

ADDING A GENOMIC HEALTHCARE COMPONENT TO A HEALTH INFORMATION MANAGEMENT CURRICULUM

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Abstract

The inclusion of genomic information will become routine in electronic health records (EHRs). Educating health information management (HIM) students about how to best manage, protect, properly release, and use this information for patient care is of utmost importance.

This study examined the usefulness of incorporating genomic modules into an existing course in quality management. Pretest and posttest results showed that students improved in all areas related to genomics in healthcare. Also, students enjoyed the class scenarios and discussion on the ethical use of genomic information. Interspersing genomic information management throughout an existing quality management class is an effective way to add this information to an existing HIM curriculum.

Key words: genomic information, pretest-posttest study, quality management, education

Introduction

Hospitals go to great lengths to ensure that patients' health information is kept private, confidential, and secure, but are hospitals prepared to protect the next wave of health information—genomic information? Genomics is the study of an organism's genome. A person's genome is essentially a complete set of the person's hereditary information. The human genome was first mapped in 2003—the result of a decade-long international research effort called the Human Genome Project (HGP).¹ The HGP allowed scientists, for the first time, to obtain a complete genetic blueprint for a human, making it, according to the National Human Genome Research Institute, “one of the great feats of exploration in history.”²

Genomic health information is on track to become an integral part of many Americans' health records. In a few years, as it becomes cheaper and easier, sequencing a person's genome may become a routine hospital test. Storing genomic information in patients' medical records allows clinicians and researchers to explore new data in disease surveillance, genetic disorders, epidemiology, and even criminal investigations. Sequencing a person's DNA also allows clinicians to design personalized courses of treatment. For instance, in oncology treatment, clinicians can utilize genetic testing to determine how a patient's genes interact to accurately predict the patient's response to treatment, prognosis, and likelihood of recurrence or relapse. Genomic health information can also track instances of disease in families through multiple generations.

While the inclusion of genomic health information in electronic health records (EHRs) will be a tremendous asset in improving and personalizing healthcare services, it comes with some

potentially devastating risks. If a person's genomic information falls into the hands of someone with malicious intent, tremendous damage could ensue because the genome exposes an individual's most identifiable aspect—the person's DNA sequence. Once this information is made known, the safety of the patient and the quality of the patient's care may be severely compromised.

Unfortunately, genomic information theft victimizes more than just the patient. If one person's genomic information is revealed, information regarding the person's biological family is also at risk. Keeping genomic information private and secure is a responsibility that is shared in many areas across the healthcare setting.

One area of healthcare that is especially vital in preserving, storing, and protecting genomic information is health information management (HIM). HIM professionals are traditionally responsible for managing protected health information (PHI). As technology progresses, the role of HIM professionals is also evolving. EHRs are quickly replacing traditional paper records in many healthcare facilities. Many HIM professionals have found new career directions related to EHRs, including designing, implementing, and troubleshooting EHR systems and teaching staff in facilities and private practices how to use them. While knowing the technical dynamics of the EHR is important, HIM professionals must also know how to preserve and protect the complex data within these systems. As it becomes cheaper and easier to map an individual's genome in the coming years, HIM professionals will be at the forefront of managing genomic information electronically. Because human genome mapping is so new, it is fair to ask: *are current HIM professionals ready for the impending surge of genomic health information?*

The answer to this question may lie at the University of Pittsburgh, where a unique effort is being made in the HIM department within the School of Health and Rehabilitation Sciences (SHRS) to teach students—future HIM professionals—about genomic information and what it will mean for their profession. Teaching the students who will soon be at the forefront of managing complex genomic health information how to preserve and protect the quality and integrity of this information is the most logical way to combat the risks associated with storing complete human genomes in EHRs. With only a few years before storage of genetic health data becomes routine in hospitals, now is the crucial time for students to learn the complexity of genomic health data and how to secure it.

Background

In 2010, the University of Pittsburgh's HIM department was awarded a grant from the National Science Foundation (NSF) to integrate computational thinking into health science education. The goal of this initiative is to prepare future HIM professionals for the accumulation of amounts of data stored digitally in the EHRs of many Americans. According to the proposal, "the digitalization of all Americans' health records and the advancement of high throughput biomedical technologies will produce huge amounts of electronic health information. properly manage and make full use of these data will greatly benefit all Americans. Every American may have the opportunity to receive

more effective, efficient, and personalized health care."

In the proposal, several teaching approaches were suggested for implementing new computational thinking courses, including a module for HIM students on the management and organization of genetic health data. One approach suggested was peer instruction. The proposal explained: "We will recruit undergraduate students who performed well in the previous semester as teaching assistants. This approach will be good both for the students who will work as teaching assistants and their fellow students. The students in the class may easily access the teaching assistants and feel comfortable about asking questions."

The structure of the HIM program at the University of Pittsburgh allows little to no room to add a course to the curriculum. Instead, the decision was made to add a four-part module on genomics to an established HIM course. The most appropriate course to accommodate this module was the HIM Quality Management course and lab. When the first author of this article, a HIM senior student, was awarded the Chancellor's Undergraduate Teaching Fellowship at the University of Pittsburgh, this student was chosen to serve as the teaching assistant for the course and teach the genomic module.

Objectives

To adequately prepare the HIM students for the future of genomic healthcare, several objectives were established for the student instructor:

1. Research current methods and examine the possible alternatives to discover the newest, most effective, and safest ways to store genomic health information in an EHR.
2. Design a module on the effective electronic management of genomic health information to be incorporated into the HIM quality management course. The lesson will be designed with the intent of becoming a core part of the quality management course for future classes.
3. Educate the HIM students so that, upon graduation, they will be prepared to fill the need for the safe and effective management of genomic health information.
4. Gain personal experience and achievement through teaching students a practical and useful application that they may utilize in their future careers.

Furthermore, several objectives were established for students in the module itself:

1. Explain the fundamental structure of DNA and eukaryotic gene structure.
2. Explain the central dogma of molecular biology.
3. Demonstrate the skills of performing searches on several molecular databases.
4. Describe the different types of molecular databases and explain their advantages and disadvantages.
5. Explain the importance of protecting personal genomic information.

6. Understand the importance of preserving the integrity of genomic information to prevent genetic discrimination.

Literature Review

It is interesting to note that in 2005–2006, most electronic medical records (EMRs) were not capable of managing genomic information. Many EMRs could not store a genome physically, nor were they capable of reflecting the biological knowledge of a genome.³ Hoffman stated that in order for the genome-enabled medical record to become a reality, three developments needed to take place:

1. The development of tools to support the capture of genomic results,
2. The development of a genomic-appropriate controlled vocabulary, and
3. The development of clinical decision support applications for genomic findings.⁴

Hoffman also stated that the ideal genome-enabled medical record would have a part for the inclusion of a structured family history (such as a pedigree visualization tool) and a part for the easy access of reference material such as PubMed.⁵ Now, more than four years later, many of these changes have yet to take place. Like Hoffman, Belmont and McGuire agree that EHRs are not ready to store genomic information efficiently. In their article, they state that all the work going into genetic counseling and personalized medicine will be futile unless a uniform EHR is created that can capture this data.⁶ Again, like Hoffman, Belmont and McGuire make suggestions that would enable the EHR to be genome friendly.⁷ Belmont and McGuire also agree that for an EHR to be genome-enabled, a common vocabulary must be used to capture, archive, and retrieve genomic information from EHRs.⁸ Another point brought up by Belmont and McGuire that was first suggested by Hoffman is the evolution of clinical practice guidelines to support genomic information so that physicians will not be overwhelmed by the sheer volume of information surrounding just one genome.⁹ While Hoffman recognized the need for these clinical practice guidelines, Belmont and McGuire offered the solution of using algorithms that can compute an individual's risk (based on the result of genetic counseling) and then be applied to clinical decision support as needed.¹⁰ Belmont and McGuire feel that by using these algorithm-triggered clinical practice guidelines, physicians will be less overwhelmed. The October 2009 proposition by the U.S. Department of Health and Human Services to change the Health Insurance Portability and Accountability Act (HIPAA) privacy rule to reflect changes in the newly passed Genetic Information Nondiscrimination Act (GINA) is a major breakthrough.¹¹

Three of the proposed changes to HIPAA would do the following:

1. Make explicit that genetic information is considered to be PHI,
2. Prohibit health plans from using or disclosing genetic information for underwriting (reviewing a person's health status to determine insurance availability) purposes, and
3. Revise provisions relating to the notice of privacy practices for health plans that perform underwriting.¹²

Genomic information and GINA are becoming major players in the healthcare field. GINA was passed in 2008 and is already affecting major statutes like HIPAA, showing that protecting genomic health information is more important than ever. The Office of the National Coordinator for Health Information Technology is continuing to work on integrating genomic information into health IT systems, which is crucial to taking genomic-enabled IT to its fullest potential in healthcare.¹³ The main focuses of genomic information technology are storage capacity and interoperability. Feero et al. state that "if sequence data for an entire genome become part of the individual's medical record for life, health IT systems will need to be developed that can store enormous amounts of data securely and offer access to that data across that person's life span."¹⁴ IT systems must be designed not only to afford genomic information the same level of security as other parts of the health record but to also go above and beyond to protect genomic information because it is so different from other information stored within the record.¹⁵ One main feature that differentiates genomic information from other health information typically stored electronically is the sheer size of a genome. One genome requires a large amount of electronic storage space, and many people will require their EHR to contain more than one genome (for example, a cancer patient having a personal genome and a genome for a tumor).¹⁶ Like Hoffman and Belmont and McGuire, Feero et al. also believe that interpreting the genomic sequence for clinical decision support should be done through algorithms.¹⁷ However, Feero et al. state that there are currently no accepted mechanisms for developing or disseminating these algorithms in the United States. Pilot projects exploring the use of EHRs to support genomic research are also discussed by Feero et al. with special attention given to the ethical implications of such novel approaches to genomic research. Feero et al. go on to state that the numerous debates regarding genomics, personalized medicine, and health IT will continue to grow; therefore, HIM professionals should be at the center of these debates to give fully informed solutions.¹⁸ Feero et al stresses the importance of having HIM professionals well educated in the topic of genomic healthcare, and at the University of Pittsburgh's HIM department, the goal of graduating students who can fill these roles is underway.

Methods

All HIM students enrolled in the Quality Management course were introduced to the genomics module. Thirty-nine students were enrolled in the class and lab during the 2010 spring semester. To

teach the HIM students about genomics and personalized healthcare, the module was separated into two 2-hour lectures and two 2-hour labs. Before the lectures began, the students were also given a “mini-syllabus” ([Appendix A](#)) that outlined the module and listed several objectives (noted previously) that the students were expected to accomplish by the end of the module. Before the module was presented, the students were asked to take a survey that asked general questions to gauge the students' prior understanding of genomics and genetics.

Questions on the premodule survey included the following:

1. Prior to this lesson, how much college-level biology have you completed?
2. In any previous biology courses, how much time was devoted to learning about genetics and/or genomics?
3. Have you taken a course devoted solely to genetics and/or genomics?
4. How familiar would you say you are with genomics?
5. Are you at all familiar with the Human Genome Project?
6. Are you aware of the important relationship between genomics and HIM?

After the students completed the premodule survey, they were asked to complete a module pretest ([Appendix B](#)). The pretest asked content-specific questions to further gauge the students' understanding of the material before the module. The pretest consisted of seven questions used to generate a score and one opinion question. After the students took the survey and the pretest and received the syllabus, the first lecture was given. The lecture, presented by the student instructor, was roughly 90 minutes in length. The content of this lecture was developed by one of the authors (LZ) and modified by the student instructor (B.L.). This lecture provided an introduction to genomics. For most of the students, it served as a review of concepts they would have learned in an introductory college-level biology course. Topics covered were the central dogma of molecular biology, DNA replication, transcription, translation, and gene splicing. The purpose of this first lecture was to refresh the students' memory of important biological concepts before the implications of knowing genomic information were discussed in more depth in subsequent lectures.

The first genomic lab presented to the HIM students was dedicated solely to the genomic databases accessible through the National Center for Biotechnology Information (NCBI) Web site. The purposes of this lab were to

1. Display the physical structure of a protein,
2. Establish the physical layout of a genome sequence,
3. Demonstrate how accessible information on genes is to the common public, and
4. Increase students' awareness of computational genetic resources available to them.

The lab consisted of a 20-minute PowerPoint presentation followed by a one-hour lab. The PowerPoint presentation was designed by one of the faculty and authors (LZ) and modified by the

student instructor (B.L.). The lab portion was designed by the student instructor. The lab required students to access different databases available from NCBI based on a unique gene that was assigned to them and to capture the NCBI output.

The second lecture in the module addressed genomic ethics. It was intended to teach students about the potential risks that can occur if genomic information is mishandled. The lecture followed the following content outline:

1. The Human Genome Project
2. Quality and safety
3. Genomic ethics
4. Genetic law
5. Risk perception and health behavior
6. Scenarios

Some of the lab's content came from Vanderbilt University in Nashville, Tennessee. Mary Reeves, RHIA, director of medical information services at Vanderbilt, provided the student instructor with a tremendous amount of information on efforts currently underway at the university in the area of genomics and personalized healthcare. Currently, the most significant genomic initiative to come out of Vanderbilt is BioVU. BioVU is a DNA databank research repository, one of the largest of its kind. BioVU currently has around 75,000 DNA samples in its repository, each of which is linked to a deidentified medical record. Additional information provided by Reeves, including an example of an informed consent document as well as a comic showing how DNA sequencing will revolutionize drug therapy, was also used in the lecture.¹⁹

The students were very responsive to and engaged in the material. They were eager to voice their thoughts and opinions on different ethical situations that were discussed. The final lab session was a time for students to bring together the information they had learned in the first two lectures and the first lab. The students participated in a discussion where they broke into groups and were given a scenario based on an ethical dilemma surrounding genomics featured on the National Human Genome Research Institute Web site.²⁰ The group had to decide what they would do in the situation and present their decision to the class. For example, one scenario stated:

Alice Smith is a 75-year old woman with four adult daughters. Two years ago, she was treated for medullary thyroid cancer, a disease known to run in families. Alice dies. Now, Lucy, Alice's eldest daughter, has been diagnosed with medullary thyroid cancer. Lucy's physician tells her that she likely inherited the genetic alteration and encourages her to warn her siblings and her children. Lucy is very angry with her mother's physician for not warning her. She feels that her cancer could have been detected much earlier if she had known she was at increased risk. Lucy considers filing a lawsuit against the physician.

The students were asked several questions regarding this vignette, such as “Did Alice’s physician have a duty to warn Alice that medullary thyroid cancer runs in families and she might wish to notify her family of their increased risk?” The students considered the ethical implications of this situation and generally agreed that Alice’s doctor should have warned her, but they also considered that maybe the doctor did tell Alice and she did not tell her daughters. The scenarios had no “right or wrong” answers, but they were a great way to spark the students’ interest in the ethics of genomics.

The student instructor also used a game of “gene bingo” to test the students’ understanding of the module. The bingo questions were formulated by the student instructor based on the content of the previous lectures. Students had to indicate the answers on their gene bingo boards. For example, if the student instructor read, “A person’s agreement to participate in a medical procedure or a research project after he or she has obtained complete information about its possible risks and benefits,” the students would put a bingo chip on the “Informed Consent” square. This was a fun way for students to review the concepts they had learned in the previous sessions.

At the end of the session, the students were asked to participate in a postmodule test. This test had identical questions to the pretest with the exception of the opinion question. Instead of writing what questions they had before the module, the students were asked what they had enjoyed most and least about the lectures and labs.

Results

After the module had been completed, the effectiveness of the methods could be interpreted. First, the premodule survey was interpreted to gauge the students’ prior experience with genomics. The results of the survey indicated that most students had taken at least an introductory course on biology, with 81 percent of students reporting to have had one to two semesters of college-level biology. Most of these introductory biology courses featured lessons on genetics, but only 5 percent of students had taken a course devoted solely to genomics or genetics. Fifty-seven percent of students were aware of the Human Genome Project but were unsure of its exact purpose or benefit.

Only 22 percent of students said they were aware of a relationship between genomics and HIM. Most of these results were expected based on the education level of the students and the prerequisites for the HIM major. However, these results were very important since they provided key insights into the students’ preparatory knowledge and experiences in the area of genomics. This information enabled the authors to better define the genomics module so that the areas in which students were not well educated could be a focus and a major part of class lectures, discussions, and assignments. The survey also included an area where students could list any questions they had regarding the module. An overwhelming majority asked what learning about genomics had to do with HIM and how it applied to them and their future HIM careers.

Pretest Results

A total of 32 students completed both the pretest and posttest, so this number was used to compute the statistics since equal pairs were needed to run the statistical analysis.

The average score on the pretest was 58.0 percent. Students performed best on question 3, with 62.5 percent of students answering correctly. Students performed worst on question 1, with only 15.0 percent of students answering correctly. The results of the pretest showed that there was room for the students to improve their knowledge and understanding of genomics in healthcare.

Thirty-nine students completed the lab, and 72 percent earned 10 out of a possible 10 points. Twenty-eight percent earned 9 points out of 10, and no students scored less than 9. The average score was 9.72. The only question students lost points on was one that required them to go to PubMed and search their assigned gene, read an abstract, and then briefly summarize it. The students performed exceptionally well on all the other questions.

Posttest Results

On average, students improved overall from pretest to posttest with an average percent gain of 24 percent and very high statistical significance ($p < .001$). The student who did the poorest on the pretest (1 out of 7) improved to 6 out of 7 on the posttest—a 71 percent increase. Students improved across all questions but especially on question 1 (“Describe the ‘Central Dogma of Molecular Biology’ in your own words”), with a 33 percent increase from pretest to posttest. However, only questions 1, 4, 5, and 7 showed statistically significant improvements, with question 1 showing very high significance ($p < .001$) ([Table 1](#)).

As previously mentioned, the students were asked in the posttest to say what they liked and disliked about the module. Some of the results are as follows:

- Many students felt that the first lecture (a review of molecular biology) was repetitive of what they had already learned in high school and college-level biology.
- Many students enjoyed the “discussion” aspect of the lecture. They felt it was easier to retain information when they were actively engaged in the topic.
- Almost all the students commented that they liked the bingo game. They said it was a great way to reiterate key topics, it was fun, and it kept them interested.

Limitations

The biggest limitations associated with this project involve the pretest/posttest methodology. These limitations include the following:

- As discussed in the results section, the posttest exactly replicated many of the questions on

the pretest. While repeating the questions is an effective way of gauging how much information the students absorbed over the course of the module, it creates a limitation if the students are able to recall answers from memory. Although students were never explicitly given the answers after the pretest, all the material was discussed during the module. On the other hand, because the students were never explicitly given the answers to the pretest, if they chose a wrong answer initially and then recalled it from memory at the time of the posttest, hence choosing the wrong answer again, another limitation is created.

- The pretest and posttest were both designed with a majority of the questions in a multiple-choice format. While a very popular method of testing, the use of multiple-choice questions may create a limitation due to the fact that on each multiple-choice question, students have a one-in-four chance of getting the correct answer by random guessing.
- A limitation is created by the high ratio of the amount of material covered to the number of questions on the pretest and posttest. Because of time constraints, a lot of material (much of it that students had never encountered before) had to be covered in only four sessions. It would have been very difficult to include enough material in the pretest and posttest to appropriately reflect the amount of content covered within the module.

While limitations in studies such as this are unavoidable, they can be combated in various ways. For instance, in addition to multiple-choice questions, some open-ended questions were included in the tests. It can be argued that the answers to open-ended questions are harder to memorize because there they lack visual cues to jog the student's memory of the answer as exist with multiple-choice questions. It is also important to note that the pretest and posttest were not the only methods of evaluation. Questions pertaining to the module were also included on the students' Quality Management course final exam, which was given several weeks after the module was completed.

Recommendations and Conclusions

Introducing a lesson on genomics into the HIM curriculum for the spring 2010 semester was a trial-and-error exercise. If the decision is made to keep a lesson on genomics a permanent part of the quality management class and lab, several changes may need to be made to future lessons to educate the students more efficiently. Based on the responses from the posttest, removing the first lecture, a review of molecular biology, should be considered because an overwhelming majority of students felt that they did not need it and that it did not apply to them as HIM majors. Instead, that time could be used for students to learn more about new concepts, rather than review ones they have learned several times in the past. It is also important to consider that the students responded well to group discussion and hands-on activities. Of course students like to play games, but the score improvements from pretest to posttest suggest that these activities are actually very beneficial to their learning.

For associate-degree programs, tutorials instead of course modules on genomics could be

provided, as is done in an introductory course for junior-level students at the University of Pittsburgh. The tutorials should provide an introduction to or overview of genomics and gene structure, the quality of genomic data when incorporated into the EHR, and privacy and genomic information security.

A deeper approach to genomic education is offered by a new course associated with the NSF grant discussed above. In spring 2010, one of the authors of this article (L.Z.) offered a new course called Genomics and Personalized Health Care to undergraduate and graduate students in the SHRS at the University of Pittsburgh. Most of the students in this course had taken general biology courses, but only one of them had previously taken a genomics course. The course focused on general instruction on genomics, gene structure and annotation, gene and disease association, and genomic information security. Other topics such as RNA and protein structure, microarray experiments, and DNA sequencing technologies were also covered. Students developed a deeper understanding of gene structure and annotation by working on novel gene annotation projects. All students were required to give two presentations, one explaining a chosen disease at the gene level and the other defending their gene annotation approach and results. Graduate students in the class were also required to write a scholarly paper on selected topics relevant to the course. By the end of the course, students submitted the successful results of their gene annotation projects to a database. A postcourse survey showed that all students claimed to have better knowledge of genes, gene annotation, and genomic resources. After taking this course, students can apply appropriate data analysis tools to various genomic data sets. [Appendix C](#) provides the contents of this genomics course for undergraduate and graduate students.

No matter how the information is delivered to students, one thing is certain—it is imperative that HIM students learn the importance of genomic health information and its relationship to HIM. There is no going back to the days before the human genome was sequenced, so students must be prepared for a future of continuing debate, discovery, and innovation surrounding genomics. Genomic competencies should be incorporated into the HIM curriculum and may include the examples outlined in [Table 2](#). The application will differ across the associate, baccalaureate, and master's levels, but genomic competencies are a must in HIM education. Genomic information is used every day to treat and care for patients with many different diseases. HIM professionals are the managers of health-related information. Therefore, genomics should be applied across the HIM curriculum and incorporated not just in one course but in many as it applies to anatomy and physiology, medical terminology, quality management, privacy and release of information, security of data, and the analysis and proper management of data and databases. As Feero et al. state, HIM professionals will be at the center of genomic issues, and “fully informed solutions to these issues could yield tremendous benefits to individuals and society for decades to come.”²¹

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