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Modernizing family health history: achievable strategies to reduce implementation gaps

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Abstract

Family health history (FHH) is a valuable yet underused healthcare tool for assessing health risks for both prevalent disorders like diabetes, cancer, and cardiovascular diseases, and for rare, monogenic disorders. Full implementation of FHH collection and analysis in healthcare could improve both primary and secondary disease prevention for individuals and, through cascade testing, make at risk family members eligible for pre-symptomatic testing and preventative interventions. In addition to risk assessment in the clinic, FHH is increasingly important for interpreting clinical genetic testing results and for research connecting health risks to genomic variation. Despite this value, diverse implementation gaps in clinical settings undermine its potential clinical value and limit the quality of connected health and genomic data. The NHGRI Family Health History Group, an open-membership, US-based group with international members, believes that integrating FHH in healthcare and research is more important than ever, and that achievable implementation advances, including education, are urgently needed to boost the pace of translational utility in genomic medicine. An inventory of implementation gaps and proposed achievable strategies to address them, representing a consensus developed in meetings from 2019–2020, is presented here. The proposed measures are diverse, interdisciplinary, and are guided by experience and ongoing implementation and research efforts.

Keywords Family health history · Implementation · Strategies · Informatics · Primary care

Introduction

Many types of risk data are collected during healthcare diagnosis, management, and research. Family health history (FHH) is a data type serving risk assessment, diagnosis, research, and preventative health, and it is universal. Before clinical genetic tests became available, FHH was the only tool for assessing risks attributable to genetic inheritance,

particularly for common diseases with genetic components. Despite technologic leaps in genomic variant detection, FHH remains the most accessible, least expensive, and practical assessment tool for this purpose, and remains the sole tool for those without consistent access to modern genetic testing. FHH is validated, recognized as standard of care, and dedicated FHH storage is a required component of electronic health records (EHR) (Ginsburg et al. 2019). Documentation requirements for billing endorse the value of FHH because it changes management and motivates behavior change. Rather than being replaced by genetic testing, FHH enhances the quality of variant interpretation by modifying prior probability, particularly for variants of uncertain significance (Allen et al. 2020). FHH serves as a crucial bridge between the observed phenomenon of inheritance and genomic variation.

The power of FHH in the hands of the primary care practitioner is often underestimated. An individual with a family history of diabetes, cardiovascular diseases, or various types of cancer has a 2- to fivefold risk for these diseases relative to one without a positive family history (Guttmacher et al. 2004; Houwink et al. 2019). When multiple family members are affected with these common diseases, and when

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this occurs at a young age, the relative risk increases further. A family history is therefore a useful tool for pre-symptomatic risk assessment for multiple common chronic diseases prevalent in primary care practices. It reinforces the role of general practitioners as family doctors. Collecting a family history opens possibilities for early primary and secondary prevention of these diseases and their monogenic disease counterparts like long QT syndrome, inherited breast and ovarian cancer risk, and monogenic forms of diabetes like maturity-onset diabetes of the young. The FHH is also a powerful tool to find, inform, and treat at risk family members pre-symptomatically.

Trends indicate that FHH is more important than ever. FHH value in familial disorders is increasing, especially in cancer, advanced by better risk guidance (Allen et al. 2020). The expanding genetic and genomic testing volume in general health care parallels new management options. On a systems level, there is renewed emphasis on managing health at the population level and on applying public health strategies to genetic and common disease (Valdez et al. 2010). State-mandated newborn screening continues to expand to uncover and treat recessive disorders undetected in most FHHs, yet once found, FHH plays a key role in disseminating risk information to family members. FHH also extends the value of meaningful genetic test results to additional family members through timely cascade screening, and it improves risk estimation for common diseases (Ginsburg et al. 2019). As such, it is a critical enabler of the public health approach to genomic disease.

Importantly, accurate FHH information serves the patient. Indeed, the information is generally supplied by the patient and their family (Lin et al. 2017). Broad community awareness of the uses of FHH in healthcare leads to greater sharing within families and higher accuracy of health information for health care providers, adding robust value for both providers and the informed individual (Allen et al. 2020). Research shows implementation can work (Wu et al. 2013, 2019) and has value (Hussein et al. 2020, Qureshi et al. 2012), yet optimal use of FHH remains elusive (Wu and Orlando 2015, Ginsburg et al. 2019). Though quality FHH research and outreach exists, in many clinical contexts (Ginsburg et al. 2019, Qureshi et al. 2012, Orlando et al. 2020), patient, system, and provider factors persist. For example, modern families may be separated geographically and receive healthcare in different systems thus impeding FHH sharing both between health records and among family members. Accessibility of genetic testing is uneven in many locations, placing additional weight on FHH accuracy and rigorous application (Lin et al. 2017).

As with any provider data tool, the effectiveness of FHH depends on user familiarity, frequency of use, the quality of its content, and the skill of its operator (Welch et al. 2015). Unfortunately, these dependencies have not been systematically

addressed and the digital promise remains woefully unfulfilled (Ginsburg et al. 2019). Disciplined collection of FHH by clinicians remains low and recording is fragmented and differently formatted.

The barriers to optimal collection and use are diverse and well documented (Welch et al. 2015). Among them, time to collect and record, inadequate EHR user interface, lack of EHR interoperability, and inadequate provider education are often cited (Houwink et al. 2014; Manolio et al. 2013; David et al. 2015). Lack of robust FHH data and barriers to its effective sharing also hamper public and preventative health programs' ability to target screening for inherited disease to those at highest risk (Valdez et al. 2010). The net result is ineffective use of key information in patient care.

Methods

As these issues exist in similar form across the globe, irrespective of healthcare enterprise type, an open membership community of professionals, scientists, and clinicians has coalesced to create a declaration with the intent of bringing the opportunities into focus and of supporting and inspiring stakeholders' demands for change.

The Family Health History Group embodies decades-long efforts begun by the US Office of the Surgeon General to connect FHH stakeholders, researchers, and thought leaders to frame the value of FHH for patients and for the healthcare enterprise and encourage related policy development and routine practice. This multi-disciplinary group with members from at least three countries is coordinated by National Human Genome Research Institute staff at the US National Institutes of Health. Members (<https://www.genome.gov/Health/Family-Health-History/Health-Professionals/Family-Health-History-Group>) from government, industry, primary and specialty health care, research, advocacy, and policy backgrounds volunteer and teleconference regularly to share learning, understand barriers in depth, discuss issues and potential solutions, and host speakers working in the FHH field.

Members of the group met multiple times between 2019 and 2020 with the intent of bringing new focus to the current needs. Discussion threads, input from expert FHH stakeholders, and consensus elements contributing to the conceptual framework of this document were collected using Google Docs. The authors, with the consent of the contributing members, then assembled these concepts into implementation gaps and achievable strategies for reducing them.

Results and discussion

The key FHH implementation gaps and domain-related strategies for reducing them are outlined in Table 1. These are neither prescriptive nor exhaustive but represent the

Table 1 Suggested strategies to reduce implementation gaps in family health history

Gap: patients' knowledge and appreciation of FHH value:

- Educate patients about the importance of inherited genetic risks and FHH's role in uncovering them (Sanghavi et al. 2019)
- Disseminate engaging, online, public-oriented, learning experiences about FHH
- Leverage existing intrafamilial interaction behaviors and patterns to facilitate FHH sharing and collection (Hood 2018)
- Exercise linguistically and culturally aware methods and apply knowledge of unique barriers in communities with health disparities and diverse backgrounds (Hood 2018; Lin et al. 2018; Sanghavi et al. 2019; Malen et al. 2016; Cerda et al. 2019)
- Increase attentiveness to FHH and enhance recollection among patients by facilitating collection, documentation, and sharing of FHH with family members and providers

Gap: providers' knowledge of FHH collection and usage:

Educate providers in how to collect and use FHH in modern medical practice, including efficient, accurate collection in limited time during patient encounters (Harding et al. 2019; Hull et al. 2020; Houwink et al. 2014; Bennett 2019)

- Deliver convenient provider training online
- Support systematic FHH expert participation in establishing training requirements and competencies, and in implementing them
- Standardize practitioner continuing medical education and require maintenance of certification in FHH
- Highlight the frequency of actionable FHHs and missed opportunities in primary care settings, including for preventative care

Gap: workforce optimization:

- System planners must anticipate heightened integration of family and genetic testing data in primary care
- Clearly delineate expectations for FHH collection and use of genetic professionals by primary care providers
- Increase the numbers of geneticists and genetic counselors in clinical care
- Ensure that support for non-geneticists is routinely accessible from genetic counselors or other FHH experts (Harding et al. 2019; Hull et al. 2020)

Gap: effectiveness of digital tools and interoperability with and among EHRs:

- Design FHH tools for patients and providers around modern smart device capabilities
- Make widely available FHH tools that increase information quality and user efficiency and that interoperate with EHRs
- Require that FHH information recorded in EHRs is coded and computable while protecting provider interface usability
- Leverage clinical decision support systems in EHRs to flag the most actionable FHH data (Lemke et al. 2020)
- Modernize electronic FHH collection and retrieval tools (Welch et al. 2020)
- Standardize FHH data representations and ensure that FHH data structures reflect the diversity of family structures
- Allow FHH tools to link among family members' records to optimize information accuracy and consistent use
- Ensure that role-specific tools, including FHH risk scoring systems, are available to health professionals trained in their use
- Digitize paper based FHH data including pedigrees and attach to discrete data stores

Gap: healthcare administration policies that facilitate FHH collection:

- Ensure that FHH collection is a reimbursed activity
- Harmonize and expand coding systems for FHH procedures and in FHH collection and data input
- Evolve unambiguous and practical guidelines for FHH collection, use, and referral for genetics care, using existing domain-specific models
- Build evidence suitable for guideline recommendations by US Preventative Services Task Force and other preventative health policy makers and enforcers

consensus of diverse FHH experts regarding major targets where concerted efforts may have the greatest yield, and which are within reach. The strategies suggested here are achievable today and have impacts that will benefit current and future generations.

Importantly, no single entity is positioned to transform the FHH field. We thus challenge healthcare leaders, administrators, policymakers, providers, informaticists, and implementers to inventory their FHH gap status and to formulate specific and achievable goals using the strategies presented here, guided by existing research. Activating such collaborative quality improvement projects is needed to realize the full potential of FHH in healthcare, research, and public health.

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Declarations

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Consent for publication Not applicable.

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