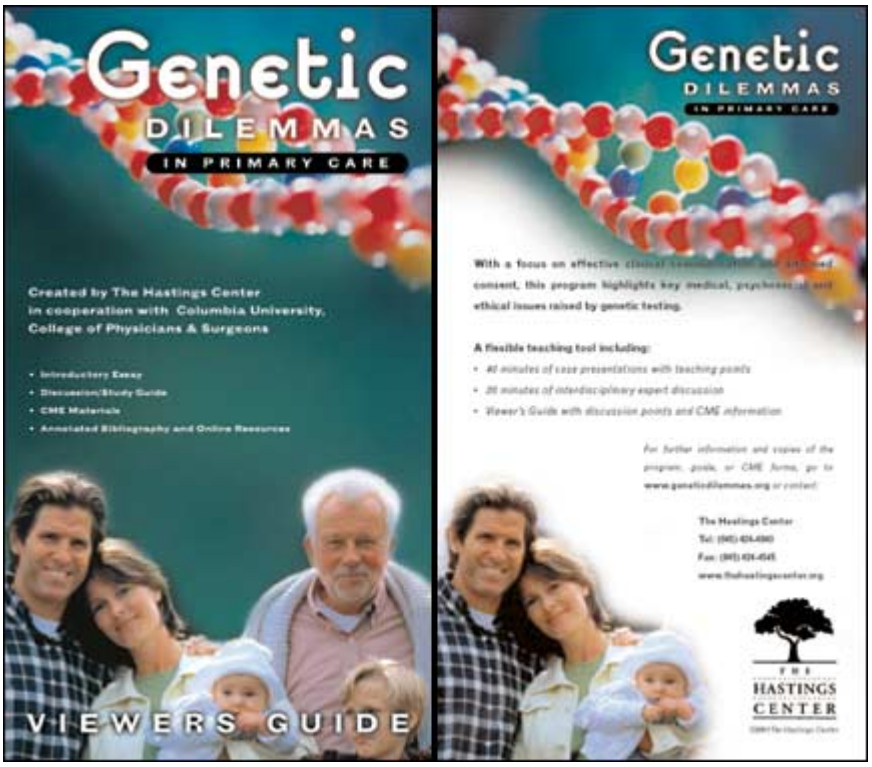
Genetic Dilemmas in Primary Care



CME Credit

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There is no mention of any specific product in the program.

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The Hastings Center, Membership Dept.
21 Malcolm Gordon Drive, Garrison, NY 10524-5555
Tel: (845) 424-4040, Fax: (845) 424-4545

Program Faculty

Core Working Group:

Larry Amsel, MD, MPH Assistant Professor of Clinical Psychiatry, College of Physicians & Surgeons of Columbia University, New York, NY, Adjunct Associate for Education, The Hastings Center

Steve Baum, MFA Steve Baum Productions, New York, NY

Barbara Bowles Biesecker, MS, CGC Genetic Counselor and Associate Investigator; Co-director JHU/NHGRI Genetic Counseling Training Program, National Human Genome Research Institute, NIH, Bethesda, MD

Daniel Callahan, PhD Director of International Programs, The Hastings Center, Garrison, NY

Diane Dreher, MA Independent Writer / Producer; Adjunct Associate for Education, The Hastings Center, Garrison, NY

Ann Fagan, Patient Representative, Conyngham, PA

Kevin Ferentz, MD Associate Professor, University of Maryland School of Medicine; Residency Director Department of Family Medicine, Baltimore, MD

Elena Gates, MD Professor, Department of Obstetrics, Gynecology, and Reproductive Sciences, University of California, San Francisco

Bruce Jennings, MA Senior Research Scholar, The Hastings Center, Garrison, NY

Trish Magyari, MS, CGC Genetic Counselor, National Human Genome Research Institute, Office of the Clinical Director, NIH, Bethesda, MD

Erik Parens, PhD Associate for Philosophical Studies, The Hastings Center, Garrison, NY

Victor B. Penchaszadeh, MD, MSPH Professor of Pediatrics, Albert Einstein College of Medicine; Chief, Division of Medical Genetics, Beth Israel Medical Center, New York, NY

Diana Punales-Morejon, MS, CGC Coordinator, Genetic Counseling, Division of Medical Genetics, Beth Israel Medical Center, New York, NY

Colleen Scanlon, RN, JD Senior Vice President, Advocacy, Catholic Health Initiatives, Denver, CO

Vivian Trakinski, BA Independent Writer/Producer, New York, NY

Expert Advisory Panel:

Steven M. Albert, PhD, MSc Associate Professor of Clinical Public Health, Columbia University, New York, NY

Bonnie S. LeRoy, MS, CGC Director, Graduate Program in Genetic Counseling, University of Minnesota, Minneapolis, MN

Larry Norton, MD Head, Division of Solid Tumor Oncology, Memorial Sloan Kettering Cancer Center, New York, NY

Artemis P. Simopoulos, MD President, The Center for Genetics, Nutrition and Health, Washington, DC

Claudio Stern, PhD Professor and Chairman, Department of Genetics and Development, College of Physicians & Surgeons of Columbia University, New York, NY

Dorothy Warburton, PhD Professor of Clinical Genetics and Development, College of Physicians & Surgeons of Columbia University, New York, NY

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Dr. Norman Relkin was tremendously helpful as a teacher and reviewer at the onset of the project. Dr. Niharika Khanna has played a key role in reviewing the final guide and helping to disseminate the program among FP Residency Directors. Wendy Child has guided our qualitative research and evaluation during every phase of work. Thanks also to Esther Modell of the Center for Continuing Education at Columbia University for her guidance and patience throughout and to Mary Ann Hasbrouck, Deborah Giordano, Joe Paino, Chris McKee, and Christine Tucciarone of the Hastings Center for their valuable advice and assistance.

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– Diane Dreher, Project Director

Primary Care Clinicians and the Dilemmas of Genetic Testing

If even a fraction of the claims made about the impending impact of genetics on clinical practice came true, the clinical genetics services would be overwhelmed. We must not miss the opportunity to prepare primary care for the new genetics.
– Jon Emery and Susan Hayflick [1]

The Human Genome Project, Scientific Reality and Public Expectations.

There is little doubt that the sequencing of the human genome, announced on June 26th, 2000 by Francis Collins of the NIH and Craig Ventner, of Celera Genomics, represented one of the most significant achievements in the history of science. The formal announcement, which included both President Clinton and Prime Minister Tony Blair, crystallized the fact that after a half century of research, genetic knowledge had reached a critical mass and was now expected to transform our understanding of our biological selves.

Although the scientific achievement is unquestionable, the nature and degree of its impact on the current and future practice of medicine, is not clear. Some experts have predicted a rapid and profound revolution in medicine.[2] Others, even some geneticists, doubt that genetics will fundamentally change the nature of medical practice.[3]

While medical experts debate the impact of genetics on medicine, the public perception is that a significant change in medicine has already begun. *The Los Angeles Time*s, for example, called the recent advances in genetics, "official recognition that a new era in medicine had begun."[4] Healthcare providers are thus faced not only with the actual developments in genetic medicine, but also with powerful perceptions that the public has about the promises of genetic medicine.

The Role of the Primary Care Provider in Genetic Medicine

As primary care clinicians are becoming more conversant with the recent advances in genetics, the key issues that many are raising include: How will genetics impact my daily practice? What kinds of questions will patients be asking about genetic medicine? What is the appropriate role of the primary care clinician in the overall delivery of genetic services? What kind of education do I need to effectively fulfill this role?

In a household survey of patients conducted by the American Medical Association, over 80% of those surveyed were confident that their primary care clinicians could

assess the risks of their developing a genetic disorder and could appropriately recommend genetic testing when indicated. About 74% were also confident that their primary care clinicians could correctly interpret the results of a genetic test.[5] Thus, the general public has a high level of confidence in their primary care clinicians and an equally high set of expectations when it comes to genetic medicine.

Two areas of genetic medicine are currently most relevant to primary care and are thus most likely to impact daily practice. They are:

1) Prediction of risk for adult onset disorders with a known genetic component, (familial breast and colorectal cancers, Huntington's Disease and others) based on family history and/or genetic testing.[6]

2) Assessment of reproductive risk by testing for common autosomal recessive conditions such as hemoglobinopathies, cystic fibrosis, and muscular dystrophy.[7]

For the near future, these issues will probably define the focus of genetic medicine in primary care. However, over the next several years, as genetic medicine becomes more pervasive, primary care clinicians are likely to become involved with:

- Testing for the genetic components of common disorders that have multi-factorial etiologies such as ischemic heart disease, asthma, or diabetes, and
- Identifying normal genetic variations that predict drug response and side effect profiles.[8]

With respect to these areas of genetic medicine, primary care practitioners will probably have specific roles within a collaborative healthcare team that includes genetic counselors, clinical geneticists, and others. With rare exception, the primary care clinician will have the closest relationship with the patient and is therefore most likely to know the patient's medical history, family medical history, and attitudes towards healthcare.[9] In light of this, Jon Emery and Susan Hayflick have suggested that in the future, primary care clinicians might take on the following:[10]

- Identifying individuals who may benefit from genetic services.
- Recognizing physical and historical features of genetic disorders.
- Providing basic genetic information, and counseling to facilitate informed decision-making and informed consent for genetic testing and other genetic services.
- Recognizing the special psychosocial needs of a family in which a genetic disorder or susceptibility has been identified.

• Knowing the full range of genetic specialists available in one's area and when referral and collaboration are indicated.

• Collaborating with genetics specialists in managing patients with complex and rare genetic disorders.

These activities encompass what we might call the science and the art of genetic medicine.

The Science and the Art of Genetic Medicine

Grappling with the science of genetic medicine, while quite challenging, is not a unique problem. In all areas of medical research, knowledge is being produced at a daunting rate and keeping up with the advances, in the traditional sense of learning all the new material, has simply become impossible. However, utilizing this new knowledge in the best interests of patients is not only possible but it is a primary challenge in health care, as it transitions to an information-based model. To accomplish this, clinicians are increasingly using a few related strategies. There has been a shift from a focus on remembering information to a focus on judiciously applying information in the service of shared decision-making. This involves:

• Easy access to the relevant medical literature, (often electronic) and critical application of this knowledge to the case at hand.

• Formal and informal consultation with, and referral to medical specialists and other health care professionals who have specialized knowledge. In genetic medicine of course, this requires familiarity with the functions of genetic counselors, medical geneticists, and other specialists who may have genetic expertise regarding particular disorders.

• A greater reliance on the team approach to health care delivery including the involvement of the patient as an informed team member. This involves knowledge of consumer- oriented information sources that can help patients become active participants in their own care.

There is an enormous amount of genetic information available electronically as well as in print. The Resource section of this guide (page 37) lists a number of helpful articles, books and websites that can assist clinicians with the science of genetic medicine. It also includes sites for patient education.

Resources for practicing the art of genetic medicine on the other hand, are less readily available, and are the focus of this educational video and viewer's guide. The art of delivering genetic services in a primary care setting involves specific psychological, familial, social, ethical, vocational, financial, and legal issues that often differ from other areas of medicine.

In 1995, the American Society of Human Genetics (ASHG) published a recommended Core Curriculum in Genetics for medical schools.[11] This curriculum and others have been used to create a list of Core Competencies in Genetics for all health care professionals by the National Coalition for Health Professional Education in Genetics (NCHPEG), a coalition of over 100 health professional organizations.[12] Over half of the 44 competencies included concern the acquisition of communication skills and changes in practice attitudes that speak to the ethical, legal and psychosocial aspects of genetic medicine.

These skills are probably best acquired in the context of specific clinical cases. The accompanying videotape presents three such cases, together with a discussion of the relevant issues by genetic experts, primary care clinicians, ethicists and patient representatives. Watching the tape in conjunction with this viewer's guide, and exploring these issues with colleagues should help clinicians begin to acquire or reinforce the skills and attitudes necessary to initiate pre-test counseling, guide a shared decision-making process about testing, obtain educated informed consent, follow up with post-test counseling, and/or determine when referral to a genetic specialist is indicated.

Challenges Inherent in Information Obtained by Genetic Tests

In general, clinicians have reserved in-depth consultation and patient counseling for difficult therapeutic decisions, and have tended to think of informed consent primarily in the context of treatment. In genetic medicine, however, the most important informed consent discussion is the one that takes place prior to ordering any tests – in the context of diagnosis. Consider some of the challenges that this presents for clinicians:

1. Genetic information does not affect only the individual receiving the test, but other family members as well. While we have always been able to make inferences about the health status of family members from family medical histories and general medical information, genetic testing makes those inferences considerably more precise, bringing some dramatic medical, ethical and psychological side effects into play. For example, the tape depicts, and a number of studies describe, how family members who do not carry a mutation can experience "survivor guilt", demonstrating that even a negative genetic test may have adverse psychological consequences.[13]

A related dilemma concerns whether a clinician has the right, or even the duty, to override her/his patient's wish to keep test results private in order to warn potentially affected family members. As the tape shows, there is disagreement about whether such a duty exists.

2. Genetic information can transform how individuals perceive themselves and are perceived by others. Even in the absence of clinical disease, a positive genetic test result has the potential to transform how a person perceives herself and is perceived by her family, employer, and health care provider. Such "reclassification" of course can have important psychological effects. It may also affect one's chances of getting or keeping a job; one's chances of being promoted; or one's access to health insurance or life insurance coverage. Anecdotal evidence suggests that genetic discrimination is an issue, but as of today, there still are no good data on how often such discrimination actually occurs.[14]

Given that comprehensive federal legislation against genetic discrimination has not been passed (as of January 2002), and that state laws vary, patients are still wise to question the consequences of testing. And in the current health care context, where medical information is shared across huge networks, providers offering tests are faced with the dilemma of being unable to ensure privacy and confidentiality for their patients. It is therefore critical for clinicians to speak explicitly with patients about the prospect that their test results might be disclosed.

3. Genetic information is essentially probabilistic, making it more complex and more difficult to communicate to patients than other types of medical information. As has been shown in numerous studies, effective communication of risk or probabilistic information to patients in the primary care setting is very challenging.[15] Genetic 'information' can paradoxically, seem to create more uncertainty than it resolves. This is seen in the program's first case about BRCA testing. To begin with, there is the phenomenon of incomplete penetrance: even if a person has a mutation associated with a given disease, she will not necessarily have symptoms of that disease. Second, the absence of a mutation does not ensure the absence of disease. Less than 10% of breast and ovarian cancer can be traced to identified inherited mutations, while more than 90% can not be.

Another source of uncertainty in genetic illness is variable expressivity. As illustrated in the tape, even when we know for certain that a fetus is destined to develop cystic fibrosis, we do not know how severe the CF will be or at what age it will first manifest. Moreover, inaccuracy of genetic tests, namely false positive and false negative results, add yet another layer of complexity to genetic medicine. As we saw in our CF case, the lack of standardization in laboratory procedures can easily lead to a false negative result. Finally, as Dr. Giardiello explains in the tape, his study demonstrated that clinicians' lack of familiarity with these tests can, and has led to serious misinterpretation, even when the test results are accurate.[16]

4. Since there are few definitive therapies, the clinical uses of genetic information are often subtle. In genetic medicine today, testing is often far ahead of therapeutics. The utility of these tests may lie either in the psychological benefit of 'knowing' one's status or, in some cases, in the preventive strategies that such testing can promote. The former is highly dependent on individual preferences and coping styles, which need to be carefully explored. The latter depends on a number of complex interacting factors as described by Evans.[17] In particular, each of the following factors tends to increase the utility of genetic testing.

1. *The test is highly predictive*
2. *The disorder involved is serious*
3. *The illness is not easily detected by standard screening or surveillance*
4. *The illness is not easily treated once manifest, and*
5. *Effective preventive and/or screening measures exist, but are too costly or difficult to recommend to the entire population.*

For example, according to Evans, a predictive genetic test for hypertension would not have a high utility, as this condition is easily screened, effectively treated once manifest, and the preventive strategies are relatively inexpensive, and probably beneficial for the entire population – not just for those at high risk. Therefore, population-based genetic testing would probably add little to the overall management of this condition.

The Process of Facilitating Truly Informed Consent

The popular conception of informed consent has rested on a model of patient-physician interaction during which patients ask for, and physicians offer professional recommendations, to which patients then usually consent. Though in the past, informed consent has often been a brief, pro forma "event," collaborative decision-making has recently been gaining ground as an appropriate model for most clinical decisions. Because the risks and benefits of genetic testing are complicated and their evaluation so contingent upon the perspective of the particular patient, the Task Force on Genetic Testing has called for what has been referred to as an expanded informed consent process or an educated consent process.[18] The point is that there are few cases where clinicians can recommend with confidence that a given patient should or should not have a particular genetic test. This is a shared decision, based on a discussion of the individual patient's values and coping styles, as much as it is based on a patient's medical status.

The difficulty, of course, is that conducting an expanded informed consent is a time-intensive process, and the current structure of health care delivery does not encourage clinicians to spend more time with patients. Despite these barriers,

clinicians are finding creative ways of delivering the appropriate genetic services by rethinking their own roles in informed consent, utilizing multiple visits and discussion aids, involving genetic counselors and other health care personnel, and using information technologies.[19] Hopefully, we will also see changes in the structure of health care delivery that address the need for these services and make them easier to deliver.

Conclusion

We began this essay with references to genetics revolutionizing medicine. Most experts who speak of this revolution are referring to a revolution of biotechnology and information science that would make medicine even more highly technological than it is today. While this may come to pass, what we hope to convey in this program is that genetics also has the potential – paradoxically – to reinforce the humanistic, empathic and communicative aspects of clinical care. Clinicians will conceivably become more aware of their patients' values, and more aware of the family, religious and social structures within which these patients live their, still very unpredictable, lives. Genetics will revolutionize medicine. It will send it back to its roots; back to its future.

– Larry Amsel, Diane Dreher, Bruce Jennings, and Erik Parens,
The Hastings Center.

References

1. Emery J, Hayflick S. The challenge of integrating genetic medicine into primary care. BMJ 2001;322(7293):1027-30.
2. Is genetics the future of medicine? Report by New York Presbyterian Hospital. New York, NY 1991.
3. Holtzman NA, Marteau TM. Will genetics revolutionize medicine? N Engl J Med 2000; 343(2):141-4.
4. Los Angeles Times 2000; Financial Desk, 27 June.
5. Goldberg S. Genetic testing: a study of consumer attitudes. American Medical Association 1998; available at: www.ama-assn.org/ama/pub/article/2304-2937.html (accessed 12/28/01).
6. Scheuner MT, Wang SJ, Raffel LJ, Larabell SK, Rotter JI. Family history: a comprehensive genetic risk assessment method for the chronic conditions of adulthood. Am J Med Genet 1997;71(3):315-24.

7. For a review of the ethical issues raised by prenatal genetic testing, see Parens E, Asch A. The disability rights critique of prenatal genetic testing: reflections and recommendations, Hastings Cent Rep 1999;29(5):S1-22.

8. Mathew C. Science, medicine, and the future: postgenomic technologies: hunting the genes for common disorders. BMJ 2001;322(7293):1031-4.

9. Donnai D, Elles R. Integrated regional genetic services: current and future provision. BMJ 2001;322(7293):1048-52.

10. Emery J, Hayflick S. The challenge of integrating genetic medicine into primary care. BMJ 2001;322(7293):1027-30.

11. American Society of Human Genetics Information and Education Committee. Medical school core curriculum in genetics. Am J Hum Genet 1995;56(2):535-7.

12. National Coalition for Health Professional Education in Genetics. Core competencies in genetics essential for all health-care professionals; 2000. available at: www.ncpeg.org/news-box/corecompetencies000.html (accessed on 12/28/01).

13. Lynch HT, Smyrk T, Lynch J, et al. Genetic counseling in an extended attenuated familial adenomatous polyposis kindred. Am J Gastroenterol 1996;91(3):455-9.

14. Rothenberg K, Fuller B, Rothstein M, et al. Genetic information and the workplace: legislative approaches and policy challenges. Science 1997;275(5307):1755-7.

15. Edwards A, Elwyn G, Covey J, Matthews E, Pill R. Presenting risk information--a review of the effects of “framing” and other manipulations on patient outcomes. J Health Commun 2001;6(1):61-82.

16. 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(12):823-7.

17. Evans JP, Skrzynia C, Burke W. The complexities of predictive genetic testing. BMJ 2001;322(7293):1052-6.

18. Holtzman NA, Watson MS. Promoting safe and effective genetic testing in the United States: final report of the task force on genetic testing. Baltimore and London: The Johns Hopkins University Press;1998.

19. Green MJ, Fost N. An interactive computer program for educating and counseling patients about genetic susceptibility to breast cancer. J Cancer Educ 1997;12:204-8.

About CME Credit

The deadline for CME credit has expired as of June 30, 2004.

This program is designed for primary care clinicians, including family physicians, internists, obstetrician/ gynecologists, pediatricians and advance practice nurses. After viewing the videotape, participants will:

- Have an increased awareness of the psychological and social implications of genetic testing.
- Have an increased understanding of the current accuracy, reliability, and implications of genetic testing in general, with specific emphasis on testing for inherited breast and ovarian cancer, cystic fibrosis and familial adenomatous polyposis.
- Understand the concept of an expanded informed consent process for genetic testing that will promote meaningful dialogue between caregiver and patient and facilitate shared decision-making.

The activity has been planned and implemented in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint sponsorship of the College of Physicians and Surgeons of Columbia University and The Hastings Center.

The College of Physicians and Surgeons is accredited by ACCME to provide continuing medical education to physicians. The College of Physicians and Surgeons designates this educational activity for a maximum of 2 hours in Category 1 Credit towards the AMA Physician's Recognition Award. Each physician should claim only those hours of credit that he/she actually spent in the educational activity.

There is no discussion of specific products in this program.

How to Obtain CME Credit

The deadline for CME credit has expired as of June 30, 2004.

Completion of both the Post-test and the Program Evaluation are required for CME credit.

1. Print out and complete the Post-test and the Program Evaluation Answer Sheets.

2. Send both completed answer sheets with a check for $20 payable to Columbia University to:

> Center for Continuing Education (Psych #902)
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Twelve correct answers are required to receive credit. Upon completion of these requirements, the College of Physicians & Surgeons of Columbia University will issue a certificate and return your corrected quiz for your permanent records.

CME Post-test Questions

Genetic Dilemmas in Primary Care

The deadline for CME credit has expired as of June 30, 2004.

Select the single best answer to each question and record your responses on the Post-test Answer Sheet (pages 21-22). When completed, mail the Post-test Answer Sheet (pages 21-22) and the Evaluation Answer Sheet (pages 26-28) to:

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You will also need to include one check for $20, payable to Columbia University, for both answer sheets.

Twelve correct answers are required.

Questions

1. A 40-year-old woman with no family history of breast cancer requests testing for the 'breast cancer gene' stating that she wants to know her genetic status so that she can potentially avoid screening mammography. In counseling this patient about genetic testing, it would be appropriate to:

    a. inform her that a negative test result for mutations in BRCA1 and BRCA2 would not eliminate her risk of breast cancer and that a woman of her age should be following mammography screening recommendations regardless of the results of genetic testing.

    b. inform her that a negative test result for mutations in BRCA1 and BRCA2 would cut her risk of developing breast cancer in half but not eliminate it altogether.

    c. inform her that although most women who have developed breast cancer test positive for mutations in BRCA1 or BRCA2, most of the women who test positive don't develop breast cancer.

    d. inform her that there is no role for genetic testing for mutations in BRCA1 and BRCA2 in the absence of a family history of breast cancer.

2. In most cases a genetic test result gives only probabilistic information. There are multiple sources of uncertainty in these tests. Which of the following is /are correctly defined:

    a. “Penetrance” refers to the likelihood that a given gene mutation will actually result in symptoms of the disease.

    b. “Variable expressivity” is a measure of the number of different gene mutations related to a given disorder

    c. “Sensitivity” is the proportion of those individuals who have the mutation who actually get a positive test result for that mutation.

    d. Both a and c.

3. Which of the following diseases follows an autosomal dominant pattern of inheritance?

    a. Inherited breast cancer

    b. Familial adenomatous polyposis

    c. Cystic Fibrosis

    d. Both a and b

4. A 35 year old woman presents with a family history that includes a mother who was diagnosed with breast cancer at age 65 and subsequently died, and a sister diagnosed at 42 who is currently in treatment. She asks if she should be tested for the “breast cancer gene.”

    a. She should be discouraged from genetic testing because the test results would make no difference in her clinical management.

    b. She should be strongly encouraged to undergo genetic testing for BRCA1 and BRCA2 as the current medical standard upon which to base your clinical approach.

    c. This patient can be appropriately managed either with or without genetic test results, so the choice to be tested should be hers.

    d. This family history is a strong indication that the patient carries a mutation in BRCA1 or BRCA2 and therefore testing can add no useful information.

5. You suspect a patient may have familial adenomatous polyposis and discuss the possibility of genetic testing with the patient. The patient responds that he has no family history of FAP and asks why he would need a genetic test. You explain that:

    a. A spontaneous mutation of the APC gene occurs in about 1/3 of cases of familial adenomatous polyposis

    b. Spontaneous mutations develop during embryogenesis in an individual and therefore FAP can present without an associated family history

    c. FAP is always associated with a family history of colon cancer but testing is done because family members may have died of other causes before the FAP manifested.

    d. Both a and b

6. In most cases involving gene testing, a patient should receive genetic counseling from a physician, nurse or genetic counselor

    a. Before the patient receives the test results

    b. When the patient receives the test results

    c. Only if the test result is positive

    d. Before a decision to undergo testing is made

7. In cases of prenatal testing, when one person in a couple tests positive as a carrier for an autosomal recessive genetic condition

    a. The other person in the couple should receive the most sensitive testing available for that condition

    b. The other person needs to undergo testing only if both members of the couple have the same ethnic background

    c. The couple should be referred for genetic counseling

    d. a and c

8. Which of the following are considered potential benefits of genetic testing for some individuals?
    a. relief from uncertainty
    b. increased sense of control by the patient
    c. targeted screening, recommendations and prevention
    d. all of the above

9. Current policies and standards endorse the genetic testing of minors
    a. whenever the parents request it
    b. only if the minor is old enough to understand the nature of the test
    c. only when there is a medical intervention available that is likely to be beneficial to the child
    d. based on the same criteria used for the testing of adults

10. Genetic testing requires an expanded informed consent process because
    a. results of genetic tests are more likely to impact family members of the patient both medically and psychologically
    b. genetic tests can give false negative as well as false positive results
    c. genetic tests may be offered even when there is no medical intervention available that is likely to be beneficial.
    d. All of the above

True or False:

11. Accurate interpretation of genetic testing for familial adenomatous polyposis requires that the specific APC gene mutation in the family must be identified in a symptomatic family member before testing an asymptomatic person.
12. There are psychological benefits for some patients who test positive for a genetic mutation, even in the absence of available medical treatment, and these benefits alone can justify the testing.
13. An appropriate informed consent process for genetic testing would include a discussion of the psychological risks involved, except in cases where there is a clear medical benefit to having the information that the test will yield.
14. Medical management based on a false negative result in a patient tested for a mutation of the APC gene causing familial adenomatous polyposis can cause significant psychological problems, but is not life threatening.
15. As of December 2001, employment discrimination due to results of genetic testing is no longer a concern, as all fifty states have now enacted comprehensive, effective antidiscrimination legislation.
16. Informing patients about the range of emotional reactions that others have experienced with genetic testing may help them to anticipate ways they may feel that they had not previously considered, as they decide whether or not to undergo testing.
17. In cases when there is a clear medical benefit associated with genetic testing, clinicians are required to reveal a positive test result to potentially affected family members, even if the patient does not wish to share the information.
18. Because of the high level of technological expertise required to perform genetic analysis, the results obtained from different genetics labs are highly uniform.

CME Post-test ANSWER SHEET

Genetic Dilemmas in Primary Care (Psych #902)

The deadline for CME credit has expired as of June 30, 2004.

Instructions: Use this answer sheet to complete the Post-test (questions start on page #16). Mail your completed answer sheet (pages 21-22) along with the Evaluation Answer Sheet (pages 26-28) to:

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Time Spent on this Activity – Hours _____ Minutes _____

Please **circle** the single best answer to each question.

1.	a	b	c	d
2.	a	b	c	d
3.	a	b	c	d
4.	a	b	c	d
5.	a	b	c	d
6.	a	b	c	d
7.	a	b	c	d
8.	a	b	c	d
9.	a	b	c	d
10.	a	b	c	d
11.	True	False		
12.	True	False		
13.	True	False		
14.	True	False		
15.	True	False		
16.	True	False		
17.	True	False		
18.	True	False		

Program Evaluation Questions for CME Credit

Genetic Dilemmas in Primary Care

The deadline for CME credit has expired as of June 30, 2004.

Please answer these questions using the Evaluation Answer Sheet (pages 26-28). When completed, mail the Evaluation Answer Sheet (pages 26-28) and the Post-test Answer Sheet (pages 21-22) to:

Center for Continuing Education (Psych #902)
Columbia University, College of Physicians & Surgeons
630 West 168th Street, Unit 39
New York, NY 10032-3702

You will also need to include one check for $20, payable to Columbia University, for both answer sheets.

Questions

1. What is your profession?

2. Highest degree completed or Degree program currently attending?

3. At what stage is your career?
   (Training completed, fellowship, resident, intern/extern, attending degree program?)

4. What is your therapeutic area of practice?
   (Family Practice, Internal Medicine, OBGYN, Pediatrics, other?)

5. Number of years in practice?

Questions 6-9: Did the material presented meet the following educational objectives?

6. Increased your awareness of the psychological and social implications of genetic testing? *(Yes or No)*

7. Increased your understanding of the accuracy, reliability and implications of genetic testing in general? *(Yes or No)*

8. Increased your understanding of the accuracy, reliability and implications of genetic testing for breast/ovarian cancer, cystic fibrosis and familial adenomatous polyposis? *(Yes or No)*

9. Increased your understanding of the concept of an expanded informed consent process for genetic testing? *(Yes or No)*

10. How would you rate this program on its relevance to your clinical practice?
    a) Excellent b) Good c) Fair d) Poor

11. Rate the program in terms of its ability to hold your interest.
    a) Excellent b) Good c) Fair d) Poor

12. Was the material presented in a clear and understandable fashion?
    a) Excellent b) Good c) Fair d) Poor

Please answer questions 13-17 using the following scale:

a) Never
b) About once a year
c) Once a month
d) Once a week
e) More often than once a week

13. How often have you initiated a conversation about genetic testing with a patient?

14. How often have you received requests or inquiries about genetic testing from a patient?

15. How often have you ordered genetic tests?

16. How often have you referred a patient for genetic counseling?

17. How often do you personally counsel patients about genetic testing?

18. After viewing this video, will you be better prepared to counsel patients and their families about genetic testing?
    a) Definitely b) Possibly c) Unlikely d) Definitely not

19. Are you likely to change your practice in any of the following areas after viewing this program? (Yes or No)

    a. More likely to seek out information about genetic testing?

    b. More likely to initiate counseling of your patients about genetic testing?

    c. More likely to refer patients to genetic counseling?

    d. More likely to expand the informed consent process for genetic testing?

    e. More likely to consider the effects of genetic testing on children and adolescents?

    f. More likely to emphasize with patients the privacy and confidentiality issues related to genetic testing?

20. Overall, how would you rate this video?
    a) Excellent b) Good c) Fair d) Poor

21. Would you recommend this video to a colleague?
    a) Definitely b) Possibly c) Unlikely d) Definitely not

22. The most valuable part of this videotape was …

23. The least valuable part of this videotape was …

24. Was the program presented objectively?

Program Evaluation ANSWER SHEET for CME Credit

Genetic Dilemmas in Primary Care (Psych #902)

The deadline for CME credit has expired as of June 30, 2004.

Instructions: Use this answer sheet to complete the Program Evaluation (questions start on page #23). Mail your completed answer sheet (pages 26-28) along with the Post-test Answer Sheet (pages 21-22) to:

Center for Continuing Education (Psych #902)
Columbia University, College of Physicians & Surgeons
630 West 168th Street, Unit 39
New York, NY 10032-3702

You will also need to include one check for $20, payable to Columbia University, for both answer sheets.

Please **fill in the blank** or **circle** the single best answer to each question.

1. _____

2. _____

3. _____

4. _____

5. _____

6.	Yes	No			
7.	Yes	No			
8.	Yes	No			
9.	Yes	No			
10.	a	b	c	d	
11.	a	b	c	d	
12.	a	b	c	d	
13.	a	b	c	d	e
14.	a	b	c	d	e
15.	a	b	c	d	e
16.	a	b	c	d	e
17.	a	b	c	d	e
18.	a	b	c	d	
19.a	Yes	No			
19.b	Yes	No			
19.c	Yes	No			
19.d	Yes	No			
19.e	Yes	No			
19.f	Yes	No			
20.	a	b	c	d	
21.	a	b	c	d	

22. _____

23. _____

24. _____

Case Review and Discussion Guide

(See also General Discussion Topics on page 33)

Case #1: Inherited Breast and Ovarian Cancer

Synopsis: Rita was 56 years old and the mother of six when she was diagnosed with ovarian cancer in 1994. Within six months of her diagnosis, her 33-year old daughter Tanya was diagnosed with breast cancer. Mother and daughter underwent gene testing at the University of Chicago's Cancer Risk Clinic and were found to carry a BRCA2 mutation. Two years later, a second daughter, Rita Jr., was diagnosed with breast cancer at age 30. In the course of assessing various treatment options, she also elected to undergo gene testing. Her results revealed no BRCA1 or 2 mutations.

Among Rita's four asymptomatic children, decisions about whether to be tested have varied. Rita's 36-year old daughter, Roslyn elected to be tested and was found to carry the same BRCA2 mutation as her mother and sister as well as a polymorphism – a variation that is considered to be normal – in BRCA1. Another daughter has also chosen to be tested but at the time of the video recording, had chosen not to receive her results. The remaining daughter and Rita's only son have so far declined testing.

Rita begins her story by saying, "Cancer is a devastating disease, not only on the body but on the family." It becomes clear that she is speaking not only about cancer, but about the genetic mutation in her family that is associated with breast and ovarian cancer.

1. Though primary care providers are accustomed to handling complex patient communication and management issues, genetic testing presents specific challenges in that the impact is on the family as well as the individual patient. What practical implications might such a difference have for handling the pretest discussion, informed consent process and posttest discussion of results?

2. Does the entire family become the "patient" when we offer genetic testing? For example, would you as a physician have any responsibilities to Rita's children or grandchildren given that she carries a mutation for a potentially life-threatening illness? If her daughters were already your patients would you want to initiate a discussion about testing even if they have no symptoms of cancer?

3. One of the daughters in this family did not choose to be tested despite the strong positive family history of cancer. Does her decision seem to be accepted as

legitimate by her family? How might a primary care physician facilitate acceptance of her choice by her mother and sisters?

4. Would it be reasonable for Rita's son to undergo BRCA1 and 2 testing? What are the potential benefits, risks and limitations of such testing that he should consider?

5. In the program Tanya stated that she decided to be tested in part for her daughter's benefit. How would you respond to a mother who had tested positive and now wanted BRCA 1 and 2 testing for her 7-year old daughter?

6. The genetic test results in this family were complex and sometimes counter-intuitive. How might such results affect a family's confidence in the health care system, and in the logic of medical science? Might the potential confusion affect future health decisions or behaviors? How can we help patients deal with the uncertainty inherent in most genetic testing?

Case #2: Familial Adenomatous Polyposis

Synopsis: Ann was 26 and the mother of two young girls when she presented with symptoms of rectal bleeding, stomach pain and weight loss. Soon after, she had endoscopic screening and was diagnosed with FAP. Despite the absence of FAP in her family history, each of Ann's eight siblings were examined endoscopically for signs of the disease. No polyps were discovered in any family member, which indicated that Ann had a spontaneous mutation of the APC gene. Ann's daughters began endoscopic screening at ages 10 and 8.

Two years later, Ann and her daughters were invited to participate in a study assessing the psychological effects of APC gene testing on children. After extensive pre-test counseling, they all consented to undergo testing. The results revealed that both girls inherited their mother's APC gene mutation. They continued regular screening. Polyps were eventually found in both girls, who subsequently had surgery.

(Note: *In cases of classic FAP, polyps typically appear around age 15 and the average age for colon cancer to develop is 39. There is also an attenuated form of FAP, characterized by fewer polyps that typically appear in patients in their 30's. In the attenuated form, colon cancer develops at an average age of* 51).

APC testing is a rare example of gene testing considered to be appropriate for children. Given that a negative test result can eliminate the need for invasive endoscopic screening and a positive result can lead to early diagnosis and better treatment, the medical benefit of testing is clear. There are, however, important psychological and social risks associated with the test for both children and adults.

1. Given the medical benefits of APC testing, many providers would be strongly inclined to recommend the test. How "directive" should a clinician be in

recommending the test? How would you describe to patients and families the psychological and social risks involved with APC testing?

2. What would you say to parents who preferred to avoid APC gene testing for their children due to potential stigma, discrimination, self-image problems, or other psychosocial effects?

3. How much (if any) autonomy should at-risk adolescents have in deciding whether they want an APC test?

4. Though this is a high functioning family that has coped extremely well with the disorder, there is nevertheless a great deal of guilt and anger on the mother's part for passing FAP to her daughters and a sense of "survivor guilt" on the part of the father who does not share this disorder. These feelings and attitudes have profoundly affected the family. If they were your patients, what, if anything would you do to manage the psychosocial effects of FAP on the family?

Case #3: Cystic Fibrosis

Synopsis: Bill and Christine were married for 7 years when Christine finally became pregnant for the first time. During her first pre-natal exam, Christine's obstetrician offered her CF carrier testing along with several other prenatal blood tests. Christine agreed to all of the tests without genetic counseling or formalized informed consent.

Christine's CF test was positive for the .F508 mutation, which accounts for 70% of CF mutations among Caucasians. She was told that Bill should be tested immediately. Bill went to his primary care provider for the testing, as required by his insurance carrier. Like Christine, he received no genetic counseling or formal informed consent. The results of Bill's test were negative.

Twenty weeks into the pregnancy an echogenic bowel – a possible indication of CF-- was discovered upon ultrasound of the fetus. A reexamination of Bill's test results showed that he was tested for only 10 common CF mutations, although testing was available for 70 mutations. Only at this point did the couple attend their first genetic counseling session. As a result of that session, Bill was retested using a lab that looked for 70 mutations and an amniocentesis was done on the fetus.

The pregnancy was at 22 weeks when Bill's test came back positive for the rare V520F mutation, which accounts for less than 1% of CF mutations among Caucasians. One week later, results of the amniocentesis confirmed that the fetus was affected with CF. According to state law, the couple had one week to decide whether or not to terminate the pregnancy. After an intensive period of research about CF and visiting with patients and families at a local CF clinic, Christine and Bill decided to carry the pregnancy to term.

Note: *As of December 2001, over 900 mutations of the gene responsible for CF have been identified. The American College of Medical Genetics currently recommends the use of a standard 25 mutation panel for routine carrier testing for CF. Mutation detection rates will vary among different population groups. Negative test results reduce, but do not eliminate the risk of being a carrier. For more information on molecular testing for CF, see* **www.geneclinics.org.**

1. In the program, the following comment is made regarding clinical discussion about genetic screening and other prenatal tests: "One of the difficult jobs that primary care providers have is to take something that seems very abstract and unlikely to happen, and not scare somebody, because it is still unlikely, but to make it real enough that the patient [sic] really is making a decision about the information that genetic test has in relationship to the health of their baby. Because we all want to have a healthy child, we protect ourselves by assuming...our children won't be affected. So we are up against...normal coping mechanisms when we...ask people to consider rare and unlikely and frightening events."

How would you explain prenatal CF testing to a pregnant woman? What are some effective ways to discuss probabilistic information with patients?

2. According to Christine, her OB-GYN suggested she "might want to see a genetic counselor." Should Christine's physician have been more directive about referring to genetic counseling after she tested positive as a carrier for CF? What impact could that have had on this case?

3. The NIH has recommended that the offer of CF testing be phased in over time for couples currently planning a pregnancy as well as couples seeking prenatal testing even if there is no positive family history. Is it feasible in a primary care setting to address issues of prenatal screening with patients before a pregnancy? What might trigger such a discussion?

4. Would you offer prenatal genetic testing to a couple whom you know would not consider terminating a pregnancy? Why or why not?

5. In trying to decide whether to continue the pregnancy, Christine and Bill made a point of meeting children with CF and their families, to get a better understanding of what it was like. The genetic counselor tried to introduce them to a variety of cases, but stated that it wasn't easy to get an accurate view of life with CF from a few meetings, particularly given the variability of expression of CF. Do you think interaction with people living with a genetic disorder is a good way to support patient decision-making? How might it work in your clinical setting? Are you aware of support groups or advocacy organizations in your community for families with specific genetic disorders?

General Discussion Topics

Genetics in Primary Care: The Challenge

Even when referral to a genetic counselor or another genetic specialist is an option, a primary care clinician will often be the first to discuss genetic testing and/or genetic counseling with a patient. How this initial discussion is handled can have a large influence on the patient's attitudes and expectations about testing, which is particularly important, given that the testing decision is often a personal one. Consider the following:

1. When, if ever, is it appropriate to be "directive" about whether a patient should be tested? Should it depend on whether treatment is available for the disorder in question? On how well the provider knows the patient and/or family?

2. What are some of your own personal values and biases that might influence your management of patients with regard to testing? (Pregnancy termination, the inherent value of information, disclosure to family, attitudes about disability and chronic disease?)

3. Are there institutional, cultural, or community biases about genetic testing that may be embedded in your clinical setting? How might they affect patient care?

4. As was asked during the roundtable discussion segment, "...how do we draw the line between a ...non-biased attitude, ...respecting the patient's choices ...versus ...the responsibility ...to protect our patients from misinterpretation, misinformation and harmful bad choices?" When might it be appropriate to recommend testing? To recommend not testing?

5. Genetic counseling is likely to be limited to a few sessions at most, while we as primary care providers will continue to see the patient over time. In addition, as we saw in the CF case, even highly educated patients do not necessarily know what to expect from genetic counseling or understand how it can help them. How can primary care providers prepare patients in advance to derive the most benefit from limited genetic counseling sessions?

6. In focus groups for this program, primary care physicians said they did not know much about what genetic counseling entails. What do you need to know? Where can you obtain this information? What are the key components of an effective collaboration between primary care and genetic counseling (if available) in your setting? What kinds of improvements could be made?

Informed Consent: Provider vs. Patient?

1. There is growing concern that noninvasive, procedurally simple prenatal tests, including those that look for multiple genetic markers in a single sample of blood, will become widely used without regard for the impact and meaning of the results on individual patients and on society. As prenatal genetic testing becomes routine, in what ways can a busy primary care or obstetrics practice realistically implement a more deliberative informed consent process for genetic screening?

2. In a 1995 article about informed consent for prenatal testing, Press and Browner discuss an inherent conflict between the bioethical and legal interpretations of informed consent.***** They describe research on the offer of the maternal serum alpha-fetaprotein test to prospective mothers in California which indicated that the moral or ethical dimensions of screening --including decision-making about abortion—were not covered in the informed consent, just as they were not covered in the CF case in this program. Women in the study were not given information about the potential physiological, emotional or familial effects of the conditions covered by the test. Rather, the testing was characterized as an unexceptional part of standard prenatal care. The result of this omission was to maximize patient consent and minimize patient refusal of the test, therefore minimizing legal risks, such as wrongful birth suits, for providers.

What are some strategies that could be implemented to effectively manage the legal risks while ensuring a truly educational process and allowing for meaningful deliberation with your patients?

***(Press N, Browner CH. Risk, autonomy, and responsibility. Informed consent for prenatal testing. Hastings Cent Rep.1995;May-Jun; 25;(3):S9-S12.)*

Privacy / Confidentiality / Duty to Warn

1. Several physicians interviewed during research for this program said they might opt to protect a patient's privacy and confidentiality by keeping test results out of their charts or using a private code. What is driving some physicians to this approach? What are the dangers of it, and what alternatives are there for protecting privacy and confidentiality of genetic test results?

2. How do privacy/confidentiality concerns about genetic testing compare to privacy/confidentiality issues that arose in the early days of HIV testing? To HIV testing today?

3. Given the life-threatening potential of familial adenomatous polyposis (FAP), which has a penetrance of almost 100%, and the availability of screening and effective treatment, would you feel a duty to warn other family members if your

patient tested positive for an APC mutation? How would you handle a patient who says they do not want to share their APC gene test results with any family members? Does your responsibility or strategy change if those family members are also your patients?

4. In the roundtable discussion, Dr. Penchaszadeh said he would refer a patient to another physician before they underwent genetic testing for a serious disorder, if the patient did not want to warn potentially affected family members of a positive result. Is this appropriate? Feasible?

Communication Techniques for Informed Patient Decision-making

1. Given the wide variability in levels of education, scientific understanding, religious and cultural beliefs, expectations of individual patients, and individual coping styles, providing adequate discussion for informed consent about genetic testing in the limited amount of time available is certainly a challenge. Could the following be useful in your clinical setting? Discuss their feasibility and possible alternatives:

- Use of existing support staff
- Use of sequential visits for discussion and deliberation over time
- Use of questionnaires to be answered at home to help patients obtain accurate family histories and clarify their personal beliefs, needs and expectations
- Use of patient waiting time for orientation and deliberation, supported by print, video or computerized software
- Use of lists of the common pros and cons of testing for a particular condition for patients to consider
- Other patient education materials, including internet-based education and visual aids to make complex probabilistic information more accessible
- Exposing patients to families already living with a particular condition as was done in the CF case to support decision-making about testing, pregnancy and/or treatment.
- Use of referral database of genetic counselors and other specialists in your area

2. A positive test result for mutations such as BRCA 1 or 2 means that one has a predisposition for breast or ovarian cancer rather than a diagnosis of cancer. But a positive result can still change a patient's sense of self or perceived health status within the larger community, reclassifying her or him from being healthy to "at risk."

Other unintended side effects of genetic information include insurance or employment discrimination and unwanted or unexpected information about paternity. How can we communicate about psychological and social side effects of testing to make sure that their potential impact will be carefully considered by patients before a testing decision is made?

3. Is it feasible, as discussed in the roundtable, for primary care clinicians to ask a patient to consider various scenarios and imagine how he/she might feel or what might happen in the family? Consider the utility of the following:

- Explore with patients how they have reacted to stressful events in the past
- Explore with patients their tolerance for ambiguity
- Discuss with patients the variety of feelings and experiences that others have had with testing
- Ask patients to discuss exactly what their expectations are and what they plan to do with the genetic information, medically, personally, and within their families

Selected Resources for Providers and Patients

Mainly for Providers: (*entries are for patients also)

American Medical Association (AMA)
http://enet.ama-assn.org/public/cme/careg.htm

Continuing Medical Education Medical Genetics Series: Identifying and Managing Hereditary Cancer Risk.

****Genes and Disease***
www.ncbi.nlm.nih.gov/disease/

Series of web pages dedicated to specific diseases, providing short synopses for medical professionals and/or patients/families. Prepared and maintained by the National Center for Biotechnology Information (NCBI), National Library of Medicine, National Institutes of Health. Links to relevant sources.

****GeneClinics™ and GeneTests™***
www.geneclinics.org or www.genetests.org

Expert-authored, peer-reviewed resource for clinical information. Concise descriptions of specific inherited disorders and current information on role of genetic testing in diagnosis, management and genetic counseling of patients with inherited conditions. Updated weekly. Funded by NIH. HRSA and DOE. Developed at University of Washington, Seattle.

GeneLetter
www.geneletter.org.

Online magazine with daily news, monthly features about scientific, medical and bioethical issues surrounding genetics and a journal watch feature. Authors and guest columnists include leading specialists in human genetics.

Genetic Testing for Cystic Fibrosis: NIH Consensus Statement;1997
http://odp.od.nih.gov/consensus/cons/106/106intro.htm

Genetics and Your Practice: CD ROM and Professional Services
www.modimes.org.

A set of professional services offered through March of Dimes including a self-paced, interactive CD ROM for provider education (CME available) as well as live presentations at conferences, meetings, or at your site; a comprehensive print curriculum; an online list of genetic service providers and a resource list. CD ROM order #09-1177-99, $9.00. (800) 367-6630.

The Hastings Center
www.thehastingscenter.org

An independent, nonprofit and nonpartisan research organization addressing fundamental ethical issues in health, medicine, and the environment. Developed and produced the Genetic Dilemmas project. *The Hastings Center Report,* a bimonthly journal with special supplements contains a wealth of material about genetics. See website or call (845) 424-4040 for list of relevant publications.

****Human Genome Project Information***
www.ornl.gov/hgmis/

A wide range of useful information about the HGP including Research, Education, Medicine, and of particular relevance to this program, Ethical, Legal & Social Issues.

National Coalition for Health Professional Education in Genetics (NCHPEG)
www.nchpeg.org

National effort to promote professional education and access to information about advances in human genetics. Comprised of an interdisciplinary group of leaders from over 100 diverse health professional organizations, consumer and voluntary groups, government agencies, private industry, managed-care organizations and professional genetics societies.

Online Mendelian Inheritance in Man (OMIM)
www.ncbi.nlm.nih.gov/Omim/

Presents current knowledge of all known human genetic diseases, based on more than 30 years of work directed by Victor A. McKusick and colleagues at Johns Hopkins University. Linked to a variety of sites and current, related literature on PubMed.

Mainly for Patients and Their Families:

Alliance of Genetic Support Groups
http://geneticalliance.org
(202)966-5557. Helpline: 1-800-336-GENE

An international coalition of more than 300 consumer and professional health organizations. Supports individuals/families with genetic conditions, educates the public and advocates for consumer-informed public policies. Special section on issues, including ethical, legal and social implications and genetic discrimination.

Cystic Fibrosis Foundation
www.cff.org/
(301) 951-4422 or (800) FIGHTCF (344-4823)

IMPACC (Intestinal Multiple Polyposis and Colorectal Cancer)
email: **impacc@epix.net**
(717) 788-1818. P.O. Box 11, Conyngham, PA 18219

A support group for families with FAP and/or hereditary colon cancer.

The Johns Hopkins Colorectal Cancer Registry
www.hopkins-coloncancer.org
(888)772-6566

One of several registries in the US for patients/families with a diagnosis of inherited colon cancer. Helps identify people at risk within a family, provides information, education and referral to genetic specialists.

National Cancer Institute Public Inquiries Office.
http://cis.nci.nih.gov
1-800-4-CANCER.

A free national information and education service about cancer.

NOAH: New York Online Access to Health, Genetic Disorders page.
www.noah-health.org

A bilingual English-Spanish resource for consumers with information from a variety of sources about genetics, genetic testing and specific diseases.

Journals (Theme issues and Articles)

British Medical Journal Special Issue on Genetics in Primary Care.
April 28, 2001; vol 322.

JAMA Theme issue on Human Genomics/Genetics.
November 14, 2001; vol 286: no.18.

Science Special Issue on the Human Genome.
February 16, 2001; vol 291.

The Genetic Resource Special Issue
Optimizing genetics services in a social, ethical and policy context: suggestions from consumers and providers 1996;10(2):1-107. (www.nergg.org). (617)243-3033. $10.00. Video also available.

Collins FS, McKusick VA.
Implications of the human genome project for medical science.
JAMA 2001;285:540-4.

Emery J.
Is informed choice in genetic testing a different breed of informed decision-making? A discussion paper. *Health Expect* 2001;4:81-86.

Geller G, Botkin J, Green M, et al.
Genetic testing for susceptibility to adult-onset cancer.
JAMA 1997;277(18):1467-1474.

Parens E, Asch A.
The disability rights critique of prenatal testing: reflections and recommendations. *Hastings Cent Rep* 1999;29(5):S1-22.

Press N, Browner CH.
Risk, autonomy and responsibility. Informed consent for prenatal testing. *Hastings Cent Rep* 1995;25(3):S9-12.

Reyna V.
Genetic testing and medical decision making. *Arch Intern Med* 2001;161:2406-2408.

Books

Baker, Diane L., Jane L. Schuette, Wendy R. Uhlmann, eds.

A Guide to Genetic Counseling. John Wiley and Sons, 1998.

Gelehrter, Thomas, Francis S. Collins, David Ginsburg.
Principles of Medical Genetics. 2nd edition. Lippincott, Williams & Wilkins, 1998.

Horwitz, Marshall.
Basic Concepts in Medical Genetics. McGraw Hill, 2000.

Touchette, Nancy, Neil A. Holtzman, Jessica G. Davis, Suzanne Feetham.

Toward the 21st Century: Incorporating Genetics into Primary Health Care.
Cold Spring Harbor Laboratory Press, 1997.

About The Hastings Center...

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