Projecting the Impact of the Human Genome Project on the Practice of Medicine

A Senior Honors Thesis (HONRS499) in Fulfillment of the Requirements for Graduation from the Honors College

by

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In 1989 the National Institutes of Health and the Department of Energy undertook a project designed to map the human genome. Humans have 23 pairs of chromosomes upon which reside between 50,000 and 100,000 genes. (Cox, 1992; Hughes, 1991) These genes are responsible for each person's anatomical structure and function and for many behavioral traits as well. This enormous undertaking has employed biologists, biochemists, engineers, computer scientists, and mathematicians in a joint endeavor to map and sequence each gene and then store the data in computer databases. The importance of this information lies in "...providing new strategies to diagnose, treat, and possibly prevent human (sic. genetic) disease." (Anon, 1992) The knowledge gained in the past five years had already impacted the medical profession. Genes for Huntington's disease, cystic fibrosis, amyotrophic lateral sclerosis (ALS), myotonic dystrophy, and over 20 other severe genetic diseases have already been mapped. (Caskey, 1993; Caskey, 1993; Cox, 1992; Hughes, 1991; Rossiter, 1993) The new technique of gene insertion (genetic surgery) has already successfully treated children with severe combined immune deficiency. (Cox, 1993) New information concerning how genes function is creating alternate treatment strategies at an ever accelerating rate. The importance of understanding genetic disease is better recognized when one considers that genetic diseases of the heart and circulatory system, lungs, kidneys, and cancers are the major causes of death in the developed world. The most recent statistical data, published in 1994, lists genetic-based diseases as the cause of 74% of all deaths in the United States in 1992. Cardiovascular diseases accounted for 42%, malignancies accounted for 24%, chronic obstructive pulmonary diseases and allied conditions accounted for 4%, diabetes mellitus and chronic liver disease totaled to 3.4%, and congenital anomalies caused 0.56% of deaths in the United States in 1992. (U.S., 1994) As such, genetics is playing an ever increasing role in modern medical practice.
Although the goal of the Human Genome Project is to provide a map of the entire sequence of genes carried in the human body, this information will only be beneficial if it is put to use for the purpose of enhancing the quality of life. One major way this information can become functional is for the medical community to welcome this new knowledge as an advance in medicine and use it for the patient's benefit. Being able to determine a person's genetic disposition to disease may be useful in the prevention and/or treatment of disease. Although the genetic information regarding the human body is being compiled in the laboratory, the functionality of this information is yet to be determined. While preferable, physicians are not immediately required to maintain a high level of competency in new technology and information. It is possible that information in medical genetics is not being adequately presented to either physicians in training or currently practicing physicians. In addition, physicians may not be taking the time to utilize the information, assuming that it is available to them. Therefore, any and all knowledge collected through the Human Genome Project (HGP) does not necessarily translate into everyday medical practice. A questionnaire was developed to determine the extent to which the HGP is affecting, or will affect, the practice of general medicine.

**Research Goal**

The goal of this research is to assess physicians' knowledge and opinions on the impact of the Human Genome Project on the practice of medicine and then compare these perceptions to what is currently being addressed in the literature about the effect of this knowledge on the current and future practice of medicine.

**Statement of Hypotheses**

Data gathered from physicians and third-year medical residents in general practice were used to test the following hypotheses: We hypothesized that physicians would report a low level of exposure to genetics-related information and education and a low level of exposure to patients with genetic-based diseases. We felt that physicians would report that they do not yet feel the effects of the Human Genome
Project in medical practice, that the Human Genome Project will take a long time to incorporate into medical practice, and that the effects of the Human Genome Project on the practice of medicine will not be as profound as the literature states. Finally, we felt that physicians would report great potential for discrimination and great need for safeguards and confidentiality, which would concur with the literature.

For the purposes of this research, we operationally define *low-level exposure* as 20% or less of the total (patients or information), *long time* as 5 or more years, and *profound* as thorough and far-reaching.
Chapter II

LITERATURE

The research process began in April 1994, by initiating a thorough review of the literature pertaining to the HGP and medicine. Medline and Educational Resources Information Center (ERIC) databases were used to develop a list of the literature that made reference to the potential uses of the genome map in medical practice. From this search, sixteen articles were found that contributed relevant information.

Of the sixteen, seven articles are full-scale analyses of the Human Genome Project, contributed to major scientific journals such as the Journal of the American Medical Association (JAMA), the American Journal of Human Genetics, Oncology, and Trends in Biochemical Sciences, by such human genetics experts as Francis Collins, C. Thomas Caskey, Belinda J.F. Rossiter, and Leroy Hood. These articles provide a complete explanation of the structure of the human genome, the process by which genetic mapping takes place, the timeline of the Project, and the goals and implications of the Project. All of the authors insist that once a gene has been identified, mutation detection techniques can be used to diagnose disease. In most cases, it is speculated that this will be the basis of medical diagnosis in the not-so-distant future. In addition, knowledge of the gene will allow for easier analysis of the disease and invention of a cure, although diagnosis will inevitably precede cure, causing an ethical dilemma concerning the use of this knowledge before a treatment is available. (Caskey, 1993; Green, 1991; Guyer, 1993; Hoffman, 1994; Keleher, 1993; Rossiter, 1992; Yager, 1991)

Another seven articles, published in similarly impressive journals, including JAMA and Science, by such qualified authors as Victor McKusick and Renato Dulbecco, are much more focused and opinionated. These short articles generally begin by describing the purpose and scope of the Human Genome Project. The authors' focus on the success the HGP has already had in finding genes associated with ALS (Lou Gehrig's Disease), Fragile X Syndrome, Alzheimer's Disease, and some cancers, to name a few. The conclusion drawn by these articles is that the map of the human genome will provide a basis by which all genetic-based traits and
diseases can be screened for and modified or cured. The idea that medicine will move from diagnose-and-treat to predict-and-manage approach (Bergman, 1993), i.e., that medicine will become more focused on prevention than cure, is prominent throughout the articles. There is an acknowledgment of the risks involved, including ethical decisions about who should receive genetic information and under what circumstances they should receive the information. The authors of these articles seem to have a generally positive feeling that, with proper safeguards, the information gained through the HGP can and will provide great benefits to the practice of health care. (Caskey, 1993; Cox, 1992; Dulbecco, 1993; Hughes, 1991; McKusick, 1993; Rossiter, 1993; White, 1993)

Rhonda Bergman's (1993) article, Quantum Leaps, is a look at the health care delivery system over the next twenty years from the point of view of hospital administration, rather than medical practice. This view of the Human Genome Project offers a perspective of amazing new technologies which will revolutionize the practice of medicine. Besides focusing on a future of lower inpatient hospital usage and lower surgical rates, Bergman feels that technology, including the genome map, will increase the length of life and the need for more geriatric health professionals, provide cures for such diseases as cancer, AIDS, and Alzheimer's disease, and enable defective and damaged genes to simply be replaced.

The last article, written by George J. Annas (1992), a noted expert in legal medicine and bioethics, focuses on the ethics of the Human Genome Project. Annas raises, or rather, duplicates questions about when and to whom genetic screening tests should be available, what information should be given, and how the costs will be covered. The discussion covers all facets of the potential uses of the genome map, from new drug treatments to somatic cell therapy to germ-line therapy. Annas makes the point, as the other authors, that the implications of the HGP are impossible to estimate from present knowledge, but that proper care and understanding of the potential effects is necessary to ensure the medical benefits of a genome map outweigh the dangers of information abuse.

In summary, the literature found on the Human Genome Project and the practice
of medicine is mostly theoretical in nature, describing the background of the HGP and citing potential uses and dangers of use.
Delineation of Problem

It was decided that a topic that combined human genetics, bioethics, and medical practice was mutually beneficial to both Dr. Jon R. Hendrix, Professor of Biology at Ball State University and Director of the Human Genetics and Bioethics Education Laboratory (HGABEL), and Mr. Marc Pinchouck, Ball State University honors student and biology/premedicine major. The project offered Mr. Pinchouck the ability to assess how this information will influence his chosen profession and offered Dr. Hendrix data necessary to revise the human genetics course offered in the Department of Biology and to revise the education program for secondary school teachers so as to reflect new concepts in human genetics and their importance to human health. By assessing opinions on the ways genetic information may affect medical practice, the stated goals may be achieved. In addition, the information to be determined was not found in any previous literature.

Selection of Criterion Groups

The groups that were studied must have met the following set of criterion at selection:

1. Must be a currently practicing physician OR a physician in the final year of medical residency.
2. Must be practicing/training in one of the following medical specialties: Family Practice, General Internal Medicine, or Pediatrics.

Selection of Subjects for Study

Three independent groups, each representing one-third of the survey population, were sent questionnaires. The first group consisted of physicians affiliated with Ball Memorial Hospital in Muncie, IN. This group was chosen primarily for ease of access. In addition, the investigators hoped that response would be increased by the proximity and potential affiliation of these physicians with Ball State University. The second group consisted of physicians affiliated with Lutheran General Hospital in Park
Ridge, IL. This group was similarly chosen for ease of access. Mr. Pinchouck is a former Summer Research Extern of Lutheran General Hospital and, therefore, has access to physicians affiliated with this hospital. It was hoped that response would be increased by his affiliation with this hospital. Ten each of family practitioners, internists, and pediatricians were selected randomly from the house staff of each hospital, yielding 60 subjects. The third group consisted of physicians in the third year of residency training. This group was chosen to gain the perspective of physicians most recently trained in medicine and closest to individual practice. It was hypothesized that these individuals may have greater knowledge of medical genetics, as medical schools may be teaching more medical genetics at present than was taught before the initiation of the HGP. The group was selected by identifying residency program directors throughout the state of Indiana. This identification was facilitated by Dr. Fred L. Ficklin, Assistant Dean, Indiana University School of Medicine. The directors were sent packets and were asked to distribute the questionnaires to resident physicians meeting the selection criteria. Twenty questionnaires were sent to directors in each of the three primary care specialties, yielding 60 subjects.

**Questionnaire Development**

A researcher-designed questionnaire was used to obtain information from practicing physicians and third-year medical residents in the primary care specialties. The questionnaire was designed (1) to obtain information from respondents so that comparison groups could be formed and (2) to test the previously stated hypotheses. *Questionnaires: Design and Use*, by Berdie and Anderson, (1986) was consulted for the overall construct of the instrument.

The questionnaire was designed to eliminate confusion and human error, and to minimize response time. Therefore, items were designed to enable use of a computer scan sheet, the General Purpose NCS® Answer Sheet, form no. 4521. All responses could be filled in on the pre-coded answer sheet, with the exception of one item. This item, the respondent's medical school, could be written in the blank space.
at the top of the form. To ease any confusion, the number one (#1) was printed at the
top of the page and the item number one on the answer sheet was marked out by
creating an overlay with Aldus Pagemaker 4.2 on an Apple Macintosh IIIVX (Appendix
B). The answer sheets were then run through a GCC Technologies BLP Elite Laser
Printer while printing this overlay. The final version of the questionnaire consisted of
three double-sided pages with a total of 35 questions.

The first 11 items elicit information pertinent to the demographics of the
individual respondent. These items include information on gender, medical school,
years in practice, and education in genetics and ethics. The remaining 24 items ask
the respondent about personal experience with genetics in medical practice and
opinions on the future use of genetics in medical practice. Items #12-18 were
designed to quantify the physician’s exposure to and understanding of genetics in
medicine. Items #19-25 were designed to assess the physician’s opinion on the
impact the HGP has had and will have on the way he/she practices medicine. Items
#26-28 were designed to assess the physician’s opinion on changes in medical
financing in response to the HGP. Items #29-31 were designed to assess the
physician’s opinion on additional time he/she has spent or will spend with patients in
response to the knowledge gained via the HGP. In addition, it was requested that
respondents elaborate on their responses to items #32-35 in the blank space provided
immediately below the items. These items were designed to assess physicians
opinions on the ethical dilemmas surrounding the information compiled through the
HGP. Item #32 asks the physician to elaborate on discrimination that may or may not
be a product of the completion of the HGP. Items #33-35 asks the physician to
elaborate on confidentiality issues that may or may not arise as a result of the
completion of the HGP.

The questionnaire was checked for construct validity by two methods. Initial
checks were provided by Jon R. Hendrix, Ed.D. Additional construct validity was
checked by sending a pilot questionnaire to Patricia Bader, M.D, medical geneticist,
Joe C. Christian, M.D, Chairman of the Department of Medical and Molecular Genetics
at Indiana University School of Medicine, and David Weaver, M.D, Professor of
Medical Genetics at Indiana University School of Medicine. The final version of the questionnaire (Appendix C), as well as the cover letters (Appendix A) and instruction sheet (Appendix B), was written with Microsoft Works 4.0 and printed on an Apple Laserwriter from a Power Macintosh 7700. The questionnaires were duplicated on gray 8.5 X 11 in. paper by the Ball State University Copy Center. The instruction sheets were duplicated in the Ball State University Department of Biology office by Mrs. Nita Strickland.

**Questionnaire Dissemination and Response**

Techniques used to enhance response rate are as follows:

1. The questionnaire was designed to look as professional as possible. It was professionally printed on gray paper to avoid a homemade appearance. The investigators did make two errors which reduced the professional appearance. First, Item #16 was truncated mid-sentence in the editing process. Not only did this make the questionnaire less professional, it also invalidated the item. Second, the investigators called the form “questionnaire” rather than “survey” as suggested by Berdie and Anderson (1986).

2. Ball State University Human Genetics and Bioethics Education Laboratory stationery was used for all cover letters. Microsoft Works 4.0 allowed the investigators to individually address all cover letters and mailing labels. Letters were one page or less in length and were signed by both Mr. Pinchouck and Dr. Hendrix. These techniques were used to impress authority and personal attention to the project.

3. Stamped, self-addresses return envelopes were enclosed with each questionnaire packet.

4. Questionnaires were numerically coded to maintain confidentiality. The codes were used to remove respondents from the data base when the response was received. The cover letter expressed the intent to maintain confidentiality through this method.

5. Questionnaires were mailed on November 10, 1994. This time frame was chosen to allow completion of the questionnaire and enough time for physicians to
respond prior to the busy holiday season between Thanksgiving and the new year.

Initial Mailing

Questionnaires were mailed to the 120 subjects on November 10, 1994. Each physician received an individually addressed, 9 X 12 in. brown envelope containing the following items (in order): an individually addressed cover letter, instruction sheet, questionnaire, and answer sheet. (Appendices B,C,D,C, respectively) The answer sheet was pre-coded in the special codes section from #001 to #120. This enabled the investigators to record response and later initiate follow-up. In addition, a self-addressed, stamped, 9 X 12 in. brown envelope was folded in half and wrapped around the other materials such that the entire packet could be placed in the envelope. In order to access residents, packets were assembled as described above. The packets were placed together in a box, and a separate cover letter to the director of residents was placed on the top of the packets. The cover letter contained much of the same information as the physician cover letters, with the addition of the request that the director randomly distribute the packets to residents under his/her direction that met the criteria as described in Selection of Criterion Groups. A copy of the directors cover letter can be found in Appendix A.

Microsoft Works 4.0 was used to create and maintain a data base consisting of the subject mailing list, subject specialty, questionnaire return and follow-up dates.

From the initial mailing, 32 (26.67%) of the 120 questionnaires were returned. Of those returned, 14 were from currently practicing physicians and 18 were from current residents.

Follow-up Mailings

On December 14, 1995, a follow-up postcard, featuring a cartoon elephant asking if the physician remembered the survey, was sent out to all currently practicing physicians who had not yet responded to the initial mailing. The postcard could not be sent to residents, as the identities of those individuals were unknown. The postcard can be found in Appendix D. The postcard was used 1) as a reminder that the
questionnaire should be returned and 2) to offer the opportunity to call Dr. Hendrix's office toll-free to request another copy of the materials. From this mailing, one request for additional materials was received. The materials were mailed and later received completed from the subject. One additional questionnaire was also returned completed. In a final act of desperation, the original packets that were sent out to the Lutheran General group were reconstructed and mailed to a contact at the hospital. The contact then distributed the packets to the mailboxes in the physician's mailroom. This approach was taken to decrease the possibility of a member of a physician's staff intercepting and throwing away the questionnaire. From this mailing, three additional questionnaires were returned, two completed and one incomplete, with the explanation that the material was out of the physician's realm of expertise. The investigators found this response somewhat startling, as it would seem that a physician with no knowledge whatsoever of the Human Genome Project or medical genetics would be at a serious disadvantage. One of the completed questionnaires was disqualified for not meeting the selection criteria. At this point, the investigators decided that every attempt had been made to enhance response, and the 35 completed questionnaires (29.17%) would be the final data, however insufficient.

**Analysis of Data**

Once the final questionnaires had been received, the answer sheets were taken to Scanning and Data Entry at Ball State University. A frequency scan was run to determine the counts and percents of each response. The answer sheets were then divided into Ball Memorial Hospital, Lutheran General Hospital, and Residents categories and scanned again to determine the counts and percents of each response by population. The answer sheets were once again divided into those which were marked A, B, or C for item #4, corresponding to family practice, general internal medicine, or pediatrics, respectively. These data were organized in a Microsoft Works spreadsheet in both numerical and percentage form and used to draw conclusions about the individual groups and the relationships between groups.

The data from each individual was entered into the Ball State University
VAX/VMS computer system as a text file and analyzed using SPSS 4.1 statistical software. Terry Schurr, Ph.D., Assistant Director of Research Computing and Professor of Educational Psychology, completed this project for the investigators. After converting alpha responses (A,B,C,D,E) to numerical responses (1,2,3,4,5), SPSS was used to determine the mean response for each item for the total group, for the family practice (F) group, and for the combined internal medicine and pediatrics (IP) groups. These groups were combined at the recommendation of Dr. Schurr, who felt the low response rate could be best adjusted for in this manner. The IP group yielded a quantity of responses which could be compared to the quantity of responses of the F group. The statistical analysis also provided 2-tail probability for the pooled variance and the separate variance, giving the investigators a value by which a comparison of the differences between the groups could be considered significant or otherwise.
In summary of the demographic background of the respondents, 26 males and 9 females responded to the survey. Seventeen (48.6%) were currently practicing physicians and 18 (51.4%) were currently finishing residency. Fifteen respondents (42.9%) claimed family practice, 12 (34.3%) were general internal medicine, and 8 (22.9%) indicated pediatrics as their current medical specialty. The average years in practice ranged from 1 to 6 years from the internal medicine and pediatrics (IP) group, but the family practice (F) group averaged 4 to 10 years in practice. These data do, however, include years in residency, which inevitably weigh the data toward fewer years of experience. The IP group claimed to be seeing, on average, 11 to 20 patients per day, while the F group averaged closer to 21 to 30 patients per day. Nearly half of the respondents work in private practice and the other half are members of a medical group. This information was obtained through items #2-7. The t-tests of the data for these items show significance between the IP group and the F group, as would be expected for demographic data. Only 1 additional item was found to have a significant difference between groups; therefore, it is assumed that the groups did not differ unless otherwise stated.

Items #8-11 were intended to clarify the genetics and ethics background of each respondent. As expected, the average number of undergraduate genetics courses taken was 1, although 28.6% of the respondents showed no undergraduate training in genetics and 20.0% responded 2 or more courses taken in genetics. The data are nearly identical for medical school courses taken in genetics. Four surveys (11.4%) were marked "0" and 6 surveys (17.1%) were marked "2" with all the rest taking 1 course. The investigators wondered whether genetics education has increased in medical school since the initiation of the Human Genome Project. The residents showed no greater amount of genetics courses than the current physicians, although this may be due to lag time between the beginning of the Project and the development
of such courses. The investigators also postulated that few physicians would report having taken an ethics class in medical school; 45.7% of the respondents reported taking no ethics classes, while another 45.7% reported taking only one class. Three respondents (8.6%) claimed to have taken two ethics courses while in medical school. Finally, when asked the question as to whether or not there is enough medical genetics taught in medical education today, there was a nearly equal division of “yes,” “no,” and “I do not know,” responses. The fact that one-third of respondents did not know if medical genetics is part of the curriculum leads us to believe that not enough is being taught, especially in light of the need to understand the information resulting from the HGP.

The remaining items, #12-35, focused on the hypotheses we hoped to test, as stated in Chapter I. The first group of questions, items #12-18, were designed to quantify the physician’s exposure to and understanding of genetics in medicine. On average, respondents reported that only 1 to 20% of their patients were being treated for a condition that is determined at least partly by genetic factors. Considering that the most recent statistical data, published in 1994, lists genetic-based diseases as the cause of 74% of all deaths in the United States in 1992 (U.S., 1994), it is possible that physicians are underestimating the number of patients with a genetic condition based upon limited understanding of the scope of genetics in medicine. Similarly, almost all of the physicians stated that between 0 and 10% of patients with genetic-based problems had an actual single-gene based genetic disease. As stated in the hypotheses, physicians are reporting a low level of exposure to patients with genetic-based diseases. Therefore, the hypothesis that physicians would report a low level of exposure to patients with genetic-based diseases is supported. This is our first confirmation that many physicians are not looking at disease on the level of a medical geneticist. Disease seems to be examined on a specific case by case basis, rather than looking at disease on the broad level of human health and genetic background.

We similarly hypothesized that physicians would report a low level of exposure to genetics-related information and education. More than half of the respondents
claimed that no more than 10% of the medical literature they have read in the past year has dealt with a genetics-related topic. Only one respondent claimed this figure is greater than 21%. Physicians overwhelmingly reported that no more than 10% of medical conferences, lectures, and rounds they have attended in the past year dealt with a genetics-related topic. Not surprisingly, physicians report that only between 0 and 20% of this genetics-related material will be helpful to their patients; although the internal medicine/pediatrics group (IP) averaged closer to the 0 to 10% range and the family practice (F) group averaged closer to the 11 to 20% range. This difference between the IP group and the F group may be explained by the nature of the family practitioner. As a physician interested in the health care of families, as well as individuals, the family practitioner may have greater interest in genetic factors in health care. The family practitioner seems to have a different frame of reference from the more specialized medical fields. We infer this frame of reference to be based upon a broader scope of medicine that includes both genetics and the environment. Physicians may need to adopt this holistic approach if medicine shifts from a “diagnose-and-treat approach to predict-and-manage” approach (Bergman, 1993).

These data on the quantity of genetics-related information and its applicability to patients show that these physicians are not receiving enough information about medical genetics and do not feel that most of their patients will benefit from the information gained through the HGP, supporting our hypotheses on these points.

Approximately two-thirds of the respondents chose medical literature as the main source of information on medical genetics. The remaining third was divided among continuing education programs, major conferences, mass media, and other resources. These results again support our ideas that physicians are not dealing with genetics information to the same extent that we believe physicians will need to, as a result of the Human Genome Project.

Items #19-25 were designed to assess the physician’s opinion on the impact the Human Genome Project has had and will have on the way he/she practices medicine. The responses to our inquiry about changes in methods of diagnosis due to
the HGP yielded some interesting data. Confirming our prediction, 16 physicians (45.7%) felt that their methods of diagnosis had not changed at all and 12 (34.3%) physicians felt they had changed only somewhat. However, we were surprised to find that 3 physicians (8.6%) felt that their methods of diagnosis had already changed quite a bit and 4 physicians (11.4%) had no opinion. The IP group reported a somewhat lower impact of the HGP on their current practice. This raises an interesting question of why family practitioners are finding methods of diagnosis changing sooner than their other general practice colleagues. Again, the researchers infer that since family practitioners are geared toward families and genetic inheritance is a factor in the medical care within a family, the family practitioner would take advantage of the new technologies produced by the HGP sooner than the internists or pediatricians. It may be beneficial for internists and pediatricians to shift to the frame of reference of family practitioners to take advantage of the knowledge gained by family history, inheritance patterns, and eventually, genome maps. The next two items asked to what extent the physicians thought their methods of diagnosis would change in the next five years, and in ten years, when the Human Genome Project is set for completion. The responses to each question gradually became more positive that change would occur. In the next five years category, only 2 physicians (5.7%) responded “not at all.” Fourteen (40%) responded “somewhat,” 10 (28.6%) responded “quite a bit,” and 5 (14.3%) responded “very much.” This shows that physicians are, for the most part, aware that changes in methods of diagnosis are very likely as the Human Genome Project continues to generate information. Additional support can be found in that not a single physician thought his/her methods of diagnosis would not change at all due to the completion of the HGP. Thirteen physicians (37.1%) thought methods of diagnosis would change somewhat, and 9 each (25.7% each) thought methods of diagnosis would change quite a bit and very much. Discouraging, however, is the fact that 4 physicians maintained “no opinion” throughout this entire line of questioning. This may indicate that some physicians are highly aware while others are unaware of the changes taking place in their field as a result of the knowledge gained through the HGP.
One of the issues brought up in discussions about the Human Genome Project is the ethical implications of being able to diagnose a genetic disease without the ability to cure the disease. When asked what they thought the typical lag time between the ability to diagnose and the ability to treat or cure would be, physicians responded cautiously. Only 14.3% thought this lag time would be less than 5 years and no physician thought this lag time would be less than 2 years. The mean response was between 5 and 10 years, although 25.7% thought the lag time would be greater than 10 years. The researchers infer from these data that physicians recognize that a lag time will occur and recognize the ethical dilemma that this lag time can produce. For example, when is testing for a disease with no cure appropriate and what ought be done with the information from a test for a disease with no cure? With the ability to test for such diseases as cystic fibrosis and Alzheimer's Disease, the need to address these issues is current and appropriate.

Items #23-25 followed a line of questioning to determine the extent to which physicians thought methods of treatment would be changed by the Human Genome Project. These three questions followed the same time frame as the questions about changes to methods of diagnosis: current, five years, and ten years. At the time of response, 65.7% of the physicians thought methods of treatment had not changed at all, 28.6% thought they had changed somewhat, and only 2.9% thought they had changed quite a bit. Another 2.9% of the respondents had no opinion. Item #23 was the only question that yielded a significant difference between the IP and the F groups [2-tail probability of the separate variance was 0.096]. Similar to the methods of diagnosis, the IP group reported almost no changes in the methods of treatment, while the F group reported much more often that some changes in methods of treatment are present in their practice. Once again supporting our idea that family practice physicians may be more in tune with the broader scope of medicine. In the next five years category, only 11.4% of physicians responded that no changes in methods of treatment would occur, 65.7% responded that methods of treatment would change somewhat, 11.4% responded “quite a bit,” and 5.7% responded “very much,” with an additional 5.7% responding “no opinion.” Finally, when asked about changes in
methods of treatment at the completion of the HGP, 2.9% thought no changes would occur, 57.1% thought some changes would occur, 20% thought quite a few changes would occur and another 20% thought many changes would occur. Surprisingly, no physicians marked “no opinion.” One reason may be that while some physicians do not currently have enough information to understand the implications of the HGP to their practice, they do understand that the HGP will have implications, and simply marked one of the options that assigns some effect on methods of treatment to the HGP. These data support our hypotheses that physicians would report that they do not yet feel the effects of the Human Genome Project on their medical practice, that the HGP will take a long time to incorporate into medical practice, and that the effects of the HGP will not be as profound as the literature states. The respondents to not seem to be in congruence with the literature in terms of the extent to which the HGP will affect medical practice. As stated by Guyer and Collins (1993), “Ultimately, the results of the HGP...will profoundly alter our approach to medical care, from treating disease that is already advanced to a preventative mode focused on identification of individual risk.”

Items #26-28 were designed to assess the physician’s opinion on changes in medical financing in response to the Human Genome Project. Three aspects of medical financing were covered: patient costs (including insurance premiums), medical practice expenses, and malpractice insurance costs. In terms of patient costs, 62.8% of physicians estimated that the HGP would change patient costs somewhat or quite a bit. Only 8.6% thought that patients costs would not change and an additional 8.6% thought patient costs would change very much. However, 20% of respondents had no opinion. Much of the literature equates information gained by the HGP with being able to anticipate disease and provide prophylactic care; i.e., doctors should be able to suggest measures to help prevent disease, eliminating the cost (both financial and physical) of treatment. Caskey (1993) remarked that “...appropriate use of preemptive care will benefit the patient and lower the cost of disease.” An average of 68.6% of physicians responding to the survey seemed to agree that costs would
change. However, due to the phrasing of the question, we do not know whether physicians expect costs to go up or down. This was a weakness in this survey item. However, 31.4% of physicians did not know whether or not costs would change or did not agree that costs would change, supporting our opinion that many physicians do not have enough knowledge of medical genetics and the Human Genome Project.

In terms of expenses that the physicians would themselves incur, physicians seemed to make an interesting relationship between the impact of the HGP on medical practice expenses and the impact of the HGP on patient costs. Eleven (31.4%) respondents thought patient costs would change moderately, while 15 (42.9%) thought that medical practice expenses would change moderately. In addition, 14 (38.0%) thought patient costs would change quite a bit or very much, but 11 (31.4%) thought medical practice expenses would change quite a bit or very much. However, the same number of physicians thought there would be no impact and one physician had no opinion. Therefore, of the respondents who felt a change would occur, more physicians thought the change in their own expenses due to the HGP would be moderate and the change to patient expenses would be changed quite a bit to very much. There does seem to be a logical correlation between medical practice expenses and patient costs, making this relationship in responses to the two items questionable. Why would patient costs change while medical expense costs would not? Also, the respondents were often in agreement that malpractice insurance costs would not change significantly. While quite a few (17.1%) still maintained no opinion, those physicians who thought malpractice insurance costs would change quite a bit or very much dropped from the previous expense items to 17.1% and those who thought no change or some change would occur rose dramatically to 65.8%. This item has some logic behind the results. Should the Human Genome Project increase the diagnostic capabilities of physicians and decrease the incidence of preventable disease, the HGP would, in effect, increase physicians competency in dealing with genetic-based diseases. A high degree of competency should have little or no effect in raising malpractice insurance costs, but the likelihood of costs actually lowering is probably very low. Therefore, the high percentage of physicians responding that
malpractice insurance costs will change very little is probably well founded. To augment the previous point, with malpractice changing very little and medical practice expenses changing only moderately, why would patient costs change greatly? This question should be addressed in a future survey.

Items # 29-31 were designed to assess the physicians opinion on additional time he/she has spent or will spend with patients in response to the knowledge gained via the Human Genome Project. We feel it is important that physicians not only understand the scientific background of a patient’s potential genetic problems, but also understand the human emotions that may surface in regards to these conditions. Patients will most likely have a need for support, empathy, and professional advice that must come from the health care provider that may be springing the potentially down-heartening or even devastating news. Bearing in mind the need for physicians to have an increasing role as genetic counselors, we probed to find out how much physicians agreed with this sentiment and to what extent they felt they were currently prepared to handle their patient’s genetic counseling needs. We were pleased to discover that the vast majority (82.9%) stated that they would need to act as genetic counselors somewhat to quite a bit. Only 1 physician (2.9%) responded that there would be no need for physicians to act as genetic counselors. Except for 1 physician without an opinion, the remaining 4 physicians (11.4%) responded that they would have a great need to act as genetic counselors. Nearly every respondent claimed there would be a need for physicians to act as genetic counselor. These data are surprising in comparison to other questions that show lesser understanding of the impact of the Human Genome Project on medicine. Although these physicians seem to understand the need for them to act as genetic counselors, and hopefully are willing to take on this role, 14.3% (1 family practice, 2 internal medicine, 2 pediatrics) claimed they were not at all prepared to handle their patients’ genetic counseling needs and 74.3% (11 family practice, 9 internal medicine, 6 pediatrics) claimed they were only somewhat prepared. The remaining 11.4% (3 family practice, 1 internal medicine) claimed to have quite a bit of preparation. These data may indicate that while
physicians are aware of the need for them to act as genetic counselors, the majority of them are currently unprepared and may need additional training or preparation in order to fulfill this role. These data also show that the family practitioners tend to be more attuned to the role that genetics plays in their practice. The questions that remain to be answered are: how to provide physicians with this training and how to convince physicians to take the time to learn, when the opportunity is offered.

Items # 32-35 were designed to assess physicians opinions on the ethical dilemmas surrounding the information compiled through the Human Genome Project. The issues we chose to address were discrimination and confidentiality. Hughes and Caskey (1991) make note that the application of DNA technology will be a challenge to patients, physicians, and insurance carriers, who will want to set premium rates based upon the use of genomic information as a “…pre-existing condition.” Rossiter and Caskey (1993) cite potential difficulties in using that genetic knowledge “…for unjustified discrimination or termination of pregnancies, and commercial pressures that might not be in the best interest of the individual.” We asked the respondents to think about the issues of discrimination in medical care, employment, and insurance, and decide whether or not they thought discrimination would be a factor in these areas. The respondents were asked to choose “yes” or “no” and then explain the response in a blank area directly below the question. Of the 68.6% of physicians who responded that complete genome mapping would lead to discrimination, many of them gave different explanations for their response. Some took the stance that insurance companies would simply use genomic information to increase insurance premiums to make money. These respondents took the “cynical” approach (note: cynical is a direct quote from one respondent) that discrimination would occur wherever it can and people will make money in any way possible. As one physician responded, “Insurance companies currently discriminate greatly in underwriting based on what is known about the health of the prospective insured. The capability to generate a genome map will only make it worse.” Some respondents even tried to justify this action as basic business and a completely natural reaction. One physician remarked,
"It will only be natural to want the strongest, healthiest, smartest, etc. persons working for you." The third and smallest group who agreed that discrimination would occur made the clarification that much genomic information will only indicate a tendency toward a particular disease. As long as insurance companies view this tendency as the likelihood that a disease will occur, discrimination is imminent. The alternative to this position was well stated by Hughes and Caskey (1991), who felt that patients with a predisposition could be "...targeted for customized care..." by physicians specializing in the area of the predisposition. Customized care would decrease an individual's chances of developing the disease and effectively lower costs. The 28.6% of physicians responding that the complete genome map would not lead to discrimination had similar ideas. Some of these respondents felt that the information should be used as a platform for intervention and correction of abnormalities. In this case, there would be no need for discrimination because the abnormality would be corrected. Others agreed that there is no need to discriminate on the basis of potential disease. One physician in this category felt that it would be discovered that environment has more effect than genes in the expression of traits. This continuing "nature vs. nurture" debate could have some effect on the extent to which society relies on genome data. If proven that genetics (nature) plays the major role in determining human predisposition to disease, the extent to which society relies on genome data would be enhanced. If proven that environment (nurture) plays the major role in determining human predisposition to disease, the extent to which society relies on genome data would be reduced. Since the roles that genetics and environment play in developing human characteristics are probably somewhere in the middle, the reliance on genome data in determining predisposition to disease will probably lie somewhere between complete and no reliance. Still others sought legislation to protect individuals from discrimination. In their explanation of the Americans with Disabilities Act of 1990 (ADA), Golden, Kilb, and Mayerson (1994) state that "...the term (disability) includes such conditions, diseases, and infections as...cerebral palsy, epilepsy, muscular dystrophy, multiple sclerosis,...cancer, heart disease, diabetes, mental retardation, emotional illness, specific learning disabilities,...and alcoholism."
All of these conditions are known to have at least some heritability and genetic influence. In the event that a genome map can be constructed to show an individual’s predisposition to such genetic diseases, it seems that legislation such as ADA may already hold the key to protection from discrimination. A few respondents had ideas about discrimination that were not mentioned in the literature. One physician remarked that the issue would be irrelevant if pre-existing conditions clauses are removed by health care reform. Another physician, of the opinion that the ability to create an individual’s complete genome map is still distant future, mentioned that “...by the time we can routinely map everyone’s genome, discrimination will be a thing of the past.”

On the issue of confidentiality, physicians were first asked to respond whether they thought the medical profession would be able to keep genomic information confidential. We were surprised to find that 42.9% of the respondents thought the medical profession would be able to keep a person’s genomic map confidential. Most of these respondents agreed that patient privacy is of the utmost importance, that genomic information should be subject to the same regulations as any other information found in a patient’s medical chart, and that medical information should only be given out with patient consent. The 54.3% of physicians responding that the medical profession would not be able to keep genomic information confidential cited the age of electronic media as the culprit of this invasion of privacy. Many physicians felt that information would be able to be accessed, legally or otherwise, by insurance companies, third-party payors, employers, or the government. Many of them mentioned the word “leak” in reference to the ability of humans to keep information confidential. One physician omitted the item, yielding the incomplete percentage total. The physicians were also asked whether there were specific circumstances under which a person’s genomic map should and should not be kept confidential. Although there was high variability of response to the item about the ability to keep genomic information confidential, 91.4% responded that there are specific circumstances in which confidentiality should be maintained. Nearly every physician felt that under normal circumstances, every individual has the right to confidentiality. There is no
direct explanation from the two physicians who responded that there are no specific circumstances under which a person's genomic map should be kept confidential. A response of "no" to the item could be interpreted to mean that confidentiality should be maintained in every instance, i.e., there are no specific circumstances for confidentiality, rather than a person's genomic map should not be kept confidential. This was a weakness of the particular item. Of the five individuals who proofread the survey, no one realized that the question could be interpreted in this manner. Again, one respondent omitted the item. When asked if specific circumstances existed under which a person's genomic map should not be kept confidential, four respondents omitted the item. The remaining respondents were split 45.7% to 45.7% to either "yes" or "no". Those responding that no circumstances existed under which a person's genomic map should not be kept confidential maintained that confidentiality is crucial and that information should only be released with the patient's consent. The other half felt that genome information should not be held confidential in cases in which withholding such information would cause harm to others, usually by transmission of life-threatening diseases. The subsets (IP and F) were split on this item. This is probably due to one physician's belief that confidentiality should be absolute while another may view family issues as an overriding factor in confidentiality. Many of the respondents who said that confidentiality should not be an absolute commented that genetic information should be available to an individual's partner and children. Some respondents mentioned confidentiality should be overlooked in cases of extremely high risk of anti-social behavior, including tendency towards murder and rape. A few respondents noted that access to genomic information should be allowed in cases of rape; however, there was no mention of which person's genome and for what use. The data accumulated on discrimination and confidentiality seems to support our hypothesis that physicians would report great potential for discrimination and great need for safeguards of confidentiality.

In summary, the data gathered helped to support the original hypotheses that physicians would report: 1) a low-level of exposure to genetics-related information and
education and 2) a low level of exposure to patients with genetic-based diseases; 3) that they do not yet feel the effects of the HGP in their practice, 4) that the HGP will take a long time to incorporate into medical practice, and 5) that the effects of the HGP on medical practice will not be as profound as the literature states. The respondents also supported our ideas that they would report great potential for discrimination and great need for safeguards and confidentiality. Thus, all of our stated hypotheses were supported by the data.
Chapter V

COMMENTS AND RECOMMENDATIONS

The project is considered by the investigators to be a success. Although the data and conclusions generated by the project are not generalizable to the entire medical community, the information brings forth some interesting points that can, hopefully, be followed up in the future. Physicians did report a relatively low level of genetics education and exposure to genetics-related information. In the event that the Human Genome Project generates the ability to increase the use of genetic information in medical diagnosis and treatment, the medical community may find itself at a disadvantage without prior understanding of the new technology and of medical genetics in general. This limited knowledge of genetics may have resulted in the low numbers of patients the physicians reported seeing with genetic-based diseases. We should note, however, that the numbers could accurately reflect a high number of physical injuries and non-genetic related infections.

In terms of the incorporation of the HGP into medical practice, the physicians agreed that the information produced to date has resulted in little change, that incorporation of any information will take a long time, and that the effects of the HGP will not be as profound as the literature states. Is this sentiment due to the nature of the HGP itself, the foreign nature of genetics, or the common human reaction to resist change? In any case, should physicians choose to be more accepting of a potentially exciting and beneficial new world of medicine, the quality of life could be improved for millions of human beings.

On the final topic of ethical issues, we were pleased that physicians recognized the potential abuses of human genomic information and felt it is important to protect individuals from the misuse of personal information. As the senior author, I cannot agree that discrimination and confidentiality will not be relevant issues. Discrimination will continue to exist until the last prejudiced person on Earth refuses to teach that doctrine to his/her children. We must deal with discrimination as a real and present issue. One important way to protect individuals from discrimination is to prepare
guidelines for maintaining confidentiality. We cannot assume that information is confidential simply because it is listed in a patient's medical chart. Medical charts are no better protected than electronic media that can be accessed by a knowledgeable computer user. There must be a better way to protect this information from invasion. Once the information is protected, we must also determine who has the authority to access the information. Should a significant other or child be allowed to know the genetic background of the partner or parent? Under what circumstances? Only under the assumption that antisocial behavior has a genetic background could we say that an individual with a predisposition to murder or rape should lose the right to confidentiality; and even then, is it fair to stigmatize a person whose upbringing may have compensated for any genetic tendency towards violence? In cases of rape, do we need the genomic information of the person who was raped? I do not see how this information would be useful. Similarly, if a simple blood test can be used for DNA comparison in a rape case, there seems to be no need for the complete genome of an accused rapist. As some of the physician responses show, we must be very careful to protect individuals' rights by setting guidelines before the technology needed to map an individual is available. Otherwise, human beings will certainly be trampled upon in the excitement to use new technology invented in the wake of the HGP.

In terms of the project itself, several mistakes should have been corrected before dissemination of the survey. Obviously, item #16 should have been a complete question in order to make the item valid and to make the researchers seem more competent. Items #26-28 should have been more specific about the nature of the change in medical financing. Of physicians who thought changes in medical financing would occur due to the HGP, it would have been useful to know whether they thought the changes would be positive or negative. We know the respondents thought patient costs would change significantly and medical practice costs would change moderately, but we do not know whether those respondents thought costs would go up or down. Had we asked for this information, better inferences could have been made from the data. The items #34 and 35 should have been recognized as offering vague responses that were confusing and did not provide the information for which we were
searching. Especially with item #34, we should have been more specific as to the meaning we wanted to attribute to each response; either "yes" there are specific circumstances under which a person's genome map should be kept confidential, or "no" it is not necessary to keep a person's genome map confidential.

Besides the individual items that should be corrected, any future attempt at such a project might consider some of the following recommendations. Another survey might probe deeper into the specific reasons that family practitioners report, in general, a more positive attitude toward the Human Genome Project and its effect on the practice of medicine. The prediction that malpractice insurance costs will change very little and medical practice expenses will change only moderately, yet patient costs will change significantly due to the information gained through the HGP, would be another interesting point to question further. In addition, the questions posed throughout this chapter would be worthwhile material for a follow-up survey.

The only additional change in the dissemination of the survey that might have enhanced response would have been to hand deliver the packets to physicians' offices in Muncie. Other than the personal contact that delivering the packets would provide, we feel that every possible effort was made to promote response. It should be noted that the investigators were warned that the population we intended to study would not produce sufficient returns. We decided that if we were professional and sincere in our efforts, physicians would not refuse to return a survey. In light of the response we received, we admit to being wrong. However, it is a sad state of affairs when an educated group of people cannot take time out of their admittedly busy schedules to fill out a survey pertaining to their own opinions. I would venture to say that most of the same individuals would fight to protect government funding for scientific research, probably on the basis that humankind does not progress without research. As the primary researcher, I have learned many important lessons through the experience of this Senior Honors Project. I have learned much about the trials and tribulations of the pursuit of knowledge. I have enhanced my ability to think, to write, to edit, and to ask for help when a task is beyond my ability. But, most of all, I have learned that I should never be too busy to help someone who values and counts on
my opinion.


Appendix A

COVER LETTERS
November X, 1994

Dr. XXXXXXXX
XXXXXXXXXXX
XXXXXXXXXXX

Dear Dr. XXXXXXX:

In 1989 the National Institutes of Health and the Department of Energy undertook a project designed to map the human genome. This enormous undertaking has employed molecular geneticists, biologists, biochemists, engineers, computer scientists, and mathematicians in a joint endeavor to map and sequence each gene and then store the data in computer databases. The importance of this information lies in "...providing new strategies to diagnose, treat, and possibly prevent human (sic. genetic) disease." New information concerning how genes function is accumulating at an ever accelerating rate. Current medical literature suggests that the completion of the Human Genome Project will revolutionize medicine in terms of both diagnosis and treatment. However, the literature mentions nothing about the individual physician's readiness for this information, about their willingness to accept and incorporate this information into his/her practice, or about the ethical issues regarding use of this information.

Only you as a physician are able to provide the information needed for our study. While the mapped human genome is doubling nearly every 18 months, medical genetics remains a relatively new and underdeveloped part of the medical school curriculum. Dr. Francis Collins, Director of the Human Genome Project, commented during his address at the Indiana University Medical Center as the 1994 Beering Award recipient, "...every physician is going to have to learn to be a genetic counselor," with the increase in information from the Human Genome Project. We are attempting to assess how well the medical community is being updated on genetic advances and whether or not physicians are being given enough information about this rapidly evolving field.

We ask you, as a current practitioner of medicine, to complete the enclosed survey and return it to us within the next two weeks. For your convenience a stamped, addressed envelope is enclosed. The results of this survey will be used to assess physicians' opinions on the issues listed above and to develop an estimation of the actual effects the Human Genome Project will have on medical practice. A code number has been attached to each survey to assist in follow-up techniques. To maintain confidentiality, all coded data linking names to responses will be destroyed after sufficient response is received and all data will be reported as grouped data.

We thank you for your time and cooperation, and look forward to receiving your completed survey.

Sincerely yours,

Marc Pinchouck, Premedical Student

Jon R. Hendrix, Ed.D
Professor of Biology, Ball State University
Director of Human Genetics and Bioethics Education Laboratory

317-285-8840 or 285-8827 Muncie, Indiana 47306-0442
November X, 1994

Dr. XXXXXXX

Dear Dr. XXXXXXX:

This past summer I worked for Lutheran General, and some of your colleagues, as a Research Extern through the Department of Research and Education. Now I am doing some research on my own and I am in need of your help.

In 1989 the National Institutes of Health and the Department of Energy undertook a project designed to map the human genome. This enormous undertaking has employed molecular geneticists, biologists, biochemists, engineers, computer scientists, and mathematicians in a joint endeavor to map and sequence each gene and then store the data in computer databases. The importance of this information lies in "...providing new strategies to diagnose, treat, and possibly prevent human (sic. genetic) disease." New information concerning how genes function is accumulating at an ever accelerating rate. Current medical literature suggests that the completion of the Human Genome Project will revolutionize medicine in terms of both diagnosis and treatment. However, the literature mentions nothing about the individual physician's readiness for this information, about their willingness to accept and incorporate this information into his/her practice, or about the ethical issues regarding use of this information.

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Sincerely yours,

Marc Pinchouck, Premedical Student

Jon R. Hendrix, Ed.D
Professor of Biology, Ball State University
Director of Human Genetics and Bioethics Education Laboratory

317-285-8840 or 285-8827 Muncie, Indiana 47306-0442
Dear Dr. XXXXXXXXX:

With the assistance and direction of Dr. Fred L. Ficklin, Assistant Dean, Indiana University School of Medicine, we identified you as the residents Program Director at your medical institution.

While the mapped human genome is doubling nearly every 18 months, medical genetics remains a relatively new and underdeveloped part of the medical school curriculum. Dr. Francis Collins, Director of the Human Genome Project, commented during his address at the Indiana University Medical Center as the 1994 Beering Award recipient, "...every physician is going to have to learn to be a genetic counselor," with the increase in information from the Human Genome Project. We are attempting to assess how well the medical community is being updated on genetic advances and whether or not physicians are being provided with enough information about this rapidly evolving field.

Only well-educated, current physicians are able to provide the information needed for our study. We are requesting that you randomly distribute the enclosed questionnaire packets to XX residents under your direction who are final-year medical residents in general practice specialties, i.e., those physicians who are nearing individual medical practice in family practice, internal medicine, or pediatrics. Please be assured that we are guaranteeing full confidentiality and are merely requesting the assistance of those individuals. Feel free to examine any of the enclosed materials. We hope that you will find them satisfactory and our endeavor worthwhile. Should this be the case, please distribute the packets as you see fit and encourage those residents to complete and return the questionnaires in the postage-paid envelopes. If not, please be so kind as to return all materials to us, in order to help us keep costs down. We sincerely appreciate your time and thank you in advance for your assistance in this project. With luck, the results of this research will be made available to the medical community via publication in an Indiana medical journal.

Sincerely,

Marc J. Pinchouck, premedical student

Jon R. Hendrix, Ed.D
Professor of Biology, Ball State University
Director of Human Genetics and Bioethics Education Laboratory

317-285-8840 or 285-8827 Muncie, Indiana 47306-0442
Dear Doctor:

I would prefer to individually address you; however, the dissemination of this survey has been done through the director of your residency program.

In 1989 the National Institutes of Health and the Department of Energy undertook a project designed to map the human genome. This enormous undertaking has employed molecular geneticists, biologists, biochemists, engineers, computer scientists, and mathematicians in a joint endeavor to map and sequence each gene and then store the data in computer databases. The importance of this information lies in "...providing new strategies to diagnose, treat, and possibly prevent human (sic. genetic) disease." New information concerning how genes function is accumulating at an ever accelerating rate. Current medical literature suggests that the completion of the Human Genome Project will revolutionize medicine in terms of both diagnosis and treatment. However, the literature mentions nothing about the individual physician’s readiness for this information, about their willingness to accept and incorporate this information into his/her practice, or about the ethical issues regarding use of this information.

Only you as a physician are able to provide the information needed for our study. While the mapped human genome is doubling nearly every 18 months, medical genetics remains a relatively new and underdeveloped part of the medical school curriculum. Dr. Francis Collins, Director of the Human Genome Project, commented during his address at the Indiana University Medical Center as the 1994 Beering Award recipient, "...every physician is going to have to learn to be a genetic counselor," with the increase in information from the Human Genome Project. We are attempting to assess how well the medical community is being updated on genetic advances and whether or not physicians are being given enough information about this rapidly evolving field.

We ask you, as an educated medical resident, to complete the enclosed survey and return it to us within the next two weeks. For your convenience a stamped, addressed envelope is enclosed. The results of this survey will be used to assess physicians' opinions on the issues listed above and to develop an estimation of the actual effects the Human Genome Project will have on medical practice. A code number has been attached to each survey to assist in follow-up techniques. To maintain confidentiality, all coded data linking names to responses will be destroyed after sufficient response is received and all data will be reported as grouped data.

We thank you for your time and cooperation, and look forward to receiving your completed survey.

Sincerely yours,

Marc Pinchouck, Premedical Student

Jon R. Hendrix, Ed.D
Professor of Biology, Ball State University
Director of Human Genetics and Bioethics Education Laboratory
Appendix B

INSTRUCTION AND ANSWER SHEETS
Instructions for Completion of Questionnaire

Thank you for taking a moment to complete this questionnaire. The questionnaire contains 34 multiple choice and 1 response item. For item #1, medical school attended, please write your response at the top of page 1 of the answer sheet, above the line that reads “General Purpose - NCS - Answer Sheet”. For the multiple choice items #2-35, please use the bubble area of the same page. Fill in the circles completely with #2 pencil, making your marks as dark as possible. Please avoid making stray marks on the answer sheet and erase cleanly any answers you wish to change. Do not mark the areas titled Name, Sex, Grade, Birth Date, Identification Number, and Special Codes. Please elaborate on items #32 and #33-35 on the questionnaire form itself, not on the answer sheet. Should you need more space for your response, feel free to attach an additional sheet. Once you have completed all the items, simply return the questionnaire and the answer sheet in the enclosed return envelope.

Thank you once again for your assistance in this project.
Appendix C

FINAL QUESTIONNAIRE
Physicians' and Residents' Questionnaire

Demographics

1. Medical School attended (please write on the top line of the answer sheet, side one)

2. Are you a:
   A..... currently practicing physician
   B..... current resident physician

3. Gender
   A..... Male
   B..... Female

4. Current medical specialty:
   A..... Family Practice
   B..... General Internal Medicine
   C..... Pediatrics
   D..... Other

5. Years in practice (current residents - Years in residency)
   A..... 1 to 3 years
   B..... 4 to 6 years
   C..... 7 to 10 years
   D..... 11 to 20 years
   E..... >20 years

6. Patients per day
   A..... 1 to 10 patients
   B..... 11 to 20 patients
   C..... 21 to 30 patients
   D..... 31 to 40 patients
   E..... >40 patients

7. Do you work in: (current residents - Do you plan to work in:)
   A..... Private practice
   B..... Medical group
   C..... Other

8. Number of undergraduate college courses taken in genetics
   A..... 0
   B..... 1
   C..... 2
   D..... 3
   E..... 4+

1.
9. Number of medical school courses taken in genetics
   A..... 0
   B..... 1
   C..... 2
   D..... 3
   E..... 4+

10. Number of medical school courses taken in ethics
    A..... 0
    B..... 1
    C..... 2+

11. Do you think there is enough medical genetics taught in medical education today?
    A..... yes
    B..... no
    C..... I do not know

Further Questions

12. In a normal business day, approximately what percentage of patients do you treat that have their condition determined completely or in part by genetic factors, e.g. cystic fibrosis or diabetes mellitus?
    A..... None
    B..... 1 - 20%
    C..... 21 - 40%
    D..... 41 - 60%
    E..... >61%

13. Of patients with genetic-based problems, approximately what percentage are actual single-gene-based genetic diseases?
    A..... 0 - 10%
    B..... 11 - 15%
    C..... 16 - 20%
    D..... 21 - 25%
    E..... >25%

14. Of the medical literature you have read in the past year, approximately what percentage has dealt with a genetics related topic?
    A..... 0 - 10%
    B..... 11 - 20%
    C..... 21 - 40%
    D..... 41 - 60%
    E..... >60%
15. Of the medical conferences, lectures, and rounds you have attended in the past year, approximately what percentage have dealt with a genetics related topic?
   A..... 0 - 10%
   B..... 11 - 20%
   C..... 21 - 40%
   D..... 41 - 60%
   E..... >60%

16. Of the genetics-related material you have been exposed to in the past year, approximately what percentage do you think will be of use to you, as a physician?
   A..... 0 - 10%
   B..... 11 - 20%
   C..... 21 - 40%
   D..... 41 - 60%
   E..... >60%

17. Of the genetics-related material you have been exposed to in the past year, approximately what percentage do you think will be helpful to your patients?
   A..... 0 - 10%
   B..... 11 - 20%
   C..... 21 - 40%
   D..... 41 - 60%
   E..... >60%

18. What is the major resource from which you receive information about medical genetics?
   A..... medical literature
   B..... continuing education programs
   C..... major conferences
   D..... mass media
   E..... other

19. To what extent do you think your methods of diagnosis of genetic diseases has been changed by the Human Genome Project to date?
   A..... not at all
   B..... somewhat
   C..... quite a bit
   D..... very much
   E..... no opinion

20. To what extent do you think your methods of diagnosis of genetic diseases will be changed in the next five years, due to information gained by the Human Genome Project?
   A..... not at all
   B..... somewhat
   C..... quite a bit
   D..... very much
   E..... no opinion
21. To what extent do you think your **methods of diagnosis** of genetic diseases will be changed by the completion of the Human Genome Project in 2005?

A..... not at all  
B..... somewhat  
C..... quite a bit  
D..... very much  
E..... no opinion  

22. What do you think the lag time period will be between the ability to diagnose vs. the ability to treat or cure genetic diseases as a result of the information gained in the Human Genome Project?

A..... 1/2 to 1 year  
B..... 2 to 4 years  
C..... 5 to 7 years  
D..... 8 to 10 years  
E..... > 10 years  

23. To what extent do you think your **methods of treatment** of genetic diseases have been changed by the Human Genome Project to date?

A..... not at all  
B..... somewhat  
C..... quite a bit  
D..... very much  
E..... no opinion  

24. To what extent do you think your **methods of treatment** of genetic diseases will be changed in the next five years, due to information gained by the Human Genome Project?

A..... not at all  
B..... somewhat  
C..... quite a bit  
D..... very much  
E..... no opinion  

25. To what extent do you think your **methods of treatment** of genetic diseases will be changed by the completion of the Human Genome Project in 2005?

A..... not at all  
B..... somewhat  
C..... quite a bit  
D..... very much  
E..... no opinion  

26. To what extent do you think that **patient costs**, including insurance premiums, will be changed as a result of the completion of the Human Genome Project?

A..... not at all  
B..... somewhat  
C..... quite a bit  
D..... very much  
E..... no opinion
27. To what extent do you think that medical practice expenses will be changed as a result of the completion of the Human Genome Project?
   A ..... not at all
   B ..... somewhat
   C ..... quite a bit
   D ..... very much
   E ..... no opinion

28. To what extent do you think that your malpractice insurance costs will be changed as a result of the completion of the Human Genome Project?
   A ..... not at all
   B ..... somewhat
   C ..... quite a bit
   D ..... very much
   E ..... no opinion

29. To what extent do you think physicians will need to act as genetic counselors as a result of the completion of the Human Genome Project?
   A ..... not at all
   B ..... somewhat
   C ..... quite a bit
   D ..... very much
   E ..... no opinion

30. To what extent do you think that you, personally, are prepared, or will be prepared, to handle your patients' genetic counseling needs?
   A ..... not at all
   B ..... somewhat
   C ..... quite a bit
   D ..... very much
   E ..... no opinion

31. To what extent do you think the time you spend with an average patient will be changed as a result of the completion of the Human Genome Project?
   A ..... Not at all
   B ..... I will spend more time
   C ..... I will spend less time
   D ..... no opinion
   continue to next page
For the following questions, please elaborate in the space provided on this form.

32. It has been suggested that obtaining a complete genome map of an individual will lead to discrimination. Do you think discrimination will play a role, once a person’s complete genome map can be constructed, especially in terms of medical care and factors related to medical care, such as employment, health insurance, and life insurance?
A..... yes
B..... no

Please explain your response to item 32 here

33. In relation to question #32, do you think the medical profession will be able to keep a person’s genome map confidential?
A..... yes
B..... no

34. Do you think there are specific circumstances under which a person’s genome map should be kept confidential?
A..... yes
B..... no

35. Do you think there are specific circumstances under which a person’s genome map should not be kept confidential?
A..... yes
B..... no

Please explain your responses to items 33-35 here

THANK YOU!
Appendix D

FOLLOW-UP MATERIALS
Did You Forget?

Remember what it was like when you were getting started? Without your help, I may not be able to graduate! Please complete and return the questionnaire I sent to you from Ball State University a few weeks ago. If you have misplaced your copy, please call 1-800-537-9604 and I will send you another copy right away. Thank you for your assistance.

Marc Pinchouck
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