



## CLINICAL SCHOLARSHIP

# Improving Utilization of the Family History in the Electronic Health Record

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### Abstract

**Purpose:** The purpose of this article is to provide an overview of Family History in the Electronic Health Record and to identify opportunities to advance the contributions of nurses in obtaining, updating and assessing family history in order to improve the health of all individuals and populations.

**Organizing Construct:** The article presents an overview of the obstacles to charting Family History within the Electronic Health Record and recommendations for using specific Family History tools and core Family History data sets.

**Methods:** Opportunities to advance nursing contributions in obtaining, updating, and assessing family history in order to improve the health of all individuals were identified. These opportunities are focused within the area of promoting the importance of communication within families and between healthcare providers to obtain, document, and update family histories.

**Findings:** Nurses can increase awareness of existing resources that can guide collection of a comprehensive and accurate family history and facilitate family discussions. In this paper, opportunities to advance nursing contributions in obtaining, updating, and assessing family history in order to improve the health of all individuals were identified.

**Conclusions:** Aligned with the clinical preparation of nurses, family health should be used routinely by nurses for risk assessment and to help inform patient and family members on screening, health promotion, and disease prevention. The quality of family health information is critical in order to leverage the use of genomic healthcare information and derive new knowledge about disease biology, treatment efficacy, and drug safety. These actionable steps need to be performed in the context of promoting evidence-based applications of family history that will be essential for implementing personalized genomic healthcare approaches and disease prevention efforts.

**Clinical Relevance:** Family health history is one of the most important tools for identifying the risk of developing rare and chronic conditions, including cardiovascular disease, cancer, and diabetes, and represents an integration of disease risk from genetic, environmental, and behavioral/lifestyle factors. In fact, family history has long been recognized as a strong independent risk factor for disease and is the current best practice used in clinical practice to guide risk assessment.

An accurate family history (FH) is fundamental to providing the highest quality of personalized health care. Amidst advancements in genomic science and translation to improved diagnostic precision, the FH still provides the most efficient and low-cost potential of providing an individualized blueprint for care. In 2002, the Office of Public Health Genomics started the Family History Public Health Initiative to increase awareness of family health history as an important risk factor for disease and to promote the use of FH in programs aimed at reducing the burden of disease in the U.S. population (Centers for Disease Control and Prevention, 2013). This was followed in 2009 by the National Institutes of Health (NIH) State of the Science Statement, which aimed to provide healthcare providers, patients, and the general public with an assessment of currently available data on FH and improving health (NIH, 2009). Although a comprehensive, three-generation pedigree used in medical genetics, counseling, and research remains a recognized important assessment for disease prevention and health promotion, significant knowledge gaps exist (Guttmacher, Collins, & Carmona, 2004; Pyeritz, 2012; Valdez, Yoon, Qureshi, Green, & Khoury, 2010). Though the American Association of Colleges of Nursing (AACN) has asserted that baccalaureate nursing programs “recognize the relationship of genetics and genomics to health ... using a constructed pedigree from collected family history information,” studies have shown that many nurses lack literacy and skill in genomics and its application to health care (AACN, 2008; Camak, 2016).

Since the original publication of the NIH State of the Science Statement, the following gaps continue to exist:

- Lack of standardized practices for documenting and updating the FH given existing time and resource restraints in our healthcare system;
- Lack of ability to access family health data across the many different healthcare settings by all members of the interprofessional team where services are obtained and, ideally, coordinated (e.g., hospitals, home, outpatient settings, clinics, schools, communities);
- Inability to incorporate nonbiological family members who share common social-behavioral, cultural, and environmental factors;
- Inability to link family histories, which are dynamic and ever changing, with genomic data to guide automated risk assessment, prevention, and health promotion strategies within electronic health information systems;
- Lack of easy-to-use and culturally, linguistically, and educationally tailored resources, which can guide families on how to start the conversation on family health

and support in acquiring, documenting, and updating an FH;

- Lack of a cohesive education strategy to support interprofessional communications of the health team’s ability to translate genomic findings into practice.

The authors and members of The American Academy of Nursing’s Genomic Nursing and Healthcare Expert Panel sought to further explore the current state of evidence on these issues in order to identify a process for advancing the utilization of FH in clinical practice. Nurses have an important role in promoting family health tracking among patients and families, the acquisition and documentation of a three-generation FH in the electronic health record (EHR), and performing updates of family health information and risk assessment on a routine basis for every individual.

To consistently support these practices across healthcare systems, action must be taken to (a) standardize collection methods and documentation content; (b) utilize the evidence-based recommendations for FH content acquisition to promote adoption of health promotion behaviors; and (c) advance interoperability of FH data among individuals, clinicians, and healthcare systems.

In order to support a standardized method of collecting FH in the EHR, a core data set and common data definitions must be developed. According to the New Standards and Enhanced Unity for Family Health History Information in the Electronic Health Record, in 2007 the critical importance of a core data set was recognized. An interdisciplinary team was formed to determine an FH core data set for primary care providers. For example, consanguinity, cause of death, and approximate dates/ages for data fields are suggested required core data sets relevant for FH collection in the EHR. The specific FH recommendations were adopted by the American Health Information Community from the Personal Health Record Workgroup. Additionally, it was recommended that studies be conducted to determine an evidence base and support pilot programs in order to evaluate the core minimum data set. Once the core data set is implemented, interoperable standards that allow collaboration between FH and health information technology can be achieved (Feero, Bigley, Brinner, & The Family Health History Multi-Stakeholder Workgroup of the American Health Information Community, 2008). In Oregon, 96% of the Federally Qualified Health Centers have already begun utilizing an FH section on their initial patient intake forms (Harris & McMullen, 2006). A senior epidemiologist from the Oregon Genetics Program in the Department of Human Services anticipates that collecting FH information may motivate patients who are at high risk for chronic conditions to engage in healthier behaviors (Zlot, Valdez, Han, Silvey, & Leman, 2010).

## Standardized Family Health Collection Methods and Documentation Standards

Nurses have an important role in promoting family discussion and documentation of FH. Numerous agencies have provided recommendations on how to begin discussions about FH, including a tool developed by the U.S. Surgeon General, My Family Health Portrait (<https://familyhistory.hhs.gov/FHH/html/index.html>).

Advantages of using these tools include wider dissemination of health information among family members and clinicians and less time required to collect the information in the healthcare setting. However, these strategies have not been routinely adopted into the delivery of health care, largely due to the potential inaccuracies of the information (Facio et al., 2010). To address this issue, metrics for the quality of FH data were developed and tested using MeTree, a patient-facing web-based family and personal health history collection and clinical decision support program (Wu et al., 2014). While using MeTree improved the quantity and quality of data collected, the study also found that the process of engaging patients to discuss FH with relatives prior to collection improved the quantity and quality of data provided by patients.

Recommendations for using specific FH tools and core FH data sets have been published (Feero et al., 2008). However, the lack of a standardized process for charting FH remains a significant obstacle to implementation. A strategy to promote quality FH data within the EHR interface would be implementation of metrics that must be routinely updated in order to qualify as complete documentation, such as those defined by Wu and colleagues (2014), including (a) three generations of relatives; (b) relatives' lineage; (c) relatives' gender; (d) an up-to-date FH; (e) pertinent negatives noted; (f) age of disease onset in affected relatives; and, for deceased relatives, the (g) age and (h) cause of death. This process would necessitate a reconceptualization of how FH is charted; instead of having one free-text box to fill in, the FH would be documented by filling in all known metrics.

Thus, the process of including comprehensive FH in EHRs can facilitate family discussions about family health, improve communication among individuals and healthcare providers, and aid verification of FH data. One method of promoting wider adoption of FH tools in the EHR is application program interface, which allows data transfer between EHR platforms. A verification protocol, similar to the process used by Wikipedia that tags verified or unverified information, could be used in the EHR to improve confidence in, and utilization of, FH. In addition, automated reminders in the EHR should be included to remind the healthcare team to ask for updates in the

FH, flag data for further investigation or screening, and link health information from other family members in the system. Automated updates of FH information may enhance the process of data collection if family member profiles are linked (e.g., a confirmed diagnosis in a father or mother would be automatically updated in their child's profile). In children, automatic FH updates could also cross-pollinate into siblings' histories, thereby drawing attention to familial risk or exposure.

Despite the utility of routinely updating FH data, there are important ethical considerations in doing so that must be taken into account. Ethical factors including who has access to the patient EHR, the accuracy of automated data that is captured, and family dynamics that affect the disclosure of significant FH may have drastic implications for the patient and additional family members. Obtaining genetic information from an FH has the ability to prevent disease, aid in reproductive planning, and even result in life-saving treatments for family members who may be unaware of any preexisting conditions. The American College of Medical Genetics and Genomics' (AMCG) recent policy statement supports the clinical utility of genetic and genomic services that may benefit the individual and family in informing them on a particular diagnosis (AMCG, 2015). Additionally, the American Nurses Association encourages further efforts to be made to the EHR that will improve patient care in the future (ANA, 2009). Moving forward, ways in which to protect patient confidentiality and improve the collection and accuracy of FH must be better defined to enhance patient outcomes.

## Ensure Return of Evidence-Based Recommendations to Promote Adoption of Health Promotion Behaviors

Obtaining an accurate three-generation FH is one critical area in which nurses and other healthcare professionals can actively engage. However, FH continues to be underdocumented and underutilized in clinical practice. Retrospective reviews of EHRs have continued to show that FH information is gathered for only a small percentage of patients and lacks sufficient detail for an accurate risk assessment (Langlands, Prentice, & Ravine, 2010; Quereshi et al., 2009). Beyond the barrier of lack of time to accurately collect, update, and document FH data, there is also a lack of access to guidelines or resources for estimating the impact of FH (Daelemans, Vandevoorde, Vansintjan, Borgermans, & Devroey, 2013). Documentation of FH in at least 20% of patients is an optional "menu-set objective" that healthcare providers may use to meet Stage 2 meaningful use

requirements for the Centers for Medicare and Medicaid Service's EHR incentive program. However, this requirement falls short of its mandate by the lack of criteria for quality assessment (e.g., documenting all chronic conditions and age at onset utilizing a three-generation pedigree) and evaluation of risk, that is, the utilization of the data to provide recommendations for health promotion and disease prevention. Ideally, automated messages could be designed in the EHR to flag patients at risk for health conditions based on FH data; however, the implementation of this approach requires provider engagement. Recently, a study that evaluated primary physician responses to automated tailored prompts that alerted them to a patient's risk to one or more of six diseases based on FH found no significant change in adding the FH risk to the problem summary lists or screening interventions (Zazove, Plegue, Uhlmann, & Ruffin, 2015). In addition, the manner in which healthcare providers are reimbursed for services, such as the time and effort for collecting and documenting an FH, has yet to be addressed. Thus, better strategies that recognize and reimburse practitioners for the value of integrating the collection and utilization of FH information to risks that are amenable to preventative interventions are needed.

Genetic and genomic educational standards of bachelor's level nurses include the basic competency of obtaining and constructing a three-generation pedigree family health history. Nurses focus on the identification of red flags as identification of basic risk assessment of individuals with an emphasis on referral to the physician regarding the assessment findings. Master's- and doctoral-level nurses are prepared to manage both real and potential risks identified based on FH information acquisition in efforts to intervene and improve health outcomes. Through collaboration between the ANA and the International Society of Nurses in Genetics (ISONG), genetic and genomic competencies and outcomes have been established for nursing across all levels of education (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). While a national strategy to support the integration and translation of genetics and genomics has been defined for nursing (Calzone et al., 2013), these competencies could ideally extend across all interprofessional team members in order to facilitate clear, nonrepetitive client communication that will support collaborative team outcomes.

Limited evidence exists on the effect of FH collected by nurses on the delivery of healthcare services and the impact on direct or indirect health outcomes. Future integrated electronic healthcare delivery systems, particularly those utilizing EHRs, may provide greater opportunities to evaluate the enormous value of nursing in collecting, documenting, updating, referring, and managing real and

potential issues of clients derived from FH at all levels of practice across all populations

Nurses work in many settings and are the most trusted healthcare professionals; thus, they are ideally suited to answer many important questions related to the FH. This includes questions related to the variance in accuracy and completeness of FH information according to the setting, the mode of collection, and the person who is collecting it. In addition, nurses are able to recognize and assess how genomic health literacy, family dynamics, and various health disorders affect an individual's awareness and ability to communicate and report his or her family health history.

Nurses with genetic and genomic knowledge are also uniquely positioned to identify and answer other important questions that will impact personalized health care. These questions may pertain to which environmental and lifestyle elements of an FH are most useful in helping patients make positive changes in health-related behavior. Nurses may also inform how FH may be best collected and integrated into practice in diverse racial, ethnic, religious, social, cultural, and economic populations. Finally, simple-to-use "real world" culturally, linguistically, and educationally tailored resources that will allow nurses and other healthcare professionals to accurately update, assess, and make evidence-based recommendations across healthcare settings are needed to make this a reality. Nurses are ideally suited to develop FH resources for implementation across a variety of healthcare settings.

The Family History Impact Trial Group has provided critically important data for informing the ways in which we help patients engage in efforts to promote or maintain health. A self-administered web-based tool, Family Healthware, provided personalized risk-tailored messages intended to influence health behaviors. Patients completed the FH history and physicians reviewed the data. The study examined the influence of risk-based alerts on physical activity, fruit and vegetable consumption, and cholesterol screening (Ruffin et al., 2011). Six months after implementation of tailored messages, they found a modest increase in self-reported physical activity and fruit and vegetable intake, but reduced likelihood of receiving cholesterol screening. Although this study did not demonstrate an appreciable improvement in health outcomes or behavior change, recent studies focused on technology show promise regarding how technology can motivate adoption of health promotion behaviors (Joseph, Keller, Adams, & Ainsworth, 2015; Nahm et al., 2015).

Nurses are often the most readily available healthcare team member and routinely spend more time with patients and families regarding their health. The delivery of evidence-based recommendations must occur within



the context of the patient and family relationship with consideration of environment and resources. Advances in understanding the importance of epigenetics in health has brought an increasing interest in incorporating FH elements to include fictive kin and other-mothers, which are nonbiological family members who play a major role in the transmission of culture, health promotion, and decision making (Spruill, Coleman, Powell-Young, Williams, & Magwood, 2014). With this knowledge, there is greater potential to identify at-risk individuals and provide opportunities for education, prevention, and early diagnosis. As common diseases cluster within families as a result of shared environment and genetics, there has also been an interest in including neighbors and communities in the family health risk assessment (Hartmann, Marshall, & Goldenberg, 2015). Housing and neighborhood factors may also be included as social and behavioral determinants of health (SBDH), indices that are also recommended for inclusion in the EHR by the American Academy of Nursing, the Agency for Healthcare Research and Quality, and the World Health Organization ([http://www.nursingoutlook.org/article/S0029-6554\(1500257-2/fulltext\);](http://www.nursingoutlook.org/article/S0029-6554(1500257-2/fulltext);) [http://www.healthypeople.gov/2020/topics-objectives/topic/social-determinants-health;](http://www.healthypeople.gov/2020/topics-objectives/topic/social-determinants-health;http://www.who.int/social_determinants/en/) [http://www.who.int/social\\_determinants/en/](http://www.who.int/social_determinants/en/)). As similar challenges are inherent in collecting, analyzing, and providing recommendation for FH and SBDH factors, it may be reasonable to address these issues together. Nurses are in a prime position to take a leading role in the implementation process for ensuring that these data are collected and addressed within the realms of health care for individuals and families.

## Integrating Family Health History With Other Genomic Tools

The potential for having an EHR that is available to patients across their lifespan and accessible to healthcare providers during every interaction with the healthcare system is becoming more of a reality. As genetic and genomic testing is becoming more readily available and more frequently used by healthcare providers, there is great potential for these data to be housed within individual EHR profiles. This advancement could provide another avenue for linking FH data with genomic risk factors and improving the early diagnosis and treatment of common chronic health conditions.

Nurses across the nation are performing cutting edge research to identify genomic risk factors and Gene  $\times$  Environment interactions that influence health outcomes. With increasing proficiency in the utilization and interpretation of big data, nurses will continue to

provide personalized health interventions and contribute to President Obama's Precision Medicine Initiative<sup>®</sup> announced in January 2015 (<https://www.whitehouse.gov/the-press-office/2015/01/30/fact-sheet-president-obamas-precision-medicine-initiative>). The integration of family health history with genetic test results will be a major issue that needs to be addressed as EHRs provide the capability of automating the analysis of genomic discoveries into personalized risk stratification. As technology continues to increase the ability to advance precision approaches for health, nurses must be prepared to interpret and discuss this information with individuals and families.

While some of these systems are becoming more automated, patients will still require assistance with knowing how to apply the information in the context of screening recommendations and options for addressing psychological, behavioral, or environmental risk factors. In a recent study using MeTree, the number of patients at increased risk for breast/ovarian cancer, colon cancer, hereditary syndrome risk, and thrombosis were examined (Orlando et al., 2014). The authors reported that the implementation of risk stratification tools in primary care will likely increase costs related to an increased prevalence of non-routine risk assessment and management resources required. As tools such as MeTree become more available, nurses are ready to be part of the solution in assisting patients and families to make the right personal choices. Nurses play an important role as frontline educators and advocates of patients and families, and documentation of FH and risk assessment should be part of routine practice.

FH remains the best means of assessing lifespan health risks and for providing patients and families with the best guidance on lifestyle choices to prevent chronic disease. Incorporating genomic risk assessments with other genomic advancements using a lifespan approach will only increase the ability of nurses to provide more precise information to patients and families and provide more momentum to improve the health of the nation.

## Conclusions

In order to further its mission of utilizing FH within the EHR as an essential fundamental health assessment tool critical to healthcare delivery and policy, we have identified opportunities to advance the contributions of nurses in obtaining, updating, and assessing FH in order to improve the health of all individuals and populations. Identified opportunities are focused within the areas of:

- Urging Medicare and healthcare insurance companies to adopt quality metrics of FH data as an essential

component of documentation; routine risk assessment should be addressed in health promotion and screening recommendations for each patient encounter.

- Promoting the importance of communication within families and between healthcare providers to obtain, document, and update family histories at all levels of practice.
- Improving the awareness and utility of existing resources and developing new resources that can guide and increase the frequency of obtaining and updating a comprehensive and accurate FH.
- Developing and evaluating new tools that can be used in primary care and nontraditional settings that will integrate FH with other genomic healthcare information to derive new knowledge about disease biology, risk assessment, treatment efficacy, and drug safety.
- Promoting evidence-based applications of FH for implementing personalized genomic healthcare approaches and disease prevention efforts.
- Determining the impact of FH on direct and indirect health outcomes, patient choices, and economic healthcare costs across the lifespan through systematic evaluation.
- Developing strategies that assist families to understand the value of passing generational information on within families and with healthcare providers as a mechanism to inform and maintain health through personal engagement.

### Clinical Resources

- Centers for Disease Control and Prevention. Family health history: <https://www.cdc.gov/genomics/famhistory/>
- Genetic Alliance. Family health history: <http://geneticalliance.org/programs/genesinlife/fhh>
- HealthIT.gov. Family health history: <https://www.healthit.gov/providers-professionals/achieve-meaningful-use/menu-measures-2/family-health-history>
- International Society of Nurses in Genetics: <http://www.isong.org/>
- Partners in Information Access for the Public Health Workplace. Public health genomics: [https://phpartners.org/public\\_health\\_genomics.html](https://phpartners.org/public_health_genomics.html)
- U.S. Department of Health & Human Services. The Surgeon General's family health history initiative: <http://www.hhs.gov/programs/prevention-and-wellness/family-health-history/index.html>

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