How to Use This Curriculum

The modules in this curriculum include teaching cases in 8 topic areas:

- Breast/Ovarian Cancer
- Cardiovascular Disease
- Colorectal Cancer
- Congenital Hearing Loss
- Dementia
- Developmental Delay
- Iron Overload
- Ethical, Legal and Social Issues (ELSI)

Each teaching case includes questions and discussion to illustrate genetics themes and diagnoses. Each module also includes references to key documents and useful web sites for additional background information. The 8 major topic areas were chosen because they are core areas of primary care practice and because they could illustrate a range of genetic issues relevant to primary care practice. ELSI is presented as a separate module, but is also incorporated into the other modules. We took this approach because of the attention that has been given in both popular and scientific press to the broader social implications of genetic medicine. We assume that many of the broader issues raised by genetics have counterpoints in primary care practice; the ELSI cases are intended to provide a basis for identifying these common themes. The discussion provided for the teaching cases are intended as a guide to the teaching points the case raises rather than as a an exhaustive review of the topic or a replacement for critical literature review. We expect that you will identify additional points that should be made and useful variations on the themes for each case.

The case-based approach, and the concept of teaching materials serving as a bridge between primary care practice and genetics, emerged from discussions of the GPC Advisory and Executive Committees in September 1999 and March 2000. Based on these discussions, we sought to provide materials that:

- represented patients who would be recognizable to primary care trainees,
- were adaptable to different teaching settings, and
- emphasized connections between genetic and primary care practice, in terms of both clinical skills and philosophy of practice (for example, the value of taking a family history; the importance of evaluating tests in terms of patient outcomes; and the core value of respecting patient preferences).

The teaching cases should be viewed as raw material, to be used in any combination or setting that proves useful. They can be used alone or as an accompaniment to didactic material, lectures or seminar discussion, either on a given topic area (e.g. colorectal cancer, developmental delay) or on a particular theme (e.g. patient confidentiality, preventive care).

In developing these materials, we used a framework for integrating genetics issues into primary care practice that emerged from discussions of the GPC Advisory and Executive Committees. The underlying themes of the framework concern the distinction between "thinking genetically" - the concept that a genetic cause will not be found unless it is considered as a possibility - and "acting genetically" - the recognition that a decision to pursue a genetic cause in a particular patient should be determined by its likelihood (estimated from the patient's presentation, and the prevalence of the genetic condition) and the value of the genetic diagnosis in caring for the patient (its treatability, the prognostic or management information provided by the genetic diagnosis, the implications of the genetic diagnosis for the family). This framework identifies the following elements as important to the appropriate use of genetics in primary care:

1. Defining populations at risk/prevalence
    - What genetic conditions are most commonly seen in primary care populations?
    - What indicators (in the patient or in the clinical presentation) increase the likelihood of a genetics explanation?

2. Determining the impact of genetic information on patient outcomes
    - To what extent does a genetic diagnosis lead to a specific preventive or management option?
    - When does genetic information provide unique prognostic information?
    - When/how can genetic information cause harm?

3. Relevance of mode of inheritance
    - Mode of inheritance is key to determining other family members at risk
    - When risk to other family members is high, there may be an obligation or a concern to help the patient inform other family members of potential genetic risk.

4. Cautious approach to genetic testing
    - Genetic tests may provide a means to make a definitive diagnosis, and may be particularly important in identifying inherited risk in family members of an affected individual.
    - However, many genetic tests are complex with limited sensitivity and/or specificity.
    - Needs for pre- and post-test counseling are often detailed, addressing issues of test interpretation (potential for ambiguous test results, as well as false positives and false negatives), implications of test results for clinical management and psychosocially, and implications of test results for family members.