Health and disease: what can medicine do for philosophy?

J G Scadding  University of London

Author’s abstract

Philosophical discussions about health and disease often refer to a ‘medical model’ of bodily disease, in which diseases are regarded as causes of illness; diagnosis consists in identifying the disease affecting the patient, and this determines the appropriate treatment. This view is plausible only for diseases whose cause is known, though even in such instances the disease is the effect on the affected person, and must not be confused with its own cause. But in fact the medical diagnostic process which progresses from recognition of patterns of symptoms and signs, through search for abnormalities of structure and/or function, towards knowledge of causation often stops short of this desirable end-point; and at whatever point it comes to a halt, its result is expressed in terms of ‘diseases’. Thus in medical discourse the names of diseases are a convenient device by which the current conclusion of the diagnostic process can be stated briefly; and they have widely varying factual implications. This nominalist analysis of the medical usages of the names of diseases has consequences for definitions of health and disease, and for some problems in medical ethics.

Reading Professor Hare’s paper (1) based on his John Locke Lecture entitled What can philosophy do for medicine? , I was reminded of the comment made by a non-medical participant at an early conference on the application of computers to medical diagnosis that until the physician is willing to investigate his own terminology, all the mathematician can do is to stand in the wings and help out in minor ways (2). This stimulated me in 1967 to set out in terms which I hoped would be comprehensible to a non-medical enquirer the results of an analysis of the meaning of medical diagnostic statements with which as a practising clinician, mainly in the field of respiratory disease, I had been concerned for many years (3). Some psychiatrists have found it relevant to their special problems of nosology (4,5,6). Their interest in an analysis of the semantic problems of medical diagnosis, undertaken to clarify discussion of some respiratory diseases, encourages me to think that it may be relevant to some of the ethical problems in relation both to individual patients and to society which today are widely discussed. I therefore venture to outline this analysis, and then to comment on its possible relevance to these problems.

The heterogeneity of ‘diseases’

Consideration of the usages of the names of diseases in medical discourse shows that they have widely differing factual implications. The simple idea that diseases are causes of illness and that diagnosis consists in identifying the disease which is causing the patient’s illness is obviously erroneous, although it is implicit in colloquial usage. I will illustrate this by a series of hypothetical scenarios, all starting with a patient consulting a physician because he has a persistent cough.

1. The cough produces mucoid phlegm, and there are episodes of more severe symptoms after upper respiratory infections. The physician diagnoses chronic bronchitis.

   The patient may take this to mean that his cough is caused by chronic bronchitis.

   The physician should know that at present the most generally accepted definition of chronic bronchitis is in terms of clinical description, and thus it is absurd to regard this disease as the cause of the symptoms by which it is defined. He should be considering possible causal factors. The patient is likely to be a smoker; if so, the physician will probably tell him that cigarette smoking is the principal cause of his symptoms.

2. Because the cough produces persistently yellow phlegm, further investigation is undertaken, and this shows that there is localised dilatation of some bronchi, leading to a diagnosis of bronchiectasis.

   The patient is likely to think that this impressively-named demon is the real cause of his symptoms.

   The physician should know that in saying ‘bronchiectasis’ he has only said ‘bronchial dilatation’ in Greek. In a few cases this is a consequence of a past episode of acute broncho-pulmonary inflammation from which the patient made an otherwise complete recovery, but in most it appears during the course of chronic broncho-pulmonary disease of various sorts;
and the mechanisms which lead to dilatation of the bronchi are diverse. In themselves, dilated bronchi cause no symptoms; the contribution of infective and vascular changes in them to the production of symptoms varies greatly from case to case, and should be individually assessed. Thus, a diagnosis of bronchiectasis is a claim to knowledge that the patient has dilated bronchi, with the implication that at least some of the symptoms are due to changes in them. It conveys no necessary implication about the pathogenesis of this anatomical abnormality; if anything is known about this, a further diagnostic term is required to specify it.

3. The patient complains of episodes of shortness of breath and wheeze as well as cough, and tests of the ventilatory function of the lungs show wide variations in expiratory airflow resistance. This leads to a diagnosis of asthma.

Again, the patient will probably think that the cause of his symptoms has been identified.

The physician should know that in applying the diagnostic term ‘asthma’ he is claiming no more than that the wheezy breathlessness is due to wide variations in resistance to expiratory air-flow in the lungs. He may be able to show that the patient’s bronchi are abnormally reactive to a variety of stimuli, but can only speculate about the causes of this abnormal reactivity. He will try to identify factors causing increases in resistance. In many instances, he will fail. If he succeeds, he may qualify ‘asthma’ by a term indicative of this factor. Thus in making an unqualified diagnosis of asthma, he is categorising the patient’s case in a group defined by a specified abnormality of function.

4. Investigation of the coughing patient includes a chest x-ray, which is abnormal, and tubercle bacilli are found in the sputum. The physician makes a diagnosis of pulmonary tuberculosis.

The patient will probably think of tuberculosis as the cause of his illness.

For the physician, the diagnosis of tuberculosis is a claim to knowledge that mycobacterial infection is the necessary cause of the abnormalities which he has found in the patient’s body, and which he believes to be the cause of the symptoms; in current nosology, ‘tuberculosis’ refers to the effects of infection by mycobacteria on an animal host, although these effects are greatly dependent upon factors of host resistance (see appendix 1). Thus, he must not confuse the causal agent, the tubercle bacillus, with its effect, tuberculosis.

Classes of defining characteristics for primary diagnostic categories

These examples illustrate the four main classes of characteristics by which diseases can now be defined. I have chosen them from the respiratory field, in which I first became concerned with semantic problems in medicine (7,8,9,10), but the analysis is generally applicable. In the order in which, historically, they entered medical practice, they are:

i. CLINICAL DESCRIPTION (SYNDROME)

Initially, diseases could be defined only by description. Patients with a recognisably similar pattern of symptoms and signs were said to be suffering from the same disease. A recognisable pattern of this sort is called a syndrome; the relationship between ‘disease’ and ‘syndrome’ is discussed below. Study of a group of patients recognised in this way will be directed towards discovery of any distinctive changes in structure or function and elucidation of causation. But even when causation is not known, and anatomical and functional changes are inconstant or not distinctive, clinical study will lead to knowledge of prognosis and may discover ways to ameliorate symptoms and possibly to improve prognosis. Thus, it is useful from the beginning to have a name by which to refer to the phenomena displayed by a clinically recognisable group of patients; and this is adopted as the name of a disease.

Most psychiatric diagnostic categories are still syndromally defined, and some categories definable only in this way remain in current use in non-psychiatric medicine. For example, migraine refers to a sort of recurrent headache whose recognition is important in several respects. The headaches are distressing and may interfere with normal activities; and while migraine is not dangerous to life, headaches of similar character may be caused by serious organic changes in the brain, so that a positive diagnosis of migraine is important. The description of the disease ‘migraine’ is based on the study of individuals who have recurrent headaches conforming to a carefully defined pattern, and in whom no structural abnormality of the nervous system is found. It includes such facts as the proportions of cases in which symptoms such as visual disturbances and vomiting precede or follow the headaches, the age- and sex-incidence, the responses to various medications, and an account of evidence relevant to precipitating factors and to possible vascular mechanisms underlying the symptoms; but these findings, being inconstant, have no place in the definition.

ii. DISORDER OF STRUCTURE (MORBID ANATOMY)

Histologically, when a disease defined by clinical description was found to be associated constantly with a recognisable morbid-anatomical change, it tended to be re-defined, implicitly or explicitly, and possibly renamed in these terms; and with the development of microscopy, in terms of microscopic structure (histology). As examples of diseases defined in these ways in current nosology, mitral stenosis and cirrhosis of the liver may be quoted. The description of each of these diseases is based on the study of patients in whom it has been established that the relevant abnormality of structure is present, and is an account of all abnormalities found to be associated with it. If the same word is used to name both the structural abnormality and the disease, it is important to distinguish carefully between these two usages; as the
name of a disease, 'mitral stenosis' refers, not to the narrowing of the mitral valve alone, but also to the symptoms and signs and changes in other organs that may be consequent upon it.

iii. DISORDER OF FUNCTION (PATHOPHYSIOLOGY)

Similarly, when a specifiable disorder of function was found to be associated with a recognisable clinical syndrome, it was likely to be used as the defining characteristic for an appropriately named diagnostic category. Many examples can be found in current nosology. They range from broad categories, such as hypertension, to deficient or excessive secretion of a single hormone by an endocrine gland. Again, it is important to distinguish between the connotations of a word when used to specify a disorder of function and when used to refer to the disease which is the consequence of this disorder; 'hypothyroidism' as the name of a disease refers, not to deficient secretion of thyroid hormones alone, but also to the consequences of this deficiency.

Diagnosis of a disease defined in morbid-anatomical or pathophysiological terms carries no necessary implications about the causation of the underlying abnormality of structure or function.

iv. CAUSATION (AETIOLOGY)

When the cause of a disease becomes known, the disease is generally re-defined, explicitly or implicitly, in causal terms. Tuberculosis, already mentioned, is an example of this. In medical discourse, 'influenza' should be used to refer only to the disease caused by an influenza virus, notwithstanding its general colloquial use to refer to any short-term febrile respiratory illness presumed to be caused by one of many possible viral and other agents. Although the common infectious fevers are nearly always diagnosed confidently on clinical grounds alone, the ultimate criterion of the correctness of the diagnosis for those whose causal agent has been identified is evidence of infection by this agent, and thus they should now be defined as the diseases caused by the relevant agents.

Groups ii, iii, and iv can be subdivided in an increasing number of ways. For instance, abnormalities of chromosome structure or number now define some genetically definable disorders, such as sub-types of Down's syndrome; biochemical abnormalities, possibly linked to single gene defects, now define some inherited diseases, previously syndromally-defined; and deficiencies of vitamins and other nutritional factors can be included in group iv as causal defining characteristics of diseases.

The advance of medical knowledge is, in general, towards causation; it often leads to re-definition of diseases, disorders of structure or of function displacing syndrome, and causation in turn displacing these as defining characteristics. It is thus inevitable that diagnostic terms of these various sorts co-exist in current nosology. Unless the differing factual implications of the names of diseases defined in these various ways are recognised, confusion is inevitable.

Another possible source of confusion is failure to recognise that when the basis of definition of a disease is changed, the set of patients specified by the new definition to provide the description of the disease is unlikely to be identical with that specified by the old; in other words, the disease defined on the new basis may not be identical with that defined on the old. For instance, myxoedema, defined syndromally, is not the same as hypothyroidism, defined functionally. And tuberculosis provides an excellent example of the effects of changes in defining characteristics with advances in knowledge (see appendix 1).

Simple and compound diagnostic categories

Another complication is that a disease may be defined by characteristics derived from more than one field of study. Such a disease should be regarded as the intersection of the sets constituted by two or more primary or simple categories, constituting a compound diagnostic category. This may be made obvious by the name used for it; for instance, pneumococcal pneumonia is the intersection of aetiologically and morbid-anatomically defined sets. It may be less obvious when an old-established name conceals the complexity of the definition entailed by current knowledge. For example, the definition of pernicious anaemia, originally described by Addison as a clinical syndrome, now requires elements referring to the abnormal morphology of the red blood cells and their precursors (meagloblastic anaemia) and to the functional disorder which leads to it (defective absorption of vitamin B12 due to gastric intrinsic factor deficiency).

A note on definitions in scientific contexts

In the foregoing analysis of current nosology, I have sought to make explicit the factual implications of medical usages of the names of diseases. This analysis is an essential preliminary to definitions of diseases in what Popper (11) calls a methodologically nominalist as opposed to an essentialist manner. Nominalist definitions do not attempt the impossible task of revealing the essence of the definiendum, but state how words or other symbols are to be related to observable phenomena. Popper points out that essentialist definitions, depending upon intuitive acceptance for their validity, have no place in science; although he claims that science could progress without definitions of any sort, he admits that this would be at the cost of excessive verbosity, and accepts nominalist definitions as convenient shorthand symbols. Indeed, it is difficult to see how problems in physics could be discussed without the universal acceptance of nominalist definitions of its concepts in terms of the dimensions length, mass, and time. Similarly, I do not see how a disease-terminology can be used in scientific discussion of medical problems without nominalist definitions of diseases. The name of a disease, as properly used in medical discourse, is a convenient
brief statement of the current conclusion of a
diagnostic process which for each patient follows the
general direction of the historical advance of medicine
from recognition of syndrome towards knowledge of
causation but often stops short of this desirable end-
point.

The medical concept 'a disease'

Is the concept 'a disease' necessary in medical
discourse? Important problems may be discussed, and
decisions may be made, especially in emergency
situations, without use of the disease terminology.
Although its results are expressed in this terminology,
one of the most successful applications of computing to
diagnosis, that concerned with acute abdominal pain,
is in effect an aid to the decision whether to operate or
not (12). Moreover, opinion among doctors about
whether words in common use for conditions which
might lead to medical consultation refer to diseases
varies, though less than among laymen (13). But
though it seems possible that a medical language which
dispensed with the names of diseases might be
elaborated, the prospect of this must be remote, and it
is certain that for the foreseeable future the concept 'a
disease' will persist in medical as well as in colloquial
discourse. It is therefore essential that the implications
of medical usages of the names of diseases should be
made explicit. The following statement attempts to do
this, in terms which allow it to be applied in all
biological contexts:

In medical discourse, the name of a disease refers to the
sum of the abnormal phenomena displayed by a group
of living organisms in association with a specified
common characteristic or set of characteristics by
which they differ from the norm of their species in such
a way as to place them at a biological disadvantage.

This general statement recognises the logical
heterogeneity of the concept 'a disease' in medical
contexts. If it is accepted, a particular disease can be
defined by specification of the common feature which
characterises the group upon the study of which its
description is based. This defining characteristic may
be of several sorts: descriptive or syndromal, morbid-
anatomical, pathophysiological, and aetiological. As
knowledge advances, syndrome tends to be displaced
as the basis of definition by more objective features,
among which, when possible, aetiology takes precedence.

Qualitative and quantitative elements in
definition

The first step in definition is qualitative. What sort of
deviation from the norm specifies the group the study of
which leads to the description of the disease? As
noted above, choice of this will depend upon the
current state of knowledge. When this choice has been
made, quantitative terms for practical application of
the definition are required. The norm for the selected
characteristic must be ascertained, and then the
magnitude of the deviation from it that will be accepted
as significant must be decided.

The difficulty of these steps varies with the nature of
the defining characteristic. It is least for diseases
defined aetiologically, for which the defining
characteristic is categorical; though even here decision
may be required about the magnitude of effects of the
causal factor that will be regarded as indicative of
disease. For diseases defined in terms of abnormal
structure or function, dimensional factors are required
also in the specification of the defining characteristic.
In scientific work, specification of these factors may
require appropriate statistical treatment of data
derived from study of relevant populations; in clinical
practice, simple criteria, such as association with
relevant symptoms and signs, usually suffice.

Factual and evaluative elements

In this discussion, I shall use 'factual' to mean capable
of being stated in objectively demonstrable terms,
preferring it to 'descriptive', because I have used the
phrase 'clinical description' and wish to consider how
far clinical description can be objective. 'Evaluative'
will be taken to mean involving value judgements; in
these, things are assessed on a scale of goodness and
badness, for which criteria are required.

I distinguish between two sorts of evaluative criteria
objective, which can be stated in terms of factual
comparisons with specified norms available to general
inspection, and in principle quantifiable; and
subjective, which cannot be stated in such terms.

The general statement seeks to maximise the factual
content in definitions of diseases for use in scientific
contexts, eliminating elements that are not objectively
demonstrable wherever possible. How far does it
succeed? This question can be considered in relation to
two elements in the statement: 1) defining
characteristics and 2) biological disadvantage.

1) Defining characteristics can be stated factually for
all diseases defined aetiologically or by disorders of
function or structure and for those defined
syndromally in which the description of the syndrome
includes objectively observable elements. For some of
those defined syndromally, symptoms constitute the
principal part of the defining characteristics; these
include many in the field of psychiatry, especially the
common affective disorders. With this exception,
which is discussed below, the demonstration of
defining characteristics does not involve value
judgements.

2) The proviso 'in such a way as to place them at a
biological disadvantage', which is required to
distinguish the sort of deviation from the norm that
will be accepted as indicative of disease from one that is
regarded as harmless, is evidently evaluative, but can
in principle be applied objectively in general biological
contexts. Assessment of disadvantage requires
comparison of those having the defining characteristic
with members of the same species not showing it and
living in a similar environment; if quantifiable criteria
can be specified, this comparison will become factual. In human medicine, disadvantage in terms of inability to undertake normal activities or threat to life is obvious for most 'organic' diseases. In other instances, agreement about objectively demonstrable criteria by which disadvantage may be assessed should resolve controversies about whether a specified group should be said to be suffering from a disease.

The assessment of 'disadvantage' in relation to the whole group having the defining characteristic rather than to the individual resolves several problems:

(a) It avoids quibbles based on contingent circumstances of individuals, such as the soldier for whom a skin disease may diminish the risk of death in battle.

(b) It makes it permissible to say that someone has a symptomless disease. Most people over middle-age have symptomless atherosclerosis. This is properly regarded as a disease, since as a group those with it have a shorter life expectancy than those who do not, although many individuals will suffer no evident ill-effects from it and live to die of some other disease.

(c) It clarifies discussion of some more general questions, such as whether heterozygosity for the sickle-cell gene should be considered a disease in areas where falciparum malaria is endemic. Homozygotes, carrying this gene on both chromosomes of pair 11, suffer severe symptoms from anaemia and a variety of changes in internal organs, leading to early death. Heterozygotes, carrying it on only one of this pair, do not suffer these ill-effects, but they differ from individuals not carrying it in two respects. They suffer from the disadvantage that if they mate with another heterozygote one in four of their offspring will suffer from sickle-cell anaemia; on the other hand, they have the advantage of being somewhat more resistant to the ill-effects of infection with P falciparum. The balance between these can be discussed for the temporal and local circumstances.

I conclude that the general statement makes it possible to define most diseases in current nosology in factual terms, without subjective value judgements. This may not be possible for some psychiatric disorders currently definable only by symptoms; but in such instances, as discussed below, advances in knowledge may reduce or eliminate the subjective element.

Some consequences of the general definition

The general statement originated from an analysis of usages of the names of diseases whose purpose was to clarify medical discourse. It evidently has wider implications. Space permits only brief mention of these and some of them lead into fields in which I can claim no special expertise.

SYNDROME, DISEASE, 'CLINICAL ENTITY'

Questions such as 'Disease or syndrome?' and 'Is this a clinical entity?' are in effect requests for statement of the current basis of definition of a category. They probably arise from a feeling that a disease ought to be definable eventually, even if not now, in causal terms. But, as has been shown, the names of diseases defined in other ways are current in medical discourse, which without them would be intolerably prolix; and with due care they can be used clearly. One can speak of the syndrome of any disease with which a consistently recognisable pattern of symptoms and signs is associated. Whether a category definable only in clinical-descriptive terms is called a syndrome or a disease does not matter, provided that verbal usages are made explicit and applied consistently. 'Clinical entity' always needs ad hoc explanation; it often seems to be the refuge of one who has not succeeded in clarifying his or her thoughts, but is nevertheless determined to put them into words.

PSYCHIATRIC NOSOLOGY

Most psychiatric diagnostic categories, especially the affective disorders that are so prevalent, are definable only in syndromal terms. I suggest that there are two main causes for dissonance about the validity of the concept of mental illness (14).

One is doubt about the possibility of an agreed objective basis for the elucidation of the symptoms which constitute the defining characteristics of such syndromes. The only evidence may be what the patient and those around him or her say about feelings and behaviour. Interpretation of some of this evidence requires value judgements for which it may be difficult to specify objectively demonstrable criteria. It is legitimate to hope that appropriate community studies may resolve this problem (15).

The other is failure to appreciate that in psychiatry, as in medicine generally, recognition of a syndromally defined category is to be regarded as the starting-point for advance towards its supersession by one or more categories defined in more precise terms, such as specific causal factors or disorders of function or of structure (6).

'HOLISTIC MEDICINE'

The general statement underlines the fact that diseases have no existence apart from that of the patients who are said to suffer from them. Acceptance of this makes medicine necessarily holistic. It is the patient whom we treat, not the disease. When we speak of the treatment of a disease, we must recognise that this phrase is an ellipsis for treatment of patients with that disease.

DIAGNOSIS AND TREATMENT

The general statement aims to maximise the factual content of medical diagnosis, minimising the need for value judgements, and providing a framework within which objective criteria for such judgements can be specified. Decisions about treatment, on the other hand, necessarily involve value judgements. The idea that medical practice proceeds from diagnosis to the treatment specified for the diagnosed disease by
textbooks and other authorities is foolishly simplistic. 'Treatment' refers to a wide range of actions with differing objectives, and the possibilities vary according to the basis of definition of different diseases. For all patients, explanation, supportive advice, and measures for relief of symptoms are possible; for those with disorders of structure or of function, correction or amelioration may be possible; and for those with diseases of known cause, it may be possible to remove or neutralise the causal factor. The doctor needs factual knowledge of these possibilities, and of the risks that some of them carry. The application of this knowledge will be affected to varying degrees by the circumstances and feelings of the patient, and thus calls for value judgements of varying difficulty. The old aphorism 'Guérir quelquefois, soulager souvent, consoler toujours' recognises both the varying possibilities that may be available, and the duty to apply them to the greatest benefit of the patient.

Thus the realities of medical practice are far removed from the pattern underlying Hare's argument about mental illness (16). All arguments based upon a supposed 'medical model of disease' are suspect, since current nosology includes diseases defined by criteria of several different sorts, having different factual implications.

THE DEFINITION OF HEALTH

Professor Hare suggests that the concept of health is necessarily evaluative. He criticises Boorse's (17) attempt to define it in entirely descriptive terms.

As a physician, I am content to define health as the absence of disease. In this, I am in apparent agreement with both Boorse and Hare. I agree also with Boorse's trenchant criticism of concepts of 'positive health'. If someone asks 'Am I healthy?', all the doctor can do is to seek for evidences of known diseases and for significant deviations of structure and function from expected norms, and reassure the enquirer if he finds none. The evaluative element in 'health', so defined, is thus similar to that in definitions of diseases, from which, with the exception of some syndromally defined categories in psychiatry, the general statement permits evaluation not based on objectively demonstrable criteria to be eliminated.

Boorse seeks to exclude subjective value judgements from concepts of health and disease by defining diseases as internal states that depress functional activity below species-typical levels; health and disease are related to norms which can be established by appropriate statistical studies. While I am in sympathy with his aim to make the concepts of health and disease, certainly for use in scientific contexts, as free as possible from subjective value judgements, I am doubtful about several points in his analysis. As criteria distinguishing disease from health, depression of functional activity, which Boorse adopts, and biological disadvantage, used in the general statement, can both be applied objectively. But I think that Boorse's definition gives undue prominence to functional deficit as a defining characteristic of diseases, and thus conflicts with current nosology, in which diseases are defined by several sorts of characteristics, among which aetiology takes precedence. For this reason, I prefer the wider criterion of biological disadvantage to distinguish deviations from the norm that should be accepted as defining characteristics of diseases. The identification of disease with an 'internal state' is confusing. This state is presumably regarded as the explanation of the phenomena constituting a disease; but at the same time, it is itself implied from these phenomena. In fact, the level to which we can take the explanatory process varies for different diseases; a state of affairs reflected in the general statement, which allows the names of diseases to have different sorts of causal implication.

The general statement about the proper use of the names of diseases is intended to make it possible to express the end-point of the diagnostic process without subjective value judgements; the judgement implicit in saying that observed phenomena constitute a disease is related to biological disadvantage, which can, in principle, be assessed objectively. I think that it can be applied successfully in most medical contexts, though difficulties persist for some syndromally defined psychiatric categories.

It is a legitimate aim of medical science to free medical concepts of disease, and consequently of health, from value judgements not based on objectively demonstrable and quantifiable criteria. On the other hand, decisions about the management of individual patients, including not only therapeutic but also complex investigative procedures, involve value judgements of varying difficulty, into which subjective elements necessarily enter.

Appendix 1

TUBERCULOSIS AS A MODEL FOR CHANGING MEDICAL CONCEPTS

Until 1882, patients who would now be said to be suffering from tuberculosis were placed in a number of different diagnostic categories; and diverse views were held about the causation of these and about their relationship, if any, to each other. Initially, they were described and defined in clinical-descriptive terms. Pulmonary consumption or phthisis was known from ancient times and named after the characteristic wasting with symptoms, such as cough and blood-spitting, referable to the lungs. Scrofula was recognised by chronic swelling of lymph-nodes in the neck; it was at one time called the King's Evil, because it was thought to be curable by the royal touch, providing an example, perhaps unique, of a disease named after its supposed cure. Caries of the spine was recognised by the deformity which it caused; its association with palsy of the legs was noted by Hippocrates, and fully described by Pott in 1779. *Tabes mesenterica* was recognised by swelling of the abdomen by enlarged
mesenteric lymph-nodes and wasting. There was uncertainty and controversy about the causation of these syndromically-defined diseases and about the relationship between them.

The first objective evidence of a relationship between them was morbid-anatomical. From the middle of the 17th century onwards small pale nodules, which were named tubercles, were recognised in affected tissues, and later these were found to have a characteristic microscopic appearance. By the early 19th century, this led to the possibility of re-definition in morbid-anatomical terms, with the new name ‘tuberculosis’, first suggested by Schönlein in 1839. But this unification was not generally accepted; the great Virchow at first denied that phthisis and scrofula should be regarded as tuberculous and opposed the idea that _lupus vulgaris_, later shown to be a form of tuberculosis of the skin, was related to scrofula.

Views about their causation varied from attribution to evil spirits through the humoral theories of Hippocrates and of ancient Hindu writers and the Galenical theory of ulceration of the lungs by ‘pituita’ dropping from the head, to constitutional predisposition. Communicability by contagion was denied by some and asserted by others. There was no objective way of resolving these controversies until Villemin in 1867 showed experimentally that tuberculosis was communicable, and Koch in 1882 demonstrated in histologically tuberculous tissues a bacillus which could cause similar changes in artificially infected animals. After this discovery, the disease ‘tuberculosis’ could be re-defined in aetiological terms. This led to the unification of a number of diseases previously defined syndromally or morbid-anatomically into a single aetiologically defined category. At the same time, some patients who would in the past have been said to be suffering from the syndromally-defined diseases phthisis and scrofula were excluded from this new category when no evidence of infection with Koch’s bacilli could be found in them. Later, the complex natural history of this infection was elucidated; it was found that many people became infected but only some showed evidence of disease of varied severity, site and outcome.

The story is not ended, for there is still argument about the definition of tuberculosis. There are two views. One would limit ‘tuberculosis’ to disease caused by one species of mycobacterium, that which causes the form of morbid-anatomically defined tuberculosis most frequent in man; this organism was originally called the human tubercle bacillus but is now named _Mycobacterium tuberculosis_. This definition leaves in limbo cases of disease that pathologists recognise as tuberculosis but which are found to be caused by another species of mycobacterium. The other is that tuberculosis should be recognised as a compound diagnostic category, defined in both morbid-anatomical and aetiological terms; full diagnostic statement would include specification of the causal mycobacterium, though it might be accepted as a convention that when this was not stated the organism had been shown or was assumed to be the human tubercle bacillus (18).

The history of tuberculosis thus illustrates how the definitions of diseases, as used in medical discourse, have changed and continue to change with advances from recognition by clinical description to knowledge of causation. It also emphasises the importance of recognising that a disease defined causally is the effect of a complex interaction between the causal agent and the affected individual.

J G Scadding MD FRCP is Emeritus Professor of Medicine at the University of London and is Honorary Consultant Physician, Brompton Hospital, and Hammersmith Hospital, London.

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