Family Health History
The Case for Better Tools

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Every medical student remembers the clinical lesson to collect a family health history. In fact, the family history remains one of the most important resources available to help clinicians identify potential health risks and personalize care of their patients. Empowered with information from the family history, clinicians can guide their patients to take preventive steps and initiate mitigating strategies to reduce disease risk. For example, if a physician knows that a patient has a history of familial hypercholesterolemia, the physician can encourage the patient to make lifestyle changes or, if appropriate, take medication to reduce his or her risk for heart disease.

Information from the family history also plays an important role in the interpretation of genetic test results. For example, if a healthy 35-year-old woman with no documented family history of breast cancer receives a *BRCA1* test result showing a variant of unknown significance, the clinician might recommend "watchful waiting"; however, if the same woman has a known family history that includes a mother with bilateral breast cancer at age 42 years and a maternal aunt

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with ovarian cancer at 53 years, the clinician can now recommend a substantially different course of action, from active surveillance to consideration of preemptive bilateral mastectomy and salpingo-oophorectomy. Despite the increasing complexity of genomics and genetic testing, the family health history continues to be a simple and powerful way to assess an individual patient's risk for disease.

Despite its importance in health care and personalized medicine, the family health history remains underutilized in clinical practice constrained by time, accuracy, and physician bias. Physicians, other clinicians, and even trainees often lack time to collect, interpret, and discuss the family history during a busy clinical visit. As a result, family history information is often incompletely collected during a clinical encounter, is gathered from only a minority of patients, and lacks the necessary detail for accurate risk assessment.[1]

Even if sufficient time could be created to record a detailed family history, a patient's self-reported family history is often suboptimal. The accuracy and consistency of self-reported family history range widely from 30% to 90%, depending on the degree of family relatedness and the reported disease.[1] Health history of first-degree relatives (eg, parents, siblings, and children) tends to be more accurately reported than that of more distant relatives (eg, aunts, uncles, cousins, grandparents). Also, certain major physical diseases, such as cancer and cardiovascular disease, will be more accurately remembered by a patient than other diseases, such as mental health disorders or autoimmune diseases. Collecting a complete and accurate family history in the clinic continues to be a significant challenge. As a result, the family history may be deemphasized during the clinical encounter compared with other modifiable health behavior changes such as diet and smoking. Nevertheless, the family history remains relevant to a patient's health.

To help overcome the challenges of accurately collecting and using the family health history, a number of groups have developed paper-based and web-based tools to help patients organize their family history outside of the clinical visit. These tools facilitate patient-directed collection using a simple interface to record family history, including type of relation, disease, age at onset, cause of death, or other potentially relevant information. One collection tool, developed by the US Surgeon General, is called "My Family Health Portrait."[2] The premise of such family history tools is 2-fold: first, patients will have greater access to family information outside a clinic visit and thus be able to collect more accurate family history information without time constraints. Second, when a patient returns to the clinic with his or her completed family history, the clinician can focus on using that information to guide a patient's care rather than collecting it.

However, evidence suggests that these tools, while acceptable for collecting family history information,[3] remain poorly adopted outside of research settings.[4] Furthermore, the use of a family history tool does not prevent inaccurate recollection of one's history.[5] Any significant deviation from the true family history interferes with genomic interpretation and assessment of disease risk, and this must be addressed if family history information is to be used effectively today and to improve patient care in the future.

A better tool is needed. Collection of family history information is in a prime position for significant innovation over the next several years. In addition to the increasing need for a detailed family history to assist in genomic and big data analysis, the changing reimbursement

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structure from a fee-for-service model to managed health will increase emphasis on clinicians to personalize preventive strategies based on family risks. Furthermore, documenting a family history in at least 20% of patients is an optional "menu-set objective" that clinicians can use to meet stage 2 meaningful use requirements for the Center for Medicare & Medicaid Services electronic health record (EHR) financial incentive program. Increasing evidence suggests that family members aware of family history play an important role in influencing positive behavior change in the family, leading to healthier decisions.[1] However, current family history tools remain archaic by today's standards and are unlikely able to meet challenges related to changing reimbursement structure and EHR incorporation or even to be readily shared with family members.

To realize the potential of the most accurate family health history, new tools should accomplish several important clinical objectives. First, any new family history tool must harness the social potential of families (ie, put the "family" into family health history). Families have a wealth of information regarding their own health; therefore, an ideal family history tool should facilitate intrafamily communication and collaboration to create a complete family history and then promote positive behavior, influencing dialogue between family members. Second, family history tools must support meaningful and simple interoperability and information exchange with EHRs. As meaningful use creates new opportunities between health information technology systems, family history tools can leverage such standards to promote the exchange of information between health care practitioners and patients.

Third, in the era of big data, family history tools must do more than just provide a description of familial disease; they must actually return health information and evidence-based recommendations to patients and clinicians to drive adoption.[6,7] For example, a strongly positive family history of premature atherosclerosis,

such as 2 or more family members with myocardial infarctions in their early 40s, should prompt a recommendation for earlier (ages 20-35 years in men) evaluation of serum cholesterol levels, rather than screening initiating in men between ages 35 and 50 years. Simple interfaces that only record information are tedious and lackluster. Providing personalized care recommendations based on information entered by patients and family members serves as an incentive that can promote usage and clinical action. Fourth, family history tools should be built in a way to support clinical research and analysis of big data. The family history can guide the treatment of a single patient and may be important to researchers studying a disease or gene. Combining well-documented family histories with genomic information for family members can be a great help in elucidating the genetic influences of a particular phenotype, and this combined information can collectively become a repository of family history big data for exploration with novel analysis techniques.

If designed correctly, family history tools will play an important role in the rapidly changing world of personalized medicine. Current tools are underutilized, produce inaccurate family history information, and miss opportunities to harness the social capital of the family to record accurate family history information and influence behavior change. Before it is widely adopted, any new tool for obtaining family history should be tested for validity, reliability, and affect on patient outcomes. Family health history could affect health care on a widespread scale if more effective and efficient tools were designed to collect and use family history information based on modern technology and access to big data. Clinicians and technology innovators should refocus efforts on developing novel solutions and family history tools that incorporate broad family usage, integrate into the EHR, provide clinical recommendations, and support clinical research.

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