Instructor: Dr. Crosby W. Jones, Jr.
Office: 003C Cavness Science Building
Office Hours: MWF 12-1; M 9-11 am; M 2-5 pm
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Book: No textbooks (but see reading assignment section below)
Attendance: Roll will be taken at each class meeting but will not be factored in to your grade except as described below.

Goal: By exposure to a classroom atmosphere where competition between academically strong peers, time management skills, scholarly analysis of scientific literature and succinct scientific writing are emphasized, the student will be more aware of his/her suitability for post-graduate studies in the field of medical genetics including graduate studies and/or health professional programs with strong human genetic components.

Student Learning Objective: For departmental, state and accreditation purposes this course will assess the following learning objective: Biology Department Learning Goal #5—the student's ability to communicate and critically evaluate information in oral and written forms.

Student Learning Outcome: Students completing this course should demonstrate good time management skills, an ability to find, recognize and analyze quality scientific information, and an ability to write scientifically in a clear, accurate and concise manner as compared to fellow classmates.

Student Learning Assessment: This learning objective will be assessed by administering problem sets throughout the semester. A total of 9 will be used. The students will be given 5 days to complete the answers for each problem set. They will access relevant scientific data from a variety of sources, analyze it critically, formulate answers to the posed questions and write answers in a concise, scientifically correct manner. Each student’s performance on a given problem set will be evaluated by the instructor as to how well it demonstrates the learning outcomes. The student will then receive a ranking on that problem set relative to fellow classmates. By the end of the semester, each student will be assigned a class ranking based on overall performance.

Lecture topics:

A. Chromosomal structure and number defects (first 7 weeks of the semester)
   1. Overview (terminology and ISCN rules, examples of the major types, origin and nature of the genetic abnormalities involved, inheritance pattern of the defects)
   2. Clinical aspects (genetic testing & diagnosis, medical therapy, genetic counseling)
   3. Selected examples discussed in depth including case studies

B. Single gene defects (autosomal, sex-linked and mitochondrial) (second 8 weeks of the semester)
   1. Overview (examples of the major types, origin and nature of the genetic abnormality, inheritance pattern of the defect including pedigree analysis)
   2. Clinical aspects (genetic testing & diagnosis, medical therapy, genetic counseling)
   3. In-depth examples (from gene defect to phenotype)

C. Multifactorial traits (Polygenic & Environment). (This topic will be interspersed during the semester)
   This section of the course will discuss genetic traits for which the environment and/or multiple genes play a major role in the expression of the phenotype. Topics may include but are not limited to the following: cancer, behavioral traits, epigenetics, teratogenesis, twin studies and tissue typing.

D. Genetic Technology (This topic will be interspersed during the semester)
   This section of the course will discuss recent advances in analyzing and/or manipulating the genetic system. Topics may include but are not limited to the following: Stem cell therapy, In vitro fertilization and related technology, CRISPR technology, pharmacogenetics, exome sequencing, personalized genetics, gene therapy, whole genome assays and microarrays.

Exams: No exams. However quizzes following book & video assignments will be required — see next page for details.
Course Grade

A = 5-10 "A"s on the problem sets and presentation plus all 3 "Extras" including at least two books

B = 6-10 "B"s on the problem sets and presentation plus all 3 "Extras" including at least two books

C = 3-4 problem sets with a grade of C or missing one of the 3 "Extras" including at least two books

D = Missing two of the "Extras" or failing to turn in one problem set or failure to do the presentation or more than 4 problem sets with a "C" or worse grade

F = Not meeting the requirements for any of the grades above

Problem sets

A problem set will be distributed on each of the 9 days indicated alongside: 9/6 (W) 11/8 (W) 9/20 (W) 11/15 (W) 10/4 (W) 11/20 (M) 10/18 (W) 11/4 (M) 11/1 (W)

For all problem sets, except the last one, your answers must be turned in at the beginning of class on the following Monday. The 12/4 (M) assignment is due on 12/8 (F) & will be returned to you on final exam day.


You will be limited to a maximum of four 8 1/2 X 11 inch pages of paper with answers written on one side only. Your left hand margin MUST be 1 inch but otherwise you can stretch margins, single-space and/or reduce font size if you need the room (it must be readable however). Word processing is required. Number your answers in order but do not re-write the question on your answer sheets. Your answer sheet must be folded lengthwise with an anonymous code on the outside when submitted (your name must not appear on any part of the problem set). Change font from problem set to problem set to maintain anonymity. Failure to follow any of these “rules” will result in a grade no higher than a “B” on that paper. Late paper submissions will receive a grade no higher than B.

Take good (written, recorded or mental) notes of all material presented in class. Many problem set questions require that you show an ability to apply information to questions, are thorough in doing so and include all relevant facts in your answer (but do not include extraneous information since you are limited in space). Most questions will be open ended / discussion / essay. In problem type questions, the better you describe and/or show your thought process at each step, the more likely you will receive a higher score.

You must cite your references in the paper if they originate from any source other than the lectures or handouts. Referenced material must be stated in your own words and not copied verbatim. Make sure you use the best references available. Wikipedia is OK to survey an answer but it should not be your primary reference for any questions. Some other suggested sources of genetic information to start you off—first and foremost are OMIM (On-Line Inheritance in Man) and GeneReviews when you know the name of the gene or the disease it causes. But you can also survey other government resources by searching Google with the following search phrases: ClinicalTrials.gov; NIH, Genetics Home Reference; NIH, Gene Tests; NIH, Genes and Disease; or NIH Online Inheritance in Animals (OMIA): PubMed: GeneClinics.org.; National Human Mutation Database www.hgmd.cf.ac.uk/docs/register.html ; MedlinePlus; WebMD.

A part of each Friday’s lecture time will be reserved to entertain questions concerning the current week’s problem set assignment. This is the only time that help on the problem set will be available from the instructor.

Each person must attempt all homework on her/ his own. However, recognizing that quality learning can come from collaborations, you are not discouraged from discussing answers with classmates. Nevertheless, written collaborations will require that the collaborators do twice the quality of work of their fellow classmates for the same grade. Hence, it is to your advantage to compile your own answers, from your own sources and in your own words.
Each problem set will be returned with comments / corrections / suggestions, etc. Your work will be evaluated relative to all class members as to how well the questions were answered. Each answer will be ranked as "top half", "bottom half", or "borderline (i.e. almost top half but not quite)", of the class for that answer. Then your overall performance on that problem set will be ranked top half or bottom half for grade considerations. The upper half will receive an "A". The lower half will receive a "B" unless the work is noticeably lacking compared to other "lower halves" on that problem set --in which case, a "C" or lower will be assigned. The top scores in the "A" category will also receive a ranking on that problem set (e.g. A-1, A-2 etc) which represents additional feedback. These latter rankings have no bearing on your final grade. At the end of the semester, a final class ranking for top performance will be determined emphasizing 3 criteria: # of total A’s on problem sets, # of top three finishes on problem sets and # of times a person had the #1 problem set.

Final Exam Day Presentation (December 13, Wednesday, 10:30 AM-12:30 PM)

Your assignment is to choose a genetic disease topic that we did not discuss this semester. You must reserve the topic by informing the instructor of your decision. Then find at least 12 good visuals (e.g., photos, graphs, tables, graphics) depicting that topic. You will present a talk during our final exam period in which you will show the class the results of your search (a powerpoint presentation will suffice to do this). Your presentation must be between 7-10 minutes.

[1] Was the presentation between 7-10 minutes long?
[2] Did the presentation include at least 12 visuals (non text) & were they incorporated into the oral presentation in a logical manner?
[3] Did the presentation include a title page and also a reference page that listed the source of all the information and visuals in the presentation?
[4] Was the presentation sent electronically to the instructor?

If you adhere to all of the above criteria in this section, you will receive an "A" for this assignment. If not, you will receive a grade no better than "B".

Choices of Extras (must include at least two books)

1. Book Assignments (14 choices of books)

You are invited to read, The Cobra Event by Richard Preston, The Genius Factory by David Plotz, Survival of the Sickest by Sharon Moalem, Experimental Heart by Jennifer Rohn, Monkey Luv by Robert Sapolsky, Another Day in the Frontal Lobe by Katrina Firlik, Genome by Matt Ridley, Mutation by Robin Cook, A Life Decoded by J. Craig Venter, Zoobiquity by Barbara Natterson-Horowitz and Kathryn Bowers and/or The Immortal Life of Henrietta Lacks by Rebecca Skloot, Mapping Fate by Alice Wexler, p53 The Gene that Cracked the Cancer Code by Sue Armstrong and The Sports Gene by David Epstein. The instructor has at least one copy of each and will check these out to you----first come, first served. It is requested that you return each book you check out no later than 3 weeks after checking it out. You are invited to buy copies of your own if that is more convenient.

A brief overview of each book: The Cobra Event and Mutation are both fiction with plots that are strongly based in real science. They are action oriented, centered around a genetic entity created using genetic engineering techniques. Survival of the Sickest, Genome and Monkey Luv are non-fiction collections of essays with each essay describing an intriguing aspect of biology (mainly genetics) in understanding human conditions (e.g. why is depression common? Why has obesity and heart disease become so common? Why is it important that new babies be held? Why do men and women differ so much in what they desire in a mate?). Experimental Heart is fiction about graduate student experiences, both personal and professional, in a biomedical laboratory. The Genius Factory is the history of the Nobel Sperm bank. A Life Decoded is the autobiography of the most famous DNA sequencer living today. Another Day in the Frontal Lobe is about the experiences of a brain surgeon who also happens to be one of the few females in that profession. Zoobiquity is particularly good for those interested in a veterinary approach or a comparative anatomy/physiology approach. The authors show that many human responses can also be seen and studied in animals. For example, homosexuality, allergies, addictions, cancer, obesity
and heart attacks. **The Immortal Life of Henrietta Lacks** is an excellent description of the history of the HeLa cell culture line that has been used for half a century in cell biology and genetics research laboratories all over the world with a special emphasis on the family from which the original cells were isolated. **Mapping Fate** was written by the daughter of a woman who died of Huntington Disease and whose sister has spent her life researching the disease. **p53 The Gene that Cracked the Cancer Code** is a history of perhaps the most protective of all genes in the human genome---the so-called "guardian of the genome". **The Sports Gene** is a discussion of the genes that influence athletic performance and how ancestry, evolution and environment influence the versions found in different human populations.

To complete a book assignment "Extra", you must arrange with the instructor to take a quiz over the book at a mutually agreed upon place & time. This quiz will pose 3 essay questions of which you will pick two to answer. The questions will be theme type questions and your answer for each question must exceed 275 words. During your quiz, you can use a 3X5 index card filled on one side only with handwritten information. The instructor must approve the card before you start the quiz. Your performance will be evaluated on the basis of 3 criteria: word count, multiple specifics from the reading and adequate answer to the question. If one or more of these criteria are not met, you will receive a "Not Pass" otherwise a "Pass" represents a completed "Extra". There are several versions of each quiz so classmates reading the same book will probably not get the same quiz. If you do not pass a quiz you can not be re-tested for that "Extra". You must return both your answer sheet and the quiz form when you are finished. The graded quiz will be available for you to review but must be returned to the instructor immediately after reviewing it. **No book quizzes can be taken Dead Week or later.**

### II. Video Assignments

You will have the opportunity to check out (for no more than 3 days) a genetic based video. Your task will be to watch the program (maximum 2 hours in length) and schedule a quiz with the instructor. The quiz will pose three essay questions of which you will choose one to answer. You can use any notes that you have hand written during the video to support your answer to the question you chose. Your performance on the quiz will be evaluated on the basis of the quality of your answer (emphasizing accuracy & depth) to the question and whether you used specific examples cited from the video. If these criteria are satisfactorily met, you will receive a "Pass", otherwise you will not be able to count this as a completed "Extra". There will be no opportunities to attempt a "re-do" of this particular video.

Your choice of videos include: "**Decoding Annie Parker**" (a somewhat fictionalized movie describing Mary Clare King’s discovery of the BRCA1 gene); and documentaries: "**Thinking Big: Medical Disorders and Dwarfism**, "**In the Womb: Identical Twins**, "**Boy in the Bubble**, "**Sexual Evolution: From X to Y**, and "**The Strength of Families: Solving Rett Syndrome**".

**University Academic Honor Code:** Angelo State University expects its students to maintain complete honesty and integrity in their academic pursuits. Students are responsible for understanding the Academic Honor Code, which is contained in both print and web versions of the Student Handbook.

**Religious holy day:** A student who intends to observe a religious holy day during the semester should make that intention known in writing to the instructor during the first week of the semester and one week prior to the absence. If this submission is completed, a student who is absent from classes for the observance of a religious holy day shall be allowed to take make-up exams or assignments scheduled for that day in accordance with syllabus policy.

**Disability Statement:** Persons with disabilities which may warrant academic accommodations must contact the Student Life Office (SLO), in order to request and to implement academic accommodations. The instructor must be given a written description of the accommodations originating from the SLO.