

Overriding Considerations

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The Case

Mrs. G visited her obstetrician for first trimester routine prenatal care. The obstetrician offered genetic testing for a variety of conditions, including Tay-Sachs and Canavan's diseases, since both Mrs. G and her husband, a healthy 35-year-old physician, were of Ashkenazi Jewish descent. Mrs. G consented to be tested and told the obstetrician that she would discuss with her husband that evening whether he wanted to have the genetic tests. The obstetrician gave Mrs. G consent forms and information to take home to her husband. The obstetrician also entered laboratory test orders in the computerized order entry system for the genetic screening panel for both Mrs. G, since she had consented to be tested, and for Dr. G, assuming he would consent to be tested. However, the obstetrician did not mention this to Mrs. G.

At home that evening, Dr. G reviewed the materials and told his wife that he definitely did not want to be tested. Several weeks later, Dr. G visited his primary care doctor for a check-up. The physician ordered routine screening laboratory tests (fasting lipid profile, complete blood count, and urinalysis) through the computerized order entry system, and the next morning Dr. G presented to the laboratory for testing. The laboratory and its computer system were the same as used by Mrs. G's obstetrician.

Unbeknownst to Dr. G, the phlebotomist drew samples not only for the routine testing ordered by his primary care doctor but also for the genetic screening, because it was listed in the computer even though Dr. G had not consented. In doing so, the phlebotomist overrode the computerized alert that prompted him to be sure the patient had consented; he assumed that the physician must have obtained consent before ordering the tests. Ten days later, the obstetrician called Mrs. G to give her "the good news" that all her screening tests were normal. The obstetrician mentioned incidentally that her husband tested positive as a carrier of Canavan's disease. This disclosure caused some distress, but no physical harm. No increased level of care was needed.

The Commentary

Some health care providers will think Dr. G's refusal to have genetic tests for serious disorders was irrational. There are, however, a number of reasons why Dr. G's decision might be rational. His decision

reveals some of the complexities of testing, particularly for individuals who belong to groups that are at increased risk of possessing certain disease-related mutations.

First, without a significant family history of the disease, he may perceive the risk of having an affected fetus as too low to warrant testing. The couple's estimated risk is 1 in 3600 for Tay-Sachs and 1 in 6400 for Canavan's. [note that site calls it *Canavan disease*] Second, the utility of testing would be limited if the G's were Orthodox Jews (whose beliefs generally prohibit pregnancy termination, even if a fetus were found to be affected).

Third, Dr. G could have concluded there was no point in his being tested unless his wife was found to be a carrier. If Mrs. G was tested on her first prenatal visit, there would still be time after the result was returned for him to be tested. If he also was positive, the couple could then consider prenatal diagnosis. Since the chance of either of them being a carrier of Tay-Sachs is about 1 in 30 and of Canavan's about 1 in 40, the chance of their both being carriers is 1 in 900 and 1 in 1600, respectively. Couples in whom both partners are carriers have a 1 in 4 chance of having an affected fetus with each pregnancy. Hence, the overall risks mentioned above of 1 in 3600 and 1 in 6400, respectively.

Fourth, once Dr. G knows he is a carrier he may feel he has an ethical obligation to inform his brothers and sisters that they each have a 50% chance of being a carrier. Sharing of important genetic information can be problematic and have significant consequences within families.⁽¹⁾ Fifth, Dr. G may be concerned about the ramifications that a positive test might have on his access to, or costs of, health care insurance, either for himself or his child. In sum, Dr. G could have had any number of rational reasons for his decision. His decision thus highlights the the complexity and potential impact of carrier testing for an individual, and the need of all patients who are offered genetic testing to completely understand the risks, benefits, effectiveness, implications, and alternatives to testing and, based on that understanding, to consent to testing.

Thus, refusing informed consent for carrier testing and other types of genetic testing is not irrational. Unfortunately, many physicians take the matter of informed consent lightly. Giardiello and colleagues found that only 17% of patients who had a genetic test for familial adenomatous polyposis had given written informed consent.⁽²⁾ In 1996, we asked a representative sample of Maryland physicians (including obstetricians) how important they thought it would be to obtain consent from a woman at high risk of hereditary breast cancer before obtaining a test for BRCA1. Only 56% of responding physicians thought it was "very important."⁽³⁾ As well, there are practical barriers to effective and complete informed consent. Ensuring adequate patient understanding of disease prevalence, risks of testing, and long-term consequences takes significant time on the part of the clinician and may not be adequately reimbursed as a physician service.

The need for informed consent must also be examined rationally. When the benefits of any intervention outweigh its risks by a wide margin, and when the risks are minimal, a formal consent process is less important.⁽⁴⁾ For procedures that have reproductive and family ramifications, which is the case for prenatal and carrier genetic testing, informed consent is always important.

What can be done to provide clinicians with the necessary tools to obtain truly 'informed' consent and protect patient autonomy? More medical education focused on patient autonomy and the importance of

informed consent in genetic testing is an important foundation. Some evidence shows that current curricula may have an impact: at two medical schools, approximately two-thirds of first and fourth-year students thought obtaining consent before testing was important.(3)

Steps must also be taken to ensure patients are better informed. We are told that Dr. G was given "information" to review at home, but it is unclear how detailed or explicit the materials were. Given physician time constraints, information technology through the use of audiotaped, videotaped, or web-based materials could facilitate patient education and understanding. One site, available both to physicians and consumers, that might be helpful is <http://www.genetests.org>. Interactive Web sites or CD-ROM programs could present alternative scenarios in a clear, easy-to-understand format. These materials could be supplemented by quizzes whose purpose is to ensure that patients understand the disclosure/consent documents they have been given before making complicated decisions. A recent systematic review of consent for participation in research trials indicated that there might be a benefit to multimedia presentations.(5) The same review indicated that one-on-one discussions with neutral educators might be the most effective means of improving understanding.

Once patients are adequately informed, how can we document that patient consent has or has not been obtained? The computerized physician order entry in this case appeared to have a mechanism to ensure consent at multiple levels, but it was overridden by the phlebotomist. Fewer than 20% of all hospitals nationally have computerized physician order entry, especially in the outpatient setting, so most systems probably do not have similar rigorous checks in place.(6) But even when there is an adequate information technology background, this case demonstrates that the procedure for documenting informed consent often needs to be strengthened. Physicians should be responsible for obtaining and documenting written informed consent from all patients. The written consent should be placed in the permanent medical record, whether electronic or paper. Signed paper copies can easily be scanned into the permanent electronic record, as some institutions have done in the case of advance directives. Phlebotomists could then be allowed to draw blood for genetic tests only if they see the written consent signed by the patient and the ordering physician.

In this case no permanent harm was done. Nonetheless, protection of patient autonomy for many forms of genetic testing, or any procedure in which reasonable people may have doubts about its safety or effectiveness, should receive priority. We must ensure patients are truly informed and subsequently establish safeguards to ensure consent has been provided.

Take-Home Points

- Some patients will have rational reasons for declining genetic testing.
- For many forms of genetic testing, physicians must obtain written, informed consent from patients and must document it in the chart.
- Patients must be fully educated and informed prior to providing consent for any genetic testing when reproductive and family issues arise and when the safety and effectiveness of the interventions that are based on test results are questionable.
- Consent safeguards should never be overridden in the absence of signed informed consent.

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References

1. Harmon A. As gene test menu grows, who gets to choose? New York Times. July 21, 2004. Section A, Page 1, Column 1.
2. Giardiello FM, Brensinger JD, Petersen GM, et al. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. N Engl J Med 1997;336:823-7.[[go to PubMed](#)]
3. James CA, Geller G, Bernhardt BA, Doksum T, Holtzman NA. Are practicing and future physicians prepared to obtain informed consent? The case of genetic testing for susceptibility to breast cancer. Community Genet. 1998;1:203-12.[[go to PubMed](#)][[go to abstract](#)]
4. Faden RR, Holtzman NA, Chwalow AJ. Parental rights, child welfare, and public health: the case of PKU screening. Am J Publ Health. 1982;72:1396-1400.[[go to PubMed](#)]
5. Flory J, Emanuel E. Interventions to improve research participants' understanding in informed consent for research: a systematic review. JAMA. 2004;292:1593-601.[[go to PubMed](#)]
6. Kuperman GJ, Gibson RF. Computer physician order entry: benefits, costs, and issues. Ann Intern Med. 2003;139:31-9.[[go to PubMed](#)]

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