

Hearing and Speech Department

AUD 940 – SEMINAR IN GENETICS AND HEARING LOSS

Summer Semester, 2007

(2 credit hours)

Time: Monday June 4, 9:00 am - 4:00 pm
Monday June 11, 1:00 - 5:00 pm
Monday June 18, 1:00 - 5:00 pm
Friday July 6, 8:00 – noon
Monday July 9, 1:00-5:00 pm

Place: H. C. Miller Building, 4th Floor Conference Room

Instructor: Judith E. Widen, Ph.D., CCC-A
Room 4001 H.C. Miller Bldg,
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Office Hours: by appointment.

Course Description:

This seminar is designed to cover the fundamentals of human genetics as related to hearing loss, including patterns of inheritance, genotypic and phenotypic characteristics of the major forms of syndromic and nonsyndromic hearing loss; genetic counseling, genetic testing, and issues related to them; resources for keeping up with this rapidly changing field.

Learning Objectives:

Knowledge: Participants will develop a basic understanding of:

1. basic human genetics terminology.
2. patterns of inheritance (Mendelian) and the importance of family history.
3. patterns of inheritance (nontraditional).
4. the clinical characteristics of the major forms of syndromic and nonsyndromic hearing loss.
5. how identification of genetic variation (forms of hearing loss) facilitates development of prevention, diagnosis and treatment.
6. the history of genetics and the deaf community.
7. the potential physical and/or psychological benefits, limitations, and risks of genetic information for individuals, family members and communities.
8. the ethical, legal and social issues related to genetic testing for genetic conditions and recording of genetic information in such cases.
9. the resources available to meet the needs of clients seeking genetic information and services.
10. the components of the genetic counseling process and indications for referral of individuals with hearing loss to genetic specialists as well as their own professional role in provision, follow-up, and quality of genetic services.

Skills: Participants should be able to

1. explain basic concepts of probability related to modes of inheritance and the influence of genetic factors in causing hearing loss.
2. gather basic family history information, identify individuals and families that would benefit from genetic services.
3. identify sources of credible, current information about genetics for self, client, students and colleagues.
4. safeguard privacy and confidentiality of genetic information of clients and families to the extent possible and respect the choices that clients and families intend to make about genetic testing.
5. educate individuals about the availability of genetic testing for hearing loss and provide basic information about the potential risks, benefits and limitations of genetic testing for hearing loss.
6. prepare clients and families for what to expect from the genetic evaluation and counseling process.
7. educate their professional peers about key genetic concepts relative to hearing loss and related ethical and policy issues.

Attitudes: Participants should be able to:

1. appreciate the sensitivity of genetic information, and the need for privacy and confidentiality.
2. recognize the limitations of their own genetic expertise and seek the help of genetics professionals as needed to education their own students, peers and clients.
3. recognize philosophical, cultural and ethical perspectives influencing utilization of genetic information and services, especially those related to the deaf and hearing communities.
4. recognize when personal values and biases with regard to ethical, social, cultural, religious and ethnic issue may impact or interfere with care provided to clients and families.
5. support client-beneficial policies, including access, choice to utilize genetic services, genetic privacy and non-discrimination.
6. seek coordination and collaboration with an interdisciplinary team of health professionals.
7. recognize that information about the genetics of deafness is not a static body of knowledge and develop effective personal strategies for keeping up with this rapidly changing field.

ASHA Certification Standards:

Students enrolled in this course will acquire knowledge and skills associated with the following ASHA standards for certification in Audiology:

- B2, B4, B8, B18
- C1, C2, C3, C4
- D1, D2, D3, D13, D14

Course Format: The format of the class will include traditional lecture format, on-line study, and active participation/presentation by class members. The course is web enhanced by use of Angel, an on-line course management tool (Angel URL: <https://elearning.kumc.edu/angel>). Please check Angel on a regular basis for announcements and updated course materials.

Textbooks and Software:

There is no required text for this course, partly because there are only a few on genetics and hearing loss and because information changes so rapidly they are quickly out of date.

Texts on genetics (in general) are published more frequently and a couple that I have found helpful are:

- Westman, J.A. (2006). Medical Genetics for the Modern Clinician. Philadelphia: Lippincott, Williams & Wilkins.
- Cummings, M.R. (2006). Human Heredity: Principles and Issues. 7th Ed. Belmont CA: Thomson Brooks/Cole.

Selected readings will be provided on Angel.

A number of helpful websites are recommended and linked via Angel. Some that you should be sure to visit and use are:

- Genetics in the Practice of Speech-Language Pathology and Audiology <http://shla.nchpeg.org/>
- University of Utah Genetic Learning Center <http://learn.genetics.utah.edu/>
- GeneClinics, University of Washington www.geneclinics.org
- Hereditary Hearing Loss Homepage <http://webhost.ua.ac.be/hhh/>
- Genetics and Deafness, Boys Town National Research Hospital <http://www.boystownhospital.org/Hearing/info/genetics/index.asp>
- McKusick's Online Mendelian Inheritance in Man (OMIM) <http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM>
- University of Kansas Genetics Homepage <http://www.kumc.edu/gec>, including Genetic and Rare Conditions Site <http://www.kumc.edu/gec/support/>

Rules for the Widen Library

You are welcome to peruse my journals and books.

You may borrow them PROVIDED that I can access them at any time, that means ...

- 1) you may use them here at the Med Center but you may not take them home, and
- 2) you must let me know What you've borrowed, When and Where you will be with the book/journal in the event I need it. There's a clipboard with a sign up sheet on the bookshelf outside my office door.

Thanks, in advance, for your adherence to these rules!

Grading Policy:

Grades will be based on:

Class Attendance & Participation	10%
Exam 1	30
Exam 2	30
Case Project	<u>30</u>
Total %	100

Course Grading Scale

91 - 100%	A
81 - 90%	B
71 - 80%	C
61 - 70%	D
60% & less	F

Disability Statement

Learning assistance, academic performance enhancement, and psychological services at KUMC are free, confidential, and available at Student Counseling & Educational Support Services by calling 913-588-6580 or visit G116 Student Center.

Any student in this course who needs an accommodation because of a disability in order to complete this course should contact the instructor and the Equal Opportunity/Disability Specialist (913-588-7813, TDD 913-588-7963) as soon as possible.

Any student with extenuating circumstances which could prevent the fullest expression of his/her abilities should contact the instructor to make individual arrangements.

Any student who does not understand and/or accept the contents and terms of this syllabus must notify the instructor in writing within one week after receiving this syllabus.

Tentative Schedule for Lecture Topics/Exams/Topics

M 6/4	Fundamentals of Genetics Chromosome Structure and Abnormalities Pedigree Construction Transmission Genetics Assign Case History Project & Teams
M 6/11	DNA Structure and Function Non-traditional Modes of Inheritance Characteristics of Non-syndromic and Syndromic Hearing Loss
M 6/18	Genetic and Epidemiologic Aspects of Deafness Hearing and Deaf Points of View Recurrence Risk
Between 6/18 and 7/6	Midterm Exam Work on Case Presentation Preparation (Reading & CD) for remainder of classes
F 7/6	Genetic Counseling: Debra Collins Genetic Counseling: including Deaf vs Hearing perspective Resources
M 7/9	Case History Presentations
After 7/9	Final Exam