circumference measurements. Therefore hip measurements (and hence W/H) should be included in every medical examination for life assurance.

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EARLY DIAGNOSIS, SCREENING AND TRUTH-TELLING: SOONER MAY NOT BE BETTER

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The clinical consultation is a meeting of patient and doctor to assess the significance of symptoms, signs, and investigations; usually in that order. In it emphasis is on detection of organic disease. This paper will not consider primary psychiatric disorders.

What is meant by early diagnosis? The aim of diagnosis is to categorise diseases. In some respects this follows the classification of plants introduced by Linnaeus but it is more complex and not based solely on appearance. It may depend on pattern recognition, analysis of the findings, or a specific test. In clinical diagnosis there may be a bias towards recognition of conditions which are known to respond to treatment.

Early diagnosis introduces a time-scale and this is determined by the natural history of the disease under consideration. For acute illnesses such as infections or myocardial infarction diagnosis within a few hours may be crucial in controlling the disease. In chronic conditions such as rheumatoid disease diagnosis is less urgent and might be considered early if within three months of the onset of symptoms.

Screening involves a search for disease in the absence of symptoms. It emphasises the fact that all disease is initially latent. Screening may be done at random or restricted to people who show a predisposition to a disease as revealed by the earlier medical or family history.

SCREENING

Screening is carried out on volunteers and it is important that its purpose is adequately explained and the possible consequences considered.

Screening when there is predisposition

When there is known to be a predisposition to disease it would be advantageous to have a diagnostic test which could be used at a stage when the patient is asymptomatic. An example of this would be the level of creatine phosphokinase in blood which is frequently raised in patients with impending muscular dystrophy. A more recent development is the molecular test on blood or other tissues used to diagnose Huntington's chorea.[1] This autosomal dominant disorder usually develops in middle age and there is no effective therapy. The test may reassure people in whom the result is negative but it creates problems for those found to have latent disease. There is a need for counselling and there may be difficulties within a family. For example, a subject may not wish to know the result of the test while his spouse is eager to have this information.[2] Huntington's chorea is one of the most clearly defined genetic disorders. Increasing knowledge

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of the genome will confirm genetic predisposition to a variety of disorders most of which are more benign than Huntington's chorea. An abnormal test does not tell us when the disease may develop in the individual or predict its clinical course.

Antenatal screening is widely used to detect foetal abnormality during pregnancy. It may be ultrasound to show structural changes, or invasive methods such as the examination of amniotic fluid. The consequences of error are particularly serious and distressing. A false positive result may lead to abortion of a normal infant and a false negative result to the birth of a severely handicapped child. The demand for antenatal screening is increasing. While normal findings may give reassurance the detection of serious abnormality raises the question whether the pregnancy should be terminated. It is for this reason that the patient should be made aware of the possible implications of the test before it is done. Some patients respond by deciding not to have the test.

Routine screening

Other types of screening utilise radiology or laboratory study of tissues or secretions. They include chest radiography to detect pulmonary tuberculosis and carcinoma of the lung, mammography for breast carcinoma, and cervical smears for carcinoma of the uterine cervix. Polyposis of the colon predisposes to carcinoma and can be detected by endoscopy. Phenylketonuria, a rare metabolic disorder, can be detected at birth by a simple chemical test on blood or urine and then controlled by a special diet.

Various difficulties both practical and ethical arise in the course of screening.[3,4] Routine screening can only be justified when there is both a reliable diagnostic test for the disease and effective treatment. Screening is expensive and also demanding of human resources particularly when large numbers of negative tests have to be reviewed using microscopy. Reliability and reproducibility are important when screening tests are used. There is some risk from implicit belief and acceptance of the result of tests. Tests may be negative and lead to false reassurance when disease is present or show abnormality when there is no disease (false positive). Intensive screening may be a cause of anxiety and may reveal features which cannot be accurately defined. This, in turn, may lead to trials of therapy with all the associated risks.

These difficulties emphasise the need to explain to the patient the nature of the tests and their possible implication. Counselling also gives an opportunity to make it clear that tests are not infallible and that probability enters into diagnosis.

What should the patient be told? The direct response to this is that respect for autonomy demands that any findings should be reported. This is helpful to the patient when the outcome is clear. When findings are equivocal it may cause anxiety which is impossible to resolve. A judgement is therefore needed as to whether the patient really wants to be told all the details or would be more content with an edited version. There are possible repercussions. A more definite diagnosis may emerge later and if it is then revealed that earlier findings have been suppressed the patient may bear a grudge or seek litigation. Others would defend the position on the grounds that professional responsibility implies that not all doubts need be shared with the patient. This latter approach merges imperceptably into paternalism. But some patients prefer to be advised rather than given all the facts.

EARLY CLINICAL DIAGNOSIS

Early diagnosis presents difficulties because symptoms may be vague, signs of disease may be absent or equivocal, and the need for investigation may not be apparent. The doctor is required to decide how far to pursue the enquiry knowing that blind investigation is often unrewarding and demands resources. A further and increasing factor in a consumer orientated society is the demand of the patient for particular tests to be done.

Attempts to rationalise the process of diagnosis can help to offset bias, clarify thinking, and increase precision. Elaborate methods are not needed in the simpler problems of diagnosis such as measles. In more complicated cases Bayes theorem[5] may improve accuracy as has been shown for abdominal pain.[6] This is based on mathematical computation of probabilities determined by the local incidence of the disease and the frequency of a particular symptom in that disease. However, it seems unlikely that this method will find wide application.

Delay in diagnosis is not always due to difficulties with medical assessment. It is common for patients to delay consultation. They may postpone it because of difficulty in getting time off work without due explanation, or distance from appropriate help. More commonly the patient hesitates because of self-denial ('it couldn't happen to me'), hope that the condition will improve with time, fear of the unknown or that serious disease may be revealed, or a general reluctance to face up to the situation. The causes of a patient delaying consultation are difficult to control. They may be helped by dissemination of knowledge about disease. Sometimes this has the unwanted effect of increasing anxiety. It may also add to the considerable difficulty of excluding disease.

Two clinical examples

The conventional primary treatment of carcinoma of the breast is surgical excision of the tumour. This might be supplemented by radiotherapy or chemotherapy. In the absence of other signs at the time of surgical removal it cannot be known whether the tumour has spread but early latent metastasis is common. If it has spread local removal is not likely to be effective and the outcome with other treatments is hard to predict. The diagnosis may be made by mammography before any lump is palpable and the hope is that this earlier diagnosis will increase the chance of eradicating the disease. That remains to be demonstrated[7] and several regimes of treatment are undergoing trial.[8]

Primary cerebral tumour presents more difficulty in diagnosis. Computerised scanning is of considerable help but may be normal in the earliest stages. The major problem is that most of these tumours are gliomas and they cannot be completely excised because of wide infiltration and the inevitable loss of brain tissue. The nature of the tumour may be suggested by radiological findings but confirmation is by biopsy. Plainly the early diagnosis of a glioma which may evolve over several years is of little help to the patient.[9] By contrast to this the early recognition of a benign tumour such as a meningioma may be followed by excision and complete relief. Control of cerebral tumours not amenable to excision is more elusive—the technology of diagnosis has outstripped available therapy.

Work on the human genome indicates that manipulation is possible and it has been applied to somatic cells.[10] Genetic manipulation of germ cells will inevitably follow but for the present a general moratorium has been agreed because of its

far reaching implications. Malignant disease may also be influenced by genetic structure.

ESTABLISHED DISEASE

Diagnosis usually becomes easier as time passes but in some patients certain diagnosis remains elusive. Difficulty arises in the case of chronic diseases which evolve slowly. Multiple sclerosis is an example.

The patient usually presents with weakness of a limb, numbness, or loss of vision in one eye (optic neuritis) and these symptoms clear in the course of a few weeks. There may be other signs of disease. When these are absent the evidence is insufficient for a firm diagnosis of multiple sclerosis and the features could be due to other disease.

Remission may continue for months or years before other features develop and the outlook is unpredictable. Evidence of more extensive disease may be obtained from magnetic resonance scanning of the brain or spinal cord.

In the situation described, when no definite diagnosis has been reached what should the patient be told? While there is no therapy that will influence the course of the disease the diagnosis is not urgent. The patient is often told that there has been an episode of inflammation of nerves. Although probably true this statement does not help to suggest the prognosis. Reassurance and subsequent remission of the symptoms will satisfy some people. If the possibility of early multiple sclerosis is discussed some patients become anxious and introspective. They may continue seeking further evidence or react by adopting the sick role when otherwise healthy.

If the possible nature of the event is concealed there may be repercussions later. For example, a patient with a mild episode of disease who recovers may conclude that that is the end of the matter. When, after subsequent marriage, there is progression of the disease and it is then revealed that the onset was signalled some time before, the spouse may feel cheated. There can be no universal answer. The information conveyed to the patient must depend on his attitude, whether he wants to have all the details or is content with an inconclusive statement.

When the diagnosis is definite a judgment may still be needed as to whether the patient wants to know or not.

CONCLUSION

Early diagnosis is theoretically desirable and opens up the possibility of effective control of the disease. In the real life clinical situation the benefits are often less clear.

Diagnosis may be delayed because the patient defers consultation, the waiting list is long, the significance of symptoms is not appreciated, or the disease remains latent. Techniques of decision-making may help clinical consultation and screening has reached the molecular level in the study of the genome. Diagnostic logic is not alone sufficient because moral factors enter at every stage and have to be considered in relation to personal feelings, emotional reaction, social setting, and the availability of resources. Early diagnosis removes some of the mystery but has little to offer a patient with a progressive condition, such as motor neurone disease, which is not amenable to treatment. A high standard of clinical expertise is essential. Early diagnosis is important in progress towards advance in rational treatment. Its value depends on the nature of the disease, effectiveness of available

therapy, resources, the cultural environment and attitude of the person affected. The art of satisfactory practice consists in balancing these various considerations while respecting the autonomy of the patient.

Early diagnosis may not help the patient but it is important in trials of new treatment. In the broadest sense all therapy is experimental but new treatment brings extra risk. There is therefore a conflict between the interest of the individual patient and the trial of new drugs.

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