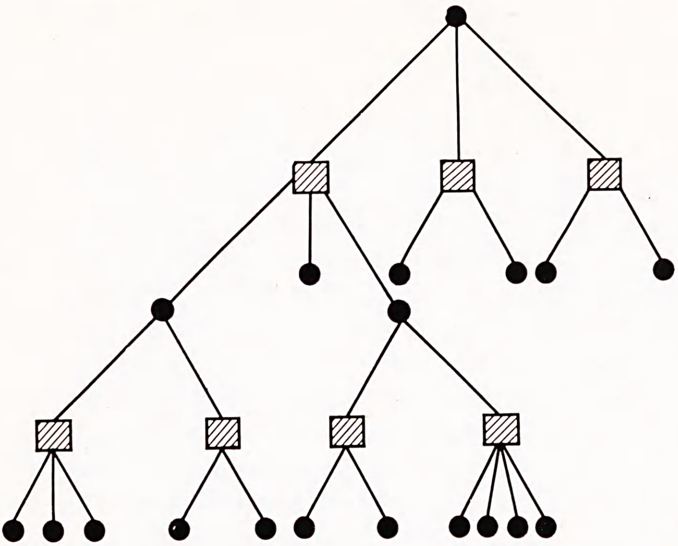

Fig. 1. Diagnostic Decision Tree. The doctor selects a facet, ▨, that comprises a set of indicants, one of which, ●, will be elicited from the patient by the test procedure. The facet could be a part of the history, e.g. whether the patient had lost weight or not, the set of indicants then being two—'Yes/No'; or part of the physical examination, e.g. for enlargement of the spleen, to which the set of indicants might be four—'No enlargement, tip of spleen palpable, moderate enlargement, gross enlargement', and so on, for all evidence elicited from the patient by any clinical, radiological, or laboratory procedure.

positive utilities of different diagnoses and negative utilities or 'costs', using cost in a very broad sense (Gorry, 1968; Aitchison, 1970; Card and Good, 1970). The introduction of these ideas alters the logic of movement through the search tree since we are now influenced, not only by the expected evidence to be elicited, but also by the cost of getting it. We are trading information for cost all the time. In terms of decision theory, movement through the search tree will cease when the cost of realising the next facet is greater than the expected gain in utility which would result from making the test. Indeed, the doctor, in considering some further procedure, may say—'I don't think it's worth while'.

The introduction of these utilities and costs would enable us to construct what we might call a Mark 3 diagnostic model, but this has yet to be done, since at the moment we have no method for estimating the utilities of states of health or the costs of investigations. Until we can do this we cannot be said to have created any model of medical diagnosis that even approaches reality.

We have briefly explored a crude but possible structure of the diagnostic process. What is the likely role of the computer if such a calculus were ever realised?

The computer excels at calculation, simple logical decisions, and the storage of information. Human beings have great ability to make decisions in very complex situations, and they excel at pattern recognition at which computers are at present primitive. If the practice of clinical medicine can be analysed as sketched above, then we can foresee the need for a doctor to elicit evidence, while the calculation of probabilities and movement through a decision tree could be guided by a computer. Management decisions, certainly of any complexity, and which rest on the imponderables from some form of pattern recognition, would have to be done by the doctor.

Clearly such a method can work only in so far as it is possible to formulate clinical problems in mathematical terms. There is no possibility of solving all the problems of diagnostic medicine in this way; the question is, can it solve any, and if so, what fraction?

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