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The diagnostic process

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The ability to cope with uncertainty, exclude the dangerous, ignore the irrelevant . . . 
(Elwyn, 1997).

When patients present with new problems, attempting to arrive at a diagnosis is perhaps the single most important consultation task for a doctor, whether in hospital or general practice. If it is possible to make a definitive diagnosis, the doctor becomes more aware not only of the specific pathophysiology underlying a patient’s complaint, but also of its probable natural history. Such understanding greatly influences the way in which plans for clinical management are formulated and implemented. The greater the confidence the doctor has in any diagnosis made, the greater is the ability to judge not only whether, but also what sort of, intervention is indicated. Thus, arriving at a diagnosis ‘is a crucial achievement which opens the way to prognosis and treatment’ (Royal College of General Practitioners, 1972).

It must be emphasized, however, that the term ‘diagnosis’ does not just refer to conventional disease labels; nor does the term ‘pathophysiology’ refer solely to organic disease. Although the search for, and identification of, organic disease (i.e. disease-centred diagnosis) is a crucially important consultation task, it is not the whole story. It is also necessary to attempt to arrive at a patient-centred diagnosis, i.e. one which includes consideration of the patient’s thoughts and feelings concerning the nature and potential causes of their presenting complaints (see previous chapter). Thus, the reader is encouraged to consider diagnoses in whole-person terms, which should include both patient-centred and disease-centred elements.

In this regard, the reader should also be aware that making a firm pathophysiological diagnosis in general practice may not be possible – even for experienced doctors – in anything up to 50% of presenting patients. In the absence of an appropriate diagnostic label, however, the ‘diagnosis’ can still be expressed in the form of the patient’s problem(s) (see also ‘The triple diagnosis’, below).

In this book, the diagnostic process and management are considered in separate chapters for the sake of convenience and clarity. It should be
remembered, however, that a diagnosis is usually a statement of probability rather than certainty, and often needs to be ‘regarded as provisional until supported by the subsequent course of the case or the response to specific treatment’ (Royal College of General Practitioners, 1972). Thus, although diagnosis often precedes and predicts management plans and actions, the diagnostic process often includes management. This is because management decisions are frequently, and justifiably, taken on the basis of an assessment of the patient’s symptoms and/or signs and/or problems, without a definitive diagnosis having been made. Although this happens in both general practice and hospital, it is especially frequent in the former.

Making a diagnosis is a complex process and involves far more than merely amassing clinical information. A wide range of skills needs to be acquired, integrated and applied (see Chapter 2). Of particular importance are skills in interviewing (see Chapter 6), clinical reasoning and accessing personal knowledge at the time when it is required. Thus:

The term [diagnosis] encompasses those processes whereby the clinician interprets clinical information [and] follows and chooses among his thoughts about what is wrong with the patient (Gale and Marsden, 1985).

It is essential to develop the ability to arrive at a ‘correct’ diagnosis as often as clinical circumstances permit. If this goal is to be achieved, you will need first of all to develop the capability to understand how diagnoses are made and why in particular circumstances you have arrived at the correct diagnosis. You are then more likely to continue to arrive at correct diagnoses on future occasions. Since many, if not most, errors in diagnosis result from errors in the diagnostic process rather than from a lack of factual knowledge, you will also be better equipped to identify possible reasons for such errors and to take appropriate action to rectify the situation. Such a process of self-learning is a powerful stimulus to improvement in clinical performance as it will enable you to cope better when, as is inevitable, you are faced with clinical problems not previously encountered.

This chapter will introduce the reader to the processes by which problems are clarified and diagnoses are formulated.

**Inductive and hypothetico-deductive methods of problem-solving**

In the final resort, doctors arrive at a diagnosis when they are able to fit a patient’s symptoms and signs into a pattern that they can recognize as representing a particular disease entity. In hospital practice there is a greater likelihood of being able to do this as compared to general practice.
Nevertheless, in any clinical context, only rarely is a distinctive pattern instantly recognizable; usually a doctor needs to embark on a search for further evidence to help to distinguish between a number of potential diagnoses. There are several ways in which this task can be accomplished. Often it can be done within a single consultation, but sometimes it requires more than one.

It is likely that you will have been taught to reach a diagnosis by using the traditional or inductive method of problem-solving (Figure 3.1). This method dictates that — irrespective of presenting complaint — a comprehensive history (including a system review) has to be taken from every patient, followed by a complete physical examination backed up by a number of investigations, many of which are of a routine nature. Furthermore, you will have been encouraged to delay the task of diagnostic formulation (i.e. interpreting the information gathered) until this mass of information has been assembled.

This approach undoubtedly provides medical students with repeated opportunities to familiarize themselves with the range of questions that may need to be asked in taking histories from patients, and gives them the necessary practice for developing their skills in physical examination techniques. In actual clinical practice, however, such an approach to clinical problem-solving is rarely used by general practitioners and infrequently by hospital doctors because 'an unfocused shotgun approach is unproductive, confusing and time-consuming' (Joorabchi, 1989).

![Comprehensive history:](image)
- Presenting complaint
- System review
- Previous medical history
- Drugs (current/adverse reactions)
- Social/family
- PLUS
- Complete physical examination
- PLUS
- Investigations
  ↓ (Then consider)
- Diagnosis

Figure 3.1 Inductive method of problem-solving
Indeed, 'excessive data collection interferes with clinical inference and reasoning by overloading the capacity of the system (i.e. the doctor)' (Hoffbrand, 1989). Consequently, the inductive approach should be reserved for the few occasions when patients present with such vague symptoms that no useful diagnostic formulations can be generated and potentially serious underlying causes cannot be excluded.

In reality, most clinicians reach diagnoses by a process of hypothetico-deductive reasoning, i.e. by educated guessing and testing (Elstein et al., 1978). Furthermore, studies have shown that general practitioners and hospital doctors both use the same 'multiple hypotheses-guided, problem orientated enquiry' (Barrows et al., 1982). Figure 3.2 provides a simplified representation of the stages involved in this process. The so-called hypothetico-deductive method is efficient as it enables doctors to solve problems with maximum time- and cost-effectiveness and minimal disturbance to patients. A professor of medicine has given further support to this view: 'In recognizing that diagnosis is fundamentally hypothetico-deductive, I am not simply contrasting it with a blank mind ritualistically collecting information, I am saying it is superior because a blank mind may miss information which is generated only in response to an idea' (Campbell, 1987).

Even before a patient enters a consulting room, whether in hospital or general practice, a clinician is likely to have access to a considerable store of information about that patient. If the doctor knows the patient well – as is likely with a significant proportion of patients in general practice – knowledge of the patient’s previous medical history, individual and family circumstances and previous patterns of illness behaviour may be readily recalled from memory.

If the patient is not known to the consulting clinician – as will be the case in the majority of consultations conducted by students – the medical record should always be selectively scrutinized before the patient enters the consulting room. With practice, this can be done in a matter of 10–20 seconds. The time invested will be repaid in the majority of consultations by arming the clinician with much valuable information, which will facilitate more efficient clinical problem-solving. The following information should be sought:

- Age, sex and social class; the latter can be gleaned from the patient’s address and/or occupation. All these factors have an influence when considering diagnostic probabilities.
- Significant previous medical history/family history: the medical records in most (teaching) practices will have these conveniently summarized. This will help you to avoid asking needless questions (for example, if a patient has had a hysterectomy you need not ask about menstruation) or may assist in the erection of diagnostic hypotheses (for example, if a patient has had an appendicectomy then appendicitis cannot be the cause of any presentation of abdominal pain).
Furthermore, knowledge of current medication may help you to link a patient’s presenting symptoms with particular drug side-effects. An awareness that a patient’s family member had previously died of cancer, heart disease, etc. may also alert you to the fact that the
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Patient's presenting symptoms may relate to anxiety about having contracted the same disease.

- Frequency of attendance: this will provide some insight into the ability of the patient to tolerate symptoms. It is usually wise to pay particular attention to any presentations from infrequent attenders.
- Details of the last consultation: the reason(s) for the current consultation may well relate to the patient's last consultation. If so, this will often signify that the patient's symptoms will not have resolved as expected or else that new ones have developed. In any event, you will be aware of what took place previously and this will make your current consultation more efficient.

As the consultation begins, the doctor is presented with information that is both verbal and non-verbal. There is now considerable evidence to indicate that: 'experienced doctors often formulate their diagnostic hypotheses in the very first few instances with the patient and they are usually correct' (van der Vleuten, 1996). In many instances a prior stage of 'pre-diagnostic interpretation' (Gale and Marsden, 1983) occurs, during which the doctor begins to assess the patient's problems in terms of broad categorizations rather than specific diagnostic entities - e.g. 'I think the problem is cardiovascular'; 'this is likely to be psychological rather than physical', 'acute rather than chronic', 'serious rather than trivial', etc.

In the next phase of the problem-solving process, the doctor seeks to gather further information by asking particular questions of the patient in an attempt to find support for, and to discriminate between, the diagnostic possibilities previously generated. Throughout the process, the doctor needs to be constantly interpreting the answers received from the patient and modifying the search for further information accordingly. The immediate aim is to try to eliminate any previously nominated diagnostic possibility that is not supported by the additional information gathered. The ultimate aim is to be left with a single definitive diagnosis: more usually, however, the doctor will be left with one or more diagnostic probabilities.

Since the history is the key predictor of eventual diagnosis (see below), the purpose of the physical examination is to seek relevant and discriminating physical signs to help confirm or refute working diagnoses (Fraser, 1994; Sandler, 1984).

If at any stage in the problem-solving process no support can be gathered and no progress can be made, the presenting problem needs to be reconsidered and a fresh assessment of the likely diagnoses made through the gathering and interpretation of further clinical information. If a diagnosis is confirmed, management decisions can then be implemented as appropriate. On the other hand, a final judgement may have to be postponed because of insufficient evidence to confirm or eliminate a diagnostic possibility. Under these circumstances, a 'non-diagnosis' may
have to be made or time employed as a deliberate diagnostic strategy. Thus, management decisions may have to be made in the absence of a firm diagnosis, and the outcome awaited. Particular aspects of this process will be considered in greater detail in due course.

**The relative contribution of the clinical history, physical examination and investigations in the diagnostic process**

Regrettably, the value of the history in the diagnosis of disease often seems to be neglected in both undergraduate and postgraduate medical education (Sandler, 1984).

Most of the clinical problems encountered in everyday clinical practice can be dealt with effectively and satisfactorily on the basis of a good clinical history (Sandler, 1984). Thus, history-taking is the key to diagnosis in the consultation. This is particularly true in general practice because of the many presentations of multiple, undifferentiated symptoms, often with a paucity of accompanying physical signs and the frequent absence of 'disease' (see Chapter 1). The results of two studies based on new referrals to outpatient departments of two hospitals in Nottingham and Barnsley confirm, perhaps more surprisingly, that it is also true in hospital practice.

Hampton et al. (1975) showed that the diagnosis can be made on the basis of the clinical history alone in 83% of new patients seen in a cardiology outpatient clinic, compared to physical examination (9%) and investigations (9%). In a more comprehensive study in a medical outpatient department, Sandler (1979) also concluded that the diagnostic value of the history far outweighed the contribution of physical examination or investigations. The history alone determined the diagnosis in 56% of all referrals made, with a range of 27–67% for alimentary and cardiovascular diagnoses respectively. Physical examination determined 17% of diagnoses, with a range of 0–24% (alimentary and cardiovascular, respectively). The corresponding figures for routine investigations were 5%, with a range of 0–17% (alimentary and respiratory, respectively) and for special investigations 18%, with a range of 6–58% (cardiovascular and alimentary, respectively). Routine haematological and urine examinations made a negligible (1%) contribution to diagnosis.

When the 180 patients with chest pain were considered separately, it is interesting to note that:

The history gave the diagnosis in 90%, and the examination was of no diagnostic value at all. Routine investigations, mainly chest radiographs and electrocardiography, helped with only 3% of diagnoses and special tests, mainly exercise electrocardiography, with 6% (Sandler, 1979).
Modern technology has made it all too easy to carry out batteries of tests. Consequently, there is ‘frequently an unfortunate tendency to rely on the results of such tests before decisions are taken on diagnosis and treatment, even though such tests are often of limited value’ (Sandler, 1984). This has been amply demonstrated above.

On the basis of these findings, Sandler concluded:

\textit{Much greater emphasis should be placed on the diagnostic \ldots value of the history. Students, and postgraduates, should be well trained in taking a good history and in drawing diagnostic conclusions from the history before embarking on the examination. This will encourage the student to seek specific examination findings to confirm or refute the diagnosis based on the history [my italics].}

This is probably the most important statement contained in this book. If all doctors could acquire and implement the necessary skills, it would result in a major improvement in the quality and cost-effectiveness of clinical practice, not least through a reduction in the ordering of unnecessary tests and investigations. It would also improve the quality of medical education by providing more appropriate role models for medical students (and junior doctors) to emulate.

A Dean of Medicine emphasized the point:

\begin{quote}
If you can take a good medical history, trust your physical examination and safely judge how sick your patient is, you can avoid excessive testing, imaging and prescribing (Federman, 1990).
\end{quote}

It must be stressed, however, that ‘a good history’ must mean an appropriate and suitably discriminating history. ‘This means asking the right question, not every question’ (Hoffbrand, 1989) [my italics]. For example, thyrotoxicosis is a condition in which dozens of clinical features may occur. If thyrotoxicosis were suspected, it would make more sense for the doctor to try and establish whether the patient had weight loss with an increased appetite and dislike of hot weather, since the presence of these three features would make the diagnosis highly likely. On the other hand, symptoms like tiredness and irritability can occur in many other conditions apart from thyrotoxicosis and are, therefore, not key symptoms in helping to discriminate between thyrotoxicosis and other conditions (see Case 2 below). As Dixon (1986) has noted with impeccable logic, ‘It makes no sense to ask a lot of history questions that will make no difference to the outcome of the consultation’.

The over-riding importance of the history in clinical medicine is further reinforced when one considers the major contribution it makes not only to diagnosis but also to determining management plans (see Chapter 4).
Generating and ranking appropriate diagnostic possibilities

There are four principal factors which influence the generation and ranking of diagnostic possibilities: probability, seriousness, treatability and novelty (Elstein et al., 1971).

**Probability**

This is by far the most important influence since, in any given clinical circumstance, the essential question a doctor must ask is: What is the most likely cause or causes of my patient’s symptoms? The probability that a particular presenting symptom or group of symptoms will result in a particular diagnosis being made is further influenced by two inter-related factors:

- The crude frequency of occurrence of the particular condition(s) suspected
- The complex interaction of patient and symptom variables and its effect on that crude frequency.

As an example, consider the presenting symptom of cough. Our knowledge of the distribution of morbidity within general practice tells us that the overwhelming likelihood is that the cough is caused by an acute infection of non-serious nature (see Table 1.1). Furthermore, consider the way that diagnostic probabilities will be influenced both by variations in the duration of the cough and the age of the particular patient (Figures 3.3 and 3.4, respectively). It is obvious that the likely diagnosis is very different for a 3-year-old with a cough – whatever its duration – compared to a 70-year-old. Likewise, the probable diagnosis of a cough in a 70-year-old will greatly vary depending on whether it has been present for 3 days or 3 months. On the basis of an awareness of probabilities, therefore, a doctor is immediately helped towards the appropriate interpretation of a patient’s presenting symptom even at this early stage of the consultation. The elicitation of the presence or absence of associated symptoms such as haemoptysis, weight loss, etc., will of course further influence and help to clarify the diagnostic probabilities.

It is important to remember, however, that the most likely underlying diagnosis need not always be of a non-serious nature. For example, if a man of 50 presents with severe crushing and central chest pain radiating into both his jaw and his left arm, accompanied by dyspnoea and sweating, the most likely diagnosis is myocardial infarction. Indeed, with such a clinical picture any other diagnosis is unlikely.

**Seriousness**

Particular consideration should be given to the possibility that a life-threatening or seriously incapacitating condition may be responsible for
Figure 3.3  Likelihood of different causes of cough relative to duration of symptom
presenting symptoms. Given appropriate circumstances, such diagnostic possibilities should merit inclusion even though disproportionate to their actual frequency of occurrence. For example, the average general practitioner is likely to encounter a malignant melanoma only once or twice in a professional lifetime. This should not stop doctors suspecting the possibility of a malignant melanoma on many more occasions – given an appropriate clinical presentation – because of the potentially catastrophic consequences of delay in making such a diagnosis.