



D. Additional Project Materials

Appendix 1D. 2005 Proposal to NHGRI-ECIB

Principal Investigator Michael F. Murray, MD submitted this proposal for “**An Employee-Based ‘Family History Project’ within a Large Urban Hospital**” to the NHGRI-ECIB in August 2005 in response to the RFP “Demonstration Project on the Integration of Family History and Genomics Education Materials and Resources into Communities.” This proposal was granted the solitary contract awarded in 2005 for this RFP, and work began immediately on the Brigham and Women’s Hospital Family History Project.

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A. Specific Aims

Employees of hospitals comprise over 3.5% of the U.S workforce, and more than 1% of the total U.S. population. The employees at each of the 5,764 U.S. hospitals thus constitute a “health care community.” We propose to employ a single hospital community as a pilot site for evaluating the dissemination and use of Family History and Genomics Educational Materials. The rationale for choosing this type of community includes: (1) hospital employees are an educationally, ethnically, and economically diverse workforce; (2) engaging the professional staff (who will be both “users” and “providers” in this model) as “early adopters” will accelerate the widespread implementation of these tools; (3) successful use of these materials by hospital employees can be subsequently scaled-up and applied to available patient populations at the same institution using the established educational and technological infrastructure; and (4) once established, a working model can be exported to hospitals and other healthcare institutions throughout the U.S. It should be noted that hospitals will become natural allies in efforts to expand the use of the Family History tool. As employers, they have a significant stake in the health of their employees. As providers, they serve to improve the health of the greater community. And as institutions, they are interested in advancing the integration of important health care information.

We propose to create a model program that will support the use of the U.S. Surgeon General’s “*My Family Health Portrait*” tool within the employee community of a large urban hospital. Brigham and Women’s Hospital (BWH) is a 735 bed hospital and a major academic medical center and teaching affiliate of Harvard Medical School. It is also part of Partners Healthcare, a comprehensive healthcare system that includes 6 hospitals and an extensive outpatient network. BWH employs 11,941 people, including 4,000 doctors and nurses and 7,000 employees who constitute the administrative, service and management staff. As such, the employees of BWH constitute an ideal community in which to test the effectiveness of focused efforts to obtain voluntary participation in a family history gathering initiative using “*My Family Health Portrait*.”

To this end, we will undertake three Specific Aims:

Specific Aim 1. To provide extensive educational support and to make available the necessary tools to all hospital employees who can then make a voluntary decision to participate in the Family History Project.

Specific Aim 2. To identify the obstacles to participation.

Specific Aim 3. To evaluate feedback strategies for both “users” and “providers.”

Following this demonstration project, the key findings will be presented at national meetings. In the most ideal circumstance, this project, denoted the Family History Project or FHP, has the potential to lead to the development of new principles for obtaining high quality family history information that can then be deployed at other hospitals throughout the U.S.

B. Background and Significance

1. The Hospital as a health care community

Hospitals are obviously a crucial component of the healthcare delivery system. In addition, it should not be overlooked that hospitals are also the place of employment for 5.3 million Americans¹. According to the U.S. Bureau of Labor Statistics, the “Health Services Sector” was the largest U.S. industry in 2002; health services provided 12.9 million jobs. Within this employment sector, hospitals constitute only 2 percent of the work establishments, but they employ 41 percent of all workers. The workforce of each of the nation’s 5,764 hospitals² can be defined as a health care community, composed of consumers and providers.

Hospital employees are a diverse group of Americans, and they perform a range of occupations within hospitals. The educational requirements alone for hospital employment range from little or no specialized education for certain service occupations to college and professional school for some professional level jobs. Among the service occupation workers are nursing aides, building cleaning workers, and food service employees. The United States Bureau of Labor Statistics categorizes hospital employees as shown in **Table 1**.

Hospital Occupational Category	Percentage of Workforce
Management	4.8%
Professionals	43.5%
Service	31.4%
Administrative Support	18.0%

Table 1. Occupational Categories within Hospitals¹

The Healthcare sector of the American workforce is unique in that while all its employees are themselves healthcare “consumers”, the professional staff have roles as both “consumers” and “providers” of healthcare. This gives the professional staff within healthcare institutions a high level of “social connectivity” which positions them to assist in the rapid dissemination of “new” ideas and practices³.

The ultimate success of a broad-based voluntary public health undertaking such as the U.S. Surgeon General’s Family History Initiative will be enhanced by demonstrating its use within the employee base of a hospital for several reasons. First, the diversity of the workforce allows for an opportunity to examine the barriers to uptake across a range of racial, ethnic, economic, and educational strata. Second, the 30-40% of employees who are also healthcare providers will be amongst the first to become educated in the process (from their consumer perspective), and they can then become allies and advocates in efforts to engage their patients. Lastly, an established demonstration model that involves hospital employees can in principle be exported and repeated within healthcare institutions in every community in the nation.

2. BWH as community for the Family History Project

a. Brigham and Women’s Hospital

BWH (<http://www.brighamandwomens.org/>) is located within the city of Boston, and is the city’s second largest employer. Metropolitan Boston has a population of approximately 575,000 people. The major industries are health care, financial services, insurance, and higher education. Like

Washington, D.C. and San Francisco, the city of Boston employs more people than it has residents, and an estimated 61% of the City's employees live outside of the city limits.

BWH is a 735-bed hospital in Boston, a major academic medical center and a teaching affiliate of Harvard Medical School. BWH employs 11,941 people, approximately 4,000 of these employees being doctors and nurses with other 7,000 employees comprising predominantly administrative, service and management staff. Despite the fact that a single institution is proposed in this demonstration project, there is a need for online and distance learning materials. A longstanding problem of scarce available real estate adjacent to the hospital campus has over the years created a situation where a significant portion of the employees work off the hospital campus at more than a dozen sites throughout Boston and its near suburbs. Effective participation from this portion of the workforce will almost exclusively require electronic access and support.

b. Resources available

BWH Healthcare and Research Infrastructure

Each year BWH provides care for approximately 41,000 inpatients, and 754,000 outpatients. More than 28,500 surgical procedures are performed annually, and the Emergency Department has 54,000 visits. As New England's largest birthing center more than 9,900 babies are born at BWH. BWH is internationally known for its treatment of complex disorders in clinical areas such as cardiac care, cancer treatment, neurosciences, orthopedics and arthritis, and women's health, as well as offering outstanding primary care services.

BWH is home to a large and complex biomedical research enterprise. There are over 500 scientists and \$240 million in research grants dedicated to exploring questions of basic and clinical science. There is research interest and expertise on genetic contributions to disease in the areas of Cardiovascular Disease, Breast Cancer, Colorectal Cancer, Diabetes, Stroke, Asthma, Osteoporosis and Renal Disease. In addition, there is an active Primary Care research group (led by Dr. David Bates) whose focus includes the optimal use of electronic medical data.

BWH Information Technology Infrastructure

BWH has three electronic tools that are relevant to this project, each of which can be further adapted to assist in the overall efforts of the institution's Family History Project. First, the Partners Patient Gateway (<http://www.patientgateway.org/>) is a patient initiated electronic tool for access to physicians, physician's offices and to the patient's medical record. As part of an ongoing ACHRQ grant to the Primary Care Research Group, a Family History Tool is coming on line as part of this electronic resource later this year to give primary care physicians decision support information in six target disease categories (cardiovascular disease, diabetes, colon cancer, breast cancer, osteoporosis, and glaucoma). However, this non-pedigree tool does not lend itself as well as Surgeon General's family history tool to procuring a detailed high quality family history. It is, however, well suited to integration with the second resource, the BWH electronic Longitudinal Medical Record (LMR) that is maintained for each BWH patient. Although beyond the immediate scope of this proposal, the Patient Gateway expertise is well suited to the goal of seeing the eventual integration of the Surgeon General's family history tool into the LMR. This would be a natural off-shoot of the project proposed here, since a significant number of BWH employees also receive their primary care at BWH. Lastly, *BWH GENE TALK* is an internet site under development by the PI to support a health professional distance-learning program, which has recently been piloted in July and August, 2005. It will be presented at a national meeting being organized by Dr. Bruce Korf for Sept., 2005, and this site will host a special area for the FHP.

BWH ELSI Infrastructure

Two distinct entities are available to this project for ongoing consultative input and advice: the Partners Institutional Review Board (IRB) and the Center for Bioethics. The key personnel on this proposal include individuals representing both of these entities: Ms. Heather Ferguson, M.S., C.G.C. is a member of the IRB, and Lisa Lehmann, M.D. is the Director for the Center for Bioethics. IRB input will be specifically sought in regards to an appropriate “informed consent” procedure for this project, as well as other issues related to the protection of human subjects. In addition, ongoing counsel from Dr. Lehmann’s Center will be sought particularly regarding clear definition of the appropriate roles for the employers and supervisors in this project.

BWH Institutional Support

It is important to note that there is considerable enthusiasm and support for a “Family History Project” of this nature at BWH. Evidence for this includes the accompanying letter of support from BWH President Gary Gottlieb, M.D., M.B.A., and the willingness of our research administration to waive the standard NIH indirect costs given this proposal’s small budget. In addition, the BWH Biomedical Research Institute will potentially provide funding (maximum \$120K) for the extension of this FHP initiative that would involve the integration of the Surgeon General’s family history tool into the BWH LMR, for those employee-patients who might voluntarily elect this option were it available. We are thus excited that there is extremely strong support from BWH for the NHGRI Family History Project.

3. Demographics of the target community

The demographics of the BWH Employee population reflect diversity in terms of ethnicity, gender, age, and geographical domicile. In particular, the ethnic composition of the complete BWH employee population is provided in **Table 2**. This breakdown is similar but not identical to that in the Boston metropolitan area, which is 54.5% White, 25.3% Black or African American, 7.5% Asian American, 0.40% American Indian with 14.4% Hispanic or Latino (of any race). One corollary of this modest disparity is that a significant number of BWH employees do not reside in the Boston metropolitan area, but rather live in areas outside the city, **Table 3**. For the purposes of this project,

Table 2. All BWH employees, by ethnicity

Ethnicity	Total No.	% Total
American Indian	18	0.15
Asian / Pacific Islander	891	7.46
Black	1923	16.10
Hispanic	885	7.41
White	7798	65.30
Not Available	425	3.56
Total No. Employees	11941	100%

Source: BWH Human Resources

Table 3. All BWH employees, by domicile

Location	Total No.	% Total
Metropolitan Boston	3974	33.28
Massachusetts (non-Bos.)	7612	63.75
Outside MA	355	2.97
Total No. Employees	11941	100

Source: BWH Human Resources

however, we regard this as a strength, because it can well be argued that the composition of a geographically defined target community of similar size to that proposed here for the demonstration project (e.g. a small town) may well be more ethnically homogeneous and less diverse than the BWH employee population described here.

In addition, the latest ethnic breakdown for BWH employees by profession, as summarized in the 2004 EEOC Report, **Table 4**. The most obvious skewing in this breakdown is the under-representation of minority employees among the professional employee group, for both male and

female genders. Conversely, minority groups are overly represented amongst service workers and office and clerical staff. Please note that the number of BWH employees listed in **Table 4** (9149) is significantly less than that listed in the other Tables shown in this section (11941). This is because the EEOC only requests employee information for weekly paid employees, and the difference (~2800 employees) reflects many professional staff who are paid on a monthly basis.

Table 4. Ethnicity by job category (Source: 2004 BWH Report to the EEOC).

SECTION D - EMPLOYMENT DATA

JOB CATEGORIES	***** MALE *****						***** FEMALE *****				
	OVERALL TOTALS	WHITE (NOT OF HISPANIC ORIGIN)	BLACK (NOT OF HISPANIC ORIGIN)	HISPANIC	ASIAN OR PACIFIC ISLANDER	AMERICAN INDIAN OR ALASKAN NATIVE	WHITE (NOT OF HISPANIC ORIGIN)	BLACK (NOT OF HISPANIC ORIGIN)	HISPANIC	ASIAN OR PACIFIC ISLANDER	AMERICAN INDIAN OR ALASKAN NATIVE
OFFICIALS AND MANAGERS	854	139	19	10	2	2	425	35	7	14	1
PROFESSIONALS	3783	420	14	20	48	0	2939	155	59	126	2
TECHNICIANS	1995	301	95	35	43	1	942	371	99	105	3
SALES WORKERS	0	0	0	0	0	0	0	0	0	0	0
OFFICE AND CLERICAL	1718	128	60	33	8	1	784	476	186	39	3
CRAFT WORKERS (SKILLED)	63	45	12	4	2	0	0	0	0	0	0
OPERATIVES (SEMI-SKILLED)	2	1	1	0	0	0	0	0	0	0	0
LABORERS	0	0	0	0	0	0	0	0	0	0	0
SERVICE WORKERS	934	80	231	122	3	0	92	255	141	10	0
TOTAL	9149	1114	432	224	106	4	5182	1292	492	294	9
PREVIOUS REPORT TOTAL	9175	1185	428	218	97	2	5170	1302	471	294	8

Table 5. All BWH employees, by age

Age Range	Total No.	% Total
14-20	265	2.22
20-29	2638	22.09
30-39	3260	27.30
40-49	3041	25.47
50-59	2100	17.59
60-69	564	4.72
70-79	66	0.55
80-86	6	0.05
Total No. Employees	11941	100%

Table 6. All BWH employees, by gender

Female	8845
Male	3094
Unknown	1

Source for Tables 5 and 6:
BWH Human Resources

Lastly, the BWH employee population is also diverse in terms of employee age, and although it deviates significantly from 50:50, by gender, **Tables 5, 6**. In sum, the demographics described above may not mirror the composition likely to be encountered in any particular geographically defined community. However, what is abundantly clear is that the BWH employee population comprises a highly diverse community that also offers several significant and potentially powerful advantages for this project in terms of its size, diversity and mix of users and providers.

4. Significance

In the future, the ability to diagnose and treat debilitating disease, and to anticipate an individual's potential to develop disease, will be enabled by knowing his or her genetic make up. As a first step towards implementing this goal at the community level, we propose to develop a systematic approach

to obtain, on a voluntary basis, detailed family histories from the 11,000 BWH employees that comprise the extended Brigham community. This health care community is socio-economically diverse and enjoins an employee population that contains both a concentrated workforce component at BWH, and a dispersed component at various BWH affiliates. As such, it will test several key aspects of family history gathering that are likely to arise in any community, including compliance, confidentiality, communication, logistics and interaction with providers.

In addition, however, use of the hospital as an initial nucleation point for gathering family history data constitutes a novel model that if successful, has the potential to be extended to other hospitals in the Partners Healthcare System (<http://www.partners.org/>), and throughout the U.S.

C. Project Plan: Engaging the Community in the Family History Project

In planning this project, we have sought to divide the task into 4 discrete, sequential phases. We have arbitrarily termed these: (1) the Pre-launch preparation phase, (2) the Pre-launch publicity and education phase, (3) the Launch and Implementation phase, and (4) the Evaluation phase. Below, we describe the key components and plan for each of these 4 phases

It is important to note that Thanksgiving Day, Thursday, November 24, 2005, has been designated as National Family History Day. Thus, this FHP will be organized to start as soon as a funding decision is made (Aug. 24, 2005). This timing will permit the FHP to take advantage of the increasing national publicity that attends National Family History Day. Nonetheless, we wish to emphasize that while a major burst of activity will occur during the Fall, the FHP will extend throughout the year, from Sept. 15, 2005 to Aug. 31, 2006, as stipulated by the NHGRI RFP.

1. Pre-launch preparation phase

a. IRB approval

There are several pre-launch preparation phase tasks of immediate importance that must be completed to maintain this timeline. The first of is the timely submission of a protocol to the Partners IRB for approval. Two major issues that need to be addressed include the provisions for confidentiality, especially given the proposed use of the on-line version of *My Family Health Portrait*, and also for the informed, voluntary consent of the employee population. Ms. Heather Ferguson, M.S., C.G.C., one of the Key Personnel on this proposal, is a member of the Partners IRB. We have contacted Jonathan Alpert, M.D. of the IRB to discuss these matters, and have been assured that the IRB is, in principle, highly supportive of this endeavor. Therefore, we do not anticipate any problems securing IRB approval.

b. Print materials for distribution

Initial visibility for the FHP amongst BWH employees will be generated by: (i) a brochure mailed to all 11,000 BWH employees, (ii) posters in the BWH Lobby (75 Francis St.), BWH Cafeteria (level 2), Ambulatory Lobby (45 Francis St.) and the PBBH Lobby (15 Francis St.); (iii) articles in the main BWH internal publications (Pike Notes and the BWH Bulletin). Peter Brown, BWH Vice President of Public Affairs and Communications, has pledged his full support for help with these matters.

Since the idea of large scale family history ascertainment can create anxiety about the use of such information, the mailed brochure in particular will serve not only as a public relations vehicle but will also provide the opportunity to address potential concerns about confidentiality. In addition, the Working Group will hold a focus group meeting with BWH employees in September to identify other potential concerns with the Project. This input will be used to inform the content and design of the

brochure. In particular, the brochure will be available in both English and Spanish and will address concerns about privacy. It will also address the potential importance of the Family History as an increasingly important public health tool in the future.

c. Establishing family history focused BWH websites

The rationale for providing a printed brochure to all employees relates in part to the fact that, just as in any community, a significant percentage of the BWH employee community may not be adept at accessing computer based information or e-mail broadcasts. On the other hand, these latter media are extraordinarily efficient vehicles for disseminating information, and we do anticipate that a large fraction of our employee base is computer literate. This same issue will arise later when we discuss paper versus web-based use of the *“My Family Health Portrait”* family history tool. Therefore, two websites will be developed as part of this project.

The first website will be the *“My Family Story”* Intranet Site. This will be a project specific site within the BWH intranet that will be easily accessible to interested employees. It will include weblinks and downloadable materials related to the task of completing one’s family history. There will be patient stories about why family history is important and how to obtain the information from relatives. There will also be elements of the site that will make it worth re-visiting, such as updates, trivia questions and learning opportunities. Once the project is underway, we will encourage participants to share interesting, funny, and important stories about their experiences in pursuing their family history. A selection from these stories will be made available to other employees after all privacy concerns are addressed. While this will be anecdotal, we believe it will also be instructive. Permission will be sought to eventually make some of these stories part of the educational materials that we will deliver as part of our final project product.

This site will initially launch with prototype “case histories” and supportive educational materials. However, we plan to have the participants make the case for “Why Family History Is An Important And Useful Tool In Health Care?” and “How To Engage One’s Family To Collect Appropriate Family History Information?” with their own stories. Lastly, there will be mechanisms to communicate with the project staff to ask for assistance or to provide feedback.

The second website that will support this project is the *“BWH GENE TALK”* Internet site, already under development by the P.I. in collaboration with the BWH Telemedicine Group. This site is intended support a health professional distance-learning program, and a “Family History” portion of the site will be added for both internal and external users. This site will include resources such as the Multilingual Family History materials described below.

d. NHGRI planning document

The NHGRI RFP requests a “planning document, outlining who will be involved in the demonstration project, and how” that is due one month after the contract is awarded (i.e. Sept. 24, 2005). We have already identified several Key Personnel for this project. The month long interval between the contract award date and the due date will allow us to further refine our plans and to identify additional personnel who will play key roles in this project. The NHGRI planning document will provide both FHP project personnel and NHGRI program staff with a milestone to ensure that the above mentioned preparatory requirements are on track.

2. Pre-launch publicity and education phase

a. Development of patient materials

Development of a Multilingual Family History Resource Library

BWH has a workforce that speaks many languages. While most employees speak English, some members of their families may not be bilingual, and some employees' capacity to understand written English may be limited. While addressing our employees' needs for Family History Tools in languages other than English, we will create a multilingual family history resource library that will eventually be posted along with other resource material as part of the Family History portion of the "BWH GENE TALK" internet site. Given our employee demographics we anticipate that at a minimum there will be requests to develop specific educational materials in Spanish (to supplement the Surgeon General's information), Haitian Creole, Kriolu (or Cape Verdean Creole), Polish, Russian, and Portuguese. Through a collaborative effort between Dr. Cynthia Morton (a key collaborator on this Project) and Peter Zhang, an M.D., Ph.D. student in Medical Genetics at Peking Union Medical College in Beijing, a Chinese language version of "My Family Health Portrait" has already been created for posting on a popular Chinese web site; we plan to add this material to our resource library.

BWH Interpreter Services

As a large urban healthcare institution, the hospital has an active "interpreter services" group who are committed to clearly and thoroughly communicating with any patient and their family who needs assistance. In addition, we will engage the BWH interpreter services to professionally translate printed materials as needed for all interested employees. We will also ask employees who translate materials for their own families to submit copies of any written or electronic translations of materials to us for review, and to potentially include in our online multilingual resource library.

b. Development of Materials for Health Care Providers

The provider as user

Our strategy includes a plan to engage every provider at BWH first as a User. This will give those providers direct knowledge of the educational materials available to users, and the potential difficulties involved in being a participant in the demonstration project. We also hope that our providers will give us useful feedback regarding how to improve the educational and operational tools. To target faculty, presentations will be made at faculty meetings for different BWH Departments (just one way in which this RFP is pan-Departmental) that will serve the purpose of making providers aware of the initiative in their capacities as both employees and providers.

BWH Electronic Medical Record Decision Support

An ongoing ACHRQ study at BWH has supported the creation of a family history feature within the electronic medical record. This feature allows for non-pedigree based family history data entry, and will provide specific decision support data to providers based on a series of "high risk" markers in the areas of cardiovascular disease, diabetes, colon cancer, breast cancer, osteoporosis and glaucoma. It is our intention that this unique tool will also support providers in their assessment of patient specific data acquired through this demonstration project. Moreover, since this Partners Gateway feature is modifiable, we expect that this tool will evolve based on findings and recommendations that emerge from this project.

3. Launch and Implementation Phase

a. Engagement of users

All 11,941 BWH employees will be invited to participate according to the participation algorithm summarized in **Figure 1**. As described below, there will be ongoing electronic and non-electronic outreach throughout the project period from October 1 to August 1, 2006. The electronic component

will involve group e-mails that provide links, make announcements and report progress throughout this period.

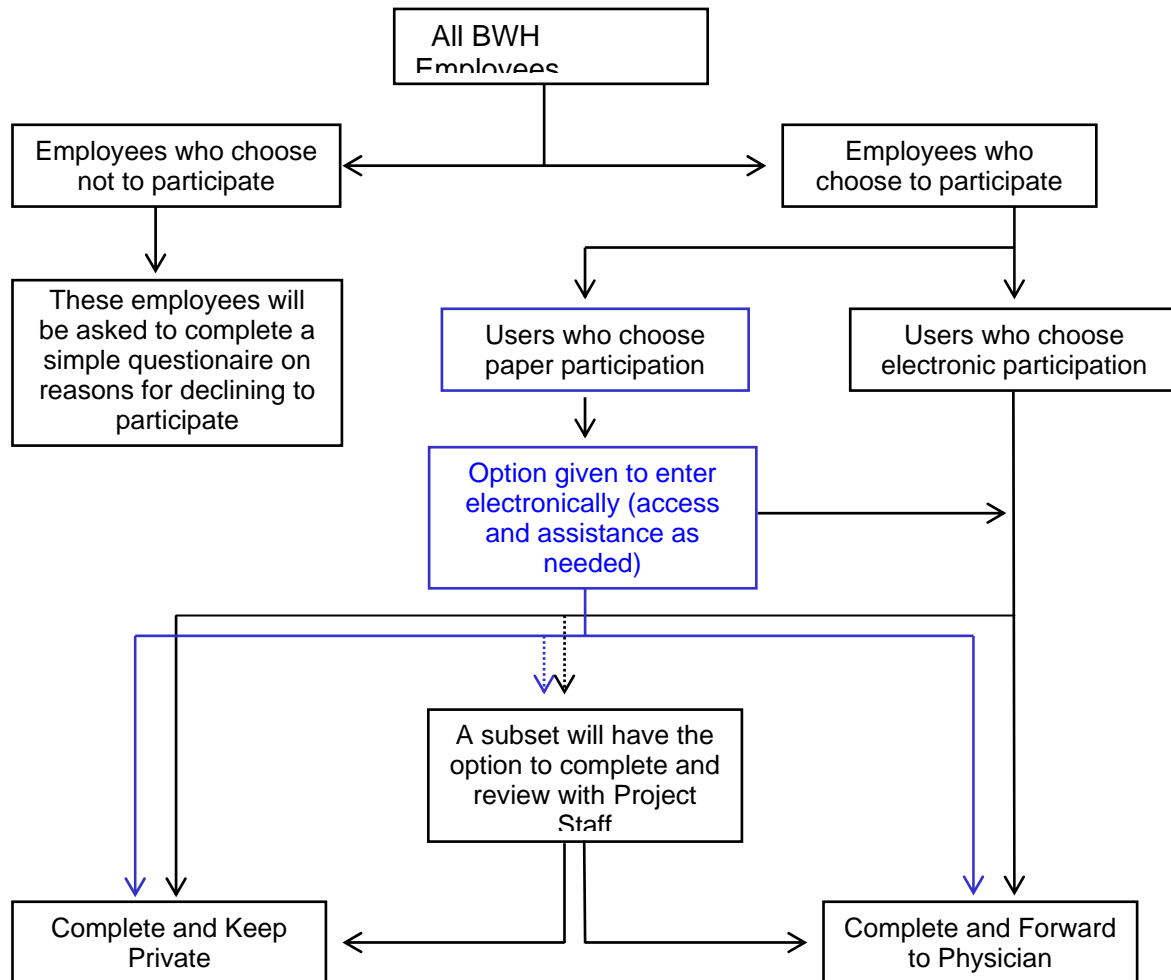


Figure 1. Employee Participation Algorithm. This completely voluntary program for hospital employees will allow for the option of declining participation in both the program and in any surveys. Translation services and assistance with using the tool will be provided to all interested employees. Review with the professional staff of the project (i.e. a medical geneticist or genetic counselor) will be offered to a subset of interested employees; a minimum sample of 100 self-identified interested persons will be engaged in this manner. Surveys of users and non-users will seek to assess reasons for decisions made by employees and barriers to participation.

The non-electronic component will involve two “family history kiosks” that will be located at different sites around the hospital at various times during the project, including the main lobby, the cafeteria, and outside the main parking facility. In addition, paper informational brochures will be distributed to employees. Project staff will also present information about the project to any employee groups that are interested in hearing more about the project (e.g. labor union meetings, new employee training sessions). Since all new BWH employees must visit Occupational Health for mandatory TB testing, the option of participating in *My Family Health Portrait* will be offered on a voluntary basis at this site, thus maintaining an up to date employee list.

One kiosk will include a centrally placed work area (table and banner) outside the BWH cafeteria that will be staffed to coincide with meals and cover all three shifts. Interested employees can register to participate in the FHP and will be given the option of whether they wish to fill out their Family Histories at home or on site, using the paper or on-line versions of the U.S. Surgeon General’s “*My Family*

Health Portrait,” available in both English and Spanish versions. We will have the electronic version of the tool installed and available on every computer workstation in the hospital. We will also distribute paper copies of the tool in English and Spanish. Project staff, including an on site genetic counselor, will be available to assist individual employees who elect to enter their family history data on-site, be it on paper or electronically. Participants may voluntarily elect to have their family history reviewed by a genetic counselor or medical geneticist, prior to forwarding to their primary care provider, which again, will be done on a purely voluntary basis.

b. Engagement of BWH and external providers

As indicated elsewhere, providers will first be engaged to participate as users. There will also be provider-specific outreach to promote participation. This electronic interface will involve the transmission of provider group-specific e-mails, with project staff responding to electronic queries. In addition, focus group meetings to address provider concerns will take place as needed, and presentations to provider meetings and clinical conferences will be offered by project staff.

Importantly, we recognize that many of the hospital employees receive some or all of their healthcare outside of BWH. All of the employees do have a medical record generated by BWH occupational health, but we know that the key parameter for this project will be where they receive their primary care. We will develop a support apparatus for outside healthcare providers that will give login access to obtain information on the family history tool, the educational support materials, and the decision support information that is available through the BWH internal electronic medical record. This login system will allow us the opportunity to begin a dialogue with these providers on how information is being generated, transmitted to them and utilized by them. There will be no breach of patient confidentiality in these exchanges since there will be no discussion or recording of patient specific medical information. This dialogue will be exclusively aimed at feedback on the project, systems evaluation and educational support for providers.

Other Educational Materials

Available resources regarding family history in healthcare are now widely available to both physicians and patients over the internet (a number of websites currently list useful links, including NHGRI's <http://www.genome.gov/11510372>). Much of the recent increase in available information is due to the efforts of many professional organizations including the National Society of Genetic Counselors (NSGC)⁴, the American Society of Human Genetics (ASHG)⁵, the American Academy of Family Practitioners (AAFP)⁶, the American Medical Association (AMA)⁷ and the Center for Disease Control (CDC)⁸. The information included on these sites includes: instructions for recording a family history, pedigree templates, medical and family history checklists, and brochures emphasizing the importance of family history. All of the information from these sources can be downloaded and printed. In addition, there are websites dedicated to specific disorders such as the American Heart Association's (AHA) site⁹ that provides specialized information and an interactive family history tool called "The Heart of Diabetes". For the purposes of this project, these resources will be used to supplement the U.S. Surgeon General's family history tool. Specifically, the weblinks will be made available through our intranet and internet sites, and printed versions of the information will be offered to those that choose paper participation.

Copies of the National Coalition for Health Professional Education in Genetics (NCHPEG) CD-ROM entitled "Genetics and Common Disorders: Implications for Primary Care and Public Health Providers"¹⁰ will be requested from NCHPEG to be made available to local providers who are interested in receiving a copy. Furthermore, if a patient or physician makes a specific request for information pertaining to a family history of a particular disease, all efforts will be made to provide up-to-date and reliable online and/or written information.

4. Evaluation Phase

a. Health “Decision Support” Trial Period

In May 2006 we plan to launch an expanded decision support initiative for both users and providers. This expanded initiative will come 5-6 months after the demonstration project is launched, and will benefit from the analysis of data collected during that initial phase. We anticipate that this expanded initiative will include specific advice for “users” on what kinds of “further data should be obtained” and “questions to discuss with your primary care provider”. For providers, decision support on an expanded number of “high risk” conditions (in addition to the six available at the start of the project) as well as decision support for cases of individuals who have an “increased risk” for target diseases (e.g., a single first degree relative with early onset disease). The process for generating the messages in this expanded decision support initiative is outlined below.

b. Surveys

We will be surveying all employees to assess obstacles to participation. We will periodically review this data and alter methodologies to encourage the maximum “user” engagement. Providers will also be surveyed regarding satisfaction and suggestions for improvement. We also anticipate having periodic face-to-face meetings with interested user and provider groups. A steering committee composed of the grant’s key personnel and others (including “non-provider” users) will be assembled to periodically review survey data and adjust practices.

c. Outcomes Reporting

In this voluntary demonstration project we will not formally attempt to track all outcomes and health interventions that are prompted by this work. However, we will have a self reporting system using unique identifiers in which patients and providers can report on: (a) physician visits, (b) health interventions, and (c) health outcomes. This pilot reporting system will give some data about the nature and range of outcomes, and possibly about the statistical requirements needed to effectively design future family history based studies.

d. Pedigree Analysis Reports

Three panels of three providers will be asked to periodically review “instructive” family pedigrees. The panels (composed of primary care providers, medical geneticists, and genetic counselors) will each review the same pedigrees, and then they will be asked to comment on what further information is needed and what recommendations should be made.

e. Expanded Decision Support Initiative

A decision support committee will meet to formulate the content of the expanded “decision support” messages for users and providers to be implemented starting in May, 2006. This committee will formulate these messages based on: (1) survey and outcomes information, (2) review of published literature and (3) recommendations made to this committee by the pedigree analysis panels. The conclusions from this work will be presented nationally to promote the free flow of ideas, and to help promote the development of a consensus regarding the future utility of family histories.

D. Timeline for Deliverables

1. Pre-launch Preparation [August 24 to November 1, 2005]

1. IRB Approval
2. Planning Document to NHGRI [September 24 2005]
3. Creation of BWH “My Family Story” intranet site
4. Development of Family History Component of the “BWH GENE TALK” internet site

2. Pre-launch Publicity and Educational Campaign

[October 1 to November 14, 2005]

1. Electronic Communication – sent to all email users, with specific emphasis on users at “off campus” sites
2. Paper Communication – via brochure and established hospital newsletters [e.g., “Pike Notes”, “BWH Nursing”, and “Medical Staff News”]
3. BWH Town Hall Meeting – the hospital has several “Town Hall” meetings per year that are open to the entire workforce, and we will plan to have one such meeting to focus on this project.
4. Employee Group Outreach – we will offer to present information at the meetings of different employee groups – e.g., Labor Union Meetings, Medical Staff Meetings, Administrative Departmental Meetings
5. Distribution of “Have you heard your story?” and “Brigham Family History Project” buttons to employees to wear on their clothing (e.g., white clinical coats)

3. Launch Day – pre-Thanksgiving Day [November 14, 2005]

1. Family History Tool and accompanying educational packet available on all hospital computer workstation desktops
2. Lobby and Cafeteria Displays

4. Demonstration Project [November 14 to August 31, 2006]

1. Initial Demonstration and Data Collection Period [November 14 to April 30, 2006]
2. Continued Demonstration, Data Collection and Expanded Health Decision Support Trial Period [May 1 to August 31, 2006]

5. Final Evaluation Report and Project Product Demonstration

[September 1, 2006]

6. Ongoing Commitment to Dissemination of the Model

1. The “BWH GENE TALK” internet site
2. Presentations at national meetings
3. Publications

7. Future Follow up to the Demonstration Project – (contingent on future funding)

1. Scale up family history demonstration to incrementally larger segments of the BWH patient population
2. Export employee-based Family History Project to other hospitals and healthcare institutions, and in our Partners Healthcare Network.

E. Literature Cited

1. US Bureau of labor statistics website: (<http://www.bls.gov/oco/cg/cgs035.htm>)

2. Source: American Hospital Association website
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3. Scott, John P. Social Network Analysis: A Handbook. 2nd Ed. London: Sage Publications, 2000
4. Source National Society of Genetic Counselors website
(<http://www.nsgc.org/consumer/familytree/index.asp>)
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10. Source Genetics & Common Disorders: Implications for Primary Care and Public Health Providers CD-ROM developed by NCHPEG (2005).

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Appendix 2D. Sub-study: Self-referral of Adult Patients for Genetic Consultation

An important sub-study of the BWH FHP was undertaken during the project year. The objective was to study the patterns and outcomes of self-referrals to genetics professionals using the subset of BWH employees who requested a genetics consultation to discuss the family health histories they created using “My Family Health Portrait”. Employees could request an appointment with a genetics professional in one of three ways: (1) a hyperlink posted on the FHP’s intranet site stated “[Request an appointment with a Geneticist](#)” and linked to the BWH FHP email address for interested employees to electronically submit their request; (2) employees were personally invited to meet with a geneticist by our staff and volunteers at the BWH FHP kiosk, during employee group presentations, data entry sessions, and during the New Employee Orientation weekly presentation; and (3) one of the questions on the BWH FHP March survey asked employees if they were interested in meeting with a geneticist to discuss their family health histories. Individual consultations took place in the Interactive Center and consisted of a 20-30 minute session with the genetic counseling intern and either a certified genetic counselor or medical geneticist. The sessions were structured to discuss the participant’s family history, clarify the employee’s concerns, and give disease-specific feedback and recommendations. There were 30 self-referrals from the pool of approximately 13,000 BWH employees (0.2%). Participants who self-referred universally sought to discuss diseases recorded in their family health history in an effort to optimize their own healthcare or that of their close relatives. Employees cited a variety of concerns prompting the Genetics consultation, including: single gene disorders (35%), common complex diseases (91%), and other genetic concerns (17%). Forty-seven percent of the sub-study participants reported that they had previously discussed their family history concerns with their primary care provider prior to the Genetics consultation.

The research conducted in the FHP sub-study was awarded a platform presentation at the 2007 American College of Medical Genetics meeting held in Nashville, Tennessee and was presented by Monica Giovanni, MS.

[1] Description of sub-study and findings

[2] Presentation by Monica Giovanni at the 2007 National Meeting of the American College of Medical Genetics

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2D1. Description of Sub-study and Findings

SELF-REFERRAL OF ADULT PATIENTS FOR GENETIC CONSULTATION

MA Giovanni, KJ Holbrook, MF Murray
Brigham and Women's Hospital, Boston, MA

ABSTRACT:

Hypothesis: The patterns of self-referral to genetics professionals and the outcome of such visits in adult patients constitute an important data set that should be explored. This information could illuminate how to most effectively position and utilize limited genetics professionals within healthcare delivery systems in order to achieve the optimal care of patients.

Study Design: A study of barrier-free patient self-referral to genetic professionals was undertaken among the employee population of a major urban medical center in Boston, Massachusetts. The study design removed specific barriers to the utilization of genetic services including: [1] need for clearance from insurers, [2] need for authorization from primary care providers, and [3] any financial disincentives since consultations were offered free of charge. This study took place within the context of a yearlong effort to support the voluntary use of a family health history tool among hospital employees. All employees were offered the opportunity to discuss genetic concerns raised by their self-gathered history with a genetics professional. These consultations consisted of a 20-30 minute session with a genetic counseling intern and either a certified genetic counselor or medical geneticist. The sessions were structured to discuss the participant's family history, clarify the patient's concerns, and give disease-specific feedback and recommendations.

Results: Over the 12-month period of the study, there were 30 self-referrals from a pool of 13,000 hospital employees (0.2%) to whom the service was available. Participants who self referred universally sought to discuss diseases recorded in their family health history in an effort to optimize their own healthcare or that of their first-degree relatives. Participants cited a variety of concerns prompting consultation, more than one in many cases, including: single gene disorders (35% of patients), common complex diseases (91%), and other genetic concerns (17%). Forty-seven percent of participants reported that they had previously discussed their family history concerns with the primary care provider prior to the genetics consultation.

Conclusion: The nature of the patient questions brought to geneticists in this study does not suggest an over-utilization or misuse of genetic services associated with the removal of the barriers to services. However, the optimal design for incorporating the consultative services of genetic professionals within healthcare delivery systems will depend on issues such as: [1] the effect of self-referral to geneticists on consumer satisfaction, [2] the capacity of the genetics workforce to accommodate this kind of new demand, [3] the relevant genetics expertise of other medical professionals (such as internal medicine subspecialists), and [4] the impact of computer-based point of care decision support on primary care-patient interactions on questions of genetic risk.

Self-Referral of Adult Patients for Genetic Consultation

MA Giovanni, KJ Holbrook, MF Murray
Brigham and Women's Hospital,
Boston, MA



Introduction

- In recent years, insurance carriers have set up barriers to the self-referral of patients to specialists within the healthcare delivery system
 - Insurance providers often require that a patient gain a referral to a genetics provider from a primary physician in order to ensure coverage for the patient visit
- Individual patients who wish to “self-refer” are often left to pay for the visit out of pocket
- It has been previously hypothesized that these barriers to services prevent the over-utilization or misuse of genetics services

Study Hypothesis

- The patterns of self-referral to genetics professionals and the outcome of such visits in adult patients constitute an important data set that should be explored
- By removing the barriers to services, the patterns of self-referral can be documented

Study Design

- A study of barrier-free patient self-referral to genetic professionals
 - Study Population: the employee population of a major urban medical center in Boston, Massachusetts.
- This study took place within the context of a yearlong employee family history project

Study Design

- All employees were encouraged to record their family health history
- Employees were then offered the opportunity to discuss genetic concerns raised by their self-gathered history with a genetics professional



<http://www.hhs.gov/familyhistory/>

Study Design

- The study design removed specific barriers to the utilization of genetic services including:
 1. The need for clearance from insurers
 2. The need for authorization from primary care providers
 3. Any financial disincentives

The Genetics Consultation

- Consultations consisted of a 20-30 minute session with a genetic counseling intern and either a certified genetic counselor or medical geneticist
- Sessions were structured to discuss the participant's family history, clarify the patient's concerns, and give disease-specific feedback and recommendations
- Participants were advised of the genetic basis of diagnosis, treatment, and prevention of disease

Results

- Over the 12-month period of the study, there were 29 self-referrals from a pool of 13,000 hospital employees (0.2%) to whom the service was available
- Participants who self referred universally sought to discuss diseases recorded in their family health history in an effort to optimize their own healthcare or that of their first-degree relatives
- Sessions consisted of discussion of diseases with both Mendelian and non-Mendelian inheritance patterns that were observed in these families

Results

- Participants cited a variety of concerns prompting consultation, more than one in many cases, including:
 - 35% of patients cited concern regarding a single gene disorders
 - Hereditary breast, ovarian, and colon cancers
 - Hemoglobinopathies
 - 91% of patients cited concern regarding common complex diseases
 - Heart disease
 - Diabetes
 - 17% of patients cited concern regarding other genetic concerns
 - Genetic risk assessment

Familial Cancer Syndromes

- From the pool of 29 participants, 6 (21%) met the criteria standards for further genetic analysis based upon personal and family histories
 - 4 patients met the *BRCAPro* criteria for Breast and Ovarian cancer testing (*BRCA1/2*)
 - 1 patient met the revised Bethesda criteria for HNPCC testing (*MLH1*, *MSH2* and *MSH6*)
 - 1 patient met the criteria for familial malignant melanoma testing (*CDK4*)

Results

- 41% (12/29) of patients reported that they had previously discussed their family history concerns with the primary care provider prior to the genetics consultation
- 35% (10/29) of patients reported that they were currently being monitored by their PCP
- 14% (4/29) reported that this monitoring was secondary to their family history

Conclusions

- The nature of the patient questions brought to geneticists in this study does not suggest an over-utilization or misuse of genetic services associated with the removal of the barriers to services

2D2. 2007 ACMG Presentation

Discussion

- Insurance carriers have set up barriers to the self-referral of patients to specialists
 - Many healthcare plans have made exceptions to these restrictions for certain specialists, such as dermatologists and obstetricians
- It is widely anticipated that applications for genetic information within the adult care arena will grow significantly over the next decade
- In such an environment, health insurance plans should examine the option of removing any prerequisites for referral to genetics services to facilitate and maximize patient self-referrals

Future Directions of Research

- Further work could examine patient satisfaction with the genetics self-referral
- Future work could focus on the optimal design for incorporating the consultative services of genetic professionals within healthcare delivery systems
- Such integration will depend on issues such as:
 1. The effect of self-referral to geneticists on consumer satisfaction
 2. The capacity of the genetics workforce to accommodate this kind of new demand
 3. The relevant genetics expertise of other medical professionals
 4. The impact of computer-based point of care decision support on primary care-patient interactions on questions of genetic risk

Genetic Consultation Sub-study [of the BWH Family History Project]

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