Development and Validation of a Primary Care-Based Family Health History and Decision Support Program (MeTree)

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ORIGINAL ARTICLE

Development and Validation of a Primary Care-Based Family Health History and Decision Support Program (MeTree)

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INTRODUCTION Family health history is a strong predictor of disease risk. To reduce the morbidity and mortality of many chronic diseases, risk-stratified evidence-based guidelines strongly encourage the collection and synthesis of family health history to guide selection of primary prevention strategies. However, the collection and synthesis of such information is not well integrated into clinical practice. To address barriers to collection and use of family health histories, the Genomedical Connection developed and validated MeTree, a Web-based, patient-facing family health history collection and clinical decision support tool. MeTree is designed for integration into primary care practices as part of the genomic medicine model for primary care.

METHODS We describe the guiding principles, operational characteristics, algorithm development, and coding used to develop MeTree. Validation was performed through stakeholder cognitive interviewing, a genetic counseling pilot program, and clinical practice pilot programs in 2 community-based primary care clinics.

RESULTS Stakeholder feedback resulted in changes to MeTree's interface and changes to the phrasing of clinical decision support documents. The pilot studies resulted in the identification and correction of coding errors and the reformatting of clinical decision support documents. MeTree's strengths in comparison with other tools are its seamless integration into clinical practice and its provision of action-oriented recommendations guided by providers' needs.

LIMITATIONS The tool was validated in a small cohort.

CONCLUSION MeTree can be integrated into primary care practices to help providers collect and synthesize family health history information from patients with the goal of improving adherence to risk-stratified evidence-based guidelines.

Family health history is widely accepted as a critical component of patient care. It serves as one of the strongest predictors of disease risk, provides information about environmental exposures, and has implications for the health of other family members. Many guidelines for screening and prevention—for instance, those of the American College of Cardiology for cardiovascular disease [1] and those of the American Gastroenterological Association for colon cancer [2]—strongly recommend (based on Level 1 evidence) that primary care providers collect family health history for disease risk stratification and risk management.

However, there are several barriers to gathering a family health history that is detailed enough to perform risk stratification and to guide clinical care: lack of time, due to competing clinical demands [3-7]; limitations in the patient's knowledge of his or her family's health history [8]; lack of reimbursement for the time spent collecting the history [9]; and lack of training in gathering family health history [3]. In addition, interpreting family health history information is difficult, particularly when this information is not available at the point of care, when it is not presented in a way that facilitates synthesis (eg, when it is scattered throughout the medical record and inconsistently formatted), or when it is not clearly actionable [9]. Unfortunately, adoption of electronic medical records (EMRs) has not improved family health history collection rates, and EMRs do not address the barriers listed previously [10]. This finding is supported by a review conducted by our group (results of which are published in this issue on pages 279-286) [11] that compares family health histories documented in paper charts (less than 4% of which were high-quality family health histories) with those documented in EMRs at the same clinic (less than 1% of which were high-quality family health histories).

Despite the limitations of EMRs, advances in technology are a key to overcoming many, if not all, of the barriers to family health history collection and use. For example, computer-based software programs that are referred to as "patient-facing" allow direct entry of family health history information by patients, which avoids the pressure to collect the data during the primary care appointment and permits patients adequate time to collect the necessary information from relatives. Such programs can also be linked to education on how to collect family histories, and they use auto-

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mated algorithms to generate clinical decision support for analysis and interpretation of the history collected.

In 2004, the Genomedical Connection—a collaboration on the part of Duke University, the University of North Carolina at Greensboro, and Cone Health System—obtained funding from the US Department of Defense to implement the genomic medicine model for primary care. The model is described in more detail elsewhere [12]. Briefly, it interweaves education, family health history collection, and recommendations for family health history risk-based prevention strategies within the clinical workflow of primary care practices. At the core of this model was the development of a self-administered, patient-facing computerized program, called MeTree, which gathers family health history from patients and generates decision support for patients and providers. In this paper, we describe the conceptual foundation, development, and validation of MeTree and compare its features with those of existing decision support programs based on family health history.

Methods

Program design goals. When we began developing MeTree in 2004, our clinical experience with family health history collection, the published literature, and a review of existing programs led us to conclude that a family health history software program should have the following characteristics, in order to be acceptable within clinical practice: It should improve clinical workflow by having patients enter their own family health history prior to an appointment with the provider; it should have point-of-care risk stratification; and it should facilitate uptake of risk-stratified preventive care recommendations that are clear, action-oriented, and evidence-based [13-15]. Therefore, we developed the following design goals for MeTree: (1) to develop a family health history collection interface that is easy for patients to use and that facilitates the collection of all the necessary components to perform risk stratification (ie, a full 3-generation pedigree with age of disease onset, current age or age at death, and cause of death for each relative); (2) to provide lay-level and technical decision support that is clinically actionable for providers and easy for patients and providers to understand; (3) to base decision support on guidelines that are widely accepted by primary care physicians; and (4) to capitalize on the patient-provider encounter to encourage discussions of preventive health and disease risk management.

MeTree description. MeTree, a stand-alone Web-based program, has 2 components: family health history collection and decision support. These 2 components were developed concurrently in order to maximize the effectiveness of each. For example, the Gail model [16, 17] was incorporated within the decision support component to identify women for whom breast cancer chemoprevention should be considered. Including this model and associated questions regarding chemoprevention and its contraindications required the addition of questions that are not routinely collected by family health history screening programs.

Family health history collection and decision support were developed by a team of 4 genetic counselors (with expertise in adult, pediatric, and cancer genetics), 3 medical geneticists, a cardiologist, a health behaviorist, 2 medical oncologists, and 3 experts in information technology. An iterative Delphi-based approach [18], along with a concurrent literature review, was used to reach a consensus regarding which professional guidelines and expert opinions to base the algorithms on and which conditions to include. Given that MeTree is intended for use in primary care clinical practices, preference was given to guidelines with which primary care physicians would be familiar (eg, those of the US Preventive Services Task Force and the American Cancer Society).

Family health history collection. The family health history collection component is the main patient interface. Patients use a Web-based survey that first establishes the family's structure, with the names and ages (current age or age at death) for 4 generations of relatives, and then identifies which relatives have been affected by any of 48 potential conditions (See Table 1). These conditions were selected by compiling a list of important familial and hereditary conditions and ranking them based on the strength of their familial risk and their importance to primary care providers. In order to maximize the effectiveness of the tool while minimizing the burden of using it, only the top-ranked 48 conditions were incorporated into MeTree.

To facilitate its ease of use, MeTree runs in full-screen mode, showing only questions and response fields, without toolbars or menus that could clutter the window. All fields are touch-screen capable, and fonts and buttons are large and easy to read. Survey questions are written at an 8th-grade reading level when possible. Use of branching questionnaire logic allows MeTree to skip irrelevant survey question screens, which minimizes the time patients need to complete the survey. Family health histories can also be updated and the algorithms rerun as needed.

Of note, a second MeTree interface was developed in 2012, employing a graphical user interface and tablet technology. Rather than relying on radio buttons and text-based input of family structure and health history, the new interface uses graphics, drag-and-drop technology for adding relatives, and drop-down lists that expand and minimize as desired to facilitate quick selection of health history by relative. Zooming, panning, and swiping facilitate rapid data entry.

Decision support. MeTree provides decision support for diseases that have a strong impact on population health, either because they are highly prevalent or because they have high morbidity and/or mortality (high clinical validity). The program also provides established risk-stratified screening and preventive care strategies that are known to have high clinical utility. Using these criteria, 5 pilot diseases

TABLE 1.
Health Conditions Included in MeTree

Cancers	Other conditions	Hereditary cancer syndromes
Brain cancer	Alzheimer disease/dementia	Hereditary breast and ovarian cancer (BRCA1/BRCA2 genes)
Breast cancer	Anemia	Hereditary nonpolyposis colon cancer (MLH1/MSH2/MSH6 genes)
Cervical cancer	Asthma	Familial adenomatous polyposis (APC gene)
Colon cancer	Blood clots in veins	Li Fraumeni syndrome (TP53 gene)
Kidney cancer	Colon polyps	Cowden syndrome (PTEN gene)
Leukemia	Diabetes	Other cancer syndromes
Liver cancer	Glaucoma	
Lung cancer	Heart attack	
Lymphoma	High blood pressure	
Melanoma	High cholesterol levels	
Ovarian cancer	Inflammatory bowel disease	
Pancreatic cancer	Lupus	
Prostate cancer	Macular degeneration	
Skin cancer (not melanoma)	Multiple miscarriages	
Small bowel cancer	Multiple sclerosis	
Stomach cancer	Osteoporosis	
Testicular cancer	Parkinson disease	
Thyroid cancer	Rheumatoid arthritis	
Uterine cancer	Seizures	
Unknown cancer	Stroke	
Other cancer, specify	Thyroid disease	

Note. Modified from Orlando et al. [19]

(breast cancer, ovarian cancer, colon cancer, thrombosis, and risk for a hereditary cancer syndrome) were chosen to demonstrate MeTree's effectiveness and acceptability. The decision support process risk-stratifies patients into one of several risk levels according to their family health history, and it then links the risk level to an action-oriented risk-management recommendation.

To foster discussion about risk, risk management, and disease prevention, decision support and structured family health history documents are given to patients (pedigree and patient report) and providers (pedigree, family health history in a tabular format, and provider report) prior to the scheduled appointment time. These documents are designed to be simple to read, with straightforward messages that are specific to the intended recipient. The patient report (Figure 1), written at an 8th-grade reading level, summarizes key points that patients might want to discuss with the provider regarding their family health history–based risk for the pilot diseases. The provider report (Figure 2) begins with an evidence-based action plan driven by the patient's estimated disease risk; this action plan is followed by a more detailed description of the criteria triggering each recommendation, along with relevant references. The increasing level of detail available in the provider report allows for just-in-time education determined by provider interest, curiosity, or need.

Decision support risk categories and their associated action-oriented risk-management strategies for thrombosis are as follows (in order of decreasing risk): genetic testing for inherited thrombophilia, with referral to genetic counseling; referral to genetic counseling alone; or no recommended intervention. Categorization for thrombosis is based on guidelines of the American College of Chest Physicians [20].

Decision support risk categories and their associated action-oriented risk-management strategies for breast cancer, ovarian cancer, colorectal cancer, and hereditary cancer syndrome are as follows (in order of decreasing risk): referral to genetic counseling, increased personal and familial risk managed by provider, and routine population-based screening. An algorithm evolved in which patients who met the criteria for genetic counseling referral were identified first, and then the patients at familial or population risk were identified. Women without a personal history of breast or ovarian cancer are selected for genetic counseling referral if they meet US Preventive Services Task Force guidelines [21]. However, these guidelines do not apply to men or to women who already have breast or ovarian cancer, so expert opinion [22] and the published guidelines of the National Society of Genetic Counselors [23] are used to select patients from these 2 groups who should be referred to a genetic counselor due to elevated risk of hereditary breast and ovarian cancer syndrome. Patients who meet the Amsterdam II diagnostic criteria for hereditary nonpolyposis colorectal cancer [24] or criteria established based on expert opinion [22] are selected for referral to discuss hereditary colorectal cancer risk.

FIGURE 1.
Example of Decision Support: Patient Report

MeTree© Personalized Profile for (ID: 12012)
based on your answers to Questionnaire #1624 on 07/08/2010

Talk to your doctor about:	Why?	More information
Referral to a genetic counselor	There's an increased chance that cancer runs in your family for these reasons. **You have:** • At least 3 members in your family with the same cancer.	
Regular colon cancer screening	Your chances of colon cancer increase with age. This is why most people should have regular screening beginning at age 50.	Several colon cancer screening tests have been shown to be effective. Talk with your doctor about the one that's right for you.

The information is based on facts you entered into MeTree©. It may not be accurate if facts are not correct. This program does not take into account all factors that may influence disease risk. Talk with your doctor about how other factors, such as health habits, influence disease risk. Based on your needs, a genetic counselor may suggest additional screenings that are not included in this report.

Note. Reprinted from Orlando et al [19].

The category "personal and familial risk management" includes patients whose risk for the 3 types of cancer does not warrant genetic counseling referral but is sufficient to merit consideration of surveillance or chemoprevention. Breast magnetic resonance imaging as an adjunct to mammography is recommended for those who meet American Cancer Society guidelines for breast cancer risk (eg, those with a lifetime risk greater than 20%) [25]. Lifetime risk is calculated using BRCAPRO, a statistical model and software program using Mendelian genetics and Bayesian updating. BRCAPRO incorporates the following information for patients and their first-degree and second-degree relatives (including those without cancer): sex; current age or age at death; diagnosis of breast cancer, second primary breast cancer, or ovarian cancer; age at cancer diagnosis; and presence or absence of Ashkenazi Jewish ancestry [26]. Breast cancer chemoprevention with tamoxifen or raloxifene is recommended for women aged 35–60 years whose 5-year breast cancer risk exceeds 1.65% [27, 28]. Five-year risk is calculated using the Gail model [16]. BRCAPRO and Gail model risk scores are included in the provider's report. Colorectal cancer surveillance is recommended for those who meet the joint guidelines of the American Cancer Society, the US Multi-Society Task Force on Colorectal Cancer, and the American College of Radiology [29]; such surveillance often involves scheduling the first colonoscopy at an earlier age and performing follow-up colonoscopies more frequently.

"Average risk" patients who do not meet criteria for genetic counseling referral or familial or personal risk management are managed according to the American Cancer Society recommendations for individuals at population risk for cancer [30].

Genetic counselors on the team are using their clinical expertise and regular literature reviews to ensure that the clinical algorithms and decision support recommendations remain current. One of the genetic counselors reviews a random sample of pedigrees, patient reports, and provider reports on a monthly basis to identify inaccuracies and misclassifications. These are reported to a working group of content and information technology experts, who review and correct the algorithm code and make content changes as necessary.

Coding. Several open-source computer software applications are incorporated into MeTree. These include PHP (version 5.4.9), a widely used general-purpose scripting language that is especially suited for Web development; Apache HTTP Server, a popular Web server; and Linux, a free UNIX-type operating system. Other programming resources include C++, which is used to calculate the Gail Score for 5-year breast cancer risk; R, an open-source statistical package used to calculate lifetime breast cancer risk using the BRCAPRO model with the BayesMendel R library; VBScript (Visual Basic Scripting Edition), a scripting tool provided with the Microsoft Windows operating system that is used

to load provider and patient visit data; and Microsoft SQL Server, a relational database management system used to model decision support algorithms.

The MeTree Admin utility is a Web-based application written in PHP that provides administrators and clinical coordinators secure access to patient data and questionnaire data. It permits updating of patient contacts (letters, phone conversations, etc), mail merging of data sources for printing introduction letters for prospective participants, printing of postquestionnaire pedigree and summary reports, and display of important data elements for patient tracking and ongoing quality evaluation. For example, it can display demographics, patients by date of visit, questionnaires completed, and patients who were no-shows or who declined participation.

Validation

To optimize the collection of family health history and the development of decision support algorithms and reports, pilot testing was carried out in several phases prior to implementation of MeTree in primary care practices. The first phase involved testing with community volunteers, and the second phase involved testing with genetic counselors. Finally, the third phase was a 3-year pilot test within 2 different primary care practices, during which feedback from providers and patients was used to optimize clinical workflow and report content [31].

FIGURE 2.
Example of Decision Support: Provider Report

02/25/2010 **MeTree Personalized Risk Profile**

MeTree ID: #1234
Questionnaire: #9999
Patient X
DOB: 5/25/1965 Age: 41
BMI: 28

ACTIONABLE ITEMS

- **Refer to genetic counseling for comprehensive INHERITED THROMBOPHILIA risk assessment & management** [1]
- **Refer to genetic counseling for comprehensive CANCER risk assessment & management** [2,3,7-9]
- **Coordinate risk management for HNPCC syndrome according to NCCN guidelines (www.nccn.org)**
- **Discuss chemoprevention for breast cancer (tamoxifen)** [5,6]

INDICATIONS

Personal History

- Venous thrombosis in unusual location (head, neck, arm or abdomen).
- Patient meets Amsterdam II criteria for clinical diagnosis of HNPCC syndrome.
- Patient's 5-year breast cancer risk (Gail model estimate = ____%) exceeds cut-off of 1.65%.

Family History

- At least 1 first-degree relative was diagnosed with colorectal cancer < age 50.
- At least 3 relatives with HNPCC-related cancers (colorectal, uterine, gastric, ovarian, renal, small bowel, pancreatic, brain).

Contraindication(s)/Other Factors to Consider:

- Patient using oral estrogen or progesterone.
- Patient has had stroke.
- Patient has had blood clot(s).
- Refer to pedigree for additional indication(s) relating to thromboembolism

NOTE(S)

NOTE(S):

- Tamoxifen's effectiveness for breast cancer chemoprevention has not been tested in women who are under age 35, pregnant, breastfeeding, or taking hormone replacement therapy.
- Tamoxifen is associated with increased risk of endometrial cancer and thromboembolic events.
- Check patient's previous tamoxifen use.

MeTree© Assessment Tool recommendations are based on information supplied by patient. They may not represent a complete clinical assessment and are not intended to supplant physician discretion in risk management. Based on your needs, a genetic counselor may suggest additional screenings that are not included in this report.

1Chest Guidelines Chest 126; 3 September 2004 Supplement 401S
2U.S. Preventive Services Task Force. Ann Intern Med. 2005;143:355-61.
3Hampel H et al. J Med Genet. 2004;41:81-91.
4Smith RA et al. CA Cancer J Clin. 2008;58:161-79.
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7Berliner JL et al. J Genet Counsel. 2007;16:241-60.
8Levin B et al. CA Cancer J Clin. 2008;58:130-60.
9Vasen HF et al. Gastroenterology. 1999;116:1453-6.
10National Comprehensive Cancer Network. 2008. http://www.nccn.org/professionals/physician_gls/
11Saslow D et al., CA Cancer J Clin. 2007;57:75-89.
12Berry DA, et al., J Clin Oncol. 2002;20:2701-2712.

Note. Reprinted from Orlando et al [19].

TABLE 2.
Features of Electronic Primary Care Decision Support Programs

Program	Diseases covered	Who enters the information?	Who receives the output?	Availability of output at point of care?	Public availability of program
MeTree	colon cancer, breast cancer, ovarian cancer, and hereditary cancer syndrome risk	Patient (online or in the physician's office)	Patient and physician	Yes	In future
Program described by Schroy et al. [36]	colon cancer	Physician	Physician	Yes	Unknown
Genetic Risk Assessment in the Clinical Environment (GRACE) [37]	breast cancer[a]	Patient (in the physician's office)	Patient, clinical nurse specialist, or physician	Yes	Unknown
Family Healthware [38]	coronary heart disease, diabetes, stroke, colon cancer, breast cancer, and ovarian cancer	Patient (online)	Patient or physician	Unknown	No
Family Health*Link* [39]	coronary heart disease, cancer	Patient (online)	Patient	No	Yes
Cancer Risk Intake System	colon cancer	Patient (in the physician's office)	Patient and physician	Yes	No
MyGenerations [40]	cancer	Patient (online)	Patient	No	Yes
HughesRiskApps [41]	breast and ovarian cancer	Patient or clinician (can be revised online or in the physician's office)	Patient and physician	Yes	Yes
Health Heritage [13]	87 diseases: including multiple cancers, diabetes, neuromuscular diseases, and cardiovascular diseases	Patient (online)	Patient	No	No

[a]GRACE is designed for use only with women who have a family history of breast cancer.

Community pilot program. The phrasing and clarity of the questions used to collect family health history were assessed via cognitive interviews with community volunteers, a technique that has been successfully used across diverse populations to ensure that health materials are understood as researchers intend [32-34]. Volunteers were acquired through convenience sampling: They were recruited from the pool of visitors and staff members entering Moses Cone Hospital and were offered a $20 gift card for participating. The volunteers were each asked to read the family health history collection questions one at a time and then tell the interviewer, a team member trained in cognitive interviewing, what the question meant to them and whether the question was clear. Usability of the family health history collection interface was evaluated by asking volunteers to complete the family health history collection and then to comment on screen layout, skip patterns, fonts, button size, and other formatting features. The amount of time required to complete the collection of family health history was tracked for each volunteer. Based on these results, the development team revised the content and presentation of the questions and then repeated the item phrasing and usability testing until saturation was reached—that is, until no new comments were given.

Genetic counselor pilot program. Ten cancer counselors and 3 thrombophilia genetic counselors, all of whom had had no prior interaction with MeTree, were recruited through local professional networks to assess the usability, quality, and thoroughness of the decision support content and the accuracy and clarity of the risk algorithms and recommendations. Each counselor entered at least 5 sample cases into MeTree, reviewed the decision support output, and completed an online survey developed by the study team. The following are examples of questions used to assess usability: "What problems do you foresee patients having with MeTree?"; "On a scale of 1 (not easy at all) to 5 (very easy), how easy will it be for your patients to understand the questions?"; and "What technical problems, if any, did you encounter?" Additional questions were used to assess content, such as: "What questions, if any, did you expect to see that pertain to cancer risk, but didn't?"; "Is the pedigree and risk report generated by MeTree more helpful than the typical referral information you receive (Y/N)?"; and "In your professional opinion, are the recommendations on the provider report consistent with the sample patient's level of risk for colon cancer?" The algorithms and reports were revised to address areas of deficiency or inaccuracy.

Clinical practice pilot program. MeTree was integrated into 2 community-based primary care clinical practices in the Cone Health system in Greensboro, North Carolina, as part of a hybrid type II implementation-effectiveness trial. (More information about how such trials are designed can be found in a 2012 article by Curran and colleagues [35].) Details of our study design can be found in the published protocol

paper [19]. The 2 practices, which have served the community for almost 20 years, care for more than 21,000 unique patients annually and are staffed by 13 primary care providers (12 internal medicine or family medicine physicians and 1 nurse practitioner). Both practices used paper charts at the time of implementation, but each converted to an EMR system (although to 2 different systems) during the 3-year pilot program. During the first 6 months of the trial, implementation research methodology was used to assess and adapt the implementation effort, and a clinical expert assessed the accuracy of the risk algorithms and recommendations.

To address the quality and impact of integration into clinical workflow, a study coordinator was embedded into each clinical practice. The study coordinators verbally conducted daily cycles of feedback from stakeholders (patients, clinic staff, and providers), and the lead investigator, an internist with training in health services research, conducted monthly cycles of verbal stakeholder feedback. The study coordinator recorded all questions asked by patients during the visit as well as any unprompted questions asked by clinic staff members and providers; the study coordinator also asked staff members and providers specific questions. All stakeholders were encouraged to provide open and honest responses to open-ended questions such as: "What has your experience been so far?"; "What could we improve?"; What barriers are you encountering?"; "Do you have any concerns?"; "Is the report content and format clear and helpful?"; and "How and when are you receiving reports?" Responses were clarified using funneling questions to elicit greater detail regarding who, what, why, how, and when; the clarified responses were then used to adapt components of the program (including usability, decision support documents, and workflow integration) that did not meet stakeholder needs. Clinic staff members (nurses and clerks) were asked the same questions as providers, but data for the 2 groups were analyzed separately.

To assess the accuracy of the programming, coding, algorithms, and report output, a genetic counselor reviewed every pedigree and its associated recommendation reports for patients enrolled during the first 6 months of the pilot program. Mismatches between pedigree input and algorithm or report output were identified and referred to a second genetic counselor for review. When both genetic counselors agreed that a mismatch was present, coding errors were identified and corrected.

Results

Community pilot program. A total of 19 individuals completed cognitive interviews during 3 iterative cycles of data collection and MeTree revision, after which saturation was reached. Among the 19 cognitive interviewees, 11 were female and 8 were male; 7 were African American and 11 were white; and 14 of the interviewees had some college or less education. Age was recorded in 5-year increments, and interviewees included at least 1 person in each age bracket from 18 to 70+ years. Interviewees suggested ways of simplifying and organizing MeTree's questions and proposed that disease definitions be added. As a result of these suggestions, longer questions were broken into multiple shorter questions, questions about maternal and paternal relatives were organized more intuitively, and pop-up boxes were added that defined diseases in lay terminology.

During 3 iterative cycles of usability testing, 22 individuals (16 females and 6 males; 7 African Americans and 15 whites) completed MeTree's family health history collection. These individuals were diverse in education (8 had less than a bachelor's degree) and age (the only age bracket that was not represented was 65–69 years). The average time to complete MeTree was 20 minutes. Comments from volunteers included recommendations to increase the size of the font and buttons, to provide clearer error messages, to employ fewer drop-down lists, to give users the option of using either a mouse or a touch screen, to add "don't know" as a response option, to emphasize important instructional words, and to make the status bar more prominent. The programming was also revised to allow users to more easily remove relatives who had been entered by mistake and to automatically save information as it is entered, thus allowing users to jump between screens.

Genetic counselor pilot program. Feedback from the cancer genetic counselors regarding MeTree's usability, question content, printed reports, and algorithms included the following recommendations: to facilitate navigation by employing skip patterns (eg, avoiding cancer-specific questions in unaffected relatives); to allow users to quickly and easily move back and forth between questions; and to ask about cancer genetic testing in the patient and his or her relatives. To address these suggestions, program developers added skip patterns throughout the program, "bread crumbs" to allow users to find earlier pages more easily, and a series of questions about specific tests for hereditary cancer syndromes.

Suggestions regarding report content and clarity, which made up the bulk of the feedback, included proposals that the following information be added: colon cancer recommendations based on polyp histology (adenomatous versus nonadenomatous); recently published guidelines on breast magnetic resonance imaging screening [25] and colorectal cancer surveillance [29]; and screening and surveillance recommendations for patients who are referred for genetic counseling, in case they decline counseling. Reports were revised accordingly.

Suggestions regarding algorithms included a recommendation that patients meeting Amsterdam II criteria be referred to genetic counseling for Lynch syndrome screening [24], and a recommendation that maternal and paternal relatives be coded separately to restrict the counting of relatives to one side of the family. This last suggestion was due to concern regarding over-referral to counseling, because some of MeTree's recommendations are based on the num-

ber of affected relatives (eg, having 3 relatives with the same cancer merits referral to genetic counseling).

Clinical practice pilot. The implementation of feedback from clinical staff was uniformly positive, and no changes were recommended. Among the 192 patients (mean age, 58 years; 58% female; 75% white) who were enrolled in the study during the first 6 months, feedback was also uniformly positive, except from some individuals older than 60 years who were uncomfortable using a computer. In those cases, no actionable feedback was provided. Providers indicated that patient flow was unaffected by integration of MeTree and that patient discussions and the clinical encounter were improved by the presence of the reports.

However, providers did make several recommendations regarding report content and organization. Initially, the beginning of the reports contained a risk management recommendation along with a significant amount of detail supporting the recommendation. In addition, to avoid offending providers, most recommendations were passively worded using terms such as "consider a discussion about . . ." All providers agreed that reorganizing the reports to highlight simple, clear, action-oriented plans was crucial to usability during normal clinic workflow. The additional details supporting the recommendations, such as the personal history or family health history triggers that elicited the recommendation; "special cases," such as when not to follow the recommendation; and a link to the guideline itself were all strongly endorsed as useful just-in-time education that the providers wanted to continue to see, but in a separate section. They also requested the addition of clinical data supporting the recommendation (including trial data, such as clinical validity and utility) and potential harms and benefits of the recommendation. Therefore, the report was modified to present a bulleted list of action items in a prominent section at the beginning of the report, followed by several sublevels of just-in-time education, each with an increasing level of detail to give providers the depth of knowledge they desired without negatively impacting their patient workflow.

Programming validation. During the genetic counselor review of pedigrees and reports for the 192 patients enrolled during the first 6 months of the clinical practice pilot, 52 participants (27%) were identified as having 73 potential mismatches between the pedigree and the provider or patient report. After review by a second genetic counselor, no error was found in 22 (30%) of the 73 potential mismatches, but the remaining 51 mismatches (70%) had errors that required programming revisions. Errors and solutions within this group were as follows: With regard to formatting, 13 (25%) mismatches involved pedigree spacing that was off and needed to be revised. With regard to clarity of the report's contents, 2 (4%) mismatches occurred because a MeTree question was not clear, and those questions were revised for clarity; 10 (20%) of the mismatches occurred because the text of the provider reports and the patient reports was unclear, so that text was revised for clarity; and 15 (29%) of the mismatches involved the pedigree being inconsistent with the reports to the provider and the patient, so relevant details were added to the pedigree, and a separate data report was generated. With regard to coding errors, 11 (22%) mismatches involved algorithm coding problems, so the coding was revised. Examples of coding corrections include adding an upper age limit to chemoprevention recommendations and adding affected maternal and paternal relatives separately. To maintain confidence in the accuracy of the coding, pedigrees were randomly reviewed for 1 year following the 6-month pilot phase, and no new errors were found.

Discussion

Primary care providers are expected to systematically collect family health history and to manage their patients' disease risks accordingly, yet many provider-level, patient-level, and system-level barriers in primary care impede the uptake of this deceptively complex activity [9]. This paper outlines the foundational goals, development, and stepwise validation of MeTree, a computerized, patient-entered family health history collection and decision support program that addresses many of the barriers to the collection of high-quality family health histories and use of this information for risk assessment. MeTree was adapted during each step of validation: testing with community volunteers for usability and understanding; testing with genetic counselors for usability, content, and accuracy; and testing in clinical practice for feasibility, uptake, and accuracy. The end result of this process is a valid tool optimized to promote uptake of family health history collection and implementation of guidelines for risk-stratified evidence-based prevention and screening in busy primary care practices.

A structured family health history is crucial to appropriate risk assessment in asymptomatic or presymptomatic individuals. Compared with other markers of disease risk in this group (ie, clinical variables), family health histories are more readily available and have higher odds ratios for predicting disease, and the collection of a family health history is frequently the first (and sometimes only) step in risk stratification. In addition, a growing number of guidelines rely on risk stratification to guide the prevention and screening strategy. Examples of conditions for which guidelines rely on risk stratification include the conditions for which MeTree provides clinical decision support (breast and ovarian cancer, colon cancer, hereditary cancer syndromes, and thrombosis) along with cardiovascular disease, cerebrovascular disease, inherited cardiomyopathies and arrhythmias, and inherited neurologic conditions. Given that primary care practices are medical homes for all patients regardless of their health status, these practices are the ideal place to carry out risk assessment and risk-guided prevention strategies, which places much of the emphasis (and the burden) on the primary care provider.

Our goal in developing MeTree was to relieve some of this

burden. MeTree has 5 of the 7 characteristics of the "ideal family history tool" described by Rich and colleagues [9]. MeTree is "patient-completed" and "adapted to patient age, gender, ethnicity, and common conditions" [9]. Our extensive pilot testing via cognitive interviewing, usability testing, genetic counselor review, and provider review showed it to be "brief, understandable and easy to use" [9]. Finally, MeTree "contains clinical decision support" at the point of care and "branches and prioritizes based on clinical significance" [9]. This combination of attributes, MeTree's focus on seamless integration within primary clinical practice workflow, and its emphasis on both the patient and the provider as recipients of different types of tailored information were unique at the time MeTree was developed. Table 2 shows a comparison of MeTree's characteristics with those of currently available electronic tools for family health history collection and decision support.

One limitation of MeTree is the lack of integration with a medical record (the most common request from providers). To our knowledge, none of the existing family health history tools directly integrate into EMR systems, although there may be institutions where local adaptions have overcome this barrier; these would be unique to the setting and permit only local integration. The reason for this lack of integration is simple, although not intuitive: Integration of a family health history collection tool into an EMR system comes with considerable limitations and complexities. Each EMR system structures family health history differently, requests different types of information (often not based on the need for risk assessment), and uses nonstandard formats (despite the push for compatibility with Health Level Seven [HL7] standards of health care informatics interoperability). In addition, compatibility with one EMR system frequently does not permit compatibility with another EMR system, or even with the same EMR system implemented in a different setting. With the current state of EMRs, the only way to directly integrate with EMRs would be to rebuild MeTree within each individual EMR system, restructuring their EMR data format in the process. The end result would be 100 different instances on 100 different systems, all of which would need algorithm maintenance and system support—an overwhelming task for an academic group with limited time and resources. Before integration can feasibly proceed, EMR systems need to adopt standards for data structure, storage, and transmission across systems.

Another limitation is that, despite extensive piloting among stakeholder groups, implementation of MeTree has been studied in only 2 clinics within a single community setting. Optimal performance in those clinics does not predict optimal performance in other settings. To promote broader uptake across a variety of settings, MeTree will need to be evaluated for uptake, fidelity, and performance in other types of settings.

The above limitations refer to the specific case of developing and validating MeTree; however, as a tool for clinical practice, its impact needs to be evaluated in the clinical environment, which we are doing as part of the hybrid type II implementation-effectiveness study that recently concluded at the pilot clinics. In order to continue to serve our goals of improving patient care and offloading provider work, we also intend to continue to do the following things: (1) optimize workflow integration by promoting family health history data standards and develop a demonstration of EMR integration; (2) add decision support for other conditions with risk-stratified evidence-based primary prevention guidelines; (3) enhance patient reporting by taking advantage of apps, online tools, and other tools to facilitate understanding and uptake of recommendations; and (4) incorporate patient behaviors, values, and preferences in the report's output to further personalize recommendations and adapt them to each patient's unique situation. NCMJ

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