

**Collins Medical Trust
Pilot Project**

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

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

As a genetic counselor, the most common ethical topics & issues relate to concerns patients have about confidentiality & discrimination; I see a severe chasm between what patients fear and what is likely to happen in the way of experiencing discrimination. This prevents using genetic information to improve health, and prevents patients from sharing genetic information appropriately with their care providers and family members. Other ethical issues relate to access – I may see the value of a genetic test but the insurance provider does not view that it is medically necessary; there is also a lot of confusion among patients & providers about genetic information vs. genetic testing and the differences/similarities of ethical concerns/issues in these areas.

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

Every patient I see is here to discuss the condition in them, their child or their family. I can perhaps best answer by saying that most of our pediatric patients are referred by their doctor, which tells me that PCPs serving this population are pretty good at recognizing the clinic features in a patient that suggest the need for genetic evaluation & referral. Among our adult patients, many are self referred or referred by their PCP because of a concern they raised with their PCP rather than their PCP raising the issue; most of these are because of a family history of cancer or a known genetic condition for which the patient may be at risk or at risk to pass on to children. There is often confusion on the part of the PCP and/or the patient about who needs to be referred for what kind of genetic service.

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?

The main barriers I recognize are time and lack of clarity about the clinical utility of such discussions. While a clinician may recognize that a patient who previously had a child with a chromosome abnormality may be at increased risk to have another affected child, it may not be clear whether, when, and in what detail discussing alternative reproductive options to avoid having another affected child and the attendant ethical issues to those options is important or relevant. A clinician may recognize that a patient with breast cancer and a family history of cancer may have an inherited cancer syndrome to explain the cancers in the family and may recognize that knowing whether there is a BRCA mutation is important to her children. However, the clinician may not appreciate the importance of knowing whether a cancer is due to a germline mutation is also extremely relevant for the patient's immediate medical decisions about cancer treatment due to differences in risks for future cancers of various types. The ethical, legal & social implications of genetic testing in that patient are important but may be less urgent than an understanding of the immediacy of the need for the genetic information to optimize cancer treatment. However, the ELSI issues in the patient's sister, who does not have cancer, have a different relevance.

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

I think that the physicians can better identify some barriers – I am sure that time is a major one. I have been impressed in my contact with PCPs, that the level of interest in genetics is high. I think specificity is an important need – when is it important to discuss these issues and for whom?