Modernizing family health history: achievable strategies to reduce implementation gaps

Wildin, R.S.; Messersmith, D.J.; Houwink, E.J.F.

SHORT COMMUNICATION

Modernizing family health history: achievable strategies to reduce implementation gaps

Robert S. Wildin[1,2] · Donna J. Messersmith[3] · Elisa J. F. Houwink[4]

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 FFH 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

✉ Donna J. Messersmith
donna.messersmith@nih.gov

1   Department of Pathology & Laboratory Medicine (Genomic Medicine), University of Vermont Larner College of Medicine, VT, Burlington, USA

2   Department of Pediatrics (Genetics), University of Vermont Larner College of Medicine, VT, Burlington, USA

3   Education and Community Involvement Branch, Division of Genomics and Society, National Human Genome Research Institute, National Institutes of Health, 31 Center Drive, Bethesda, MD 20892-2070, USA

4   Department of Public Health and Primary Care (PHEG), Leiden University Medical Center, Leiden, The Netherlands

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. FFH 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.

Data Availability Not applicable.

Code availability Not applicable.

Declarations

Ethics approval Not applicable.

Consent to participate Not applicable.

Consent for publication Not applicable.

Competing interests The authors declare no competing interests.

References

Allen CG, Duquette D, Guan Y, McBride CM (2020) Applying theory to characterize impediments to dissemination of community-facing family health history tools: a review of the literature. J Community Genet 11(2):147–159

Bennett RL (2019) Family health history: the first genetic test in precision edicine. Med Clin North Am 103(6):957–966

CerdaDiez M, Cortés DE, Trevino-Talbot M, Bangham C, Winter MR, Cabral H, Norkunas Cunningham T, Toledo DM, Bowen DJ, Paasche-Orlow MK, Bickmore T, Wang C (2019) Designing and evaluating a digital family health history tool for Spanish speakers. Int J Environ Res Public Health. 16(24):4979

David SP, Johnson SG, Berger AC, Feero WG, Terry SF, Green LA, Phillips RL Jr, Ginsburg GS (2015) Making personalized health care even more personalized: insights from activities of the IOM Genomics Roundtable. Ann Fam Med 13(4):373–380

Ginsburg GS, Wu RR, Orlando LA (2019) Family health history: underused for actionable risk assessment. Lancet 394(10198):596–603

Guttmacher AE, Collins FS, Carmona RH (2004) The family history–more important than ever. N Engl J Med 351(22):2333–2336

Harding B, Webber C, Ruhland L, Dalgarno N, Armour CM, Birthistle R, Brown G, Carroll JC, Flavin M, Phillips S, MacKenzie JJ (2019) Primary care providers' lived experiences of genetics in practice. J Community Genet 10(1):85–93

Hood SM (2018) Enhancing cultural considerations in networks and health: a commentary on racial differences in family health history knowledge and interpersonal mechanisms. Transl Behav Med 8(4):550–553

Houwink EJ, Muijtjens AM, van Teeffelen SR, Henneman L, Rethans JJ, van der Jagt LE, van Luijk SJ, Dinant GJ, van der Vleuten C, Cornel MC (2014) Effectiveness of oncogenetics training on general practitioners' consultation skills: a randomized controlled trial. Genet Med 16(1):45–52

Houwink EJF, Hortensius OR, van Boven K, Sollie A, Numans ME (2019) Genetics in primary care: validating a tool to pre-symptomatically assess common disease risk using an Australian questionnaire on family history. Clin Transl Med 8(1):17

Hull LE, Gold NB, Armstrong KA (2020) Revisiting the roles of primary care clinicians in genetic medicine. JAMA 324(16):1607–1608

Hussein N, Malik TFA, Salim H, Samad A, Qureshi N, Ng CJ (2020) Is family history still underutilised? Exploring the views and experiences of primary care doctors in Malaysia. J Community Genet 11(4):413–420

Lemke AA, Thompson J, Hulick PJ, Sereika AW, Johnson C, Oshman L, Dunnenberger HM (2020) Primary care physician experiences utilizing a family health history tool with electronic health record-integrated clinical decision support: an implementation process assessment. J Community Genet 11(3):339–350

Lin J, Marcum CS, Myers MF, Koehly LM (2017) Put the family back in family health history: a multiple-informant approach. Am J Prev Med 52(5):640–644

Lin J, Marcum CS, Myers MF, Koehly LM (2018) Racial differences in family health history knowledge of type 2 diabetes: exploring the role of interpersonal mechanisms. Transl Behav Med 8(4):540–549

Malen R, Knerr S, Delgado F, Fullerton SM, Thompson B (2016) Rural Mexican-Americans' perceptions of family health history, genetics, and disease risk: implications for disparities-focused research dissemination. J Community Genet 7(1):91–96

Manolio TA, Chisholm RL, Ozenberger B, Roden DM, Williams MS, Wilson R, Bick D, Bottinger EP, Brilliant MH, Eng C, Frazer KA, Korf B, Ledbetter DH, Lupski JR, Marsh C, Mrazek D, Murray MF, O'Donnell PH, Rader DJ, Relling MV, Shuldiner AR, Valle D, Weinshilboum R, Green ED, Ginsburg GS (2013) Implementing genomic medicine in the clinic: the future is here. Genet Med 15(4):258–267

Orlando LA, Wu RR, Myers RA, Neuner J, McCarty C, Haller IV, Harry M, Fulda KG, Dimmock D, Rakhra-Burris T, Buchanan A, Ginsburg GS (2020) At the intersection of precision medicine and population health: an implementation-effectiveness study of family health history based systematic risk assessment in primary care. BMC Health Serv Res 20(1):1015

Qureshi N, Armstrong S, Dhiman P, Saukko P, Middlemass J, Evans PH, Kai J, ADDFAM (Added Value of Family History in CVD Risk Assessment) Study Group (2012) Effect of adding systematic family history enquiry to cardiovascular disease risk assessment in primary care: a matched-pair, cluster randomized trial. Ann Intern Med. 156(4):253–62

Sanghavi K, Moses I, Moses D, Gordon A, Chyr L, Bodurtha J (2019) Family health history and genetic services-the East Baltimore community stakeholder interview project. J Community Genet 10(2):219–227

Valdez R, Yoon PW, Qureshi N, Green RF, Khoury MJ (2010) Family history in public health practice: a genomic tool for disease prevention and health promotion. Annu Rev Public Health 31:69–87

Welch BM, Dere W, Schiffman JD (2015) Family health history: the case for better tools. JAMA 313(17):1711–1712

Welch BM, Allen CG, Ritchie JB, Morrison H, Hughes-Halbert C, Schiffman JD (2020) Using a Chatbot to assess hereditary cancer risk. JCO Clin Cancer Inform 4:787–793

Wu RR, Orlando LA, Himmel TL, Buchanan AH, Powell KP, Hauser ER, Agbaje AB, Henrich VC, Ginsburg GS (2013) Patient and primary care provider experience using a family health history collection, risk stratification, and clinical decision support tool: a type 2 hybrid controlled implementation-effectiveness trial. BMC Fam Pract 14:111

Wu RR, Orlando LA (2015) Implementation of health risk assessments with family health history: barriers and benefits. Postgrad Med J 91(4):508–513

Wu RR, Myers RA, Sperber N, Voils CI, Neuner J, McCarty CA, Haller IV, Harry M, Fulda KG, Cross D, Dimmock D, Rakhra-Burris T, Buchanan AH, Ginsburg GS, Orlando LA (2019) Implementation, adoption, and utility of family health history risk assessment in diverse care settings: evaluating implementation processes and impact with an implementation framework. Genet Med 21(2):331–338