The Basis of Clinical Diagnosis

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Second Edition

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R.A.P. G.D.P.

Preface

This book is an introduction to clinical studies for medical students. The approach was chosen to emphasize the relationship of symptoms and abnormal physical signs to the underlying disorders of structure and physiology. For this reason, considerable emphasis has been placed on the introduction to each system, providing an outline of the structure, mode of action and the manner in which disease interferes with normal function, producing symptoms and signs. We hope that this will help the student to appreciate more easily the functional significance of the patient's history and clinical features, leading to an earlier understanding of the pathological conditions underlying the abnormalities found on examination. The illustrations used often show more advanced stages of disease to make recognition easier; later the student should be able to appreciate the early indications of these conditions by being aware of the classic changes.

There are separate sections in each chapter on the investigation of the patient which are designed to indicate the more relevant procedures required to establish a particular diagnosis while only briefly considering more complex tests. The interpretation of results is discussed. This section of the book is intended to acquaint the student with the various methods of investigation which are available, suggesting how and when they should be applied and the order in which they should be used. Some outline schemes are included to indicate the appropriate series of tests useful in certain common clinical problems. We consider that these will be of particular value to the more senior student and the house physician.

Our aim is to focus on the basis of a disease process, relating this to the consequent clinical features while avoiding exhaustive differential diagnosis; nevertheless, we have regarded it worthwhile to recapitulate a number of common problems in the form of lists. These are not meant to be exhaustive but are structured to provide a framework on which the reader can base his learning. This approach should enable the student to understand and assimilate more easily the detailed information available in textbooks of medicine.

This book is also of value as an introduction to clinical medicine for senior nurses, laboratory workers, physiotherapists, radiographers and non-medical graduates in research who wish to gain a wider knowledge outside their particular field of interest.

The writing of this book was encouraged by our colleagues, but the subject matter gradually evolved from our own experience as undergraduates and later as clinical teachers.

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1 Introduction

HISTORY AND EXAMINATION

The approach to the patient

The student should appreciate that his objective in clinical examination is to obtain complete information about the present and past illnesses and the environmental background which will enable him to make an accurate diagnosis. At the beginning of clinical training the student may be apprehensive and feel at a considerable disadvantage when he approaches a patient for the first time. The anxiety usually stems from the fact that he is asking the patient to co-operate in his training but is unable to offer anything in return. This attitude is natural, but the unease is usually dispelled by the realization that most people are willing or even anxious to discuss their illness if approached with genuine interest and understanding. A close relationship often develops and the student must be aware of the effect of this on both the patient and himself.

Opening remarks such as 'Do you mind if I ask a few questions about your illness?' or 'I am a medical student, my name is, would you mind if I ask about the symptoms which made you visit your doctor?' will help pave the way.

Judging the correct social and intellectual level at which to conduct the interview immediately helps to establish a satisfactory relationship as this puts the patient at his ease, gains his confidence and prevents undue emotional attachment or antagonism.

The intellectual level of the history taking may be assisted by the patient's appearance and the way he tells his story; it is vital that the student should adapt the phrasing of questions to the degree of comprehension of the patient. Initially he will be guided by previous social experience, but repeated history taking will allow him to develop a satisfactory technique.

The attitude of the interviewer is of the utmost importance in maintaining the patient's confidence. A hint of disapproval, moral judgement or excessive familiarity will destroy the rapport which has been established. The patient's reaction may then be one of hostility, tearfulness or withdrawal, making further progress difficult or even impossible.

Introduction to history taking

The manner in which the history is taken has to be developed independently by each student, drawing on his own social and medical experience so that he learns to be at ease with any patient. This creates mutual confidence and the correct atmosphere which must be maintained throughout physical examination, investigations and subsequent treatment.

From the outset the student should try to relate the patient's story to a disturbance of the function of one or more of the body systems. Experience will enable him to guide the interview to bring out other related points indicating a particular disorder. Increasing knowledge of clinical medicine and pathology can be incorporated into this approach, allowing the questioning to become more precise. If this is done skilfully the diagnosis can often be made from the history alone.

The behaviour of the patient in response to disease is modified by personality, past illnesses and his social and family background. The influence of these factors on the illness can only be appreciated by careful enquiry which will reveal some of the emotional aspects of the patient's character and indicate the part these are playing in the current illness.

History taking depends entirely upon an ability to communicate clearly and accurately. A direct account may be unobtainable if the patient is semiconscious or mentally or emotionally disturbed. In these circumstances, observation of his behaviour together with questioning of relatives may be sufficient to arrive at a probable diagnosis. A patient speaking only a foreign language imposes another barrier to accurate communication as an interpreter is essential. Great patience and care is necessary to ensure that the translator asks the exact questions posed and interprets the answers accurately. Often the interpreter is a relative or friend who may tend to give answers he thinks are correct without first asking the patient. Even if the examiner has a good grasp of the language, important nuances may be lost. Local dialects may pose certain difficulties of interpretation even in one's native language.

It is important that the words used to describe any symptom have the same precise meaning to both patient and examiner. If there is any doubt, the patient should be asked to explain what he means. A common example is 'indigestion', which can be used for any one of the following: oral or rectal flatulence, epigastric discomfort, abdominal fullness or pain. Confusion frequently arises over the use of quasi-medical jargon by the patient. For example, the term 'chronic' is often used inaccurately to describe a severe quality of pain rather than the duration of the symptom. Particular care is necessary in obtaining the story from children and the aged. Anxious relatives may try to answer all the questions but, although their story is important and often vital, whenever possible the subject should be encouraged to give an account of the illness.

THE HISTORY

Main complaint

The patient should be allowed to speak freely and without interruption. An intelligent and observant individual usually needs little assistance in describing his presenting and principal symptoms. Others require help in starting their story with a question such as 'Tell me how the illness started' or 'What was it that you first noticed wrong?' The shy patient may need further prompting

while the <u>verbose</u> subject must be carefully but firmly directed to keep to the point of the story. In particular, patients should be dissuaded from describing the details of 'diagnoses' made by friends and relatives.

The interrogation regarding the main complaint

At the end of the patient's own story, the student may have an accurate account of the main complaint. Usually further questioning will be required to elucidate a number of points, such as the time relationship between the onset of various symptoms, the order in which they developed and which particular feature has been most troublesome. Sometimes the patient requires help with the description of the character of a symptom, especially pain. Here the doctor may be able to suggest a number of alternatives; for example, 'Is the pain burning or throbbing in character, or like a pressure?' Nevertheless, it is important to avoid leading questions as far as possible, particularly to a suggestible subject. To prevent being misled by the over-co-operative patient, several questions should be framed which require a negative answer. Following the narrative and the interrogation the examiner should have a clear indication of all facets of the main symptoms. Taking pain as an example the following facts must be elicited:

Site.

Quality—dull, aching, sharp or stabbing, boring or pressing.

Intensity—persistent, intermittent, waxing or waning (colicky).

Duration-minutes, hours or days.

Radiation—spread of the discomfort, deep or superficial.

Precipitating and aggravating factors—linked with the onset of pain, such as food, exercise or respiration.

Relieving factors—such as posture, food or defaecation.

Each prominent symptom of the main complaint should be considered in this way and then interrelated with regard to time and severity. It is often helpful to recapitulate the history to the patient, particularly if this is complicated, to make sure the details are correct and in the right sequence.

The examiner who hurries and fails to give sufficient time for the full story, relying on answers to direct leading questions, may elicit a completely false history.

Other body systems

Following an account of the main complaint, it is essential to enquire about the function of all the other body systems. The following questions, framed in a suitable manner, should be asked in a systematic way to avoid omissions.

General health

Well-being. Is your general health good? Has it deteriorated since the onset of the illness, or compared with six months or a year ago?

Do you become easily tired?

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'Appetite. Is your appetite good; has it changed recently?

Diet. What do you normally eat? (A brief description of the meals taken each day should be obtained and any food fads recorded.)

Weight. Has your weight changed? What was your weight before the illness started? Have your clothes become loose or tight?

Gastrointestinal

Do you have any difficulty in chewing or swallowing?

Do you have any abdominal pain or discomfort related or unrelated to food?

Do you vomit or regurgitate fluid or wind?

Are your bowels regular; are the faeces formed?

What is their colour?

Is blood or mucus passed at any time?

Cardiorespiratory

Do you have soreness in the throat or ears?

Is there any discharge from the nose or ears?

Do you have a cough; if so, is there any sputum (phlegm) or blood?

Are you short of breath? What degree of activity produces this?

Do you have any pain in the chest?

Have you noticed your heart thumping (palpitations)?

Do your ankles swell at any time?

Urogenital

Do you have any pain or difficulty in passing urine?

Do you ever pass water involuntarily?

Do you wake at night to pass urine?

Has the urine changed in colour or volume?

Do you have any pain in the loin or abdomen?

Is there any urethral discharge?

Do you have any difficulties with sexual intercourse?

Menstrual history

At what age did your periods start (and finish)?

Are they regular? What is the time interval?

For how many days do you lose, and how much (the number of pads)?

Are they painful?

Are you on the contraceptive pill?

Have you noticed any change in the breasts?

. Endocrine

Are you unduly affected by heat or cold?

Do you perspire excessively?

Have you noticed any swelling in the neck?

Has the hair growth changed in character or quality?

Is your libido normal?

Reticuloendothelial

Have you noticed swelling in the neck, under the arms or in the groins? Do you have a tendency to excessive bruising or bleeding?

Central nervous system and special senses

Do you suffer from headaches, faints or giddiness? Is your memory good? Has it altered?

Are your vision and hearing and sense of smell normal?

Do you have any weakness of the limbs, pins and needles, numbness or cramps?

Do you have difficulty with walking or balance? How many hours do you normally sleep? Do you wake at night?

Psychiatric assessment

Has your temperament changed? Are you easily upset, angry, tearful or depressed or unduly anxious?

Musculoskeletal

Do you have pain, stiffness or discomfort in the back, neck, joints or limbs? Do your joints swell?

Skin

Have you noticed any rash, lumps or irritation of the skin? Has the texture or colour of the skin altered?

The past history

Some aspects of past disorders may have been mentioned when discussing the main complaint. Now, all the past illnesses, operations or accidents should be noted, whether or not they appear directly relevant to the present condition. This should include any childhood ailments such as rheumatic fever, chorea (St Vitus dance), nephritis, whooping cough, hay fever, allergies or asthma which often have significant sequelae in later life. A record should be made of all previous hospital admissions with their approximate dates and the nature of any operation performed, as these may be related to subsequent disease. For example, gastric resection many years previously may be responsible for anaemia, steatorrhoea or metabolic bone disease due to inadequate intestinal absorption, while secondary malignant deposits can appear years after a primary tumour has been removed.

Accidents may play an important part in subsequent ill health; a fractured skull can lead to epilepsy or persistent headaches, while premature arthritis may be precipitated by a fracture involving a joint. Patients should be asked specifically if they have ever suffered from chest disease, including pneumonia

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or heart disease such as coronary thrombosis, since serious illnesses and hospital admissions are sometimes forgotten. Often the patient is unaware of the exact nature of previous illnesses or suppresses the information even though this was explained at the time. Questions concerning past treatment sometimes give an indication of the original diagnosis. In other cases, relatives may be helpful but the previous case notes should be obtained whenever possible. A history of venereal disease is often withheld but is suggested by the story of a course of injections following a lumbar puncture, without a history of meningitis.

Residence abroad, particularly in the tropics, should be recorded together with an account of any illnesses during this time. Even a brief stay in a tropical country may be sufficient to contract a serious illness such as cerebral malaria or typhus which only becomes manifest after returning home. Immigrants from these regions may arrive with such disorders or more chronic conditions prevalent in their country of origin. Inherited haemoglobin abnormalities are common.

A specific enquiry should be made regarding previous psychiatric illnesses. The necessity to ask more than a few brief questions will often be indicated by the history of the present complaint and the manner in which it was recounted. The patient is often reluctant to discuss past admissions to psychiatric hospitals but will usually admit to a 'nervous breakdown' or suffering from 'nerves'. If discussion of these illnesses causes distress, the subject can be postponed for consideration during the taking of the family history when emotional attitudes often become clearer.

Drugs and other medication

The increasing use of drugs for treatment and the problem of addiction make it essential to ask if any drugs are being taken, how often and by what route. The patient will usually give a description of drugs that have been prescribed but may not know the names, why they are being given or the possible side effects. The drug must be identified; this is often difficult with unmarked white tablets, and the prescribing doctor or chemist should be contacted.

Unless asked, patients often fail to mention the taking of drugs, tonics or patent medicines for constipation, headaches or other minor symptoms. It should be ascertained whether a woman of child-bearing age is taking a contraceptive pill. Note should be made of any drugs recently discontinued as these may still be exerting an effect.

Cards are often issued to patients receiving long-term therapy with anticonvulsants, antidiabetic agents, anticoagulants or steroid hormones.

Addicts often deny or grossly under-estimate their use of drugs but their general appearance and behaviour whilst giving their history often suggests the possibility of addiction.

Family history

In some families there is an increased incidence of certain illnesses as a result of genetic or environmental factors. For this reason the health of relatives and the

cause of any family deaths should be determined. Direct questions should be asked regarding the occurrence of illnesses such as diabetes, rheumatoid arthritis, hypertension, allergic conditions, psychoses or cancer. These disorders do not have a predictable hereditary tendency but there is an increased incidence in certain families. Other diseases such as gout and pernicious anaemia have a recessive hereditary basis so that there is often a family history of the disease. A dominant mode of inheritance is well demonstrated by hereditary spherocytosis and the rare Huntington's chorea. Abnormal genes carried on the X chromosome are responsible for sex-linked conditions such as haemophilia and colour blindness which are transmitted to male members of the family by unaffected females. Recent studies of human lymphocytes have revealed antigenic determinants which are inherited in a Mendelian manner (human lymphocyte antigens, HLA). Certain diseases are associated with an increased frequency of specific determinants. For example, nearly all patients suffering from the rheumatic condition of ankylosing spondylitis have the HLA-B27 antigen, although only a small proportion of the population with this determinant develop the disease. This indicates that other factors are necessary to initiate the illness in these potentially susceptible individuals.

In addition to a full history of the state of health of blood relatives, an account should be obtained of any illnesses occurring in relatives by marriage or others living in the same household. This is particularly important when an infectious disease such as tuberculosis is suspected.

The emotional relationships within the family or home exert a marked influence on the patient's mental and physical health. Tensions or conflict may precipitate a mental disorder or aggravate organic disease. For example, the mother of a family deserted by her husband is likely to develop an anxiety or depressive state; alternatively, this stress may precipitate an organic illness such as peptic ulceration. In contrast, a closely knit, happy family atmosphere provides considerable physical and mental support, minimizing the impact of a serious illness.

Patients are often reluctant to mention emotional difficulties within the family when asked questions such as 'How are things at home?' or 'Have you any family worries?' Often more revealing are oblique questions such as asking a housewife 'Who is looking after your children while you are ill?'

Personal, social and environmental history

At this stage, towards the end of the history, it is more likely that the patient will be willing to discuss his personal habits and attitudes. Questions regarding this aspect of his life follow naturally and interrelate with those concerning the family circumstances.

The framing of personal questions is largely determined by the age, appearance and demeanour of the patient. A pensioner should be asked about his housing conditions, whether he lives alone, the content of his diet and consumption of alcohol or cigarettes. In the case of the teenager or young adult more emphasis will be placed on their relationships and ability to fit into the

community. An attempt should always be made to assess accurately the use of alcohol, cigarettes or drugs.

The patient's attitude to work and the relationship to colleagues and friends will be a guide as to whether he is friendly, reserved or withdrawn. A good inc cation of social instability is the frequent changing of jobs or absenteeism.

The working conditions may be directly responsible for an illness, as in exposure to industrial dust or poisonous chemicals such as weed killers which can damage the lungs, the nervous system or skin. The physical nature of the occupation may lead to conditions such as premature arthritis. Contact with toxic substances can occur in the home by the excessive or careless use of detergents or other household chemicals in the kitchen or garden, or while occupied with personal hobbies such as photography.

Additional questions to the mother of an unwell child

Did you have any infections during pregnancy or take any drugs?

Did vaginal bleeding occur during the pregnancy or were there complications at delivery?

Was the child born at full term and delivered normally?

Was labour unduly prolonged?

Did the infant have breathing difficulties or jaundice?

Was the subsequent progress from birth normal, passing the 'milestones' (walking and talking) at the expected times?

A note should then be made of inoculations and previous illness suffered by the child.

Evaluation of the history

At the completion of the history, the student should have a clear account of the illness from which it should be possible to make a provisional or differential diagnosis. The physical examination will usually confirm this diagnosis or distinguish between the two or three alternatives which may have been considered. In many surgical conditions the history is less helpful and physical examination is of paramount importance in making the diagnosis. Often, further questions are prompted by unexpected physical findings and these may clarify obscure points in the previous history. For this reason the student should realize that the history taking continues during the physical examination.

In addition to making a provisional diagnosis, it is important to know how the patient is reacting emotionally to the illness. This is gauged from the personal, family and social history. Such an assessment is necessary when considering the subsequent management of the patient. Environmental factors may precipitate or aggravate physical or mental illness. Conversely, physical disease itself may provoke an emotional illness in a susceptible subject.

Occasionally there are gross inconsistencies between the patient's complaints and the findings on examination. For example, the obese subject may stead-fastly deny over-eating. This type of discrepancy requires an explanation which

can usually be obtained by going back over the dietary and psychological history. All possible sources of calorie intake should be considered; these include snacks, sweets, cakes, sweetened beverages or alcohol taken in addition to the main meals. The discrepancies may remain despite a minute evaluation of the diet, but a member of the family or another observer may be able to point out inaccuracies in the patient's story. Self-deception of this nature suggests an emotional basis for the obesity and a detailed psychological history is indicated.

The interaction between physical symptoms and mental illness takes various forms. A stoical subject may minimize his serious condition while the anxious or hypochondriacal patient describes minor aches in minute detail. The depressive person may mournfully recite the features of intractable constipation. Completely bizarre complaints or delusions should lead one to suspect a psychosis. An example of this is the schizophrenic who, with complete indifference, describes intense abdominal pain which he regards as the effect of rays from outer space.

The student will learn to recognize the obvious psychiatric disorders. Greater experience is necessary to assess the influence of emotional factors in physical illness and to distinguish these from mental disorders presenting with physical symptoms (psychosomatic illness).

Having made a provisional diagnosis, it is informative to ask the patient what he considers is wrong. This may sometimes explain or confirm that an anxiety state with magnified physical symptoms is due to a fear of cancer.

PHYSICAL EXAMINATION

Introduction

A complete physical examination must be carried out on every patient, although it may be clear from the history that the abnormal signs are likely to be confined to one system. The signs of disease vary from the gross and obvious to the slight abnormality which is often difficult to distinguish from the normal. For this reason the closest attention should be paid to the system which the history indicates is most likely to be affected. Frequently, disease in one system produces secondary changes in other parts of the body which are the only physical indication of the underlying condition.

Methods

There is no ideal method of physical examination but there are two basic approaches. In the first a set routine is adopted starting with the examination of the head and proceeding downwards, whilst in the second the region thought to be affected is examined first followed in turn by the other systems. Both techniques have obvious disadvantages if adhered to rigidly; in the first the systems are examined piecemeal, while the second is time consuming and disconcerting to the patient as he has to uncover the same region several times while each separate system is studied.

At the beginning of his clinical training the student will probably find that examination of each system in turn helps to prevent serious omissions. Later he should evolve a more efficient technique which is easier for the patient and himself. In doing this he must ensure that he does not omit essential parts of the examination.

The scheme we adopt uses part of both methods; thus the head, neck and arms are examined first, followed by the back then the front of the chest and axillae. This order allows the nervous or shy female gradually to accept exposure with minimal embarrassment. When the chest has been completely examined it can be covered. The abdomen is then examined, and attention then turned to the legs. Finally a rectal examination is performed. In certain circumstances this order may need to be changed, as for example in the 'acute abdomen' when this is usually examined first.

The initial impression

Obvious physical signs may be apparent when the patient is first seen or on close observation whilst taking the history. This initial impression may be vital in making the correct diagnosis; for example, the facial immobility of early Parkinsonism or the restless activity of the mild thyrotoxic may be the only physical abnormality. Unless these slight changes are recognized initially, they may be overlooked during the subsequent detailed physical examination. It is vital to be able to recognize the patient who is physically ill and in many instances this is obvious: greater experience is necessary to recognize the early manifestations of physical or mental ill health. The initial physical impression given by the patient should relate to the subsequent story. If not, he has failed to give an accurate account or the history taking has been incomplete. In either case the history must be carefully reviewed.

A scheme of examination

The following method of physical examination is recommended and should be carried out in a warm well-lit room. The scheme is only outlined below but reference is made to the subsequent chapters dealing with the detailed examination of the individual systems.

Before proceeding, the patient should be weighed and asked to produce a urine specimen for routine examination. The temperature, pulse rafe and respiratory rate are usually recorded by the nursing staff. During undressing and weighing, observation may reveal abnormalities such as breathlessness, poor balance or spinal deformity.

Head, neck and arms

The head and neck should be inspected, looking particularly for pulsations, swellings and any skin or conjunctival changes. At the same time the pulse can be felt at the wrist, and subsequently the surface of the hands and forearms examined.

(See also chapters on cardiovascular, skin, central nervous, reticuloendothelial and musculoskeletal systems.)

Mouth

The lips and the inside of the mouth including the mucosa, teeth, tongue, palate, tonsils and pharynx are inspected using a torch and spatula after removing any dentures. The lower part of the pharynx can be seen by depressing the tongue with the spatula and at the same time asking the patient to say 'ah'. This allows the palatal and pharyngeal movements to be observed and if the pharyngeal wall is touched a 'gag' response will indicate a normal reflex. (See also chapters on central nervous, gastrointestinal and reticuloendothelial systems.)

Nose

Deformity of the shape of the nose may be due to collapse or displacement of the nasal cartilage. Clear nasal airways can be confirmed by asking the patient to breathe in through the nose while the examiner occludes each nostril in turn. Deviation of the septum will be seen when looking into the external nares. Discharge, bleeding or a foreign body may be visible. A nasal speculum is used to inspect further inside the nostrils.

(See also chapter on respiratory system.)

Eyes

Generalized redness of the lid (blepharitis) and infected hair follicles (styes) are common. A cyst of a tarsal gland (meibomian cyst) can cause a rounded swelling in the lid. Permanent inversion (entropion) or eversion (ectropion) of the lid may follow long-standing inflammatory fibrosis and lead to soreness and watering of the eye.

The conjunctivae are examined by drawing down each lower lid in turn, when any undue pallor associated with anaemia can be observed. Inflammation of the conjunctiva (conjunctivitis) produces discomfort, redness and often a discharge from the lower lid. A search should always be made for a foreign body which may be lodged beneath the upper evelid or in the cornea. Ulcers of the cornea can be demonstrated by instilling a drop of sterile 1% fluoroscein solution onto the lower conjunctiva. A subconjunctival haemorrhage which spreads across the sclera, sparing the cornea, can result from a blow, but often occurs spontaneously. A bulging of the conjunctiva, sometimes extending over the lower lid, can arise from an accumulation of oedema fluid in the lax space beneath the membrane (chemosis); it appears as 'a tear which never drops'. The fluid may collect when the vascular drainage of the orbit is impaired and the eye is displaced forward (p. 376), or may be part of generalized fluid retention. Localized redness at the junction of the cornea and sclera occurs in conjunctivitis but may indicate inflammation within the eye (iridocyclitis); adhesions of the iris to the cornea or lens cause irregularity of the pupil. Pain and spasm of the eyelids (blepharospasm) develop and are usually progressive in iridocyclitis, contrasting with attacks of intense orbital discomfort often associated with blurred vision occurring in the elderly. This latter disorder is termed glaucoma, a condition in which inadequate drainage of the aqueous fluid leads to circumcorneal congestion and a raised intraocular pressure. The tension is