The Dilemma of Confidentiality in Huntington Disease

Courtney Wusthoff, *University of California, San Francisco, School of Medicine*

AS GENETIC TESTING ADVANCES, PHYSICIANS FACE DILEMMAS in the disclosure of results: Is genetic information the confidential property of tested individuals, or do biological relatives also have rights to this information? Faced with a dilemma such as that posed by a mother's refusal to inform her daughter of her Huntington disease (HD), the physician first must clarify his or her responsibilities to both patients. Obligations to the mother include confidentiality, respect for autonomy, and nonmaleficence. Recently, the American Medical Association (AMA) affirmed the importance of keeping genetic information confidential.[1] While the AMA policy acknowledges that there are instances in which genetic information should be disclosed to relatives, it clearly states that only in exceptional circumstances would a physician be allowed to break confidentiality to do so. In addition to confidentiality, the physician must respect the mother's autonomy. The mother has the right to choose whether to share her diagnosis with family members and to what degree she would like them involved. For the physician to override this decision would constitute paternalism—substituting his or her own judgment for the patient's. Finally, the mother has forbidden the physician to inform the daughter, as she feels disclosure would distress both her and her daughter. The physician's first responsibility, to do no harm, suggests that he or she must comply with this request. The act of breaking the mother's trust alone could be considered harmful and could be perceived by the mother as abandonment. Then she would not only be ill but also in conflict with her physician.

However, the physician also has responsibilities to the daughter, including a duty to be honest with her. In this situation, such a duty would require the physician to fully inform the daughter of factors to consider in planning a pregnancy, including her family history of HD. Beneficence may also dictate that the physician share this information with the daughter. She could benefit from knowing of her own risk of HD; she may choose to be tested for the mutation and consider her life plans accordingly. If she learns of her risk prior to making decisions regarding reproduction, she may opt to not have children. She may also consider options such as prenatal diagnosis, ovum donation, and adoption.[2] Indeed, one study found that among HD carrier couples, one third chose not to have children and one third chose prenatal diagnosis.[3] It may even be argued that the daughter's autonomy depends on such knowledge. As her mother is the only source of information about her genetic risk, and her risk is much elevated beyond that of the general population, the daughter must be informed of the risk to truly exercise self-determination.

Unfortunately, the obligations to the mother and those to the daughter seem to conflict. The ideal solution to such a conflict would be a compromise in which the physician avoids breaking confidentiality with the mother yet also allows the daughter to become aware of all information relevant to her decision regarding pregnancy.

Such an outcome might be accomplished with time and open communication with both mother and daughter. The physician could explore the mother's motives for secrecy. First, it must be ensured that her request reflects her true wishes. One feature of HD is personality change, including obstinacy. The physician must determine that the mother has sufficient mental capacity to make an informed decision.[4] Assuming that she does, the physician could then discuss the full implications of secrecy for her daughter. It would also be worthwhile to discuss the fact that eventually, her symptoms will become more apparent, and she may require greater care and support from her family. As the illness will probably become obvious at some point, it is worth learning what the mother hopes to gain by keeping it a secret now. The physician might also suggest degrees of compromise, such as allowing him or her to disclose that there is a family history of HD without stating that the mother has been diagnosed. Such a conversation need not be coercive or pejorative, but rather seek to address these relevant issues in a straightforward fashion.

It is possible that as the mother comes to terms with her diagnosis and considers the implications of strict confidentiality, she will want to disclose her diagnosis to her daughter. One model of understanding how patients handle a diagnosis of HD describes an "incipient stage" of coping.[5] In this preliminary stage, the patient has not fully accepted the implications of the diagnosis and can respond only with shock, anxiety, or denial. Patients in this stage may alienate themselves from others and resist confronting their own diagnosis. It is important to recognize that such feelings may only be present for a few months. One study found that depression and anxiety are most common in the first 2 months after diagnosis, but for most patients, such symptoms resolve within 1 year.[6] Eventually, as the mother goes through the process of grieving and accepts the diagnosis, she may no longer deny the reality or impact of disease and may be more willing to share her diagnosis. In time, she may see that sharing her diagnosis could have great benefits for herself, in securing emotional support and relieving guilt and shame, as well as for her daughter, who would be able to make her own informed decisions about testing and life planning. The patient must be given the opportunity to come to this stage of acceptance. If the physician were to disclose the diagnosis to the daughter immediately, he or she would deprive the mother of the opportunity to talk with her children herself. Furthermore, he or she would betray the trust inherent in the patient-physician relationship and may even

cause harm to the daughter by creating the impression that her mother has deceived her. Instead, the physician may facilitate disclosure by the mother through ongoing discussion of the consequences and issues surrounding her diagnosis of HD.

Meanwhile, it would be appropriate to advise the daughter that she should gather information about her family history prior to attempting pregnancy so she will be prepared to make decisions about prenatal diagnosis. She should also be advised to assess the resources she has for starting her family and, perhaps, to wait a few months to complete this evaluation before becoming pregnant. This would be honest and good advice for any woman considering pregnancy and would not betray the confidence of the mother. Furthermore, it would allow the daughter and mother valuable time for discussion that might lead to disclosure by the mother.

While such a solution would be ideal, the mother may remain adamant that she does not want to disclose the family history. The physician must then consider whether it is permissible or even required to inform the daughter. The American Society of Human Genetics (ASHG) states that if attempts to encourage disclosure by the patient have failed, then disclosure of genetic information to relatives is only permissible if such disclosure would serve to ameliorate or prevent a highly likely and foreseeable harm to an identified individual.[7] The President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research and the Institute of Medicine Committee on Assessing Genetic Risks have adopted similar guidelines.[7,8] This case, however, only partially fulfills these criteria. Furthermore, disclosure would contradict principles required to maintain the patient-physician relationship.

The affected relative, the daughter, is certainly identifiable, as she is the physician's patient. However, there is no known way to prevent, treat, or cure HD. The results of testing would not modify the daughter's chances of developing the disease. As such, the only use of the test is to provide information to a person who has made an autonomous decision to know his or her status. While some persons might consider the information valuable for life planning, others might consider it harmful. As there is no treatment for HD, knowing genetic information might cause stigma, psychological distress, or potential discrimination with no subjective benefit. Some patients have responded to a positive HD test result with anxiety, depression, and even suicide attempts. Even those found not to carry the mutation sometimes respond with feelings of survival guilt in the face of an affected relative.[4] All persons have the right not to know their HD status if that is their desire. Ultimately, only the person to be tested can best estimate the subjective value of such information; it would not allow prevention or treatment of the disease in question. As such, disclosure would fail to meet the criteria set forth under the ASHG guidelines.

Thus, the physician must not break confidentiality to disclose the mother's information. Although doing so might benefit the daughter, it also might result in harm. It would violate confidentiality, thus betraying an underlying principle fundamental to the relationship. In this case, it would demonstrate to both mother and daughter that they cannot rely on the physician to keep confidentiality or respect requests for autonomy. On the other hand, if the physician does respect the mother's request, the principles of confidentiality and respect for autonomy are upheld and the relationship is reinforced. While the daughter may be upset that she did not have more information while planning her family, the physician will have been consistent in maintaining the standards expected in a patient-physician relationship. At the very least, the daughter might proceed with confidence that her own requests will be equally respected. The physician's course of action may not be the daughter's preference, but ethical considerations must outweigh a desire to please individual patients.

The complexities of this case reinforce the need for thorough pretest counseling. Scenarios such as this one are not strictly hypothetical; similar cases have been reported in the literature.[9] The best means of handling such situations is to have a clear policy before genetic testing is performed as to who will share the results obtained and under what circumstances the physician will find an obligation to breach confidentiality and inform relatives. By doing so, the physician can both act to inform relatives in their best interests and maintain honesty in his or her relationship with the tested patient. No duties are broken, and the tested patient gives truly informed consent.

REFERENCES

1. AMA Policy H-140.899. Disclosure of Familial Risk in Genetic Testing.
2. Hakiman R. Disclosure of Huntington's disease to family members: the dilemma of known but unknowing parties. *Genet Test.* 2000;4:359-364.
3. Evers-Kiebooms G, Decruyenaere M. Predictive testing for Huntington's disease: a challenge for persons at risk and for professionals. *Patient Educ Counseling.* 1998;35:15-26.
4. Adams J. Confidentiality and Huntington's chorea. *J Med Ethics.* 1990;16:195-199.
5. Bloch M, Adam S, Fuller A, et al. Diagnosis of Huntington's disease: a model for the stages of psychological response based on experience of a predictive testing program. *Am J Med Genet.* 1993;47:368-374.
6. President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research. *Screening and Counseling for Genetic Conditions: A Report on the Ethical, Social, and Legal Implications of Genetic Screening, Counseling, and Education Programs*. Washington, DC: President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research; 1983.
7. American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure. ASHG statement: professional disclosure of familial genetic information. *Am J Hum Genet.* 1998;62:474-483.
8. Andrews LB, Fullarton JE, Holtzman NA, Motulsky AG, eds. *Assessing Genetic Risks: Implications for Health and Social Policy*. Washington, DC: National Academy Press; 1994.
9. European Community Huntington's Disease Collaborative Study Group. Ethical and social issues in presymptomatic testing for Huntington's disease: a European community collaborative study. *J Med Genet.* 1993;30:1028-1035.