

Collins Medical Trust Pilot Project

“New Tools for Family Physicians: Enabling Informed Decisions about Ethical and Social Issues Raised by Genomic Medicine:”

Advisory Committee Responses to Four Project-Related Questions:*

1. Which genetics topics and related ethical issues do you encounter most in your clinical work?

- Advanced maternal age (to do amniocentesis or not to determine if there are genetic abnormalities and if there are, to terminate the pregnancy or not).
- Cystic fibrosis screening in pregnancy.
- Risk of certain cancers, like colon cancer, and educating people regarding the need for screening and maintaining their compliance with that screening. (OAFP Advisor #1)

- Developmental delay, prenatal screening issues (abnormal quad screens, FAHx heart defects, FHx of mental retardation cystic fibrosis, thalassemias, sickle cell)
- Asthma question CF, hearing loss, cardiovascular issues, cancer syndromes.
- Ethical-prenatal testing—to do or not and if positive, to continue pregnancy or not.
- Older-test or not for Alzheimer’s Huntington’s breast/ovarian cancers.
- Hemochromatosis, especially with abdominal pain. (OAFP Advisor #2)

- Issues around paternity
- Inherited risk of breast, ovarian and other cancers.
- Confidentiality of genetic testing results—who has a right to know the results?
(OAFP Advisor #3)

- Prenatal screening (for chromosomal anomalies and other defects such as cystic fibrosis.)
(OAFP Advisor #4)

- Knowing how to advise patients regarding their personal risk of cancer based on family history, and knowing who would benefit most from genetic counseling and/or screening
(OAFP Advisor #5)

1. Breast Cancer – BRCA1/2 gene mutation
2. Alzheimer’s & ApoE testing
3. Genetic engineering of foods
4. Hereditary forms of colon cancer
5. Prenatal screening for various inherited diseases (OAFP Advisor #6)

1. Prenatal care issues, such as cystic fibrosis, sickle cell
2. Breast cancer/ovarian CA concern
3. Hemachromotosis
4. When to test and who should pay (patient v. government v. third party)
5. Insurance company issues and potentially not accepting Pts based on info from genetic testing
6. If other family members should be made aware of information obtained on genetic info of another family member, it may affect them (OAFP Advisor #7)

-Alzheimer Dementia, genetic counseling for family members, Huntington disease (OAFP Advisor #8)

2. Under what circumstances does the need to discuss genetics with patients most often arise? Who initiates the discussion?

-Usually the need arises during a planned visit and usually I initiate the discussion.

-Initiated by physician or sometimes by patients who read things on Internet.

-In context of annual exam or when discussing family history.

-For kids, when developmental delay is picked up.

-During a pregnancy—I most often initiate but sometimes the pt does if they have a specific question

-Parent or sibling of a pt of mine is diagnosed with breast cancer – pt initiates.

-Family histories of cancer such a breast, ovarian, colon as well a prenatal or preconceptual screening. I usually initiate the question.

-During prenatal care, either in response to a patient concern, or in response to an abnormal triple screen.

-Also during the treatment of a family member for a potentially genetically acquired illness, most often cancer.

-Patients usually initiate the discussion in a non-urgent situation, often based on excessive fear of cancer (i.e., my Great Aunt had colon cancer and I have heard that it is hereditary)

-Breast cancer screening - I have a large well-female population, and a good majority of them bring up genetic testing for “the breast cancer gene” at their well-woman exam when we start t talk about mammograms. Usually these women have some first- or second-degree relative with breast cancer, and are terrified of acquiring the disease. We spend a good amount of time talking about the BRCA 1 and 2 genes, and what is involved in genetic testing. Occasionally, I send these women to see a genetic counselor.

-Living in Oregon, many people are very health-conscious & environmentally aware. I get asked about genetically engineered foods almost once a day – specifically, patients want to know the potential cancer risks associated with consuming such foods over a lifetime.

-A topic I often initiate with my pregnant patients (or ideally, if a patient is desiring pregnancy) is one of genetic screening for inherited diseases. Most of my clinic patients are currently Caucasian, so we talk more about cystic fibrosis than about Tay-Sachs, for example. I have not encountered anyone desiring karyotyping for the purpose of sex selection in Oregon, but when I was working in Chicago, this was a much bigger topic & had a number of patients desiring a male infant.

-When establishing care with a new pt, I always ask about family history. During this history taking, sometimes the need to discuss genetics comes up, usually initiated by me, as a physician

-During prenatal care, the issues of inherited illnesses are part of standard questionnaire and initiated by the physician

-Pts with a strong family history of cancer sometimes will initiate discussion of genetic testing (i.e., is there genetic testing available?) Usually these are the well-publicized cancers like breast, ovarian, prostate

-The topic comes up when the risk factors for developing AD (Alzheimer Dementia) is

discussed, and this leads to a family member asking, what is their risk of developing the disease? I bring up the genetic connection, and the subsequent question about the risk follows

3. In your experience, what are the barriers to discussing the ethical, legal, and social implications (ELSI) of the new genetic technologies with patients? How relevant are such discussions to providing good care?

- Usually the greatest barrier is education, both on the part of the patient and myself. The education piece for the patient is understanding enough science to understand the topic at hand. For me, it's knowing that is out there and how reliable and quick the testing is. I think this is fundamental to providing good care. If I can't communicate to the patient at a level he/she understands or if he/she can't grasp the concepts I'm presenting, then there's no way the discussion can continue.
- Time. Patients expecting physicians to guide their decisions as opposed to genetic counselors trained to completely let patient make choice.
- Lack of physician knowledge of latest info to give patient all pros and cons.
- State by state insurance discrimination differs
- Inability to quickly access latest info—many Web sites, not all comprehensive.
- No reimbursement for counseling—usually takes longer than 15-minute visit.
- A “negative” genetic test may just mean that the mutation or gene is not yet discovered, not that the patient does not have the illness.

- Lack of understanding (on both MD and pt) of what genetic tests are out there and what they are and are not capable of finding out.
- Lack of understanding of just what these “ELSI's” are.
- If a genetic test is under consideration the ELSI are hugely important.

- My own knowledge about the types of testing available; the test's accuracy; the subsequent need or desire for patient's to share this information with others in their families; the uncertainty about how insurers and employers may view such information.
- It is critical that the primary care provider be able to knowledgeably discuss these implications. If he and she can't, then I don't think that the patient has been afforded informed choice/informed consent about having or not having the testing done.

- I feel poorly equipped to adequately advise patients of their actual risks based on current knowledge.
- I also am under-educated in advising patients of current privacy laws and addressing their concerns about their genetic privacy.
- I also need additional information about current concerns regarding genetically modified foods.

- I don't feel there are barriers to discussion. I feel the barriers exist if one wishes to take action on the discussion. The most common is by far insurance. Will insurance pay for the counseling and/or testing, and furthermore, will patients be insurable after this testing? Many of my patients cannot or can only barely afford health insurance now, and are constantly changing jobs, changing and/or losing health insurance. Thus, this issue of insurance and insurability at premiums that are reasonable for a low-to middle-class family become real issues.

- Limited time during appointments to discuss the full ramifications, risks v benefits of testing
- Having resources/ability to perform full patient education versus pt's reliance on media and public hysteria in making decisions

- Lack of training/education for most physicians to discuss with pts each specific test
- Such ELSI discussions are extremely important for providing good care given that the info can alter one's life dramatically. Moreover, with the healthcare situation in America like it is today, it is important that we not overburden the system with expensive use of tests that have not been well established in their helpfulness to patients.

- What is going to be the psychological impact on the family member knowing that they may be at some risk? This is the barrier which I am not comfortable with. Is the family member better off not knowing their risk status, especially from a genetic standpoint, where we as physicians cannot change the risk? Is that sound practice?

4. What, specifically, is needed on the physician education/training side to help overcome these barriers?

- More information on what is available, what it means to the patient, and how to talk about genetics geared towards multiple education levels.

- Reimbursement

- Motivation for physicians to do this—ease of access to expand knowledge, test forms that give knowledge and are used to order tests, results that are interpretable (CDC and Mt. Sinai working on this already)

- Education about the capability of current genetic testing.

- Case discussion of ELSI of genetic testing

- Understanding the diseases, the screening and diagnostic tests available and when to screen.

- Knowledge about how to present the tests to patients and how to discuss the implications of testing—for positive/negative-equivocal results.

- How to talk to others in the patient's family, if requested to do so, about screening or the results of a specific test.

- how to manage the office systems to ensure excellence in patient care is offered in this area.

- How to follow and appropriately consult about patients at any phase of the process, from information gathering and testing to follow-up.

- Reliable, UNBIASED, easy-to-access information.

- CME that fits into my schedule (online would be great) and high quality patient information.

- I think direct education about the specific genetic tests available, who needs to be tested (i.e., for BRCA testing), how much the tests cost, and most importantly, what information will be gained from the specific test. Will the test give me and my patient a yes/no answer, such as the genetic test for Huntington's Disease, or will the answers be fuzzier, like the increased risk for Alzheimer's and ApoE? I still believe many physicians are cloudy on what the tests can really tell us. After we are informed, we can talk to patients about what it may or may not mean for them to be tested.

- The issue of insurance and insurability is much larger than the scope of this Questionnaire. I can only hold out hope for some form of universal health coverage in the future.

- Handouts/websites for physician education regarding proper use of tests, issues to counsel about specifically for each test. These resources should be easily accessible, easy to understand, and to the point (i.e., dos not take a lot of time to understand and get through)

-Place to refer pts to for more in depth counseling if a physician feels that he or she does not have the time or ability to sufficiently counsel pts, or patient handouts discussing the issues

-Dealing with uncertainty in a positive manner

***See list of OAFP project advisors for physician identifiers.**