Omitting family history from the hospital admission

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Family history has a role, but who should be responsible for exploring and recording it?

The increasing age, number and comorbidities of hospital inpatients has increased the load on emergency departments and necessitated significant redesign, including the introduction of short-stay and medical assessment units. These units are diverse in their casemix, but common factors include higher acuity of illness and expedited discharge. Obtaining a complete history of a patient’s acute illness and longstanding comorbidities, as well as his or her social and psychological issues, represents the ideal standard of care. Obviously, however, there are tensions between providing holistic care and continuity of care to the patient and achieving the rapid turnover required in such units.

Genetic markers and tests are increasingly available for an expanding range of conditions. Genetic counselling has moved from specialised clinics into the mainstream practice of many disciplines. The inheritance of disease is rarely a simple algorithm, and these new genetic tools provide complexity rather than clear direction. Relevant guidelines are uncommon outside cancer medicine. Family history is a frequent criterion for determining further genetic testing. For example, the Amsterdam criteria for diagnosis of hereditary non-polyposis colorectal cancer (HNPPC) include a family history of at least three relatives with HNPPC-associated cancer. There can be harm in failing to interpret genetic tests correctly, and the complexity of many conditions demands a high level of knowledge. “Genetic literacy” is a term that has been used to describe competence in this area. However, it seems unreasonable to expect all doctors to be skilful at all times in eliciting and interpreting the family history and then appropriately counselling and testing each patient.

Family history is an older tool than genetic testing and is poorly defined, applied and understood. Even now, there is not enough evidence to gauge its reliability and role. The family history can aid stratification of a patient’s risk of heritable conditions, and it has diagnostic utility for disorders with classic Mendelian inheritance, but it may be less useful in disorders with multifactorial inheritance or more complex genetic expression. In this issue of the Journal (page 682), Langlands and colleagues report that family history is not recorded in the case notes of most medical short-stay patients. They argue that a family history offers potential health gains for the patient and relatives and suggest that there should be increased focus on this element of the medical history. However, this seems unrealistic in the context of increased workload and time pressures, particularly in a hospital short-stay unit.

The acute admission is not an ideal setting for detailed and accurate history taking; patients are usually unwell and access to their family is compromised. The family history recorded is often inaccurate or misleading, not only because the level of health literacy among patients is variable but also because familial clustering is not distinguished from heritable disease. The accuracy of reporting of family history is rarely studied, but it has been shown that it can be poor in patients with cancer or cardiovascular disease.

The primary care setting affords better opportunities to explore and record family history and to make adjustments after clarification with relatives. Certain conditions (eg, malignant hyperthermia, Huntington disease) drive consideration of genetic testing of the affected individual and sometimes lead to testing of family members. The counselling required should form part of an ongoing relationship with the patient and family. As Langlands and colleagues state, the family history may be a casualty of increasing numbers of acute hospital admissions. Perhaps it is a justifiable casualty in the acute health care environment, as long as information is elicited accurately afterwards. Ideally, a patient should have his or her acute illness diagnosed and managed within the acute admission, with a clear plan then delineated for follow-up, which includes notification of those who will be responsible for doing so. It is important to have a use for any family history information once it is accurately obtained. In future, the acquisition of a family history must embrace the developments in our understanding of genetic disease. Without diminishing the role of specialised genetic units, primary care clinicians and specialists in chronic care will need to assume greater responsibility for exploring family history. Screening assessments can identify those requiring a more comprehensive review.

We would argue that, under present circumstances and with doubt hanging over its sensitivity, specificity and effect on health outcomes, the family history is a justifiable omission from many acute hospital admissions. The concept of holistic care is a noble one and, if we are to work within a new paradigm of shorter hospital inpatient stays, we will need to develop a strategy for preserving this concept. Certain diseases, such as unprovoked venous thromboembolism, should trigger an immediate focus on family history, but a routine family history is best ascertained when people are not acutely unwell. If we are serious about disease prevention and the role of genetics in modern medical management, more guidance is needed in terms of which patient groups will benefit from genetic testing and how any positive results will be managed. An integrated approach should include guidance for screening that is based on a better defined family history that has been obtained in the non-acute setting. This approach requires protocols for disease-specific genetic testing and specialist referrals for further assessment and management. A recent National Institutes of Health conference offers hope in this regard.

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