HUMAN GENETICS
Bi 428/528 Winter, 2021
MW 6:40 pm-8:20 pm

Professor: Dr. Debbie Duffield
e-mail: duffieldd@pdx.edu

REQUIRED TEXT: Korf & Irons – Human Genetics and Genomics (4th ed.). This will be available via an Online PDF. Go to library.pdx.edu, click on course reserves, then enter this title search "Human Genetics and Genomics 4th Edition -- Online PDF" - log in with odin account. **If you want a hardcopy, you can also buy it used (an inexpensive way to go) on Amazon. All additional material will be on D2L.**

PLEASE NOTE: Bi 341 or equivalent is required for this course.

General objectives: To acquaint students with terminology, current research and issues in a variety of areas of human genetics, including: pedigree and chromosomal analysis, sex determination, development, metabolic pathways, gene families and gene organization, genome mapping, imprinting, epigenetics, bioethics, cancer genetics and immunity. Our emphasis will be to look at techniques used to define normal, as well as, abnormal patterns and disorders.

Class organization: Because of the COVID pandemic, this class will be predominantly guided self-learning, although there will be a multitude of journal reading assignments, required participation in a weekly discussion of these articles on the Wed class time, special assignments, a midterm and a final. We will be using D2L as our platform. The "on line" required in-class participation will be the discussion group on Weds at 6:40 pm on Zoom. Specific assignments are listed in the weekly assignments below. Weekly instructions will be given by email from me EACH MONDAY. **Grading**: Weekly reading summaries &/or assignments (150 pts), Pedigree Assignment (50 pts), Midterm (150 pts), Final (150 pts).

Organization of the weekly discussion groups: you will be assigned to a discussion group based on your last name. These group discussions will be on Zoom and one of you in each group can volunteer (for extra credit) to be the leader and set up the Zoom meeting and take attendance. Discussion Groups: 1 = A through K; 2 = L through P; 3 = R through Z. This process will be explained further in your weekly emails from me.

Journal Article Summaries: All students are required to read a current journal article for weeks 2,3,4,6,8 (CRISPR) and 9. You will present a short synopsis of your article in the Group Discussion **and** email me a 1 pg. summary of the article (see email address above). Summaries **must include the full reference (authors, title, journal, volume, pages), as well as a brief summary.**

PROPOSED TOPICS WILL GENERALLY FOLLOW KORF AND IRON (4th Ed) with additional material given in D2L by week

PRELIMINARY SYLLABUS – THIS MAY BE REFINED AS WE GO

Date: Weeks 1-10 Topic

Week 1: Jan 4-6: Pedigree Analysis, Mendelian Disorders

1. Reading Assignment: Korf and Irons 1,2,**3**: D2L = Week 1.
2. **Special ASSIGNMENT (due Wed 6:40 pm discussion hour):** Explore "Online Mendelian Inheritance in Man" (OMIM) to find a description of a *single gene Mendelian disorder*. Prepare a 1 pg. summary to include: a) a description of the disorder, b) the mode of inheritance, c) the gene responsible and where it is located on the chromosomes, d) the frequency of occurrence (prevalence), and e) if available, the molecular – developmental - physiological consequence of the gene defect. In your discussion group (required attendance) on Wed, you will be asked to briefly describe what you have found and then submit your summary to me by email (see Discussion Group Assignments above).
3. Pedigree Assignment: This will be sent to you by email on Monday (1/4) and is due by Friday (1/8) 6:00 pm back to me by email.

Week 2: Jan 11-13: Chromosome structure and nomenclature, syndromes

1, Reading: Korf and Irons 6,9: D2L = Week 2.
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 3: Jan 18-20: Sex determination

1. Reading: D2L = Week 3
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 4: Jan 25-27: Development and Epigenetics/Imprinting

1. Reading: Korf and Irons 6, 12: D2L = Week 4
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 5: Feb 1-3: Midterm and Metabolic Pathways/Inborn Errors of Metabolism

1. **Midterm: You will be given the midterm by email on Monday (2/1). It is due back to me by Wednesday (2/3) 6:00 pm.**
2. Reading: Korf and Irons 11: D2L = Week 5

Week 6: Feb 8-10: Metabolic pathways/Inborn Errors of Metabolism (cont.)

1. Reading: Korf and Irons 10, 11, 13, 14: D2L = Week 6
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 7: Feb 15-17: Mapping & Linkage Analysis, **Mapping Complex Disease**
1. Reading: Korf and Irons 4, 5: D2L = Week 7
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 8: Feb 22-24: The human genome and Genome-wide Association Studies
1. Reading: Korf and Irons 4, 14: D2L = Week 8
2. Find and summarize a current journal article relating to this week's topic for presentation in your weekly discussion group time (required attendance: see Discussion group assignments and Journal Summary format). Submit your summary to me via email.

Week 9: Mar 1-3: Bioethics
1. Reading: Korf and Irons 10, 13: D2L = Week 9.
2. Discussion time will be used to participate in a panel on bioethics.

Week 10: Mar 8-10: Cancer Genetics
1. Reading: Korf and Irons 15, 16, 17: D2L = Week 10
2. For Discussion group: What is CRISPR - be able to summarize basically how it works. Find and report on one example of how CRISPR has been used in human genetics. Submit your summary as usual.

*** * MONDAY MAR 15: FINAL EXAM * ***
You will be given the exam at 9:00 am Monday (3/15) on D2L
and must submit your answers by 6:00 pm Wednesday (3/17)

Additional books of interest:

Garrett, L., The Coming Plague: emerging diseases in a world out of balance. 1994 (and as applicable now as then!).

Rhodes, R., Deadly Feasts: tracking the secrets of a terrifying new plague. 1997.

Ridley, M., Genome: the autobiography of a species in 23 chapters. 1999.

Strachan & Read – Human Molecular Genetics 4