Securing and Documenting Cancer Family History in the Age of the Electronic Medical Record

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KEYWORDS

- Family health history • Pedigree • Genetic risk assessment
- Electronic medical records

KEY POINTS

- Family health history (FHHx) is one of the least expensive, most useful, and most underused methods to assess genetic risk and target the need for a genetic evaluation.
- In an era of increasingly comprehensive genetic testing, family history is still vital to the interpretation of many genetic test results.
- FHHx information sufficient to assess the need for a genetic consultation can be obtained reasonably quickly.
- Documenting family history electronically may inform clinical decisions regarding a diagnosis, screening, and prevention, as well as identify the need for referral for genetic evaluation, which might include genetic testing.

Case example: Maria is a healthy 33-year-old woman with no cancer history referred for a surgical consultation after requesting prophylactic mastectomy because of her strong family history of breast cancer.

Most individuals are aware when a health condition, whether it is cancer, cardiovascular disease, asthma, obesity, or a mental health disorder, runs in a family. What most individuals do not do is distinguish among the various factors, be they environmental, lifestyle, or genetic, that can impact the development and management of those conditions. Collecting and documenting an FHHx is one of the least

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expensive, most useful, and most underused methods to target the need for a genetic evaluation for risk assessment. FHHx is valuable when making a differential diagnosis or identifying surveillance and prevention options. Especially in an era when genetic testing is moving from single gene analysis to multigene panels and exome/genome sequencing, an FHHx may become even more vital in interpreting those laboratory results. Even for families without an identifiable genetic mutation, the FHHx can be important in changing surveillance, such as the first-degree relatives of a 47-year-old individual with colon cancer who are now candidates for earlier colon screening.

FHHx has long been recognized as an important component in prediction, prevention, and management of common, complex diseases, including cancers.[1] Half of all families have a positive FHHx for one or more common chronic diseases. Depending on the condition, a positive FHHx increases a person's chance of developing a particular disease from 2 to 10 times.[2] Consequently, FHHx has always been part of a comprehensive medical intake, because it represents the complex interaction of genes, environment, and other lifestyle risk factors for diseases shared among relatives.

However, several barriers have been identified that hinder the collection of an FHHx, not only at the provider level but also with patients and health care systems in general.[3–6] It takes time, it is not clear to most clinicians as to which questions are the most useful, and even when asked, patients are not always aware of the health nuances that are relevant to a genetic assessment.[2,7–9]

At present, a significant health care system barrier to the use of documented FHHx is the lack of consistent structured data elements.[10] Examples of structured data elements include gender, age, age at death and cause, diagnosis, age at diagnosis, and sometimes screening behaviors or treatment options. Without these data elements that can be queried by the electronic health record (EHR) system, clinical decision support (CDS) that assesses the FHHx data to assist with risk assessment or referral for genetic evaluation is not possible. Given the importance of FHHx for effective screening and prevention, several federal agencies, including the Centers for Disease Control and Prevention and the National Institutes of Health (NIH), have emphasized the importance of research and development of FHHx tools.[11] At present, many teams are exploring methods of incorporating FHHx into EHRs using structured data and interoperable formats.

This article reviews the process of collecting an initial family history within the EHR and updating it during subsequent clinic visits. This process can be facilitated if the information is collected, stored, accessed, and updated in a designated section of the EHR, and the data could be more valuable to the clinician if tied to CDS. With complete family history information documented in the medical record, the surgeon can then assess if the patient is a candidate for a comprehensive genetic evaluation or if alternative or enhanced screening, medication, or surgical options should be considered.

REASONS FOR COLLECTING AND DOCUMENTING A CANCER FAMILY HISTORY

There are many reasons to collect FHHx. Family history may clarify the cause of a disease through recognition of a hereditary cancer syndrome. FHHx can influence eligibility for genetic testing or inform screening and management decisions for both affected patients and their unaffected extended family members. Often, the purpose of the FHHx is to assess the need for a genetic referral. Therefore, an initial FHHx is usually cursory and targeted to the condition of interest to address that limited