Multi-source development of an integrated model for family health history

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ABSTRACT

Objective To integrate data elements from multiple sources for informing comprehensive and standardized collection of family health history (FHH).

Materials and methods Three types of sources were analyzed to identify data elements associated with the collection of FHH. First, clinical *notes* from multiple resources were annotated for FHH information. Second, questions and responses for family members in patient-facing FHH *tools* were examined. Lastly, elements defined in FHH-related *specifications* were extracted for several standards development and related organizations. Data elements identified from the notes, tools, and specifications were subsequently combined and compared.

Results In total, 891 notes from three resources, eight tools, and seven specifications associated with four organizations were analyzed. The resulting Integrated FHH Model consisted of 44 data elements for describing source of information, family members, observations, and general statements about family history. Of these elements, 16 were common to all three source types, 17 were common to two, and 11 were unique. Intra-source comparisons also revealed common and unique elements across the different notes, tools, and specifications.

Discussion Through examination of multiple sources, a representative and complementary set of FHH data elements was identified. Further work is needed to create formal representations of the Integrated FHH Model, standardize values associated with each element, and inform context-specific implementations.

Conclusions There has been increased emphasis on the importance of FHH for supporting personalized medicine, biomedical research, and population health. Multi-source development of an integrated model could contribute to improving the standardized collection and use of FHH information in disparate systems.

Key words: family health, medical history taking, electronic health records, genomics, narration, models, theoretical

RESEARCH AND APPLICATIONS

BACKGROUND AND SIGNIFICANCE

Since the advent of the genomic era,[1] there has been renewed interest and emphasis on the importance of family health history (FHH) for individualized disease prevention, diagnosis, and treatment.[2–13] FHH has been described as a simple yet invaluable tool for risk assessment and is incorporated in a number of recommendations by the U.S. Preventive Services Task Force (eg, screening for BRCA-related cancer risk in women,[14] lipid disorders in adults,[15,16] and osteoporosis[17]).[5,18,19] While the role of FHH is clearly recognized for personalized medicine and population health, numerous barriers to its optimal collection and use have been described including: limited time and resources; insufficient knowledge for interpretation by providers; uncertainty of family composition and health history by patients; and lack of standards for data elements, terminology, structure, interoperability, presentation, and clinical decision support rules.[18,20–24] In response to these challenges, multiple initiatives have emerged that emphasize the importance of FHH and the need for more effective use (eg, U.S. Surgeon General's Family History Initiative,[25] Centers for Disease Control and Prevention Office of Public Health Genomics (CDC/OPHG) Family History Public Health Initiative,[26] National Institutes of Health State-of-the-Science Conference on Family History and Improving Health,[27] and Talk Health History Campaign supported by The American Society of Human Genetics and Genetic Alliance[28]).

Increasing efforts over the last decade have focused on the development and use of computer-based tools for facilitating the collection, maintenance, and analysis of detailed FHH.[23,29–36] The electronic health record (EHR) provides an important mechanism for documentation by providers where related efforts include a core Stage 2 Meaningful Use measure for structured data entry of FHH[37] and several natural language processing (NLP) tools for extracting FHH information from clinical notes.[38–42] Consumer-oriented resources include personal health record systems and patient-facing FHH tools where

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these tools range from those that have been developed and evaluated as part of federal initiatives (eg, U.S. Surgeon General's My Family Health Portrait[43–45] and CDC's Family Healthware[46,47]) to those from university-affiliated research efforts (eg, MeTree,[48–52] Health Heritage,[53] and OurFamilyHealth[54]). The development and use of standards to support interoperability across systems continues to be emphasized[55] and FHH-related specifications include the HL7 Clinical Genomics FHH (Pedigree) Model[56–58] and a minimum core data set defined by the American Health Information Community (AHIC; now known as the National eHealth Collaborative (NeHC)) FHH Multi-Stakeholder Workgroup.[59]

In our previous work, the adequacy of existing standards (HL7 Clinical Genomics FHH Model and HL7 Clinical Statement Model) was evaluated for representing FHH information in clinical notes from University of Minnesota-affiliated Fairview Health Services.[60] These notes were analyzed to identify different types of FHH statements and elements of information within these statements such as disease, family member, living status, negation, and uncertainty. Data elements identified in the notes were combined with those in the two HL7 models to create a 'Merged Family History Model' (henceforth referred to as the 'Integrated FHH Model'). A subsequent study involving analysis of free-text comments within the primarily structured family history module of the EHR system at Fletcher Allen Health Care (FAHC), the academic health center affiliated with the University of Vermont, served to validate the Integrated FHH Model.[61] Findings from these early studies highlighted the need for further refinements to accommodate the full breadth of FHH information documented in the EHR and other key sources.

OBJECTIVE

Building upon the aforementioned efforts, the objective of this study was to enhance the Integrated FHH Model for informing the standardized collection and use of FHH in disparate systems. Multiple sources were explored to identify a comprehensive set of data elements and characterize the complementary nature of these sources as well as potential gaps in individual resources.

MATERIALS AND METHODS

The general approach involved analysis of three types of sources to identify data elements associated with the collection of FHH: (1) clinical *notes*, (2) patient-facing FHH *tools*, and (3) FHH-related *specifications*. Data elements identified from these notes, tools, and specifications were subsequently combined and compared.

Analysis of clinical notes

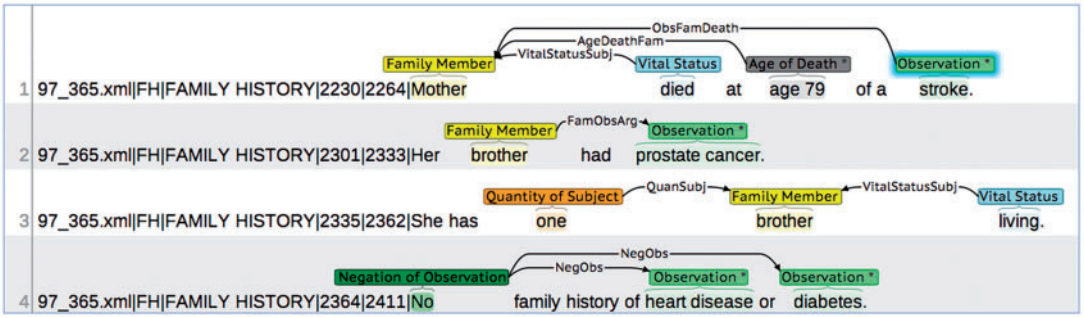
Clinical notes from three resources were collected and analyzed for FHH information: (1) MTSamples.com, (2) the Open Clinical Report Repository, and (3) FAHC. MTSamples.com (MTS) is a public web repository of almost 5000 sample transcription reports, including 491 notes categorized as 'Consult—History and Physical' (as of October 2012) that were used in this study.[62] The Open Clinical Report Repository was developed as a community resource to support NLP research and development.[63] This repository contains nine types of reports from the University of Pittsburgh Medical Center (UPMC), including history and physical notes from which a sample of 200 notes was obtained. Finally, 200 transcribed notes categorized as consult or evaluation notes were obtained from the legacy clinical information system at FAHC.[64] Collectively, these three sets included note types that typically include family history sections and covered both the inpatient and outpatient settings as well as a range of specialties (eg, cardiology, general medicine, oncology, and pediatrics).

Two open-source tools were used for manual annotation of FHH sections, sentences, and statements in each set of notes where a statement is defined as individual discrete items of information within a sentence. For example, the sentence 'mother and sister had breast cancer' includes two statements: (1) 'mother had breast cancer' and (2) 'sister had breast cancer.' Annotation of sections and sentences involved use of the General Architecture for Text Engineering (GATE).[65] A GATE annotation schema and guidelines were developed for defining two types of annotations: (1) *FHH sections*—section headers associated with FHH sentences (eg, 'Family History,' 'FHX,' or 'History of Present Illness') and (2) *FHH sentences*—consecutive sentences including FHH information.

For each annotated sentence (extracted from the GATE Extensible Markup Language (XML) output), the brat rapid annotation tool (BRAT) was then used to annotate FHH statements based on an annotation schema defining the set of entities, entity attributes, and relationships between entities.[66,67] The initial version of this schema was based on the first version of the Integrated FHH Model described earlier. For example, the entity *Observation* can be used to annotate a word or phrase describing a particular clinical observation (eg, 'diabetes,' 'breast cancer,' or 'CABG'), has an attribute for specifying the *Observation Type* (ie, 'Disease,' 'Procedure,' 'Medication,' 'Lab Test,' or 'Other'), and can be linked to other entities such as *Family Member* (eg, 'mother'⇒'diabetes') or *Onset Date* (eg, '1980'⇒'breast cancer'). Annotation guidelines were also developed that included descriptions and examples for each entity, attribute, and relationship (see online supplementary appendix A). Figure 1 depicts the annotation of FHH information in a set of sentences using the defined annotation schema and guidelines.

An iterative process was used for annotating each set of notes using GATE and BRAT. A subset of 100 MTS notes was initially annotated and annotations were revised based on multiple review sessions to achieve consensus between four annotators before separately proceeding with the remaining 391 notes. Throughout this process, the BRAT annotation schema and guidelines underwent several revisions to accommodate for additional entities and relationships. For example, the entity *Quantity of Observation* and relationship to *Observation* were added after encountering a statement referring to 'multiple strokes.' For the UPMC and FAHC sets, a subset of 10 (5%) notes was first annotated and reviewed by two annotators to achieve consensus before proceeding with the remaining notes.

Figure 1: Annotation of family health history (FHH) statements in clinical notes. Each line represents a sentence where the first five fields (separated by '|') represent information extracted from the GATE XML output (filename, annotation type, section header, and start and end positions). FHH information is annotated in each sentence ('*' indicates an attribute value and shadowing indicates a free-text note). Arrows represent relationships between entities where directionality matters (eg, 'ObsFamDeath' indicates that the Observation is the cause of death for the Family Member).



Analysis of patient-facing FHH tools

A series of literature and web searches was performed to identify patient-facing FHH tools. Preliminary searches focused on publications including lists of tools[12,21,32] and web resources such as the American Medical Association (AMA) Family Medical History,[68] CDC/OPHG FHH,[69] and Talk Health History Campaign.[28] A list of 24 tools was generated from these searches, including those focused on pediatric patients as well as specific diseases such as diabetes and heart disease. The following criteria were then used to select an initial subset for further analysis: general audience or adult-oriented, general health or cancer-specific, and available as an interactive web-based tool or in a web-accessible paper-based format (eg, PDF form). The resulting subset of eight tools included: (1) Adult Family History Form (AMA),[68] (2) Cancer Family Tree (University of Nebraska Medical Center),[70] (3) Colon Cancer Risk Assessment (Cleveland Clinic),[71] (4) Does It Run in the Family? Toolkit (Genetic Alliance),[72] (5) FHH Toolkit (Utah Department of Health),[73] (6) Family Health*Link* (The Ohio State University Wexner Medical Center),[74] (7) Family Healthware (CDC (version obtained under a Material Transfer Agreement for this study; not the publicly-accessible web tool)),[46,75] and (8) My Family Health Portrait (U.S. Surgeon General).[43]

For each tool, two reviewers analyzed questions and responses related to family structure or individual family members; questions related to the patient were excluded (eg, patient demographics and health history). For example, the question 'Is family member still living?' from the Cancer Family Tree tool and 'Living?' from My Family Health Portrait both correspond to living status. Depending on the response, additional questions are asked where responses are either selected from a pre-defined list of values or provided as free text (figure 2).

Analysis of FHH-related specifications

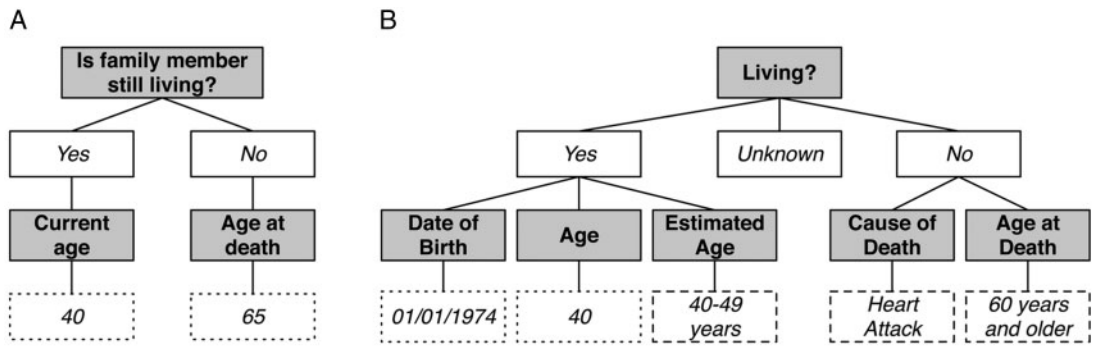
In addition to revisiting HL7 International, three additional standards development and related organizations representing both national and international efforts were explored for FHH-related specifications (subsequently analyzed by two reviewers): Health Information Technology Standards Panel (HITSP), AHIC, and openEHR. For HL7, the latest HL7 V.3 Implementation Guide: Family History/Pedigree Interoperability, Release 1 was reviewed for elements defined in its Family History Model and related vocabulary (eg, HL7 V.3 Vocabulary for RoleCode that includes a list of family members).[58,76] These elements were supplemented with those from the HL7 Implementation Guide for CDA Release 2: IHE Health Story Consolidation, Release 1.1, specifically for Family History Section, Family History Organizer, Family History Observation, Family History Death Observation, and Age Observation.[77]

Within the HITSP/C154 Data Dictionary Component, V.1.0.1 that defines the library of data elements for standards-based exchange, only those listed in the Family History Module were included.[78] From the FHH Multi-Stakeholder Workgroup Dataset Requirements Summary presented to the Personalized Health Care Workgroup of AHIC, data items associated with each core dataset requirement were considered elements and included with the exception of those associated with basic desired functionality (eg, 'Free text shall be minimized for data entry of family history' or 'Capture data that allows for generation of a pedigree').[79] Finally, the openEHR Clinical Knowledge Manager (CKM) was used to search for any archetype related to family history where three relevant archetypes were identified: Exclusion of Family History, Family History, and Risk of condition based on family history.[80] Each of these archetypes was analyzed to create a combined list of elements for openEHR.

Integration and comparison of FHH data elements

The analysis of three sets of notes, eight tools, and specifications associated with four organizations resulted in 15 separate lists of FHH-related information that were used to define a set of data elements for the second version of the Integrated FHH

Figure 2: Questions and responses for living status from patient-facing family health history (FHH) tools. From the University of Nebraska Medical Center's Cancer Family Tree (A) and the U.S. Surgeon General's My Family Health Portrait (B). Gray shaded boxes indicate questions and white boxes indicate responses (solid border indicates all possible responses; dashed line indicates example response selected from a list of available values; dotted line indicates example free-text response).



Model. A consensus-based process was used to standardize each 'source list,' which involved creating a 'master list' of elements with preferred names and mapping information in each of the source lists to the corresponding preferred element names. For example, 'Vital Status' in the BRAT annotation schema for clinical notes (figure 1), the questions 'Is family member still living?' and 'Living?' from two patient-facing FHH tools (figure 2), 'deceasedInd' from the HL7 models, and 'Deceased?' from one of the openEHR archetypes were mapped to the preferred element name *Living Status*. Once standardized, inter- and intra-source comparisons were conducted to characterize the contributions of each source type (notes, tools, and specifications) as well as sources within each type.

RESULTS

In total, 891 clinical notes (1071 sentences and 1658 statements) from three resources, eight patient-facing FHH tools, and seven FHH-related specifications associated with four organizations were examined. Table 1A presents the number of FHH sentences and statements annotated in each set of notes, table 1B provides brief descriptions and the estimated number of questions (general and specific to individual family members) for each tool, and table 1C includes the estimated number of elements or requirements defined in each specification. See online supplementary appendices A, B, and C for the full mappings of elements for notes, tools, and specifications, respectively.

Integration and inter-source comparison of data elements

The resulting Integrated FHH Model consisted of 44 data elements organized into four sections: (1) *Source*—one element for source of information; (2) *General*—two elements for representing general statements about FHH; (3) *Family Member*—29 elements for describing family members such as relation type, demographics, and living status; and (4) *Observation*—12 elements for describing specific family member observations such as diseases, procedures, genetic tests, social and behavioral factors, or general health status.

Table 2 depicts the distribution of elements across the three types of sources and also highlights the addition of 28 elements in the second version of the model (V2) compared with the first version (V1). Figure 3 further highlights the contribution of each source type in terms of common and unique elements. Of the 44 elements, 16 (36%) were common to all three source types (eg, Current Age, Age at Death, and Age at Onset), 17 (39%) were common to two source types (eg, Multiple Birth Status, Ancestry, and Date of Onset), and 11 (25%) were unique to one source type (eg, Quality of Relationship, Multiple Birth Order, and Strength of Observation).

Intra-source comparison of data elements: notes

The clinical notes from MTS, UPMC, and FAHC contributed 25, 17, and 26 elements to the Integrated FHH Model, respectively (table 3). The elements Half Relationship, Step Relationship, and Degree of Relationship were included due to values annotated for Family Member such as 'half sister,' 'stepdaughter,' and 'first-degree relative' (denoted with '*' in table 3). Similarly, based on analysis of the General Statement annotations, initial categories for General Statement Type were identified such as: Nonsignificant/Noncontributory (eg, 'family history noncontributory' or 'there is no significant family history'), Unchanged (eg, 'reviewed and unchanged'), Unknown/Unavailable (eg, 'not available at this current time' or 'unobtainable'), Negative (eg, 'negative history' or 'none'), and Positive (eg, 'family history is positive').

The most frequent elements across the three sets were Observation, Observation Type, Family Member, and Living Status. Other more frequent elements included Cause of Death and Negation of Observation (eg, 'no family history of heart disease' or 'negative for heart disease') for MTS; Certainty of

Table 1: Summary of sources

(A) Notes

Source	Note type(s)	No. of notes	No. of notes with FHH	No. of FHH sentences	No. of FHH statements
MTSamples.com (MTS)	Consult–H&P	491	270 (55%)	541	850
UPMC	H&P	200	136 (68%)	198	273
FAHC	Consult; evaluation	200	130 (65%)	332	535
Total		891	536 (60%)	1071	1658

(B) Tools

Source	Affiliation	Target conditions	Target audience	Format	Availability	No. of questions*
Adult Family History Form	American Medical Association	General	Adult	PDF form	Public	6–8
Cancer Family Tree	University of Nebraska Medical Center	Cancer	General	Web tool	Public	8 (2)
Colon Cancer Risk Assessment	Cleveland Clinic	Colon Cancer	General	Web tool	Public	4–8
Does It Run in the Family? Toolkit†	Genetic Alliance	General	General	PDF guide/form	Public	8 (1) 12 (1) 7 (2)
FHH Toolkit‡	Utah Department of Health	General	General	PDF guide/form	Public	10 12
Family Health*Link*	The Ohio State University Wexner Medical Center	Cancer and heart disease	General	Web tool	Public	7–8 (3)
Family Healthware	Centers for Disease Control and Prevention	Six including cancer	General	Web tool	MTA§	6 (12)
My Family Health Portrait	U.S. Surgeon General	General	General	Web tool	Public	15 (12)

(C) Specifications

Source	No. of elements/ requirements¶
HL7 V.3 Implementation Guide: Family History/Pedigree Interoperability, Release 1—US Realm (April 2013)	25
HL7 Implementation Guide for CDA Release 2: IHE Health Story Consolidation, DSTU Release 1.1 (US Realm) Draft Standard for Trial Use (July 2012)	11
HITSP/C154 HITSP Data Dictionary Component V.1.0.1 (January 31, 2010)	26
AHIC PHC Workgroup FHH Multi-Stakeholder Workgroup Dataset Requirements Summary (March 2008)	44
openEHR Archetype 'Exclusion of Family History' (March 2, 2010)	3
openEHR Archetype 'Family History' (December 15, 2010)	16
openEHR Archetype 'Risk of condition based on family history' (March 2, 2010)	18

*Number or range of questions/instructions specific to individual family members (number of general questions/instructions related to defining family structure (eg, quantity and relationships)).

†Three components included: (1) A Guide to FHH, (2) FHH Questionnaire, and (3) Healthcare Provider Card.

‡Two components included: (1) 10 Questions to Ask Your Family and (2) Health Family Tree Tool.

§Accessed through a Material Transfer Agreement (MTA) for the purposes of this study.

¶Estimated number of elements/requirements based on family history-related components of each specification.

AHIC, American Health Information Community; EHR, electronic health record; FAHC, Fletcher Allen Health Care; FHH, family health history; H&P, history and physical; HITSP, Health Information Technology Standards Panel; PHC, personalized health care; UPMC, University of Pittsburgh Medical Center.

Table 2: Data elements in the Integrated FHH Model

Element	Model–V1 (16)	Notes (27)	Tools (29)	Specifications (37)	Model–V2 (44)
Source					
Source of information		•		•	•
General					
General statement		•			•
General statement type		•			•
Family member					
Side of family	•	•	•	•	•
Family member	•	•	•	•	•
Genetic relationship			•	•	•
Half relationship		•	•	•	•
Step relationship		•	•	•	•
Degree of relationship		•		•	•
Quality of relationship				•	•
Adoptive status			•	•	•
Foster status			•	•	•
Multiple birth status	•		•	•	•
Multiple birth order				•	•
Consanguinity				•	•
Name			•	•	•
Sex			•	•	•
Gender	•		•	•	•
Place of birth			•	•	•
Date of birth	•		•	•	•
Current age	•	•	•	•	•
Race	•		•	•	•
Ethnicity	•		•	•	•
Ancestry		•	•		•
Partner status				•	•
Living status	•	•	•	•	•
Date of death	•	•	•	•	•
Age at death	•	•	•	•	•
Cause of death		•	•	•	•
Certainty of family member		•		•	•
Negation of family member		•			•
Quantity of family member		•	•		•

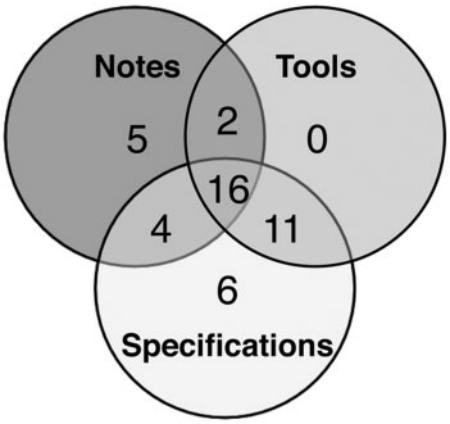
(continued)

Table 2: Continued

Element	Model–V1 (16)	Notes (27)	Tools (29)	Specifications (37)	Model–V2 (44)
Observation					
Observation	•	•	•	•	•
Observation type		•	•	•	•
Date of onset	•	•		•	•
Age at onset	•	•	•	•	•
Certainty of observation	•	•	•	•	•
Negation of observation	•	•	•	•	•
Quantity of observation		•	•	•	•
Relevance of observation		•			•
Severity of observation				•	•
Strength of observation		•			•
Status of observation				•	•
Temporality of observation		•	•	•	•

(n) indicates total number of elements.
FHH, family health history.

Figure 3: Common and unique family health history (FHH) elements across source types.



Observation (eg, 'probably healthy' or 'questionable history of coronary artery disease') and General Statement for UPMC; and Current Age and Quantity of Family Member (eg, 'multiple brothers and sisters' or 'three half-sisters') for FAHC. Ancestry (eg, 'mother is of English descent') and Date of Onset were unique to FAHC where the former was primarily found in genetic consultation notes.

Intra-source comparison of data elements: tools

Collectively, the eight patient-facing FHH tools provided a total of 29 data elements where the range was a minimum of nine elements and a maximum of 22 elements (table 4). Similar to the notes, several elements such as Side of Family and Observation Type were included due to values for Family Member (eg, 'maternal grandmother') and Observation (eg, list of diseases or lifestyle factors). Across the tools, these four elements were found in ≥75% of the tools along with Name (of the Family Member), Age at Onset (of the Observation), and Certainty and Negation of Observation. Five elements were found to be unique to a particular tool: Step Relationship, Foster Status, and Sex for the AMA's Adult Family History Form; Temporality of Observation (related to cigarette smoking) for the Utah Department of Health's FHH Toolkit; and Multiple Birth Status (responses to question 'Was this person born a twin?') for the U.S. Surgeon General's My Family Health Portrait.

Intra-source comparison of data elements: specifications

A total of 37 elements were contributed by the FHH-related specifications from HL7, HITSP, AHIC, and openEHR that provided 26, 24, 24, and 18 elements, respectively (see online supplementary appendix D). Similar to the notes and tools, several elements were included based on potential values specified for Family Member. In particular, the HL7 Vocabulary for RoleCode included values such as 'maternal grandparent,' 'natural child,' 'half sibling,' 'step child,' 'adopted child,' 'foster child,' and 'twin,' thus resulting in the inclusion of Side of Family, Genetic Relationship, Half Relationship, Step Relationship, Adoptive Status, Foster Status, and Multiple Birth Status as elements for HL7. Elements common to all four organizations included: Family Member, Name, Gender, Date of

Table 3: Comparison of FHH elements across notes

	MTS (n = 850 statements) (n = 25 elements)			UPMC (n = 273 statements) (n = 17 elements)			FAHC (n = 535 statements) (n = 26 elements)		
Element	No.	Percent	Unique values	No.	Percent	Unique values	No.	Percent	Unique values
Source									
Source of information	3	0.4	3	–	–	–	2	0.4	2
General									
General statement	93	10.9	38	45	16.5	21	45	8.4	28
General statement type	*	*	*	*	*	*	*	*	*
Family member									
Side of family	73	8.6	14	1	0.4	1	57	10.7	12
Family member	510	60.0	71	120	44.0	23	407	76.1	54
Half relationship	*	*	*	*	*	*	*	*	*
Step relationship	*	*	*	–	–	–	–	–	–
Degree of relationship	*	*	*	–	–	–	*	*	*
Current age	30	3.5	22	–	–	–	72	13.5	43
Ancestry	–	–	–	–	–	–	5	0.9	5
Living status	161	18.9	9	45	16.5	5	80	15.0	5
Date of death	3	0.4	3	1	0.4	1	4	0.8	3
Age at death	92	10.8	57	20	7.3	11	34	6.4	25
Cause of death†	115	13.5	1	31	11.4	1	52	9.7	1
Certainty of family member	2	0.2	1	–	–	–	4	0.8	3
Negation of family member	9	1.1	2	–	–	–	3	0.6	2
Quantity of family member	82	9.7	21	19	7.0	7	70	13.1	15
Observation									
Observation	720	84.7	289	216	79.1	107	460	86.0	249
Observation type	720	84.7	4	216	79.1	3	460	86.0	5
Date of onset	–	–	–	–	–	–	8	1.5	7
Age at onset	24	2.8	19	8	2.9	7	22	4.1	17
Certainty of observation	80	9.4	21	54	19.8	7	20	3.7	11
Negation of observation	98	11.5	7	41	15.0	5	63	11.8	6
Quantity of observation	9	1.1	6	–	–	–	4	0.8	4
Relevance of observation	9	1.1	4	1	0.4	1	5	0.9	3
Strength of observation	88	10.4	6	25	9.2	5	32	6.0	7
Temporality of observation	16	1.9	11	–	–	–	10	1.9	9

Values are number and percentage of statements.
*Included due to values for General Statement or Family Member.
†Determined from relationship between Family Member and Observation.
FAHC, Fletcher Allen Health Care; FHH, family health history; UPMC, University of Pittsburgh Medical Center.

Table 4: Comparison of FHH elements across tools

Element	Adult Family History Form	Cancer Family Tree	Colon Cancer Risk Assessment	Does It Run in the Family? Toolkit	Family Health History Toolkit	Family Health *Link*	Family Healthware	My Family Health Portrait
	(19)	(10)	(9)	(19)	(14)	(10)	(11)	(22)
Family member								
Side of family	○	○	●	○		○	○	○
Family member	●	●	●	●	●	●	●	●
Genetic relationship	○						●	○
Half relationship	●		●			○	○	○
Step relationship	●							
Adoptive status	●							●
Foster status	●							
Multiple birth status								●
Name	●			●	●	●	●	●
Sex	●							
Gender		●			●			●
Place of birth	●			●	●			
Date of birth	●			●	●			●
Current age	●	●		●	●			●
Race				●				●
Ethnicity	●			●				●
Ancestry				●		●		●
Living status	●	●		●		●		
Date of death	●			●				●
Age at death		●		●	●			●
Cause of death	●			●	●			●
Quantity of family member			●				●	●
Observation								
Observation	●	●	●	●	●	●	●	●
Observation type	○	○	○	○	○	○	○	○
Age at onset		●	●	●	●	●	●	●
Certainty of observation			●	●	●		●	●
Negation of observation		●	●	●	●	●	●	●
Quantity of observation	●			●				
Temporality of observation					●			

Elements for family members of patient only; some may also be collected for the patient in addition to other patient-specific elements.
(n) indicates total number of elements; ● indicates explicit element; ○ indicates included due to values for Family Member or Observation.
FHH, family health history.

Birth, Age at Death, Cause of Death, Observation, Observation Type (included due to values for Observation), and Age at Onset. Seven elements were found to be unique to a particular organization: Source of Information for HL7; Multiple Birth Order and Temporality of Observation (related to dates for genetic tests) for HITSP; and Quality of Relationship (eg, 'estranged' or 'close'), Place of Birth, Partner Status, and Certainty of Family Member for AHIC.

DISCUSSION

The results of this study highlight the value of multi-source development of an integrated model for FHH and provide guidance for the next steps. Through examination of multiple sources, a representative and complementary set of FHH data elements was identified. Compared with the first version of the Integrated FHH Model, the number of elements in the second version almost tripled, increasing from 16 to 44. The additional elements were related to demographics and relation types for family members, observation types and details, and general statements. Broader implications of this work include raising awareness of potential gaps in existing systems and documentation practices with various stakeholders (eg, developers of EHR systems and patient-facing FHH tools, standards development organizations, policy makers, and end-users such as clinicians and patients) for guiding enhancements that could ultimately lead to more comprehensive and standardized structured data entry of FHH.

Diversity in content and format was observed across the clinical notes, patient-facing FHH tools, and FHH-related specifications. While the integration process aimed to address many of these variations, there were some aspects and details that were not incorporated, which could be accommodated in future versions of the model and annotation schema for clinical text from EHR systems. For example, within the clinical notes, there were sentences including related or complex observations such as 'pneumonia as a complication to Alzheimer disease' or 'blindness secondary to diabetic retinopathy' that could be annotated in several ways (eg, as a single observation, two separate unlinked observations, or two separate linked observations). In addition, observations categorized as 'Other' suggest the need for additional categories such as General Health (eg, 'healthy' or 'well') and Exposure (eg, 'second-hand smoke exposure' or 'positive asbestos exposure') in addition to Disease, Procedure, Medication, and Lab Test. Further analysis and comparison of FHH documentation, in both structured and unstructured formats in the EHR, with respect to different roles and specialties (eg, primary care providers, prenatal care providers, oncologists, pharmacists, and medical geneticists and genetic counselors) or particular conditions (eg, cardiovascular disease and cancer) may be valuable for enhancing the general Integrated FHH Model with additional elements as well as informing the inclusion and priority of elements in context-specific implementations of the model.

Several tools included a response of 'don't know' or 'unknown' for questions suggesting the need to capture the certainty of other elements in addition to Certainty of Family Member and Certainty of Observation (eg, Certainty of Ancestry for 'What best describes your birth mother's ancestry' in Family Health*Link* and Certainty of Living Status for 'Living?' in My Family Health Portrait). Other tools included questions such as 'other information of significance' in the AMA's Adult Family History Form and 'other health concerns' in Genetic Alliance's Does It Run in the Family? Toolkit where the free-text responses would need to be analyzed in order to determine if existing elements are sufficient or additional ones are needed. Finally, there were some tools that included detailed questions related to the social history of family members (eg, smoking status and amount, frequency of alcohol use, and frequency of vigorous routine exercise in the Utah Department of Health's FHH Toolkit) where the Integrated FHH Model could include elements that link to separate models for capturing details specific to different social, psychosocial, behavioral, and environmental factors (eg, alcohol use, drug use, living situation, marital status, occupation, residence, and tobacco use).[81,82] There have been efforts by standards development organizations such as HL7 and openEHR to develop models for some of these factors that could be adopted and enhanced (eg, HL7 Tobacco Use Observation[77] or openEHR archetypes for Alcohol Use and Alcohol Use Summary[80]).

Analysis of patient-specific questions in the FHH tools as well as additional general, specialty-specific, or condition-specific tools (eg, MeTree,[48,49] OurFamilyHealth,[54] Myriad Genetics Family History Tool,[83] and March of Dimes FHH Form[84]) and specifications from national and international organizations (eg, Clinical Data Interchange Standards Consortium (CDISC)[85,86] and other HL7 implementation guides) could serve to further validate and enhance the Integrated FHH Model with additional elements. For example, while the two HL7 specifications reviewed in this study did not explicitly include elements for Partner Status and Multiple Birth Order, these are specified in the HL7 Reference Information Model (RIM) (maritalStatusCode in the HL7 Person class and multipleBirthOrderNumber in the HL7 Living Subject class).[87]

Other next steps include creating formal representations of the Integrated FHH Model in accordance with existing national and international information modeling initiatives (eg, Clinical Element Model (CEM) used by the Strategic Health IT Advanced Research Project for Secondary Use of EHR Data (SHARPn),[88,89] Clinical Information Modeling Initiative (CIMI),[90,91] and Federal Health Interoperability Modeling (FHIM) Initiative[92]). As part of this process, the model would be enhanced with clear definitions, attributes, cardinality, and value sets for each element. For example, across the sources, sex and gender[93,94] as well as race, ethnicity, and ancestry[95,96] were used interchangeably, which should be distinguished for appropriate use. For attributes, seed lists could be generated from those defined in existing specifications (eg, HL7 and AHIC) such as identifier, code, coding system, certainty, negation, status, and sensitivity. In addition, logic could be associated with particular elements that may be inferred or computed based on values for other elements to potentially minimize data entry effort (eg, if Family Member = 'grandmother,' then Sex = 'female' and Degree of Relationship = 'second-degree relative').

Analysis of the various sources in this study resulted in multiple lists of values for elements. For example, different lists of family members were observed across the tools (eg, the Cancer Family Tree tool had a list of seven types of relatives, while Family Health*Link* included 20); varying lists for observations were generated from the notes (eg, 245 values categorized as diseases for MTS, 95 for UPMC, and 213 for FAHC);

and different age and date formats were noted across the sources (eg, both specific and estimated ages such as '34,' '30s,' '30–39,' and 'young age' for Age at Onset). A key part of the information model development process is alignment with terminologies, coding systems, data types, and units.[97] Similar to the integration of data elements, efforts are needed to merge and map these values to standardized terminologies and coding systems such as those specified in the HL7 specifications examined in this work (eg, Logical Observation Identifiers Names and Codes (LOINC),[98] Systematized Nomenclature of Medicine—Clinical Terms (SNOMED CT),[99] and HL7 V.3 Vocabulary and Data Types[87,100]) to support semantic interoperability.

CONCLUSION

There has been increased emphasis on the importance of FHH for supporting personalized medicine, biomedical research, and population health. The multi-source development of an integrated FHH model in this study contributes as an initiative for improving the standardized collection and use of FHH information in disparate systems.

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CONTRIBUTORS

ESC and GBM conceptualized the overall study design. GBM led development of the annotation schema, TJW and EWC annotated the clinical notes, EWC and ESC analyzed the tools, and TJW and ESC analyzed the specifications. INS contributed to the integration and mapping of data elements across sources as well as interpretation of results. YW provided support for the annotation of clinical notes, including development of scripts for processing the annotations. ESC compiled the results and drafted the manuscript. All authors were involved with the consensus-based processes used for enhancing the annotation schema/guidelines and Integrated FHH Model, as well as reviewing and editing the manuscript.

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COMPETING INTERESTS

None.

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Supplementary material is available online at http://jamia.oxfordjournals.org/.

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