



**EPC Biotechnology, Genetics and Ethics Subcommittee  
Recommendations to EPC  
June 20, 2002**

Members: .....2  
 EPC Charge to Subcommittee: .....2  
 Operationalized Charge: .....2  
 Subcommittee Activities: .....2  
 Subcommittee Recommendations: .....3  
 Attachment A: American Society of Human Genetics (ASHG) Report from the ASHG  
 Information and Education Committee: Medical School Core Curriculum in Genetics  
 .....5  
 Attachment B: Genetics in Primary Care (GPC): A Faculty Development Initiative ..... 6  
 Attachment C: Curriculum Recommendations Chart .....7  
 Appendix A: General clinical skills that all students should master; however, achievement  
 of these skills is critical to an understanding of genetics: ..... 11  
 Appendix B: Current Curriculum Conditions, Presenting Situations, or Cases relevant to  
 Genetics ..... 13  
 Problems with familial tendencies that provide opportunities to cover genetics in the Family  
 Medical History ..... 13  
**Family Medicine** ..... **13**  
**Internal Medicine** ..... **13**  
**Neurology** ..... **13**  
**Obstetrics/Gynecology** ..... **13**  
**Pediatrics** ..... **13**  
**Psychiatry** ..... **13**  
**Surgery** ..... **13**  
 Inherited Disorders with known genetic/molecular mechanisms or specific genetic testing ..... 13  
**Family Medicine** ..... **13**  
**Internal Medicine** ..... **13**  
**Neurology** ..... **13**  
**Obstetrics/Gynecology** ..... **13**  
**Pediatrics** ..... **13**  
 Inborn errors of metabolism ..... 13  
 Chromosomal disorders—sex chromosomes ..... 13  
 Chromosomal disorders—Autosomes ..... 13  
 Genetic disorders ..... 13  
 Teratogenic agents ..... 13  
**Psychiatry** ..... **13**  
**Surgery** ..... **13**  
 Curricular issues ..... 14  
**Medical Humanities** ..... **14**  
**Current Assessment** ..... **14**  
**In cases or seminars now** ..... **14**  
**Other miscellaneous** ..... **14**  
 References ..... 15



## EPC Biotechnology, Genetics and Ethics Subcommittee Recommendations to EPC June 20, 2002

### Members:

David S. Steward, MD, Chair  
Terri Cameron, MA, Staff  
Peter Borgia, PhD  
Kimberly Espy, PhD

Jodi Huggenvik, PhD,  
David King, PhD  
Margaret Kovach, PhD,  
Michael Schneider, MD

### EPC Charge to Subcommittee:

*... to assess the effect of the biotechnology revolution on medical school curricula, to make curricular recommendations that address the realms of basic science (genetics, etc.), clinical application, and ethics (genetic discrimination, etc.), as well as the law and humanities related to the issues, and to assure the vertical and horizontal integration of biotechnology issues across the four years of the curriculum.*

### Operationalized Charge:

*Develop a set of curriculum recommendations for how genetics and topics related to genetics (ethics, counseling, etc.) should be presented to students. These recommendations should include a content outline and delivery and assessment methods. Past and current genetics curricular efforts at SIUSOM should be reviewed, as well as those at other medical schools. Recommendations from appropriate national, regional, and state groups should also be studied.*

*Recommendations developed by the group should be compiled into a report that includes:*

- What students need to know regarding Biotechnology, Genetics and Ethics*
- The curriculum activities necessary to ensure that students are exposed and learn to Biotechnology, Genetics and Ethics learning issues and clinical skills*
- How student knowledge, attitudes and skills regarding genetics should be assessed*
- The curriculum changes potentially necessary to ensure that students are exposed to and learn Biotechnology, Genetics and Ethics learning issues and clinical skills*
- The faculty or group of faculty responsible for oversight of Genetics issues across the four-year curriculum*
- A plan for faculty education to broaden faculty awareness of issues related to Genetics, to ensure that tutor guides for cases have appropriate Genetics learning issues, and to ensure that clinical faculty can assist students in making the connection between clinical activities and genetics issues*

### Subcommittee Activities:

The EPC Genetics Subcommittee met monthly from December through June, reviewing current genetics content, activities, and assessments in the SIUSOM curriculum, student performance on SIUSOM and national assessments such as USMLE and NBME subject tests, genetics content at other medical schools, and curriculum recommendations from national genetics groups (Attachments A and B).

Based on this review and discussions at subcommittee meetings, a chart of curriculum recommendations was developed (Attachment C), which outlines content to be covered, the year in which it should be covered, and the educational methods best suited for coverage.

## **Subcommittee Recommendations:**

Based on student performance in genetics content throughout the curriculum and historically on the USMLE, the EPC Genetics Subcommittee strongly recommends that genetics content receive increased focus in the SIUSOM curriculum. In a review of literature, two documents were found to be useful as templates for developing genetics curricula: *The American Society of Human Genetics (ASHG) Report from the ASHG Information and Education Committee: Medical School Core Curriculum in Genetics* (Attachment A) and *Genetics in Primary Care (GPC): A Faculty Development Initiative* (Attachment B).

### **1. Establish a mechanism to ensure comprehensive coverage of genetics content throughout the curriculum.**

The curriculum chart (Attachment C) outlines both the information the Subcommittee feels students should be exposed to and master during each year of the curriculum and the curriculum activities necessary to ensure that students learn this information. The Subcommittee did not feel that any overall curriculum change would be necessary to implement the curriculum recommendations; however, cases will need to be reviewed to ensure that there are sufficient core learning issues in the content outlined in the attached chart and that patient activities, seminars, and resource sessions are scheduled for content not appropriate for case learning issues. It was noted that, in addition to ensuring that genetics learning issues are included in PBLMs as appropriate, mini-cases could be used to focus on specific genetics learning issues across the four years of the curriculum. Appendix A and B provide additional resources for faculty implementing genetics content into their courses. Appendix A is a listing of general clinical skills that all students should master but that are critical to an understanding of genetics. Appendix B provides a series of conditions and presenting situations identified by clerkship directors as current or potential opportunities for emphasizing genetics content. This information should serve as a guide to faculty as they choose cases for Years 1 and 2 as a means of establishing core content for students to prepare them to recognize genetics issues in the clinical activities they will have a in Years 3 and 4.

Finally, as Genetics content is integrated fully into the curriculum, it may be necessary to decrease content in other areas to ensure that it is not simply added-on and therefore minimized in importance.

### **2. Ensure that Genetics learning issues are adequately assessed**

To ensure that genetics content receives appropriate emphasis in the curriculum, genetics learning issues should be assessed as part of each course's assessment and data regarding genetics curricular initiatives and student performance on genetics content is monitored and reported to the EPC on a regular basis. In addition, genetics should be assessed in performance-based examinations wherever appropriate and possible, such as during assessments that evaluate the family medical history. The current status of assessment of genetics content is included in Appendix B.

### **3. Ensure that ethical issues related to genetics content are discussed at every opportunity**

It is a philosophy of science that, where appropriate, ethics is always included as a major issue in any scientific endeavor. In particular, the genome project has included a discussion of ethics as part of all documentation related to the project. The foundation for teaching common ethic problems in genetics should be established early in Year 1 using cases and topical issues as a part of the Doctoring Curriculum. In addition, ethical issues that do not relate specifically to genetics should be included wherever possible in all case and patient discussions.

### **4. Develop a faculty development program that ensures that SIUSOM faculty have opportunities to keep abreast of the clinical and research activities in Genetics and can discuss those activities with students**

Since Genetics content is complex and constantly changing, a faculty development program that includes regularly-scheduled interdisciplinary conferences that focus on genetics content, including basic science, clinical, legal, ethical, and current research issues, should be established. Other opportunities for faculty development in Genetics could include CME events and departmental meetings. The person(s) charged

with oversight of the Genetics curriculum initiative could be charged with scheduling faculty development events that tie into curricular initiatives.

5. **Provide regular reports to the EPC regarding curriculum and assessment efforts, as well as student performance on internal and external Genetics exam content**
6. **Appoint Genetics Director to assume overall responsibility for Genetics in the curriculum**

A person or unit needs to be assigned oversight responsibility that includes systematically reviewing cases throughout the curriculum, making recommendations regarding learning issues and resource sessions necessary to support student Genetics content learning issues, monitoring curriculum and assessment activities and reporting on these activities to the Educational Policy Committee on a regular basis, serving as a resource to faculty for as they develop curriculum activities and assessments, and developing ongoing faculty development programs.

**Attachment A: American Society of Human Genetics (ASHG) Report from the ASHG Information and Education Committee: Medical School Core Curriculum in Genetics**

**Attachment B: Genetics in Primary Care (GPC): A Faculty Development Initiative**

**Attachment C: Curriculum Recommendations Chart**

KEY FOR FORMAT COLUMN RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)		Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?
		YEAR 1				YEAR 2				YEAR 3				YEAR 4			
KNOWLEDGE Objectives (Students should know:)																	
1.1	What genes are, how they are organized and controlled, what they do, and how they segregate	+	+	+	RS/SDL												
1.2	Nature of mutations and permutations and how they contribute to human variability and to disease	+	+	+	RS/SDL												
1.3	Patterns of inheritance characteristic of autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive traits	+	+		RS/MC												
1.4	Factors that affect development of the phenotype in single-gene disorders, including variable expressivity and incomplete penetrance	+	+	+	RS												
1.5	The basis of mitochondrial diseases and the expected pattern for mitochondrial (maternal) inheritance	+/-	+	+	RS												
1.6	How genes are organized into chromosomes, how chromosomes replicate in mitosis and meiosis, and how they are transmitted from parent to child	+/-	+	+	RS/MC												
1.7	Clinical manifestations of common numeric, structural, and mosaic chromosomal anomalies	+	+	+	RS/SDL												
1.8	The concepts and clinical importance of genetic imprinting and uniparental disomy	+	+		RS/MC												
1.9	The principles of population genetics and the public health implications of genetic epidemiology	+/-	+	+	RS												
1.10	How polymorphisms, gene linkage and human gene mapping are used in medicine	+/-	+	+	RS/MC												
1.11	The basic principles of inborn errors of metabolism and of pharmacogenetic variations and their general clinical manifestations					+/-	+	+	RS/SDL	+/-	+	+	RS/SDL				

<b>KEY FOR FORMAT COLUMN</b> RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)		Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?
		<b>YEAR 1</b>				<b>YEAR 2</b>				<b>YEAR 3</b>				<b>YEAR 4</b>			
1.12	Multifactorial nature of most human traits, both normal and abnormal, and the principles of multifactorial inheritance					+/-	+	+	RS/SDL	+/-	+	+	RS/SDL				
1.13	The mechanisms of teratogenesis and the effects of major human teratogens					+/-	+	+	RS	+/-	+	+	RS				
1.14	How to recognize and classify congenital anomalies and the approach to diagnosis of multiple congenital anomaly syndromes					+/-	+	+	RS/SDL	+/-	+	+	RS/SDL				
1.15	The role of genetics in the pathogenesis of neoplasms and in the predisposition to malignancies					+	+		RS/SDL	+	+		RS/SDL				
1.16	How evolutionary principles can be used to understand human biology and disease						+	+	RS		+	+	RS				
1.17	How disease frequency varies in different ethnic groups					+/-	+	+	SDL	+/-	+	+	SDL				
1.18	Common molecular and cytogenetic diagnostic techniques and how they are applied to genetic disorders	+	+	+	RS/MC												
1.19	The procedures available for prenatal genetic diagnosis and the kinds of disease that can be detected prenatally					+	+		RS/SDL	+	+		RS/SDL				
1.20	Appropriate methods of genetic counseling						+	+	RS/activity		+	+	RS/activity				
1.21	The advantages, limitations, and dangers of predictive testing for genetic disease					+	+	+	SDL	+	+	+	SDL				
1.22	The existence of and justification for screening programs to prevent genetic disease					+	+	+	RS/SDL	+	+	+	RS/SDL				
1.23	Approaches to treatment of genetic diseases (new gene therapies)					+/-	+	+	RS/SDL/MC	+/-	+	+	RS/SDL/MC				
1.24	Organizational and economic aspects of the health care system with regard to genetics						+	+	RS		+	+	RS				
1.25	Legal and ethical issues involved in the practice of medical genetics						+	+	RS/SDL		+	+	RS/SDL				
1.26	History of use and misuse of human genetics						+	+	RS/SDL		+	+	RS/SDL				
1.27	The genetics of antibiotic resistance and microbiologic virulence						+	+	RS/SDL		+	+	RS/SDL				
1.28	Immunological genetics						+	+	RS/SDL		+	+	RS/SDL				
<b>SPECIFIC SKILLS: Students should be able to:</b>																	

<b>KEY FOR FORMAT COLUMN</b> RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)		Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	
		<b>YEAR 1</b>				<b>YEAR 2</b>				<b>YEAR 3</b>				<b>YEAR 4</b>				
2.1	Elicit a comprehensive medical genetic history and construct an appropriate pedigree					+	+	+	MC/clinic		+							
2.2	Carry out a comprehensive physical examination for major and minor anomalies, with special attention to surface anatomy and anthropometric measurements							+			+							
2.3	Formulate an appropriate differential diagnosis and course of investigation, including the use of specialized tests through biochemical cytogenetic, and molecular genetic laboratories								MC/SDL/		+							
2.4	Effectively use information systems, including library and electronic resources, in the evaluation and management of patients with genetic diseases, the diagnosis of multiple congenital anomaly syndromes, and the recognition of teratogenic exposures					+/ -	+	+	RS/handout		+							
2.5	Understand cytogenetic, biochemical and molecular laboratory reports					+/ -	+	+	SDL		+							
2.6	Recognize the need for referral to a genetics specialist and/or counselor for specific conditions such as: Huntington's disease, breast cancer (high risk), ovarian cancer (high risk), colon cancer (high risk), multiple endocrine neoplasia syndromes, hemochromatosis, other heritable cancer syndromes, deafness, muscular dystrophy, mitochondrial disorders)					+/ -	+	+	SDL		+							
	<b>Management (students should be able to)</b>																	
2.9	Provide patients with access to diagnostic and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of such tests										+							
2.10	Make available to patients appropriate treatments, including dietary, pharmacological, enzyme-replacement, transplantation, and gene therapy										+							
2.11	Communicate genetic information in a manner that is suitable for each particular patient and family										+							
2.12	Understand the normal developmental stages of human behavior, maturation, and intelligence, in order to facilitate counseling of varying individuals										+							

	<b>KEY FOR FORMAT COLUMN</b> RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?
		<b>YEAR 1</b>				<b>YEAR 2</b>				<b>YEAR 3</b>				<b>YEAR 4</b>			
	<b>ATTITUDES (students should:)</b>																
3.1	Appreciate the importance of disease prediction and prevention										+						

**Appendix A: General clinical skills that all students should master; however, achievement of these skills is critical to an understanding of genetics:**

<b>KEY FOR FORMAT COLUMN</b> RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)		Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?
		YEAR 1				YEAR 2				YEAR 3				YEAR 4			
<b>KNOWLEDGE Objectives (Students should know:)</b>																	
A1.24	How novel scientific discoveries can be evaluated in a clinical context and can be applied appropriately to the care of patients						+	+	SDL		+	+	SDL				
<b>SPECIFIC SKILLS: Students should be able to:</b>																	
A2.1	Understand and be able to explain the informed consent process, realizing that this issue is more than just a signature on a form										+						
A2.2	Appreciate the role of biomedical research and develop techniques for critical analysis of current scientific developments						+	+	SDL		+						
A2.3	Coordinate information from multiple sources into a coherent and rational plan of management										+						
<b>Management (students should be able to)</b>																	
A2.4	Tolerate and encourage reiteration of information because of patient anxiety or unfamiliarity with the concepts being presented										+						
A2.5	Apply appropriate techniques for conveying bad news										+						
A2.6	Recognize patients' defense mechanisms and be able to determine when it is better to leave them intact and when they may need to be breached										+						
A2.7	Cope emotionally with responses of patients										+						
A2.8	Interpret their own attitudes toward ethical, social, cultural, religious, and ethnic issues and develop an ability to individualize each patient or family member										+						
A2.9	Resolve varying cultural, social, and religious attitudes in relation to issues such as contraception, abortion, parenting, and gender roles										+						

	<b>KEY FOR FORMAT COLUMN</b> RS: Resource Session MC: Mini-case SDL: self-directed learning (with or without PBLM)	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	Current	Future	Improve?	Format?	
		<b>YEAR 1</b>				<b>YEAR 2</b>				<b>YEAR 3</b>				<b>YEAR 4</b>				
A2.10	Utilize community support services and agencies appropriately																	
	<b>ATTITUDES (students should:)</b>																	
A3.1	Respect patients' religious, moral, and ethical beliefs and biases, even if they differ from the students' own beliefs																	
A3.2	Present all available options fairly, accurately, and nondirectively																	
A3.3	Be aware of both the importance of confidentiality and the difficulties that confidentiality poses when relatives are found to be at risk for a serious and potentially preventable disease																	
A3.4	Be aware of the advantages and potential hazards of referring patients and families to community or national resources																	
A3.5	Recognize the limitations of their own skills and seek consultation when necessary																	

## Appendix B: Current Curriculum Conditions, Presenting Situations, or Cases relevant to Genetics

### **Problems with familial tendencies that provide opportunities to cover genetics in the Family Medical History**

#### **Family Medicine**

- Tay-Sachs
- Congenital cleft palate
- Recurrent miscarriage/abortion
- Other problems as listed in other specialties

#### **Internal Medicine**

- Diabetes Mellitus, Type 1, and Type 2
- Coronary artery disease
- Hypertension
- Hyperlipidemia
- Abdominal aortic aneurysm
- Inflammatory Bowel Diseases
- Osteoporosis

- Autoimmune disorders (RA, SLE)

#### **Neurology**

- Epilepsy
- Inherited neuropathies
- Dementia/degenerating disorders

#### **Obstetrics/Gynecology**

- Endometrial cancer
- Osteoporosis
- Other routine family history
- Gestational diabetes
- Recurrent spontaneous abortion

#### **Pediatrics**

- Growth delay, developmental delay

#### **Psychiatry**

- Alcoholism
- Drug dependency
- Depression/suicide
- Schizophrenia
- Dementia

#### **Surgery**

- Inflammatory bowel disease
- Gall bladder disease
- Abdominal aortic aneurysm

### **Inherited Disorders with known genetic/molecular mechanisms or specific genetic testing**

#### **Family Medicine**

No additions

#### **Internal Medicine**

- Cystic fibrosis
- Hemochromatosis
- Alpha-1 antitrypsin deficiency
- Cancers—BRCA-1, BRCA-2, HNPCC
- Various therapeutics—HIV treatments; Her2neu in breast CA; rate of metabolism of drugs, others
- Thalassemia
- Sickle Cell Disease/Trait
- Hypertrophic Cardiomyopathy
- Hypercoagulable states (prothrombin gene mutation, Factor V Leiden)

#### **Neurology**

- Huntington's
- Inherited ataxia
- ? Alzheimer's
- Muscular Dystrophies (Duchenne, Becker)

#### **Obstetrics/Gynecology**

- Recurrent/spontaneous abortions

- Perinatal testing for various conditions ("Triple screen")(amniocentesis)
- Ovarian/Breast Cancer

#### **Pediatrics**

##### **Inborn errors of metabolism**

- Congenital adrenal hyperplasia
- MCAD deficiency

##### **Chromosomal disorders—sex chromosomes**

- Turner (XO)
- Trisomy X
- Disomy Y (XYY)
- Klinefelter (XXY)

##### **Chromosomal disorders—Autosomes**

- Down (Trisomy 21)
- Trisomy 13
- Trisomy 18

##### **Genetic disorders**

- Marfan's syndrome
- Galactosemia

- Phenylketonuria
- Hypothyroidism
- Hemophilia
- Achondroplasia
- Polycystic Kidneys
- Osteogenesis imperfecta
- Fragile X
- Prater-Willi

##### **Teratogenic agents**

#### **Psychiatry**

- Chromosomal disorders (Down, Fragile X)
- Others related to behavior

#### **Surgery**

- Sepsis response
- MEN syndromes (ret -protooncogene)
- Colon cancer and polyps
- Breast cancer
- ENT cancers
- Lung cancers

## **Curricular issues**

### **Medical Humanities**

- Confidentiality
- Informed Consent
- Bias (of all kinds) in Medicine
- General issues of insurability
- General medico-legal issues

### **Current Assessment**

- Some coverage in MCQ portions of exams in Year 1 and 2
- One case in the Pediatrics Clerkship
- Family medical history in Pediatrics Clerkship
- Some coverage in Surgery Clerkship exam
- Potential coverage in Family and Community Medicine Exam

### **In cases or seminars now**

- “Genetics Streamer” in Pediatrics Clerkship
- Some mention in Surgery Clerkship
- Possibly in psych—not known
- Possibly in medicine—in selected seminars, but unknown
- Mentioned in perinatal sessions in OB, but not clear exactly what.
- Mentioned in some seminars in neurology—Burns
- Covered in several areas in Surgery Clerkship — colon cancer, breast cancer, MEN
- Family medicine distance-learning cases might include some family history issues
- May also be covered in Family medicine in regards to counseling

### **Other miscellaneous**

- Complaint about lack of embryology
- Faculty development positively received but must be tailored to needs
- Faculty genetics czar well received, especially if offers support to curriculum.

- All CD's willing to expand but not wholesale—needs to fit into current system.
- Pediatrics, Obstetrics and Gynecology, and Psychiatry Clerkships are currently contemplating major changes in curriculum
- Internal medicine—flexible, and national clerkship curriculum known to be lacking in genetics.
- Genetics in Primary Care project—including the faculty development document.
- Suggestion that all seminars/case discussions be accompanied by a lit search related to “genome”
- Multidisciplinary grand rounds or conferences suggested—Logisitics may be an issue
- Role of referral, consultant—what does a genetic counselor do? Maybe covered in Pediatrics?
- ELSI issues not addressed directly
- No overview of the genome project

## References

- Liotta, Lance A., Kohn, Elise C., and Petricoin, Emanuel F. "Clinical Proteomics: Personalized Molecular Medicine," *JAMA* 286 (18): 2211-2213, November 2001.
- Phillips, Kathryn A., Veenstra, David L., Oren, Eyal, Lee, Jane, and Sadee, Wolfgang. "Potential Role of Pharmacogenomics in Reducing Adverse Drug Reactions: A Systematic Review," *JAMA* 286 (18): 2270-2279, November 2001.
- Hill, Adrian V S. "Immunogenetics and Genomics," *The Lancet* 357: 2037-2041, June, 2001.
- Taylor, Matthew, R. G., Alman, Amy, and Manchester, David K. "Use of the Internet by Patients and their Families to Obtain Genetics-Related Information," *Mayo Clinical Proceedings* 2001; 76:772-776, August, 2001.
- Korf, Bruce. "Medical Education in the Post-Genomic Era,"