Family history: the neglected risk factor in disease prevention

Michael J McKay

To the Editor: I agree with Emery and colleagues[1] that family history can add much to downstream clinical interventions that provide tangible benefits to the presenting patient and his or her kin. The pedigree chart has some advantages over simple narrative recording of the same details, including the ability to instantly visualise relationships between individuals and the ease with which the chart can be updated and annotated.[2]

There is a familial aggregation (commonly an affected first-degree relative) in up to 25% of presenting cancer patients, while around 5% will harbour a highly penetrant genetic predisposition to cancer. In the cancer clinic, an acceptably detailed family cancer pedigree can typically be obtained in even less time than the 30 minutes suggested by Emery et al.[1]

Steps in drawing pedigrees have been outlined elsewhere[2,3] and typically involve collecting information such as simple demographics, naming and symbolising different cancer types, and recording age of onset and age of death (if relevant) for the different individuals, starting with the presenting patient. The time required to construct a three-generation pedigree would typically be around 10 minutes, making it an attractive addition to routine history taking. If a cancer pattern emerges, the pedigree should be extended as far as possible. Of course, diagnosis verification (through death registries, etc) would be required before surveillance, prophylaxis and therapy decisions are addressed in the familial cancer setting.

A convenient refresher for doctors who do not routinely draw pedigrees might be to first construct their own family tree, with reference to the symbols and relationship illustrators commonly used.[2,3] It is likely that pedigrees in most clinics will be drawn by hand, at least initially, although there are software programs for pedigree creation available (eg, Family Tree Builder; <http://www.myheritage.com/family-tree-builder>).

Over the years, I have informally asked my specialist oncology trainees to routinely construct family pedigrees. I do not recall, among these highly clinically skilled doctors, one that was able to correctly draw a family pedigree until we had worked on it together. Given the benefits of family pedigree analysis across many disease categories, a little practice in pedigree drawing may be a useful exercise for many of us. Even without the requisite confirmation of disease status of the individuals represented, a simple pedigree chart, combined with quick reference to information on the potential clinical significance of any patterns it shows (eg, by consulting National Health and Medical Research Council guidelines),[4] can help prioritise referrals to busy familial cancer clinics.

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Timothy R Bates, Elissa B Poulter, Frank M van Bockxmeer and Gerald F Watts

To the Editor: Langlands and colleagues show that family history is poorly taken in patients presenting to an acute medical unit at a major tertiary hospital.[1]

However, their article and its companion editorials[2,3] do not adequately stress the settings in which family history taking may be both easily achievable and most cost-effective. The nihilism that Thomas and Thompson[2] convey about recording family history in the acute setting is of more concern, given that a tertiary hospital may offer the best opportunity to initiate the process of case detection for a number of heritable and lethal diseases, such as autosomal dominant familial hypercholesterolaemia (FH), the most common monogenic cause of premature coronary artery disease (CAD).

We previously demonstrated that a family history of cardiovascular disease was almost never recorded by coronary care unit medical staff at Royal Perth Hospital.[4] For 509 patients aged <60 years presenting with symptomatic CAD to the coronary care unit, we found that 70% had insufficient clinical data documented in the medical records to enable a diagnosis of FH. In a follow-up study of 103 patients with premature CAD admitted to the coronary care unit, a nurse practitioner was able to record a positive family history of premature cardiovascular disease in a primary relative for 43% of patients, of whom 95% had phenotypic FH based on a recognised clinical diagnostic tool (the Dutch Lipid Clinic Network score).[5]

Patients detected in this way in Western Australia are now referred to a statewide FH program run by staff from a lipid clinic.[5] In this program, where detailed pedigree drawing and family tracing is performed by trained nurses, we have found a causative mutation for FH in up to 85% of patients with a clinical phenotype strongly suggestive of FH. Additionally, we find that for every index case so detected, we can additionally diagnose at least three new cases of FH in the patient's relatives, many of whom are young. This method of case detection and subsequent treatment with cholesterol-lowering therapies is highly cost-effective and, more importantly, enables therapy to be targeted at younger patients, thereby maximising the potential for preventing CAD.

Our experience illustrates that for a lethal condition such as FH, an accurate family history recorded by a nurse can spark a cascade of action that leads to a definitive diagnosis of FH in the index patient and the subsequent detection of otherwise undiagnosed FH in the community, with significant associated cost savings.[6] Hence, we propose that nursing staff can efficiently bridge this gap in medical care while we are getting our house in order by training medical staff to effectively take a family history.

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Simon E P Hauser

TO THE EDITOR: I read the article by Langlands and colleagues[1] with some dismay, and a sinking heart. Their report is further evidence of the dangers of moving away from the basic skills of comprehensive history taking and performing a detailed physical examination. Interestingly, in the United States, the debate regarding performing a physical examination has come full circle, from virtually ignoring its importance to now telling us how vital it is and how to perform it.[2] But it is the taking of a comprehensive history that, as one of my mentors told me, "is where the money is", and a detailed family history is an integral part of this.

Eliciting a detailed family history is arguably more important for paediatric patients, who have a longer potential life span and hence have more to gain from this information. In my paediatric practice, for example, I see numerous overweight children, some of whom have a strong family history of hypercholesterolaemia, vascular disease or type 2 diabetes mellitus, which places them at considerable risk of cardiovascular disease in their adult years. A detailed family history may also "unmask" the genetic contribution to a child's history of deafness or learning disability.

Time constraint is the main impediment to taking a comprehensive family history, but it is worth keeping in mind that it is time well spent and that, in the paediatric population, it may make a significant contribution to the long-term health of the child. With the impending advent of personalised genomic screening, there will be an even greater imperative to formalise the gathering of family history details.[3,4]

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