

Virtual Mentor

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ETHICS CASES

Genetic Profiling of Medical Students

Commentary by John Mahoney, MD

Dr. Shepherd is a medical school professor charged with incorporating pharmacogenomics and genetic testing into her medical genetics curriculum for first-year students. To personalize the experience, she partners with a direct-to-consumer genetics testing company, for which she has consulted, to develop a modified and discounted genome test that examines four genotypes for nondisease states, including genes involved in the metabolism of certain macronutrients, medications, and alcohol. With approval from the medical school administration, the cost of the test is rolled into each student's tuition and notices are sent to all incoming medical students informing them of the study and requesting their informed consent. Once consent forms are received, the testing organization will solicit the specimens. The deidentified reports will be filed with the medical school's genetics department and made available to the participating students for reference and discussion during their medical genetics course.

Several weeks after the notices are sent out, Dr. Shepherd receives an e-mail from Lacy, a newly accepted student who is finishing a master's in genomic sciences. Lacy writes that, though she lauds the intention behind the project, she has objections to its implementation. She worries that medical students may feel pressured into participating in the project for fear of adverse academic consequences. She voices concerns about discovering and revealing genetic information, even if the information is relatively benign, and especially without the appropriate counseling. She is particularly concerned with the lack of clarity about what other student information might be collected and how privacy will be protected. She ends her communication by saying that she will probably participate if she can get her questions answered but worries that other incoming students may not fully appreciate the implications of the project and may not feel comfortable obtaining appropriate information or abstaining.

Commentary

This case describes a plan by a creative professor to stimulate student interest in studying genetics by using results from students' own specimens for analysis and discussion. A thoughtful student raises concerns about consent, coercion, and privacy. This commentary addresses the nature of the genetic profiling tests and ethical considerations for the instructor and school and identifies some unanswered questions about genetic screening.

Genetic Profiling Tests

The testing proposed here is a subset of the typical personal genomic testing that is marketed to consumers. Unlike clinical testing for a specific monogenetic disease, in this approach the tests typically result in profile information about the relative risk of developing a condition. The profile results are of limited clinical utility, particularly if they are interpreted without a correlation to a patient's overall health and medical history and if they yield a relative risk that is indistinguishable from that of the general population [1]. Consumers are not usually given in-depth, personalized pre- and posttest counseling or interpretation assistance as they would be when working with medical geneticists.

There are a range of motives and justifications for genetic profile testing. Proponents can reasonably argue that any information about current health or future diseases could potentially be useful, particularly if it can be obtained noninvasively and at modest cost. Businesses that sell testing kits or services have been effective in marketing them to the general public, but the benefit of such testing is nowhere near as clear as that of diagnostic testing in the clinical setting.

Ethical Considerations

The ethical concerns in the scenario include loss of privacy, an increased risk of future harm, coercion to consent, testing without counseling, and the consequences of how students are billed for the testing. Among these, it is useful to organize them in terms of their magnitude, related to the consequences of the worst possible or likely outcome.

Loss of privacy. Using this approach, a student's privacy might be regarded as being at risk of real and lasting harm. Today's world is replete with scientific discoveries, but also with security breaches, malevolent hackers, cyberattacks, and industrial espionage. Even without an overt breach of security, some number of staff at the testing company will have access to the students' results. These factors, alone or in combination, could counteract the measures taken to protect the students, which compels us to consider the possible impact.

The consequences of a privacy breach fall along a spectrum. At minimum, it invalidates the trust placed in the professor, university, and testing company. Although the proposed profile will not test for disease states, a student could be identified as being destined to develop a significant metabolic condition (the case mentions alcohol metabolism, for example). Thus, loss of confidentiality could place the student at risk for a gamut of discriminatory outcomes or stigmatization, including employment and insurance discrimination. Legal statutes are in place to help prevent this type of discrimination, but statutes cannot shield a person from all possible harms [2].

New gene-disease associations are being discovered continually, so a gene or sequence that is now thought to be inconsequential may in future be found to be diagnostic or predictive [1]. There is risk, therefore, that a student may ultimately be

confronted with genetic information that he or she had not chosen to know. The psychological impacts of such possibilities, including anxiety about how a result could affect career and family, adds to the stress that medical students already face.

Possible future knowledge of harm. Medical education often includes learning activities in which students' bodies are involved—students often practice physical examination skills on each other, examine their own blood or urine, or, as ultrasound instruction is introduced, practice on each other. Any of these activities could reveal a significant abnormality, such as a previously undetected blood dyscrasia or a congenital renal malformation. One difference between these activities and performing genomic screening testing is that the genomic testing generates a permanent third-party record that may later affect the student. The anonymity of the testing leaves students in the dark about what information is recorded about them and opens the door for anxiety about the unknown. While a basic science professor might perceive this as a minor and dismissible concern, it may not be trivial for a student.

For the school and professor, there are ethical implications to gathering this data while not being in a position to readily share any vital findings, now or in the future. The implications of what we may be able to do with this information could be far-reaching. An uncertain and changing future should at least be anticipated and consideration given to protecting the students from future harm.

For example, in this case, the professor believes the tests are for nondisease states. Interpretations of genetic testing results are already being revised as new gene-disease associations are discovered; in the not-too-distant future, one of these patterns may be found to be inextricably linked to a serious disease condition [3]. In a typical clinical practice, a geneticist might become aware of a new gene association, prompting a review of existing data and records. If this review identifies a patient result on file with the newly-significant finding, the practice contacts the patient.

Such reinterpretation could be done with data already used by the professor without the original specimens or costly reprocessing. A curious professor might choose on his or her own to review the data on hand to see if it revealed a profile with the newly significant finding—but it is not clear whether the professor or school has the same obligation as a clinical practice to notify if a significant abnormality surfaces.

An additional dilemma is how to communicate with the person with the abnormal result, since the professor does not have individually identified results. Is there an ethical or moral obligation to contact all students to advise them that they *may* be at specific risk and should proceed to be individually tested? This would appear to be desirable, but it would be fair to ask if it is realistic. At a minimum, this type of situation should be anticipated and plans made for handling it, which should be explained in the informed consent process. For example, if the university and professor decide that they will not undertake profile reinterpretation even as clinical knowledge evolves, this should be disclosed to the students.

That the professor and the entire class will have the set of data from the student testing raises another concern. It is possible that future discoveries will reveal new linkages between this genomic data and physical or ethnic characteristics (for example, between “macronutrient gene 1,” eye color, and ethnic background). These linkages may be sufficient to identify individuals, thus breaking the confidentiality that had been promised [3].

Coercion to consent. Lacy is justifiably concerned about being coerced to consent. Students generally understand the preciousness of their place in the medical school class and may perceive that their success is dependent on the goodwill of those with power (the professor and school administration). Given that students are totally dependent upon their professors and administrators to succeed in medical school, concern over the impact of declining to participate is entirely reasonable.

With this perceived or real vulnerability and significant power differential, substantial safeguards should be in place to prevent the professor and school administration from knowing who has opted out. Under the circumstances, there is no opportunity for students to ask consent-related questions, let alone do so in a safe environment. These young students are vulnerable to feeling pressured to participate along with the group, perhaps more so than would a group of experienced physicians engaging in similar coursework as part of a continuing education program. One might wonder if the school would even attempt to push faculty into this type of activity, in contrast to taking student participation for granted and giving only minor attention to a consent process.

Absence of counseling. Beyond coercion concerns, the consent process falls far short of accepted contemporary practices for counseling prior to genetic testing. In clinical genetics practices, extensive counseling provides patient with a solid foundation for making thoughtful and well-informed consent decisions [1]. In this case, this in-depth counseling is unavailable and impractical. The students are being treated like consumers who have volunteered for testing. Yet these students are being strongly encouraged to be tested by medical professionals at a medical school—entirely unlike consumers.

The lack of appropriate counseling may suffice to make proceeding with genomic testing unethical. An alternate viewpoint that the professor might voice is that, since no personal results will be reported to the student, there is no need for counseling of any sort—no risk of personal adverse findings, so no risk of adverse psychological or other impact.

Financial matters. An additional concern raised in this case is that students are being compelled to pay for the testing as part of their tuition. It is not entirely clear if there has been a disclosure to the students that they will be paying for testing for which they may elect not to consent. Including the fee in the students’ bills without awaiting their consent communicates the professor’s and school’s overall attitude—

they seem to have together decided that the students will participate and will pay and that the consent from individual students is a mere formality that can be taken for granted.

This approach might be more acceptable if it did not involve medical testing. The nature of the testing significantly changes the degree to which students must have an opportunity to exercise autonomous decision making, and the school must be sure it is acting in the students' best interests. Alternatively, if students were truly given the option of choosing to be tested, and only paying if they were being tested, the students' views on testing might be influenced. Having to pay an additional, optional fee might cause some students to opt out for financial reasons and others to feel more invested in the activity than if it were free. An optional fee might also cause them to think more seriously about the testing before consenting to participate.

Advice

This case sheds light on a broad range of ethical considerations associated with genomic profiling testing, and aspects of the power differential between educators and their students. Though the professor is probably attempting to provide a well-intentioned stimulus to learning, the potential negative consequences are many and the educational benefits are unclear. An educator might ask why this professor should bother with testing this class of students if the results are anonymous. It is not entirely clear whether it will really enhance learning. As proposed, students must pay for the tests, and there is the possibility that the tests could produce data that will cause alarm or harm, as described above. Yet no single party benefits from the testing, except perhaps the testing company. Considering that the professor has a prior consulting relationship with the testing company, there is the possibility that the professor has a conflict of interest that should be disclosed or avoided altogether [4]. If I were mentoring the professor, I would give advice on several aspects of the plan.

In general, I would urge Dr. Shepherd to pause and reconsider the proposed plan from a student's point of view. I would guide her to seek an alternative approach that does not place the students in an untenable position or that employs confidential, sensitive data when a safer alternative is available. The most basic alternative is to use existing data rather than test the students.

If testing were to proceed, the professor should arrange to work with a testing company with which she has no financial connection, rather than one she served as a consultant. Alternatively, she should provide the medical school and participating students with a clear disclosure of the possible conflict of interest. A process must be arranged in which the students have unimpeded access to thorough pre- and posttest counseling. The consent process must permit students to accept or decline testing without any possible reprisal for choosing to not be tested. This should include preventing faculty who may be grading the students from having knowledge of whether they consented to testing. To provide all possible security for their information, the professor and testing company should use anonymous sample submission and results retrieval, comparable to the way some HIV testing programs

are conducted. The professor and medical school should provide students with information about how they will communicate with students if there are relevant discoveries in the future.

If I were speaking with Lacy, my advice would include referring her to the dean of students or another student advocate for assistance with a tactful method of declining to participate. Students in this case may face a no-win scenario and be forced to choose which way they wish to lose.

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