Making a diagnosis in primary care: symptoms and context

DIAGNOSIS can be difficult. It is especially difficult in primary care where serious diseases, such as cancer or heart disease, are rare, there is a greater reliance on symptoms, and general practitioners (GPs) are constantly bombarded with guidelines that ignore the primary care context.

The positive predictive value (that is, the probability that the disease is present if the patient has a symptom or a positive test result) often makes the most intuitive sense to clinicians and yet is a constant source of misunderstanding between GPs and our secondary care colleagues. It is imperative to be aware that the positive predictive value is affected by the prevalence: as the prevalence falls, the number of false positives tends to increase, resulting in a lowering of the positive predictive value.1 The effect of prevalence can also be readily understood in relation to the odds ratio version of Bayes’ theorem: posterior odds = likelihood ratio x prior odds (see Box 1 for definitions).

Thus, in a low prevalence population like primary care the posterior odds of disease will be lower than in a higher prevalence hospital-based population, even if the same clinical features, such as symptoms and biochemical tests, with identical likelihood ratios could be applied. For example, although chest pain represents a common presenting symptom to both GPs and hospital-based doctors, Buntinx et al noted that gastrointestinal disorders, musculoskeletal problems and psychopathology are identified more frequently in general practice; whereas serious lung diseases and cardiovascular diseases are identified more frequently in the hospital emergency department.2 Furthermore, Sox et al have been able to demonstrate that the prevalence of coronary artery disease was much reduced in two primary care populations compared to a referred arteriography population, even when patients with virtually identical clinical histories were compared.3

Tversky and Kahneman contend that diagnostic decision making is often partially dependent on a number of heuristics or ‘rules of thumb’, in order to recall or to understand knowledge.4 A failure to appreciate that we may be using such heuristics can result in important cognitive errors being made by clinicians working in a primary care context who have spent a significant period employed or studying in secondary or tertiary care settings. For example, GPs may produce a distorted range of differential diagnoses through failure to take into account the relative prevalences of conditions in a primary care setting, misassign probability estimates or judge the odds of an event by the ease with which it can be remembered. In a scenario-based study of family physicians, Bergus and colleagues were clearly able to demonstrate how family physicians deviated from the Bayesian standard of reasoning by the inappropriate use of heuristics.5

As GPs we also encounter a much broader range of problems, many of which are presented in an undifferentiated fashion. Partially as a result of this, decisions made in general practice are dissimilar from those made in specialist settings — the precise diagnostic labels are often less important than deciding on an appropriate course of action. In primary care, diagnoses may be framed in terms of dichotomous decisions: treatment versus non-treatment, referral versus non-referral, and serious versus non-serious. In a well-known general practice-based study of respiratory illness, Howie concluded that a specific diagnostic label may be merely a justification of antibiotic treatment rather than a reason for it.6

It is suggested by some of our specialist colleagues that most of the clinical problems encountered in everyday general practice can be dealt with effectively on the basis of a good clinical history.7 However, although we can be reasonably comfortable about the reliability and validity of some elements in the medical history, such as the age and sex, there is a lower level of confidence in relation to the past medical history or the family history. Moreover, when enquiring about symptoms these must be considered in the context of an individual’s psychological and cognitive state. Patients vary in their ability to recall symptoms; some may fabricate symptoms and others have a tendency to combine separate, similar symptoms into a single generic event (referred to as telescoping).8 Within primary care, conditions will often be seen at an evolutionary stage when the characteristics of symptoms are changing; by the time the patient reaches the specialist clinic the description of the symptom may have become more fixed and, moreover, the patient will have had additional time to reflect on his or her story.9 The perceived cause of the symptom may also lead to underreporting; women with wanted pregnancies are more likely than those with unwanted pregnancies to describe the symptoms of amenorrhoea, breast tenderness and morning sickness.10 To compound matters further, patients and doctors will differ in their interpretation of the meaning of some common symptom terms, such as diarrhoea, constipation and heartburn. For example, patient definitions of heartburn vary from a burning sensation

**Box 1. Bayesian terminology.**

- **Likelihood ratio**
  - The ratio of the probability of an event (such as a symptom or a positive test result) in diseased persons to the probability of that same event in non-diseased persons.

- **Prior (pre-test) odds**
  - The odds of disease before acquiring additional information (such as identifying a symptom or acquiring a positive test result).

- **Posterior (post-test) odds**
  - The odds of disease conditional upon another event having occurred (such as the development of a symptom).
Behind the breast bone or a dull ache in the stomach to the passage of wind through the mouth, excess saliva or a feeling of the heart thumping in the chest.11

One particular problem for those of us working in primary care settings is that the vast majority of symptoms seem to defy a clear-cut organic explanation. Kroenke and Mangelsdorf demonstrated that no specific physical disorder could be established as the cause in 30–75% of instances, even after careful investigation.12 The article by Berger and colleagues reported in this issue of the Journal echoes these findings and further emphasises that symptoms are not invariably associated with organic disease.13 The authors could not confirm that biliary pain was consistently related to gallstone disease, although this is often the only feature that is used to determine the requirement for surgical intervention.

Obviously, it is always necessary to exclude organic disease when presented with a symptom of possible organic significance, such as unexplained weight loss, chest pain or palpitations. However, there is also a need to avoid undertaking investigations beyond those that are absolutely necessary. Depression and anxiety often present with somatic symptoms that may resolve with effective treatment of these disorders,14 but in our society there is a stigma associated with psychological illness. This has an impact on all of us and it is all too easy to collude with patients and their families in order to avoid leaving any ‘organic’ stone unturned. In a further article in this issue of the Journal, Armstrong and Earnshaw found that GPs tended to avoid items from the somatic subscale of the general health questionnaire in diagnosing psychological problems.15 They suggest that this may indicate a temptation to pursue an organic diagnosis at the expense of a psychological explanation. In such circumstances there is a risk that patients receive extensive investigations that are of limited value and potentially damaging both physically and psychologically, irrespective of the additional healthcare costs incurred. According to McWhinney, a symptom is best understood as the patient's way of communicating with us, and frequent attendance with the same symptom, large numbers of symptoms, or our inability to make sense of the presenting symptom, should alert us to avoid thinking solely about organic disease when attempting to reach a diagnosis.16

Making a diagnosis in primary care can be perplexing. It is particularly complicated if GPs choose to ignore their surroundings. Symptoms are not synonymous with organic disease and neither is primary care medicine merely a faded memory of hospital-based practice. When seeking to make a diagnostic decision there is a need to consider symptoms in the context of primary care and from the perspective of the patient.

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References


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Learning from Kaiser (part 2). Is integration the answer?

In 2002 the BMJ published a paper claiming that Kaiser Permanente, the long established and much respected health maintenance organisation based in California, gave better value for money than the National Health Service (NHS) in the United Kingdom (UK).1 In order to make the comparison the authors had to make a number of assumptions. Numerous letters later pointed out where many of these assumptions might have been mistaken and how they almost certainly tilted the scales against the NHS. Despite this, the paper proved influential among policy