



# Medical Genetics

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## **Departmental academic coordinator:**

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When you have questions, concerns, or suggestions, please contact the secretary to call for an appointment.

## **Course Description, Objectives, and Format**

Medical Genetics is one of the basic science courses that comprise the preclinical curriculum of the first three years of medical school. The overall goal of these courses is to provide students with the knowledge and understanding of the scientific principles that are the basis of current approaches to the diagnosis and management of disease. The application of these scientific principles and knowledge to the practice of medicine, including the development of life-long learning and problem-solving skills, is emphasized. This goal is consistent with the objectives of the United States Medical Licensing Examination (USMLE), Step 1.

The Medical Genetic course consists of 35 lectures and covers topics that are integrated with the concurrent Pharmacology, Pathology, Integrated Problems, and Introduction to Clinical Medicine courses.

Lecturers from both basic science and clinical disciplines teach the fundamental principles of genetic and how these principles apply to the diagnosis and treatment of these diseases.

### **Specific learning objectives for Medical Genetics:**

At the end of your undergraduate teaching you will be expected to be able to:

- Ø Recognize patterns of inheritance.
- Ø Have knowledge of several Mendelian and chromosomal conditions.
- Ø Recognize the genetic and environmental contribution to multifactorail conditions.
- Ø Learned approaches which can be used for the diagnosis of genetic disease and carrier detection.
- Ø Learned different forms of DNA testing: prenatal diagnosis, predictive testing, and diagnostic testing.
- Ø Be able to write a DNA report and chromosome report.
- Ø Be familiar with the practice of the genetic counseling clinic.
- Ø Know when and where to get genetic advice and information.
- Ø Be familiar with the major ethical issues.

### **Lectures**

Each lecture is accompanied by a PowerPoint presentation. Information from the presentation and assigned reading is important for mastering the learning objectives which are the primary focus of exam questions.

<b>Topics</b>	<b>Lectures</b>	<b>Hours</b>
Introduction	Distribution of marks, exams, and final exams The role of genetic in medicine Impact of genetic diseases Major types of genetic diseases	1
Revision	The structure of the cell DNA Chromosome structure	1

	Transcription and translation The genetic code	
Mutation	Mutagens and mutagenesis Mutations: types of mutations Structural effects of mutations on protein Functional effect of mutations on protein	3
<b>Exam</b>		
Patterns of Inheritance	Mendel or single gene inheritance Polygenic and multifactorail inheritance	4
Genetic disorders	Single gene disorders	1
	Biochemical genetics	1
	<b>Exam</b>	
	Genetic factors in common diseases	3
	Cancer genetics	2
<b>Exam</b>		
DNA techniques	Techniques of DNA analysis: Nucleic acid probes, Nucleic acid hybridization	1
	restriction mapping, DNA sequencing	1
	mutation screening techniques	1
	Diagnosis mutational analysis of single gene disorders	1
	Diagnosis of multiple gene disorders	1
	Cytogenetics	Chromosome abnormalities Numerical abnormalities
Structural abnormalities Mosaicism and chimerism (Mixoploidy).		1
Chromosome disorders: incidence of chromosome abnormalities		1
disorders of the autosomes disorders of the sex chromosomes		1
Methods of chromosome analysis: Karyotype analysis, FISH, CGH		2
<b>Exam</b>		
Clinical genetic	Carrier detection and presymptomatic diagnosis	1
	Prenatal and preimplantation diagnosis of genetic disease	2
	Genetic counseling	1
	Treatment of genetic diseases	3
	Ethical issues in Medical genetics	1

## Course Learning Objectives

By the end of this course, students will be able to apply their basic background in medical genetics to the practice of medicine, including the effective diagnosis, treatment and prevention of genetics disease. Detailed learning objectives are provided for each lecture.

## **Prior Knowledge and Skills Required for This Course**

The ability to fully comprehend and appreciate the fundamentals of medical genetics requires a background in basic biology, biochemistry, and immunology.

### **Roles and Responsibilities of Students and lecturers**

#### **Students are expected to;**

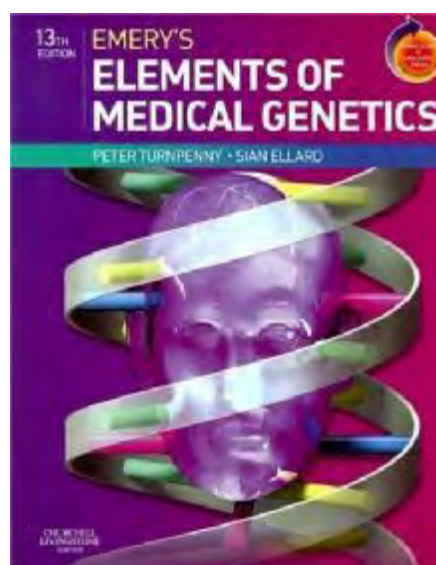
1. use all available resources to accomplish the learning objectives in each lecture and case-based discussion and exercise session, including:
  - a. attending all lecture and case-based discussion sessions.
  - b. reading textbook assignments.
  - c. participating in lecture and case sessions by answering questions posed in class and asking questions when information is unclear or more information is needed.
  - d. performing assigned exercises working individually or in groups, as directed.
  - e. submitting completed assignments on or before the stated deadlines for timely feedback.
  - f. optimizing their learning strategies by trying the suggested “tips” and/or other ideas, and working with others.
  - g. asking for help from the course manager when they need it or even think they might need it.
2. notify the course manager as soon as they can if they are seriously ill or have an emergency that prevents them from attending the case sessions or an exam.
3. provide constructive feedback regarding the course on evaluation forms that will be provided at the end of the semester.
4. adhere to the faculty academic and professional rules.
5. recognize that the study of microorganisms is critically important to everyday life and is totally fun.

### **Lecturer is expected to:**

1. provide clear and informative lecture notes with learning objectives that focus on important points,
2. give clear, informative, and stimulating 50-minute lectures with PowerPoint or other visual aids to enhance the learning experience for students.
3. answer questions either in or outside class or via e-mail or telephone.
4. compose thoughtful and fair exam questions that assess student learning and application of the course content.
5. directing the case sessions and facilitators to provide an effective learning experience in small group, team-oriented sessions.
6. providing answers and explanations to student inquiries regarding any aspect of the course.
7. providing advice and assistance to students for improving their learning strategies and performance in the course.
8. reviewing and implementing appropriate changes in the course based on student feedback and evaluations.

### **Learning Resources**

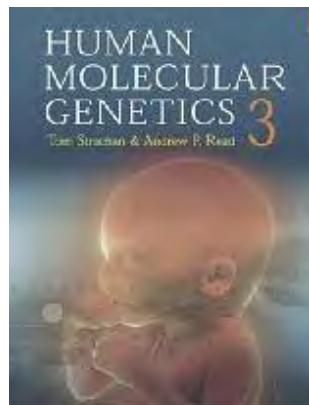
Required Textbook:



**Emery's Elements of Medical Genetics: With Student CONSULT Online Access (Paperback) by Peter Turnpenny and Sian Ellard**

By dividing the text into three sections Emery's covers everything from the basic principles of genetics to clinical genetics. This book can be delved into throughout your training. The layout is spacious; the painful task of trawling through reams of dense text is avoided. In addition, summary boxes, a full glossary, and case-based and multiple choice questions, with answers, to test your understanding.

This rigorously updated, succinct, straightforward and well-illustrated, Emery's Elements of Medical Genetics is the perfect resource to help you master the genetics you need to know! Generations of students have appreciated its unique balance of basic science, genetics as applied to medicine, and clinical genetics, making it a favourite among students and lecturers for over 30 years.



**Human molecular genetic by tom Strachan and Andrew Read.**

This book provide authoritative guidance on the principles underlying human molecular genetics. The content and organization has been fully revised.

With this fully updated third edition, Human Molecular Genetics enters the post-genome era. The content and organisation have been thoroughly revised to take account of the new knowledge and new emphases in human molecular genetics now that we have the human genome sequence.

Introductory chapters cover DNA structure and function, chromosomes, pedigrees and the basic techniques of PCR, cloning and hybridization, and now include a new chapter on cells and development. Later chapters detail our new understanding of the structure, organization, expression and evolution of the human genome, and the progress of the many parallel genome projects.

Sections on variation, mutation and disease relate this knowledge to Mendelian diseases, complex multifactorial diseases and cancer. Mapping and identification of disease genes,

molecular pathology and molecular diagnosis are treated in depth. The final sections cover functional genomics, the role of animal models and new approaches to genetic treatment of disease.

Overall, Human Molecular Genetics provides a complete, up-to-the-minute account of human molecular genetics in the post-genome era, from basic principles to current practice. Human Molecular Genetics is carefully designed to engage and motivate students with clear yet detailed full-colour illustrations, special topic boxes (including coverage of ethical, legal and social issues such as eugenics, genetic testing and human cloning) and a constant emphasis on understanding principles rather than learning detail.

## **Tips for Learning**

1. Briefly review your lecture notes before the lecture to get an idea of the material that will be covered, the degree of difficulty of the material, and how much detail is included in the notes. Look at the learning objectives to get an idea of the most important information that you are responsible for learning and that will serve as the focus for exam questions.
2. Attend the lectures. Each lecture is composed of PowerPoint presentation, and sometimes interactive class activities. The lecture presentations re-enforce, enhance, and clarify the lecture concepts.
3. Keep up! Review the lecture as soon as you can after the lecture to make sure you understand the material; pay particular attention to the learning objectives. Read the required textbook for additional and alternative presentations of information. If you have questions, or just don't get it, ask for help.
4. Be an "active learner"! Consolidate the most important concepts and facts into a form that **YOU** are most likely to understand and retain, i.e. a summary chart or flow diagram, a patient case study that includes all of the major features of an organism—include a stick diagram of the patient with the classic symptoms and arrows showing the organism's route of entry, spread in the body and modes of transmission. Be creative and make it fun! Divide the work with your study partners and share your study-aids. Try giving a minilecture to yourself (on the car or while you're walking) or to your study partners—



can you discuss the important points in your own words without looking at your notes? You will remember your own version of the information better than trying to memorize your lecturer's version.

5. If you do not do well on the first exam, please contact your lecturer immediately to determine how to improve your learning strategy.

## **Examinations**

There will be four exams in MCQs format with one best answer. Exam questions will focus on the learning objectives students are expected to master from material presented in the lectures and textbook

Final theory Exam (in week 17) cover material from lecture 1 through 15.

After each exam (especially the 1<sup>st</sup> one!), evaluate your performance and learning/study strategies. Did your performance reflect the effort you made and your confidence in knowing the material before the exam? Analyze the questions you missed, along with the challenges and responses, and try to figure out why you missed each one, e.g. couldn't remember the information, misunderstood the information, couldn't apply your knowledge to a problem solving question. Once you identify specific problems, you can implement specific solutions. If you want help with this type of evaluation, contact your lecturer.

## **Summative Evaluation and Grading**

Final grades are based on grades earned for each of the 4 periodical exams, and final theory exam.

Letter grades are based on the following final numeric grades:

**A** Excellent 90 - 100

**B** Very Good 80 – 89

**C** Good 70 – 79

**D** Pass 60- 69

**F** Fail 59 and below