Evidence-based diagnosis in general practice: needs both robust evidence and sophisticated electronic health record systems

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General practice is the point of first contact with the health care system in many countries whether or not general practitioners have a ‘gatekeeper’ role. It is characterized by early disease (and consequently, non-specific and atypical presentations), a wide range of potential diagnoses, co-morbid illness (associated with an increase in chronic conditions treated in primary care) and low prevalence of serious morbidity. These characteristics underline the diagnostic difficulties and conflicting demands that GPs face. Often as gatekeepers of the health care system, GPs are under pressure to reduce ‘unnecessary’ investigations and referrals. At the same time, they are required to detect serious conditions early on—but are not always successful. Missed malignancies and myocardial infarctions account for most claims of diagnostic error made against GPs (diagnostic error being the commonest cause of litigation against GPs both in the UK and USA). Failure to refer appropriately is a second major contributory factor in many successful claims against GPs and may well be influenced by diagnosis. Nevertheless, diagnostic error in primary care is under-researched, possibly due to the difficulties in measuring its true rate and impact and the traditional view that ‘GPs don’t diagnose, they manage’.

From a probability theory perspective, diagnosis is a process of revising the prior probability of a particular condition in the light of new evidence (Bayes’ Rule). The new, updated probability is called a posterior probability. In order to use quantitative information in diagnosis, it is necessary to know the diagnostic value of symptoms and signs (known collectively as likelihood ratios) and rules for the combination of likelihood ratios. Likelihood ratios are obtained from diagnostic cohort studies where information on the presence or absence of these diagnostic cues is collected independently of investigations to reach a final diagnosis in a prospective cohort. Broekhuizen et al. have conducted a systematic review of diagnostic studies in Primary Care for the diagnosis of Chronic Obstructive Pulmonary Disease (COPD). However, the use of this information in real-life diagnosis is complicated by the fact that most diagnostic cues are not independent; therefore, adding up likelihood ratios of correlated cues would inflate the posterior probability of the disease. For example, cough, dyspnoea and wheeze all relate to the same underlying pathology and are not independent. There are two approaches to this problem of combining likelihood ratios. First, a regression method can be used to obtain a mathematical model that adjusts the final probability for cue correlation, as shown in three of the COPD studies. In clinical use, the model would be simplified by taking logs of the regression coefficients, the log coefficients being added together to form a score, or Clinical Prediction Rule. Unfortunately, clinical prediction rules do not transfer well from one patient population to another and are often complex to calculate, which limits their use in everyday practice. An alternative approach is to create a Bayesian network that allows for a matrix of cue correlations. Much more data are required to drive a Bayesian network than a clinical prediction rule, and such tools could only be deployed as part of a computerized decision support system.

A long-standing aim of eHealth technologies has been to increase the quality and quantity of data from routine health care, using them to provide better evidence for clinical practice. While it is possible to do this via a prospective cohort study, the lack of routine and reliable coding of diagnostic cues in clinical practice is a significant barrier. One European project that has addressed this issue is the Transition Project. Since 1984, the Transition Project has been formally capturing presenting symptoms, based on episode-oriented epidemiology, and using the International Classification of Primary
Care (ICPC) and a specific electronic health record (eHR) system (TransHis). The project has made significant achievements: (i) the analysis of complete and longitudinal epidemiological databases from family practice based on routine-episode data; (ii) the development of rules for the comparison of international data from family medicine, including the development of a reliable and feasible ICPC2–ICD10 conversion; (iii) the development and implementation of an alphabetic ICPC2–ICD10 thesaurus (in Dutch, English, French, Serbian and other languages) for use in eHRs in family practice. However, a lack of interoperability between TransHis and other eHR systems has limited its use to only a few practice networks in Malta, The Netherlands and Serbia.9,10

The use of computer-driven ‘expert’ systems has long been seen as a potential solution to the failings of human cognition in medical diagnosis. While the earliest systems such as deDombal’s work on Bayesian diagnosis of abdominal pain were developed in the absence of an eHR, more recent systems such as Computerized Physician Order Entry Systems have at least had the opportunity for integration. A systematic review of the literature on computerized decision support by Garg et al.11 noted that computer-driven prompts and reminders have been shown to be very effective in preventing prescribing errors and in prompting structured chronic disease management. However, previous attempts to develop decision support systems for diagnosis were largely ineffective because they were:

1. stand alone systems having to be accessed separately from the computer running the eHR and not integrating with the eHR (if any);
2. lacking sufficient evidence base for diagnostic cues and therefore sufficient credibility with the clinician users;
3. not working in conjunction with clinician’s working patterns and reasoning processes;
4. being opaque to the user and
5. not providing any additional, useful information about the patient.

The accumulation of a greater amount of diagnostic evidence that can form the basis of either clinical prediction rules or Decision Support Systems (DSS) is greatly advanced by systematic reviews such as that of Broekhuizen et al. and the increasing evidence now available in the Cochrane Library under the guidance of the Cochrane Diagnostic Test Accuracy Working Group.12 Several DSS have been evaluated and shown to produce accurate and relevant answers including Isabel, available for use in paediatric and adult medicine, and Problem-Knowledge-Couplers, developed to aid patients in self-diagnosis, but none of these is in common use, either in the USA or Europe. Studies have shown that clinicians usually fail to take advantage of DSS for diagnosis.13

We need to make DSS as automatic and integrated as prompts and reminders. Part of the problem is that decision support has not been considered part of the ‘usual’ functionality of an eHR system. A foremost consideration in the design of the modern eHR system should be ease of use and integration into the clinician’s workflow. Systems that add functionality to the eHR, such as prompts, reminders, alerts, recall lists and templates for coded data entry (e.g. when linked to pay for performance systems such as the UK Quality and Outcomes Framework), are well accepted by clinicians. The challenge is to integrate diagnostic decision support seamlessly into the clinical workflow.14

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References