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A large, semi-transparent purple-tinted image of a laboratory or classroom setting. In the foreground, a young woman with glasses is looking down at a book or document. Behind her, another student is visible, and in the background, a man with a beard is looking towards the camera. The scene is filled with books and laboratory equipment.

Report VI
**Contemporary Issues in Medicine:
Genetics Education**

Medical School Objectives Project

June 2004

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Preamble

Although genetic factors have been recognized to play a role in health and disease since the beginning of the twentieth century, until recently the contribution of genetics to medical practice was limited to a set of important, but rare, disorders. Recent events, especially the sequencing of the human genome, have introduced new approaches to diagnosis and therapy, and have broadened the scope of genetics to include common disorders and preventative strategies of public health significance. With the general recognition that genetic medicine will play a much larger – indeed, central - role in the work and knowledge of the practicing physician of the future, the challenge to medical education today is to determine the level of knowledge about genetics that students graduating from medical school need to acquire. Genetics cuts across all areas of medical practice, creating a challenge in providing coherent exposure and an opportunity to integrate learning across multiple disciplines. Furthermore, practical applications of genetics in medical practice are only beginning to emerge, and will likely mature at different rates in different areas. Genetic medicine, therefore, presents a rapidly moving educational target, with great promise, but relatively few examples of current application. Two groups, The National Coalition for Health Professional Education in Genetics and The Association of Professors of Human and Medical Genetics/ American Society of Human Genetics (NCHPEG and APHMG/ASHG), have given considerable thought to the core competencies required of the generalist health provider. The task here is to give that same consideration to the education of the general physician.



Introduction

The panel was tasked with 1) developing learning objectives for genetics education within the general institutional structure; 2) identifying educational strategies as vehicles to achieve the stated objectives; and, 3) giving consideration to the likely pace at which new applications of genetics in medicine are likely to unfold. The convention followed by other MSOP expert panels has been to identify a consensus about the appropriate attitudes, knowledge and skills for a selected topic, and suggest educational strategies.

Students and physicians should see medicine through a “genetic lens,” as suggested by Barton Childs, understanding that a person is the product of many things, including the environment, social influences, and genetics, the later having evolutionary, familial, and physiological components. Using this lens, a doctor should ask “Why is this person presenting for care for this illness or condition at this time?” and “What risks does this individual face that will determine future health problems, and what is the likelihood of successful intervention?”

Students have concerns about what their professional obligations are, and will be, regarding genetics. The question for medical educators is how to prepare students to exhibit appropriate professional attitudes related to genetic information and diagnosis. The paucity of role models within this field makes the transmission of professionalism, *particular to genetics*, difficult. This is further challenged by the reality that the paradigm by which genetics will influence medical practice is not yet written, so we face a likelihood of major change, yet the way that change will unfold is not known. Nevertheless, students must recognize the power of genetic information and the importance of incorporating such information into the appropriate care of patients, and for that, they must have the foundation with which to maintain currency and competence as the application of genetics to medical practice becomes ever more pervasive. The power and predictive element of genetic information make for a complex experience for patients, who will likely have to make decisions based on as-yet-unrealized potentialities expressed in the language of probabilities. As is certainly the case with other specialty fields, professionalism in genetics practice encompasses a substrate of skills from which to address ethical dilemmas and privacy issues uniquely raised by genetic information.

Historical backdrop

Individuality is a product of variation and the interplay between genetics, environment, and social influences. Understanding this interplay allows physicians to think about patients in a more sophisticated and useful way. Concomitant with an appreciation of the genetic basis of health and disease must be an appreciation and understanding of the historical context of genetics information and the past misinterpretation and misuse of genetics. The eugenics movement serves as the underpinning for contemporary misunderstanding and mistrust of genetics. Keeping the proper historical perspective will allow physicians to encourage patients to develop their own understanding of genetic information and its implication for their current and future health.



Attitudes

There is a genetic contribution to all common and rare disorders, as well as to overall health status. More important is the basic truth that the contribution of genetics knows no disciplinary boundaries, and as such affects all fields of medicine. The crosscutting nature of genetics and the diverse manifestations of genetic disorders challenge current structures of care and provide an opportunity to develop a collaborative, patient and family centered model. Students need to have an appreciation of the potential power of genetics to provide new paradigms for prevention, diagnosis and treatment. They also need to have respect for the limitations of the current knowledge base and an understanding of its risks and implications. Physicians should expect changes and advances to occur during the course of their entire professional careers.

Graduates should have an appreciation of the potential psychological impact of genetic information on their patients, as well as families and the community, and the key role of privacy issues. Privacy is no more important here than in any other field of medicine, but there is a heightened concern of patients about the potential volatility and misuse of genetic information, about which physicians need to be sensitive. Currently, much more is known about testing than treating what tests may reveal, and although this is not necessarily unique to the field of genetics, its effect is accentuated by the potential emotionality with which patients receive genetic information about themselves or family members. This is particularly challenging given public access to information and misinformation, the pro-

liferation of information systems, and the enormous range of patient capability to understand the information and the reliability of information sources.

Prior to graduation, a medical student should have demonstrated to the satisfaction of the faculty appreciation of the following:

- the potential for genetics to contribute to the development of new approaches to prevention, diagnosis, and treatment of disease
- the potential for genetics to expand understanding of the basic pathophysiology of all human disease
- the possibility of using a genetic approach to provide personalized health care with a much greater focus on prevention
- current limitations in the existing knowledge base
- that the principles for use of genetic information in decision making are largely the same as for other areas of medicine
- the rapidity of the advancing front of knowledge
- that genetic information may have implications not only for an individual patient, but also for a family, and in some cases an entire community
- the potentially disconcerting nature of genetic information particularly as it relates to interpretation of predictive tests
- the need to reduce public fear and misinformation about genetics information
- the diversity in public understanding of genetic information and evaluation of information sources
- the need for continued learning and receptivity to advances in knowledge and changes in medical practice

Knowledge

Genetics represents both a basic and a clinical science, with extraordinarily rapidly advancing fronts of knowledge and application. Students will require a knowledge base of fundamental principles and approaches to serve as a founda-

tion for continued learning. The science of genetics may be approached at multiple levels: the flow of genetic traits through families, the structure and function of the genome in cells, the forces at work in populations that mold gene frequencies, and the integration of genetics information into medical practice.

Prior to graduation, a medical student should have demonstrated to the satisfaction of the faculty understanding of the following:

- **principles of genetic transmission, including**
 - modes of single gene inheritance
 - chromosomes and chromosomal abnormalities
 - multifactorial inheritance and the role of genetic factors in common disease
 - concepts of penetrance, expressivity, pleiotropism, genetic heterogeneity, mosaicism, and new mutation
 - phenomena of imprinting and anticipation

- **molecular biology of the human genome, including**
 - structure and roles of major macromolecules involved in information transfer from DNA to protein
 - structure, function, and regulation of genes
 - organization of the human genome
 - nature and types of genetic variation, mutations, and polymorphisms
 - mechanisms of replication and repair of genetic information
 - structure and function of chromosomes and roles in meiosis and mitosis
 - basis for genotype-phenotype correlations
 - the role of genetic factors in health

- **principles of population genetics, including**
 - gene frequency and Hardy-Weinberg equilibrium
 - mutation and selection
 - polymorphism, single nucleotide polymorphisms (SNPs), haplotypes, and haplotype mapping
 - genetic drift and founder effect
 - consanguinity/inbreeding
 - concepts of genetic linkage and association





- **integration of genetics into medical practice, including**
 - role of genetic factors in determining rare and common disorders
 - importance of somatic genetic change in disorders such as cancer
 - use of population screening for disease risk or carrier status
 - use of cytogenetic, biochemical, and molecular genetic tests in diagnosis
 - use of microarrays, expression analysis, etc. in prognosis and therapy
 - role of genetics in determining response of individual to environmental factors or pharmacological agents
 - major strategies in prevention and management of monogenic and chromosomal disorders
 - role of genetics in modification of risk for common multifactorial disorders

Skills

Skills important to all physicians include the ability to take a multi-generation history and to appreciate that the family history is a dynamic and evolving source of information. The general skill set specific to genetics should include an understanding of the principles of risk calculation and the ability to recognize signs of genetic disease. Physicians also need to have the ability to interpret correctly the results of genetic tests and to use these properly as a component of medical decision-making.

Prior to graduation, a medical student should have demonstrated to the satisfaction of the faculty the following abilities:

- take a multi-generation history, recognize patterns of inheritance, and do basic genetic risk calculations
- perform a physical examination with special attention to signs of major genetic disorders
- recognize when to refer a patient for genetic screening, testing and counseling
- interpret results of genetic tests and explain them to patients and family members
- explain and obtain informed consent for genetic testing
- access and critically assess appropriate literature

Educational Strategies

Strategies should aim to provide tools for life-long learning. Too often genetics suffers from the misconception that it is an esoteric field, a perception that is perpetuated by use of obscure illustrations in the learning setting. It is critical that examples provide a broad view of genetics and are designed to impart the overarching influence of genetic factors in health and disease. Important, as well, is defining an evaluation strategy that will ensure that the objectives, as outlined in the preceding sections, are met by graduating students. Educational strategies include, but are not limited to:



Mechanisms

- Standardized patients, as well as panels of actual patients to discuss patient experience of genetic disease
- Presentation of problem based or real life cases and examples
- Integration of basic science and clinical medicine specific to genetics

Clinical Application

- Inculcate a culture where genetics is “seen” on wards and in clinics and is experientially based, by having students routinely look for genetic factors at work
- Identify basic mechanisms related to clinical decision making vis-à-vis genetic influences, i.e., why is this condition in this person being evidenced now?
- Use common illustrations so that genetics is not marginalized
- Students should learn to consult primary literature and on-line databases

Evaluation

- Maintain an on-line portfolio to demonstrate student knowledge over the 4 year curriculum
- Create curriculum map of genetics integration to demonstrate coherence of information by ensuring that components are identifiable and knowledge acquisition is “trackable”
- Genetics curriculum should include targeted information that engages learners at all levels



What Do Physicians Need to Know, and When Do They Need to Know It?

Advances in genetics have engendered high expectations of major advances both on the part of the public and within the medical profession. Although genetics may well effect a major transformation in the way medicine is practiced, for example by enabling an individualization of care and much greater emphasis on behavioral modifications geared to disease prevention, it is not clear when such changes will occur or what form they will take. This creates a special educational challenge, since the manner by which genetics will be integrated into medical practice is still largely undetermined. It is difficult to motivate students to learn things on the promise that they will be important in the future, and it is also difficult to find current case examples and role models. The integration of genetics into medical practice will likely occur gradually, and at different rates in different areas of medicine. In order to provide some guidance in this area, the panel has considered major types of advances that may be expected in three domains – prevention, diagnosis, and treatment – and three time horizons – when current students do their clinical rotations, when they enter residency, and when they begin practice.

What Do Physicians Need to Know, and When Do They Need to Know It?

| | Clinical Years Now | Residency 5 years + | Practice 10 years + |
|------------|---|---|---|
| Prevention | <ul style="list-style-type: none"> Newborn screening for inborn errors of metabolism and other disorders Carrier screening for hemoglobinopathies, lysosomal storage disorders, cystic fibrosis Presymptomatic testing for breast, ovarian, colon cancer Limited proteomic screening for cancer | <ul style="list-style-type: none"> Expanded newborn screening with tandem mass spectrometry Increased number of prenatal carrier screens Expanded scope of cancer screening and presymptomatic testing Limited use of screening panels for common disorders, such as cardiovascular disease or dementia | <ul style="list-style-type: none"> Wide array of disorders subject to newborn screening Routine use of proteomic screens for very early detection of common cancers Increasing use of screening for risk for common disorders to achieve risk stratification and implement prevention strategies |
| Diagnosis | <ul style="list-style-type: none"> High resolution cytogenetic analysis for constitutional changes and cancer Molecular diagnostic tests for limited number of monogenic disorders Prenatal diagnosis by amniocentesis and CVS | <ul style="list-style-type: none"> Use of microarrays to diagnose subtle chromosomal abnormalities Increasingly routine use of molecular testing for wide range of monogenic disorders Increasing use of expression microarrays in histopathological diagnosis Use of new modes of prenatal testing, such as preimplantation testing | <ul style="list-style-type: none"> Use of panels of molecular tests to stratify common disorders such as asthma or hypertension Routine molecular characterization of tissues in pathology Use of panels of tests to achieve precise diagnosis of monogenic and chromosomal disorders |
| Treatment | <ul style="list-style-type: none"> Limited pharmacological treatment of monogenic disorders (e.g., lysosomal disorders) Limited use of pharmacogenetic testing (e.g., TPMT) New forms of chemotherapy based on knowledge of cancer biology Experimental gene therapy protocols | <ul style="list-style-type: none"> Increasing array of monogenic disorders amenable to treatment Expanded panel of pharmacogenetic tests (e.g., CYP2D6) Increasing number of new cancer-specific therapies Continued experimentation with gene therapy Use of expression arrays to determine treatment strategies for certain diseases | <ul style="list-style-type: none"> Routine use of pharmacogenetic profiling Stratification of common disease and selection of specifically targeted therapies Limited routine use of gene therapy Use of expression arrays to determine treatment strategies widespread |

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