

# Collection of Family Health History for Assessment of Chronic Disease Risk in Primary Care

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**BACKGROUND** Family health history can predict a patient's risk for common complex diseases. This project assessed the completeness of family health history data in medical charts and evaluated the utility of these data for performing risk assessments in primary care.

**METHODS** Family health history data were collected and analyzed to determine the presence of quality indicators that are necessary for effective and accurate assessment of disease risk.

**RESULTS** More than 99% of the 390 paper charts analyzed contained information about family health history, which was usually scattered throughout the chart. Information on the health of the patient's parents was collected more often than information on the health of other relatives. Key information that was often *not* collected included age of disease onset, affected side of the family, and second-degree relatives affected. Less than 4% of patient charts included family health histories that were informative enough to accurately assess risk for common complex diseases.

**LIMITATIONS** Limitations of this study include the small number of charts reviewed per provider, the fact that the sample consisted of primary care providers in a single geographic location, and the inability to assess ethnicity, consanguinity, and other indicators of the informativeness of family health history.

**CONCLUSIONS** The family health histories collected in primary care are usually not complete enough to assess the patient's risk for common complex diseases. This situation could be improved with use of tools that analyze the family health history information collected and provide risk-stratified decision support recommendations for primary care.

Primary care providers routinely see patients who are at risk for, or are affected by, common complex diseases, such as coronary artery disease, cancer, and diabetes [1-5]. Family health history is one of the strongest predictors of the patient's risk for common complex diseases, and collecting this information can dramatically improve identification of at-risk individuals [4]. For example, having 1 first-degree relative with breast cancer results in a woman's risk for the disease being 1.8 times higher, and having 2 first-degree relatives nearly triples her risk [6]. Overall, 82% of primary care patients have a familial risk for at least 1 common disease—coronary artery disease, stroke, diabetes, breast cancer, colon cancer, or ovarian cancer [5, 7, 8]—and that risk alters the prevention recommendations for the patient. For instance, 15%–20% of patients meet family health history criteria for beginning colonoscopy screenings before age 50 years [9].

Because primary care providers are frequently a patient's first point of contact with the health care system, they are well positioned to identify patients who are at increased risk for disease and to implement appropriate prevention strategies in order to lower risk or detect disease earlier [2]. Professional [10-12] and evidence-based [13] guidelines are widely available; these guidelines can be used to collect family health histories for common diseases, such as colon cancer [14-17], breast cancer [17-20], heart disease [12], and

diabetes [21, 22]. Such guidelines have been endorsed by primary care organizations [10, 23, 24], yet they are underutilized in primary care settings [25-27]. Barriers to their use include the time required to collect an accurate family health history, the need to balance the patient's agenda with the physician's goals, the difficulty of finding information about family health history within the chart, and the lack of procedures for quickly collecting and analyzing family health history data [2, 28-31]. Using patient-collected information about family health history and incorporating it into the electronic medical record (EMR) might help to overcome these barriers [32, 33].

Making recommendations based on familial risk requires an accurate and detailed family health history [26]. Key elements of such a history are that it covers 3 generations (grandparents; parents, aunts, and uncles; and half siblings and full siblings) and that it includes age of disease onset, relationship to the patient, and age and cause of death (if deceased) for each individual [29, 34-36]. It is also impor-

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tant that the family health history make note of common diseases that are *not* found in the family (a negative, or “unremarkable,” family history) [34]. Although most primary care providers collect a family health history, the documented elements vary [25, 26, 31].

The primary goal of this study was to assess the completeness of family health history data in the medical charts of primary care providers and to evaluate the utility of these data for providing patient risk assessments. The chart review was performed in selected primary care practices in a midsized community in the Southern United States.

## Methods

Paper and EMR charts were reviewed in 1 internal medicine and 2 internal medicine/family medicine community-based practices serving patients with a range of socioeconomic and insurance statuses. The size of the practices ranged from 4 providers seeing 1,700 patients per month to 9 providers seeing 4,000 patients per month.

**Data instrument.** Our chart review checklist consists of 32 questions (including 14 multiple-choice questions, 9 dichotomous [yes/no] questions, and 9 fill-in-the-blank questions) and a chart on which the reviewer can circle whether particular types of information are “always,” “sometimes,” or “never” documented for various relatives. (See Appendix 1; online version only). The checklist is divided into sections dealing with the patient’s demographics and personal health history, the patient’s insurance, date(s) of personal and family health history collection, family health history data, and specialty referrals based on family health history. Fifteen of the questions on our checklist (marked with an asterisk) were adapted from the 2006 chart audit tool developed by the Michigan Department of Community Health’s genomics team (D. Duquette, unpublished observations, 2012). Our checklist was piloted and modified to ensure that all of the necessary information would be obtained.

### APPENDIX 1. 2009-2010 Baseline Family History Chart Review

This appendix is available in its entirety in the online edition of this article. Please go to the NCMJ Web site <http://www.ncmedicaljournal.com/archives/?74402>.

The checklist and methodology were approved by the institutional review boards of the University of North Carolina at Greensboro, Moses H. Cone Memorial Hospital, and the US Army Medical Research and Materiel Command.

**Sampling.** Administrators from each practice provided a list of patients with outpatient paper charts, sorted by physician and appointment date, who were seen for a new visit or well visit between May 1 and November 1, 2007. In order to draw from patients throughout the list, every third chart

was reviewed, starting with the chart corresponding to a random digit provided by the Web site Random.org. At least 25 patient charts were reviewed for each provider. A chart was excluded if the patient was younger than 18 years or if the entire chart was not located onsite. Paper charts were abstracted by 2 genetic counselors between December 2008 and April 2010. Because 1 practice converted to an EMR system in 2010, an additional list of patients with outpatient EMR charts was provided; 2 study coordinators and the project director reviewed every other chart in this list in July and August of 2012. No identifying information was recorded, and standardized criteria for answering questions and interpreting family health histories were applied to reduce interobserver variability. Data quality was assessed by a genetics counselor who reviewed entries to correct errors. All data were analyzed in September 2012.

**Statistical analysis.** Abstracted data were entered into REDcap (Research Electronic Data Capture), a secure online survey and database storage tool. IBM SPSS Statistics software (version 19) was used for statistical analysis and reporting. Descriptive statistics were used to characterize the demographic characteristics of the patients and the health care providers and the characteristics of family health history collection.

To determine whether the family health history was informative enough to perform risk stratification and to alter a patient’s recommendations for prevention, this study used a set of quality indicators, which were subdivided by whether or not any of the patient’s family members were deceased. These quality indicators included: whether the family health history was updated during subsequent visits, whether a negative family health history was mentioned (eg, “no family health history of cancer”), whether the sex of affected relatives was noted, whether the age of the affected relative at disease onset was noted, and whether the affected relative’s lineage was noted (ie, whether the affected relative was on the maternal or paternal side of the family). Two additional quality indicators were used to assess family health history when at least 1 relative had died: the cause of death and the deceased relative’s age at death. Because each quality indicator is needed to perform an accurate risk assessment, family health histories were deemed highly informative (of high quality) only when the chart contained all 5 quality indicators (if the chart made no mention of any relative being deceased) or all 7 quality indicators (if any relative was deceased).

## Results

A total of 399 paper charts and 100 EMR charts were abstracted and entered into REDcap. Data from the paper charts were cleaned and corrected when necessary, resulting in the removal of 9 records.

### Patient and Physician Characteristics

Physician characteristics are presented in Table 1, and patient characteristics are presented in Table 2. The median

**TABLE 1.**  
**Characteristics of Providers Whose Patient Charts Were Reviewed (N = 16)**

| Characteristic           | Number of providers (%) |
|--------------------------|-------------------------|
| <b>Sex</b>               |                         |
| Male                     | 4 (25.0)                |
| Female                   | 12 (75.0)               |
| <b>Years in practice</b> |                         |
| ≤21 years                | 8 (50.0)                |
| >21 years                | 8 (50.0)                |
| <b>Medical specialty</b> |                         |
| Family medicine          | 7 (43.8)                |
| Internal medicine        | 9 (56.3)                |
| <b>Race</b>              |                         |
| White                    | 13 (81.3)               |
| Asian                    | 2 (12.5)                |
| Hispanic                 | 1 (6.3)                 |

patient age was 53 years. The most commonly noted diseases were cardiovascular disease (CVD) and cancer. In personal disease histories, hypercholesterolemia was the most frequently mentioned type of CVD; it was noted in the charts of 180 (76.3%) of the 236 patients with CVD. Nonmelanoma skin cancer was the most commonly noted form of cancer; it was reported in 27 (55.1%) of the 49 patients with cancer. The average number of years a patient had been seen in the practice was 9.76 ( $\pm 8.38$ ) years, and the median length of time a patient had been seen in the practice was 8 years.

#### Family Health History Within Paper Charts

**Location.** More than 99% of paper charts contained some family health history data, which was scattered over several areas of the chart. In 306 of the 390 charts (78.5%), family health history was located in the physician's notes. In 280 (71.8%) of the charts, the family health history was found on the patient's self-completed intake form; in 97 charts (24.9%), it was on the front summary page of the chart; in 63 charts (16.2%) it was found in the consult notes; in 16 charts (4.1%) it was found in a note from the patient; and in 1 (0.3%) of the charts, it was found in a nurse's note. Frequently, family health history was noted separately in 2 places. None of the examined charts contained a family health history in pedigree format.

**Relatives assessed.** When we looked at the health history of affected family members, we found that the health history of the patient's parents was documented in 339 (86.9%) of 390 charts, while only about half as many charts (168 [43.1%]) contained the health history of siblings. Only 131 charts (33.6%) contained the health history of grandparents; 67 charts (17.2%) contained the health history of aunts or uncles; and 32 charts (8.2%) contained the health history of children.

**Diseases collected.** A total of 390 charts were reviewed to assess whether the patient's family health history mentioned either the presence of a disease (positive history) or

the absence of a disease (negative history); we looked for diseases such as CVD, cancer, stroke, diabetes mellitus, arthritis, or depression. For example, more than three-quarters of charts had a positive family history of CVD, whereas only 23.3% of the charts mentioned the absence of CVD in the family health history. Table 3 shows the diseases and conditions for which data were collected. Table 4 shows the number and proportion of charts that recorded each of the quality indicators mentioned previously; these quality indicators are also discussed below.

**Quality indicators.** One quality indicator is whether the family health history has been updated. After an initial visit, the number of years before the first family health history was recorded in a patient's chart ranged from 0 to 32 years, with a median of 0.0 and an interquartile range of 1.00. On average, the most recent family health history had been collected or updated within the past 0.18 years (standard deviation = 1.06). Of the 390 charts reviewed, 287 charts (73.6%) had been updated; 180 (62.7%) of these charts had all updated changes, 53 (18.5%) of them had some updated changes, and 54 charts (18.8%) indicated that patients had been asked about updates but no changes had been made.

**TABLE 2.**  
**Characteristics of Patients Whose Charts Were Reviewed (N = 390)**

| Characteristic                | Number of patients (%) |
|-------------------------------|------------------------|
| <b>Sex</b>                    |                        |
| Female                        | 200 (51.3)             |
| Male                          | 187 (47.9)             |
| Missing data                  | 3 (0.8)                |
| <b>Race</b>                   |                        |
| White                         | 251 (64.3)             |
| African American              | 59 (15.1)              |
| Hispanic                      | 4 (1.0)                |
| Asian                         | 4 (1.0)                |
| Other                         | 4 (1.0)                |
| Missing data                  | 68 (17.6)              |
| <b>Type of insurance</b>      |                        |
| Commercial                    | 269 (68.9)             |
| Medicare                      | 54 (13.8)              |
| Medicaid                      | 4 (1.0)                |
| Self-pay                      | 1 (0.2)                |
| Unable to determine           | 55 (14.1)              |
| Missing data                  | 7 (1.7)                |
| <b>Medical conditions</b>     |                        |
| Cardiovascular diseases       | 236 (60.5)             |
| Hypercholesterolemia          | 180 (46.1)             |
| Hypertension                  | 157 (40.2)             |
| Other cardiovascular diseases | 28 (7.1)               |
| Cancer                        | 49 (12.5)              |
| Skin cancer                   | 27 (6.9)               |
| Breast cancer                 | 11 (2.8)               |
| Prostate cancer               | 6 (1.5)                |
| Other type of cancer          | 5 (1.2)                |

**TABLE 3.**  
Types of Family Health History Identified in Reviewed Charts<sup>a</sup> (N = 390)

| Medical conditions of relatives | Charts with positive FHH<br>Number of charts (%) | Charts with negative FHH<br>Number of charts (%) |
|---------------------------------|--------------------------------------------------|--------------------------------------------------|
| Cardiovascular diseases         | 338 (86.7)                                       | 91 (23.3)                                        |
| Hypertension                    | 242 (62.0)                                       | 22 (5.6)                                         |
| Heart attack                    | 150 (38.4)                                       | 10 (2.5)                                         |
| Hypercholesterolemia            | 69 (17.6)                                        | 4 (1.1)                                          |
| Cancer                          | 266 (68.2)                                       | 195 (50.0)                                       |
| Breast cancer                   | 83 (21.2)                                        | 34 (8.8)                                         |
| Colon cancer                    | 67 (17.2)                                        | 62 (15.9)                                        |
| Lung cancer                     | 64 (16.4)                                        | 0 (0)                                            |
| Stroke                          | 71 (18.2)                                        | 4 (1.1)                                          |
| Other conditions                |                                                  |                                                  |
| Diabetes                        | 200 (51.2)                                       | 48 (12.4)                                        |
| Arthritis                       | 71 (18.2)                                        | 14 (3.6)                                         |
| Depression                      | 39 (10.0)                                        | 12 (3.1)                                         |

Note. FHH, family health history.

<sup>a</sup>Totals do not sum to the sample size because of missing data.

Of the histories that were being taken for the first time, 37 (66.1%) were for patients who were new to the practice.

Another quality indicator is whether a negative family health history is reported. Almost half (173 [44.4%]) of the charts explicitly recorded a generalized negative statement regarding family health history for a specific disease or disease group (eg, "family history negative for cancer").

A third quality indicator is whether the sex of the affected relative is reported in the family health history. In 366 (93.8%) of the charts, a positive family history of a specific disease or disease group was noted. In these charts, the sex of the affected relative was the most frequently collected quality indicator, having been specified in 356 (91.2%) of the charts reviewed. In some cases, the sex of the affected relative was known because of the words used to describe the relative (ie, aunt, uncle, mother, father, sister, brother). The sex of the affected relative was noted in 92% of the instances in which the relative was a parent, aunt, uncle, sibling, or grandparent. The sex of the affected relative was noted in only 3 (17.6%) of the instances in which the relative was a cousin and in only 6 (24.0%) of the instances in which the individual was described as a "relative."

Age at disease onset for an affected relative was the least frequently collected quality indicator, having been collected in only 71 (18.2%) of the 366 family health histories that recorded a positive family history. Specifically, age of disease onset was documented in family health histories for 11 (6.6%) of the siblings mentioned, 14 (10.8%) of the grandparents mentioned, 4 (6.3%) of the aunts or uncles mentioned, and 50 (14.8%) of the parents mentioned.

The fifth quality indicator for a family health history is whether the lineage of the affected relative is reported. Of the 366 family health histories that recorded a positive family history, 255 (69.7%) did not include information about

the lineage (ie, maternal or paternal side) of affected family members (Table 4). More than half of the 366 charts (233 [63.7%]) did not mention an affected second-degree relative, and 44 charts did not mention an affected first-degree relative.

If the family health history includes mention of deceased relatives, then 2 additional quality indicators should be evaluated: age at death and cause of death. A deceased relative was documented in 227 (62.0%) of the 366 records with a positive family health history. In 172 (75.8%) of those 227 records, the affected relative's age at death was recorded, either for all deceased relatives (94/227 [41.4%]) or for some of them (78/227 [34.4%]). Of the 227 charts that noted a deceased relative, 213 (93.8%) listed the cause of death, either for all deceased relatives (165/227 [72.7%]) or for some of them (48/227 [21.1%]).

**Quality of family health history.** Less than 4% of patients had family health histories that could be used to perform a risk assessment. The group of 227 family health histories that mentioned a deceased relative included more "moderately informative" histories and fewer "less informative" histories than did the group of family histories that did not include mention of any deceased relatives (Table 4). The number of quality indicators present in each group is shown in Table 4, and Table 5 shows the number of charts in which each of the first 5 quality indicators was reported. Among the charts that did not mention any deceased relative, 61 charts included 4 of the 5 quality indicators; the indicators that were most frequently absent were the age of the affected relative (missing in 28 [45.9%] of the charts), negative family health history information (missing in 16 [26.2%] of the charts), and the lineage of the affected relative (missing in 15 [24.6%] of the charts). Similar results were observed for the charts that mentioned one or more deceased relatives.

**TABLE 4.**  
**Characteristics of Reviewed Charts in Family Health History Project**

| Characteristic                                  | All charts (N = 390)<br>Number of charts (%) | Charts with FHH<br>that do not mention<br>deceased relatives<br>(n = 163)<br>Number of charts (%) | Charts with FHH<br>that do mention<br>deceased relatives<br>(n = 277)<br>Number of charts (%) |
|-------------------------------------------------|----------------------------------------------|---------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------|
| <b>FHH status</b>                               |                                              |                                                                                                   |                                                                                               |
| Updated                                         | 287 (73.5)                                   | 92 (56.4)                                                                                         | 186 (81.9)                                                                                    |
| Not updated                                     | 91 (23.3)                                    | 49 (30.0)                                                                                         | 38 (16.7)                                                                                     |
| Missing data                                    | 12 (3.2)                                     | 22 (13.6)                                                                                         | 3 (1.4)                                                                                       |
| <b>Negative FHH</b>                             |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | 173 (44.3)                                   | 70 (42.9)                                                                                         | 92 (40.5)                                                                                     |
| Not recorded                                    | 213 (54.6)                                   | 71 (43.5)                                                                                         | 135 (59.5)                                                                                    |
| Missing data                                    | 4 (1.1)                                      | 22 (13.6)                                                                                         | 0 (0)                                                                                         |
| <b>Affected relative<sup>a</sup></b>            |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | 366 (93.8)                                   | 135 (82.8)                                                                                        | 226 (99.6)                                                                                    |
| Not recorded                                    | 24 (6.2)                                     | 6 (3.6)                                                                                           | 1 (0.4)                                                                                       |
| Missing data                                    | 0 (0)                                        | 22 (13.6)                                                                                         | 0 (0)                                                                                         |
| <b>Sex of affected relative</b>                 |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | 356 (91.2)                                   | 129 (79.1)                                                                                        | 222 (97.8)                                                                                    |
| Not recorded                                    | 10 (2.5)                                     | 12 (7.3)                                                                                          | 5 (2.2)                                                                                       |
| Missing data                                    | 24 (6.3)                                     | 22 (13.6)                                                                                         | 0 (0)                                                                                         |
| <b>Affected relative's age at disease onset</b> |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | 71 (18.2)                                    | 27 (16.5)                                                                                         | 44 (19.3)                                                                                     |
| Not recorded                                    | 256 (65.6)                                   | 108 (66.2)                                                                                        | 182 (80.1)                                                                                    |
| Missing data                                    | 63 (16.2)                                    | 28 (17.3)                                                                                         | 1 (0.6)                                                                                       |
| <b>Lineage of affected relative</b>             |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | 111 (28.4)                                   | 50 (30.6)                                                                                         | 58 (25.6)                                                                                     |
| Not recorded                                    | 255 (65.3)                                   | 91 (55.2)                                                                                         | 169 (74.4)                                                                                    |
| Missing data                                    | 24 (6.3)                                     | 22 (14.2)                                                                                         | 0 (0)                                                                                         |
| <b>Cause of death of affected relative</b>      |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | NA                                           | NA                                                                                                | 213 (93.8)                                                                                    |
| Not recorded                                    | NA                                           | NA                                                                                                | 14 (6.2)                                                                                      |
| <b>Age of affected relative at death</b>        |                                              |                                                                                                   |                                                                                               |
| Recorded                                        | NA                                           | NA                                                                                                | 172 (75.8)                                                                                    |
| Not recorded                                    | NA                                           | NA                                                                                                | 55 (24.2)                                                                                     |
| <b>Number of quality indicators present</b>     |                                              |                                                                                                   |                                                                                               |
| 0                                               | 2 (0.6)                                      | 2 (1.5)                                                                                           | 0 (0)                                                                                         |
| 1                                               | 37 (10.2)                                    | 14 (10.4)                                                                                         | 2 (0.9)                                                                                       |
| 2                                               | 119 (32.9)                                   | 41 (30.4)                                                                                         | 6 (2.7)                                                                                       |
| 3                                               | 131 (36.2)                                   | 49 (36.3)                                                                                         | 32 (14.3)                                                                                     |
| 4                                               | 61 (16.9)                                    | 26 (19.3)                                                                                         | 86 (38.6)                                                                                     |
| 5                                               | 12 (3.3)                                     | 3 (2.2)                                                                                           | 65 (29.1)                                                                                     |
| 6                                               | NA                                           | NA                                                                                                | 24 (10.8)                                                                                     |
| 7                                               | NA                                           | NA                                                                                                | 8 (3.6)                                                                                       |
| <b>Quality of FHH</b>                           |                                              |                                                                                                   |                                                                                               |
| Not informative <sup>b</sup>                    | 2 (0.6)                                      | 2 (1.5)                                                                                           | 0 (0)                                                                                         |
| Less informative <sup>c</sup>                   | 156 (43.0)                                   | 55 (40.7)                                                                                         | 40 (18.0)                                                                                     |
| Moderately informative <sup>d</sup>             | 192 (53.1)                                   | 75 (55.6)                                                                                         | 175 (78.4)                                                                                    |
| Highly informative <sup>e</sup>                 | 12 (3.3)                                     | 3 (2.2)                                                                                           | 8 (3.6)                                                                                       |

Note. FHH, family health history; NA, not applicable.

<sup>a</sup>"Not recorded" means a characteristic was not asked about or was not checked in the chart; "missing data" means data is completely missing from the dataset.

<sup>b</sup>Not included as an indicator of quality.

<sup>c</sup>Pedigrees were labeled "not informative" if they contained none of the quality indicators.

<sup>d</sup>Pedigrees were labeled "less informative" if they did not mention any deceased relatives and contained 1-2 quality indicators, or if they did mention a deceased relative and contained 1-3 quality indicators.

<sup>e</sup>Pedigrees were labeled "moderately informative" if they did not mention any deceased relatives and contained 3-4 quality indicators, or if they did mention a deceased relative and contained 4-6 quality indicators.

<sup>f</sup>Pedigrees were labeled "highly informative" if they did not mention any deceased relatives and contained 5 quality indicators, or if they did mention a deceased relative and contained 7 quality indicators.

**TABLE 5.**  
**Number of Charts Containing Each Type of Quality Indicator, Cross-Tabulated with the Number of Quality Indicators Present in the Chart (Number of Charts = 390)**

| Quality indicator                                 | Number of indicators present |     |     |    |    |
|---------------------------------------------------|------------------------------|-----|-----|----|----|
|                                                   | 1                            | 2   | 3   | 4  | 5  |
| FHH status updated                                | 3                            | 91  | 112 | 59 | 12 |
| Negative family history recorded                  | 2                            | 20  | 82  | 45 | 12 |
| Sex of affected relative recorded                 | 32                           | 116 | 131 | 61 | 12 |
| Affected relative's age at disease onset recorded | 0                            | 3   | 23  | 33 | 12 |
| Lineage of affected relative recorded             | 0                            | 8   | 45  | 46 | 12 |

Note. FHH, family health history.  
 Totals do not sum to the sample size because of missing data.

### Family Health History Within EMRs

Out of 100 EMR charts, 97 (97%) documented some amount of family health history. No patient had a structured 3-generation pedigree. Interestingly, the EMR's family health history collection tool was not utilized for any of the charts we reviewed. In all cases, family health history was recorded in the free text section of the clinic note or on the patient intake form, and family health history was included for only a select few relatives.

### Discussion

The inability to use family health histories in primary care poses a barrier to the practice of genomic medicine and limits physicians' ability to achieve benchmarks set by programs such as Healthy People 2020 [37]. Several problems were encountered with the charts analyzed in this study, including a lack of uniformity in the collection methods, variations in the location within the chart where family health history information was reported, and missing information about essential elements of the family health history (as presented in Tables 4 and 5). Without all of these elements, an adequate risk assessment cannot be performed.

Family health histories that included information about deceased relatives were more likely to be "moderately informative" than were those without any mention of deceased relatives. The algorithm used to assess the quality of the family health histories for the deceased-relative group included 2 additional indicators (age at death and cause of death of the affected relative). More than two-thirds of the family health histories in the deceased-relative group recorded these 2 indicators, thereby raising the mean quality of the family health histories and the quality of the information. It may be that when the death of an affected relative is recorded in a family health history, health care providers are more likely to seek details on the age at death and cause of death.

Although more than 97% of family health histories were updated, almost 23% contained incomplete information. Missing information included information that was previ-

ously collected but was not brought forward during subsequent visits and was thus lost. Some new EMR programs have addressed this problem.

In 80%-90% of the charts we reviewed, the age at diagnosis of an affected relative was never mentioned. Diagnosis of, or death from, a disease at an earlier age than expected can indicate a hereditary form of disease and is included in clinical algorithms to evaluate a patient's disease risk [13, 14, 19, 38]. Thus, there is a need for greater awareness about the importance of age at diagnosis for risk assessment. Age at diagnosis is particularly important when using family health history to identify those most at risk for heart disease and cancer, the 2 leading causes of death in the United States [39]. Interestingly, using an EMR did not increase the amount or quality of information collected by the practitioner. Because EMRs presently do not prompt the physician to collect all of the elements necessary for an assessment of disease risk based on family health history, it remains uncertain whether family health histories will be fully utilized for these widespread diseases.

The family health histories of cancer documented in these charts resemble those found in other studies [5, 31]. The vast majority of patient charts had a documented positive family health history; in more than 45% of cases, however, diseases that were not found in the family (ie, negative family history) were not explicitly mentioned. If a negative family history was mentioned by a consulting physician, it typically was not updated in the primary care provider's chart and was not considered when assessing the patient's risk for disease. Documentation of a negative family health history is almost as important as documentation of a positive family health history, and primary care providers may benefit from greater awareness of its value for interpreting family health history data [34].

Several studies have estimated the chance of having an increased risk for disease on the basis of the family health history taken by the patient's primary care provider [7, 8]. Based on the estimated frequency of at-risk patients seen in previous studies, we would have expected approximately 180 individuals in this study to have a family health history that established a strong or moderate risk for breast and ovarian cancer or colon cancer, which would suggest the need for referral to a genetic specialist. However, no such referrals were made. Although patients were sometimes referred to a specialist, such as an oncologist or a gastroenterologist, charts rarely mentioned that a patient was at high risk for disease based on family health history; this is probably because such an evaluation would be time-consuming and difficult for the primary care provider, even if the family health history were accurate and complete. Most charts had a patient intake form that allowed patients to self-report family health history. Several studies have found a bias for patients to overreport or underreport diseases, specifically cancer [40-42]. This may be another challenge to the effective utilization of family health history information.

**Overcoming barriers to collection and use of family health histories in primary care.** To remove several of the deficiencies we observed in the collection and interpretation of family health histories, it would be helpful if clinicians had a tool that creates a structure containing all of the key elements of family health histories (eg, a pedigree), provides decision support for providers [5, 32, 43], and is compatible with EMRs. A family health history decision support tool would need to be able to do the following things: allow patients to fill out a form about their family health history using a secure online system prior to their appointment; update family health histories without dropping previously collected information about family members; provide a single location for family health history within a chart (especially if the tool is compatible with the EMR); easily identify the number of affected and deceased family members; allow providers to quickly see whether a family has a negative disease history; and use clinically established information to provide an assessment of disease risk and recommendations regarding medical management.

## Limitations

Limitations of this study that may reduce the generalizability of its findings to other populations include the small number of charts reviewed per primary care provider, the fact that the sample of primary care providers were all from a single geographic location, the failure to differentiate between patient-generated and physician-collected family health histories, and the inability to assess other quality indicators, such as consanguinity. The latter limitation results from confusion about the information included in charts. For example, a chart might be unclear as to whether the number of family members in the family matches the number of relatives mentioned in the family health history (eg, a patient might have 3 siblings, only 1 of whom is mentioned in the family health history), or there might be uncertainty about some pieces of information (eg, the family health history might mention a brother with heart disease and later refer to a brother with diabetes, without explaining whether these are 2 references to the same person or references to 2 different brothers). Another problem is that the chart review checklist does not collect certain additional types of information (eg, consanguinity). Lastly, there is evidence in the literature that patient-provided family health histories can differ from physician-collected histories. Most charts contained a patient intake form that had been used to collect family health history data, and this form was considered part of the family health history in the patient record.

Despite these limitations, our findings are consistent with those of previous studies, suggesting common factors in the use of family health histories across multiple settings. This study also did not allow for assessment of conversations between patients and providers; therefore, any information about family health history that was discussed in these conversations but was not mentioned in the clinic

notes would not have been collected. The impact of verbally communicated but undocumented family health history information is unknown but is important to consider, especially in primary care settings where relationships are often long-lasting and the time available for documentation is often limited.

## Conclusion

Despite the value of family health histories for disease risk assessment, the histories collected in primary care practices usually lack some of the crucial information needed to perform a risk assessment for hereditary cancer syndromes and other chronic disorders [26]. If the use of family health histories for disease risk assessment is to succeed in primary care, collection must be easier and more complete. This study indicates a need for the adoption of family health history collection tools that can analyze the collected information and provide risk-stratified decision support recommendations. **NCMJ**

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