MEDICAL GENETICS PROJECT

Overview

The Medical Genetics Project is a small group exercise. You and your group will research information about a specific genetic disease and teach your findings to your classmates and faculty in a formal oral presentation. This project has several goals. It will give you an opportunity to further develop your ability to search for, and critically evaluate, scientific evidence for the principles and concepts covered in the course. This goal is important since there is now a vast array of powerful technologies and databases that are specific for medicine and basic medical sciences. These resources require significant practice to locate and use. The project will also add to your knowledge about a subset of Genetics Disorders that were selected by the Loyola Genetics Subcommittee as illustrating important principles in human genetics, patient care, and societal issues. Finally, this project will give you the opportunity to practice teaching difficult medical concepts to your piers, and ultimately to patients.

The different assignments for the Genetics Projects are spread throughout the course. Your first responsibility is to attend **a special lecture by the librarians at 2:30 on Aug. 8 in Tobin Hall** (190). The librarians will show you how to do a search of the National Library of Medicine "Medline" database using the Ovid search engine. You will then be asked to do a preliminary search for publications related to a particular aspect of your assigned genetic disease (see ***Literature Search*** below). The following steps should be used:

1. Select the appropriate database and timeframe.
2. Analyze the specific question that you are trying to answer in your search by writing it down on a sheet of paper and then listing the key concepts that will be used in your search.
3. Choose the appropriate MeSH subject heading for each concept.
4. Use the Explode option if appropriate.
5. Effectively narrow the results as needed:
    a. Select subheadings
    b. Use the correct Boolean logic to combine concepts
    c. Apply appropriate limits (i.e., age groups, language)
    d. Use Focus
    e. Add additional MeSH headings
    f. Search efficiency (i.e., did not limit every search statement, did not use unnecessary search terms)
6. Identify relevant citations in your search that will help to answer your question.

Turn in a copy of your search strategy along with 3-5 references to the librarians by Aug. 16 for their analysis and comments. You can drop it off in the library or e-mail it to ResearchServices@lumc.edu.

A required class meeting is scheduled for Wed,, Aug. 31, from 11:30-12:30 in Leischner Hall (Room 390) to discuss additional Genetics Project requirements and to answer your questions. Your group should meet as often as necessary to complete the project (including the Sept. 14 Small Group session). Student presentations are scheduled in different rooms on Sept. 21 from 11:30-3:40, with a lunch break in the middle. You should plan on attending all student presentations in your room, in part because there will be questions on Exam 3 on the content of these talks. See the table below for the genetic disease assignments for each small group and a detailed schedule. Each talk is scheduled for 25 minutes followed by a 5-minute question and answer period.

Assignments and Schedule

Location	Time	Presenting Groups	Topic
Room 345	11:30 – 12:00	Group 1 (Room 340)	Cystic fibrosis
	12:05 – 12:35	Group 2 "	Familial breast cancer
	12:40 – 1:10	Group 3 "	Familial hypercholesterolemia
	1:15 – 1:55	Lunch Break	
	2:00 – 2:30	Group 4 (Room 350)	Fragile X syndrome
	2:35 – 3:05	Group 5 "	MERRF
	3:10 – 3:40	Group 6 "	Acute intermittent porphyria
Room 375	11:30 – 12:00	Group 7 (Room 370)	Cystic fibrosis
	12:05 – 12:35	Group 8 "	Familial breast cancer
	12:40 –1:10	Group 9 "	Familial hypercholesterolemia
	1:15 – 1:55	Lunch Break	
	2:00 – 2:30	Group 10 (Room 380)	Fragile X syndrome
	2:35 – 3:05	Group 11 "	MERRF
	3:10 – 3:40	Group 12 "	Acute intermittent porphyria
Room 445	11:30 – 12:00	Group 13 (Room 440)	Cystic fibrosis
	12:05 – 12:35	Group 14 "	Familial breast cancer
	12:40 – 1:10	Group 15 "	Familial hypercholesterolemia
	1:15 – 1:55	Lunch Break	
	2:00 – 2:30	Group 16 (Room 450)	Fragile X syndrome
	2:35 – 3:05	Group 17 "	MERRF
	3:10 – 3:40	Group 18 "	Acute intermittent porphyria
Room 460	11:30 – 12:00	Group 19 (Room 470)	Cystic fibrosis
	12:05 – 12:35	Group 20 "	Familial breast cancer
	12:40 – 1:10	Group 21 "	Familial hypercholesterolemia
	1:15 –1:55	Lunch Break	
	2:00 – 2:30	Group 22 (Room 480)	Fragile X syndrome
	2:35 – 3:05	Group 23 "	MERRF
	3:10 – 3:40	Group 24 "	Acute intermittent porphyria

Project Details

Grading: The Medical Genetics Project contributes 32 points to the total of about 330 points (~10%) that make up the final course grade (see Section 4). There will be 2 multiple-choice questions for each of the six diseases (12 points) on Exam 3. There will also be 10 points designated for evaluation of your presentation (slides/oral delivery) and 10 points for evaluation of your written documentation of your literature research (see below).

Literature Search: In addition to the preliminary Ovid search described above, your small group should do a more detailed literature search on your assigned disease. Among the topics that you can research are disease etiology, pathogenesis, phenotype and natural history, management, inheritance risk, and family, social, legal, and ethical issues that relate to the specific disorder. **Each student in the group should be responsible for one aspect of the disease. Your group should make these assignments before doing the preliminary Ovid search.** It is not necessary or even desirable to research and present all possible topics. The nature and amount of current medical knowledge about each disorder varies, so it should become apparent from your research which issues are most important to stress for your assigned disease.

It is important for your group to work together to teach each other information and then to incorporate this information into an organized and non-redundant oral presentation. **However, *each member* of the group should do their own literature search. You should document your search strategy since you are being asked to submit an annotated bibliography to the course directors** (see below). The process of developing a good search strategy is a learning experience that will take time and involve some failure.

Acceptable sources of information are textbooks, review articles, online resources (e.g, NIH, scholarly societies, patient advocacy groups, etc.), evidence-based medicine reviews, and original research articles. You can get help in finding reliable information from the LUMC Library Website link "Resources for Medical Students," which contains catalogs, online textbooks, databases, etc. For resources specific to the assigned genetic diseases, see "MCBG Search Guides" (http://library.luhs.org/hslibrary/resources_for/MCBG_guides.html). **As one of your strategies, you must do a search of the current primary research literature in Medline, using Ovid (or PubMed) as the search engine. You should not rely solely on secondary sources (reviews).** Use the tab "Training/Education" on the Library Website and click on "Guides and Tutorials" to get detailed instructions on 1) how to use databases, 2) how to conduct a literature search, and 3) how to evaluate the reliability of the information you find. You and your group can get personal help by contacting Jeanne Sadlik (Coordinator for References & Education Services, Health Science Library) (X6-5304; jsadlik@lumc.edu) or a member of her staff.

The following are some useful websites that specifically reference genetic diseases:

www.geneclinics.org: presents detailed scientific reviews of specific genetic disorders, as well as labs and clinics that test for and treat specific disorders by locality (state).

www.ncbi.nlm.nih.gov/sites/entrez/?db=OMIM: Online Mendelian Genetics in Man (OMIM) website and database. "Search morbid map" (listed on left side of page) generates an alphabetical list of diseases, the affected genes, and their chromosomal locations

http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gnd : Genes and Diseases website. The navigation tool on the left side of the page links viewers to specific classes of diseases. Clicking on a specific chromosome at the top identifies known diseases, associated genes, and their locations on that chromosome. Clicking on the disease gene gives some basic information about the disease.

List of genetics sites: http://library.luhs.org : "E-Resources," "Resources by Subject," "Genetics."

PubMed, Medical Genetics Search: From the LUHS library webpage (http://library.luhs.org/)select PubMed under the heading "Quick Links". From the PubMed page select "Clinical Queries" under the heading "PubMED Tools". At the Clinical Queries page you can carry out a "Medical Genetics Search" at the top right of the page. Enter the disease in the search window and select a category such as "Diagnosis", "Clinical Description", "Management", "Genetic Counseling", etc. You can also search the same disease under the heading "Clinical Study Category" at the top right of the page. The search categories here are "Etiology", "Diagnosis", "Therapy", "Prognosis", etc. Finally, you can search for a systematic review of a genetic disorder under the heading "Find Systematic Reviews" in the top middle of the page. Each search produces a list of papers with the opportunity to view abstracts, if available. One can also click on an author's name and retrieve a list of related publications by that author.

Abstract Submission: Your group should prepare a clear and concise abstract (less than two-thirds of a page single-spaced), which summarizes your research findings and the content of your presentation. Make sure that the abstract contains your group number. Send it as a Word or pdf file by e-mail to Dr. Frankfater (afrankf@lumc.edu) by Sept. 16. The abstracts from all groups in your presentation room will be collated and distributed at the session.

Posting abstract/slides: Post your abstract and your group's PowerPoint slides to Moodle by Sept. 19 (see Section 12). This information will then be available to your classmates for studying for the 12 questions that will appear on Exam 3.

Presentation: Your group's presentation should be 25 minutes long, and will be followed by a 5-minute audience question period. Although each member can discuss a specific aspect of the genetic disease, make sure that the individual talks are well integrated. Your group should practice aloud ahead of time to make sure that there is no unnecessary duplication of material, and that the talk fits into the 25-minute time frame. Groups that go significantly overtime will have points deducted. You should carefully read the ***Tips for preparing slides***, and ***Tips for giving your presentation*** sections below for additional information.

Documentation of the literature search: Each student will be evaluated on his or her ability to conduct an effective literature search and to critically evaluate scientific and medical literature (see **Form 6**). Therefore, you should submit to the course directors an annotated bibliography that documents these abilities. The bibliography should include 8-10 items from a variety of sources. At least two of these should come from an Ovid or PubMed search of current research literature. In a *few sentences*, provide the following information for each reference:

a) The type of reference (e.g., textbook, a journal literature review article, primary research article, clinical trial data, meta-analysis, evidence-based practice recommendations, web-based scientific society or patient advocacy information).
b) A statement of the key information provided in the reference.
c) How you found the reference. For Ovid or PubMed references, indicate the search terms used and how you manipulated the terms.
d) The reliability of the information. Document that the information is current. If you suspect that the authors may have biases or conflicts of interest, so indicate.

You should prepare your own materials as a separate file identified by name and group. However, the course directors would appreciate it if the files of all members of a group were submitted to them in a single e-mail. Groups 1-12 should send their files to Dr. Simmons. Groups 13-24 should send them to Dr. Frankfater. These files are due no later than 5 PM on Friday, Sept. 23. Failure to submit the abstract (Sept. 16), post the abstract and slides on the Webforum (Sept. 19), or submit the annotated bibliographies (Sept. 23) by the indicated deadlines will result in the deduction of points from either the Presentation grade or the Research and Analysis grade, and may result in a 'Meets with Concerns' for the Professionalism Competency.

Evaluation: The Medical Genetics Project will be evaluated in three different ways. You will be tested on your knowledge of the six genetic diseases through 12 multiple choice questions on Exam 3. Your presentation will be evaluated by the faculty members who attend your session (see **Form 5).** The course directors will collate these evaluations and assign up to 10 points toward your final grade (see **Form 6**). (Some students who are at the session will also be asked to evaluate your group presentation, but these evaluations will not be used systematically to determine your grade.) Finally, the

Documentation of the literature search materials will be evaluated by the course directors who will assign up to 10 points toward your final grade using **Form 6**. (We will read these!)

Satisfactory completion of the Medical Genetics Project will result in a "Meets Expectations" in the **Lifelong Learning Competency** in the End-of-Course Competency Assessment Form (**Form 7**). This project will also contribute to the evaluation of the other core competencies (**Medical Knowledge**; **Interpersonal and Communication Skills**; and **Professionalism)**.

Tips for preparing slides:

a. *Use a simple, consistent background for each slide*
b. *Use large letters for text (16 font or above).*
c. *Identify a single main idea for each slide and use it as a title.*
d. *The title and other text should consist of key words or short phrases, not long sentences. (Fill in the information with your oral narrative.) Each slide should contain no more than 6 key words/phrases. Limit the number of slides that have just bullet points.*
e. *Enhance your message with pictures, graphics, etc. (The best slides may have little or no added text. "A picture is worth a thousand words!") Keep figures relatively simple and easy to understand.*
f. *Incorporate features such as color, animation, transitions, arrows, etc., that focus attention and emphasize concepts, but don't overdo it.*
g. *Limit the total number of slides to 3-4 per group member.*
h. ***Put your name in small print at the bottom right-hand corner of each slice so that the course directors can identify the author for grading purposes.***

Tips for giving your presentation:

Each presenter should begin by identifying herself/himself by name. This is particularly important because some faculty members who will be evaluating the presentations may not know each presenter personally. The first presenter should provide an overview of the genetic disorder and a description of the topics that will be discussed by the group. Each presenter should begin by stating their topic and end with a "take-home" message(s). The last presenter should end by briefly summarizing the major "take-home" messages of all previous presenters.

Some general guildlines are:

a. *Devise strategies that elicit audience interest and aid recall (e.g, involving the audience in active learning such as asking questions, eliciting responses, etc.)*
b. *Tell the audience what you are going to say, say it, and then tell them what you have just said. (tell-say-tell)*
c. *Explain the features of each slide. Relate the content to previous and subsequent slides.*
d. *Use a pointer to keep the audience focused.*
e. *Speak loudly enough to be easily heard in the back of the room. Make your voice expressive and interesting. Speak slowly and distinctly so that you can be easily understood.*
f. *Whenever possible, maintain eye contact with all sections of the audience.*
g. *Exhibit a relaxed appearance and display a suitable enthusiasm for your topic.*

Project a professional image:

a. *Dress and groom in a manner appropriate for a professional conference.*
b. *Demonstrate respect for the audience and respect for the content of each presentation.*
c. *Attribute sources where appropriate. For figures, put a small abbreviated citation nearby.*

Form 5: Medical Genetics Presentation Assessment: to be completed by lecturers, facilitators and selected students in attendance at the genetic presentations. (However, the presentation grade will be determined solely by faculty members.)

Form 6: Course Directors' Grade Sheet, Medical Genetics Project: Part A indicates the grade point distribution for evaluation of the Genetics Project ***Presentation***. The number of points given will be determined by faculty evaluations (Form 5). **Part B** indicates the grade point distribution for the ***Research and Analysis*** portion of the Genetics Project. The number of points given will be based on evaluation of the annotated bibliography that you submit to the course directors.

FORM 5 MEDICAL GENETICS PRESENTATION ASSESSMENT

GROUP NO: _____TITLE_____

ASSESSOR: (students can remain anonymous) _____

Instructions for Individual Assessment: For each student, provide a Global Rating for each main category (white box) using the following scale:

3 = Meets expectations
2 = Meets expectations with concern
1 = Does not meet expectations

Put a checkmark in a gray box where the student needs specific improvement.

Individual Presentation Attributes	**Student Name**					
Communication - Verbal						
Identified themselves by name						
Clear introduction, middle, and conclusion.						
Effectively explained important features of each slide.						
Effectively used pointer to keep audience focused.						
Clear voice (loudness, no mumbling) throughout presentation.						
Rate of speech appropriate throughout presentation.						
Varied voice intonations to keep audience interest.						
Demonstrated no nervous habits that distracted from presentation.						
Did not read notes excessively or at all.						
Communication - PowerPoint Slides						
Each slide had a clear title that identified a single main idea						
Key words were used in place of lengthy sentences						
Pictures, graphics, etc. were used to focus attention and emphasize concepts						
Professionalism						
Appropriate dress and grooming.						
Attributed sources where appropriate.						
Demonstrated respect for audience.						

Group Presentation Rating	
Presentation had a clear organizing theme, with clear main ideas and overall conclusion.	
Relationships between the topics within the presentation were clearly explained and made sense.	
All slides within presentation had a consistent background/theme.	
Each presenter understood the previous and subsequent presentations and how they related to their own topic.	
Enthusiasm for presentation was high and maintained throughout presentation.	
Group effectively involved the audience in active learning.	
Presentation took up the time allotted (not too long or too short)	

FORM 6 **COURSE-DIRECTORS' GRADE SHEET:**
MEDICAL GENETICS PROJECT

Student Name _____Group No._____

PART A: PRESENTATION (based on faculty evaluations – Form 5 – of the genetics presentation)

Competency: Interpersonal and Communication Skills

Outcomes Assessed:

- *Use written and verbal language effectively.*
- *Use information technology to manage information and communicate findings to other students.*
- *Facilitate the learning of other students.*
- *Communicate essential information effectively.*

Competency: Professionalism

Outcomes Assessed:

- *Behave professionally.*

Abilities:	**Possible Points**	**Points Earned**
Communication – Verbal (teaching fellow students)		
Communicated effectively:	2	
Worked effectively as part of a group (group rating)	2	
Communication – PowerPoint Slides	3	
Professionalism	3	
TOTAL	**10**	

PART B: RESEARCH and ANALYSIS (based on submitted annotated bibliography)

Competency: Lifelong Learning, Problem-solving, and Personal Growth

Outcomes Assessed:

- *Locate, appraise, critically review and assimilate evidence from scientific studies and medical literature.*
- *Use information technology learning resources to manage basic science information, access online information, and support your own education.*
- *Demonstrate an investigatory and analytic thinking approach.*

Abilities:	**Possible Points**	**Points Earned**
Write an annotated bibliography that demonstrates an effective use of information technology.		
Provide 8-10 citations from a variety of sources.	2	
Briefly indicate the key information in each reference.	2	
Describe how you found each reference.	2	
Indicate search terms used, and how the search was narrowed.	2	
Document that each reference is current and reliable.	2	
TOTAL	**10**	