Biology 564 HUMAN BIOCHEMICAL GENETICS
Spring 2006
Course Syllabus

Course Reference books: There is no required text book for the course. You are encouraged to use the following reference books:

Inborn Metabolic Diseases: Diagnosis and Treatment by Fernandes et al. (3rd edition, 2000, Springer)
This book is stocked in the Matador Bookstore under the course ticket.

The Metabolic and Molecular Bases of Inherited Disease, edited by Scriver et al. (8th edition: 4 volume set, 2001) OR (7th edition: 3 volume set,1995)
Both editions are in the library in the reference section.

Course Website: http://webteach.csun.edu/

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Class Schedule: F: 8:00 am – 10:50 am, LO 1117

Office Hours: WF: 12:30 pm-1:45 pm or by appointment.

COURSE OBJECTIVES:

In the post-genome era, knowledge of the genetic and biochemical bases of inherited diseases is expanding rapidly. As new genes are identified and biochemical functions of already known genes are unraveled, there promises to be a surge in information available to clinicians and medical professionals as they care for patients with inborn metabolic disorders. This course is designed to familiarize future geneticists, biochemists, clinicians and genetic counselors with current state of knowledge in the field and prepare them to keep up with the newest developments.

On completion of this course, students will demonstrate:

- Knowledge of the basic principles of metabolic pathways as they influence normal physiology and disease.
- An understanding of the genetic causes of inborn errors of metabolism, inheritance patterns and heterogeneity.
- Information regarding the molecular bases of common biochemical disorders.
- Knowledge of the biochemical techniques and tests used for the diagnosis and management of inborn errors of metabolism.
- Awareness of treatment options and dietary management of metabolic disorders.

COURSE REQUIREMENTS:

This course is designed for advanced students majoring in Biology, graduate students in genetics or genetic counseling, or students wishing to pursue a career in biomedical genetics. Prerequisites are courses in genetics (BIOL 360), cell biology (BIOL 380) and biochemistry (CHEM 365 or 464). Students are required to attend class regularly, and participate in discussions. Prior permission is required in case of any absence from class for non-emergency reasons.

METHOD OF EVALUATION: Various formats will be used to evaluate how well a student has grasped the subject:

Homework: There will be periodic homework assignments that will be due the following week, and these will account for 30% of the final score.
Mid-term exams: There will be two mid-term exams, each accounting for 30 % of the score.
Guest Lectures: We will have guest speakers who are professionals in various subfields of human biochemical genetics. We are privileged to have them devote their valuable time to talk to us. You are encouraged to read about the topic in advance of their lecture and ask questions afterwards. You are required to hand in a one-page summary of the presentation the week after the lecture. These reports will account for 10% of your final score.

Grades will be based on total points earned. No plus/minus grades will be assigned.

A: $\geq$ 90%
B: 89-80%
C: 79-70%
D: 69-60%
F: $<$ 60%

BIOLOGY DEPARTMENT WITHDRAWAL POLICY: Unrestricted withdrawals are permitted only until the end of the third week. Thereafter, requests to drop a class will be honored only when a *verifiable* serious and compelling reason exists and when there is no viable alternative to withdrawal. *Poor performance is NOT an acceptable reason for withdrawal.* During the last three weeks of the semester withdrawals will not be approved except when a student is withdrawing from ALL classes for verifiable medical reasons.

CHEATING AND PLAGIARISM: All forms of cheating and plagiarism (the claiming of the work of others as your own) are expressly forbidden by University rules and will not be tolerated. Any student observed cheating will be subject to disciplinary action by the University and may receive a grade of F in the course.

Spring 2006: BIOL 564: Human Biochemical Genetics
8:00 am-10:50 am; Friday; Rm LO1117

	Date	Time	Topic	Reading Fernandes *et al*.
Feb.	3	8:00am	Introduction.	
	3	9:30am	Basic Enzymology	
	10	8:00am	Overview of Carbohydrate Metabolism / Glycogen Storage Disorders	
	10	9:30am	Disorders of Fructose Metabolism	
				Part II
	17	8:00am	Disorders of Galactose Metabolism	
	17	9:30am	Diabetes and Hypoglycemia	
	24	8:00am	Overview of Amino Acid Metabolism	
	24	9:30am	Disorders of Amino Acid Metabolism-I	
				Part IV
March	3	8:00am	Disorders of Amino Acid Metabolism-II	
	3	9:30am	Disorders of the Urea Cycle	
	10		***Midterm Exam- I***	
	17	8:00am	*Guest Lecture: Richard Boles, M.D.* *Head of Mecical Genetics, CHLA* *Topic: Metabolic Disorders and Counseling Issues*	
	24	8:00am	Overview of Nucleic Acid Metabolism	
	24	9:30am	Disorders of Purine Metabolism-I	
				Part VIII
	31	8:00am	Disorders of Purine Metabolism-II	
	31	9:30am	Disorders of Pyrimidine Metabolism; Porphyrias	
April	7	8:00am	Overview of Energy Metabolism / Pyruvate Metabolism and the TCA Cycle	**Part III**
	7	9:30am	Disorders of Beta Oxidation and Electron Transport Chain	
	14		**SPRING BREAK**	
	21	8:00am	Overview of Lipid and Lipoprotein Metabolism	
	21	9:30am	*Guest Lecture: Jheem D. Medh, Ph. D.* *Assistant Professor of Chemistry, CSUN* *Topic: Disorders of Lipoprotein Metabolism*	**Part VII**
	28	8:00am	Dyslipidemias	
	28	9:30am	Disorders of Cholesterol and Bile Acid Synthesis and Storage	
May	5	8:00am	Organelle Functions, Glycolipids and Complex Carbohydrates	**Part X**
	5	9:30am	Lysosomal Storage Disorders; other Organelle Function Disorders	
	12	8:00am	*Guest Lecture: Mehdi Jamehdor, M.D.* *Physician Director of Genetic Testing at SCPMG* *Topic: Clinical Management of Inborn Errors of Metabolism*	
May	19		***Midterm Exam- II***	