562: Human Cytogenetics 2015

Location: 2121 Mechanical Engineering

Time: Thursdays 8:30-10:20am

Instructors:
Jennifer Laffin, PhD, FACMG
(jjlaffin@wisc.edu)
Kimberly Anderson, MS, CGC
Chris Vlangos, PhD

Course Objective: Med Gen 562 is an introduction to clinical laboratory genetics with particular focus on the relevance of human cytogenetics and molecular genetics to the practice of medical genetics. The course content will include human chromosome structure, function, nomenclature, and current clinical laboratory methods. Following completion of the course and assigned readings, students are expected to understand the mechanisms that lead to genetic alterations and their relevance to the genetic disease.

Course Design

- Review Learn@UW content in preparation for class discussion
- Quiz on topic (session 1-5 quizzed are available until the midterm and sessions 6-11 are available until the final exam)
- Classroom case-based discussion

Learn@UW: Learn@UW is an integral part of Medical Genetics 562. It is important you read through the required activities for each session. Sessions may require readings and viewings (online ppt's, Prezi's, digital videos etc.) that should be completed prior to class.

Readings on Learn@UW are designated as required or supplemental. Students will be responsible for understanding the material presented in the required readings. Undergraduates are not responsible for the graduate student specific required readings, but may be useful in clarifying or expanding topics.

Graduate Students:
Each week you will be required to participate in the course discussion board. Discussion will be focused on graduate student specific required readings and is a place to discuss things learned and additional questions.

Grades: UW Graduate School grading system [A, A/B, B, B/C, C, D, F] based on the following distribution:

- **40% Weekly Quizzes**: Quizzes (11 total) will be available within Learn@UW and based on classroom and required reading content. You will have one chance to complete the quizzes. They will be automatically graded once submitted.
- **10% Group Project and Presentation**
- **20% Midterm Exam**- all material, including online content, up to exam date will be covered
- **30% Final Exam**-all course material will be covered

Classroom Discussion Date	Topic
1/22	Session 1:Introduction and Brief History of Diagnostic Genetics (Quiz 1) • Cytogenetics • Molecular Genetics • Germline vs Somatic Genetic Variation
1/29	Session 2:Genome Structure and Function (Quiz 2) • Structure (Chromosomes and DNA) • Function
2/5	Session 3:Genetic Variants- Cytogenetic (Quiz 3) • Aneuploidy • Reciprocal Translocations • Inversions • Markers • Complex rearrangements • Chromosome instability • Fragile Sites and Heteromorphisms

562: Human Cytogenetics 2015

Classroom Discussion Date	Topic
2/12	Session 4:Genetic Variants-Molecular (Quiz 4) • Missense • Nonsense • Indel/frameshifts • Inversions • Insertions • Copy number variants • Imprinting/Epigenetic modifications
2/19	Session 5:Laboratory Methods and Nomenclature (Quiz 5) • Nomenclature o Cytogenetic (ISCN) o Molecular (HGVS) • Basic laboratory procedures o Cytogenetics ▪ Cell culture and chromosome analysis ▪ Fluorescence in situ hybridization o Molecular ▪ DNA (genomic studies) ▪ RNA (expression studies)
2/26	**Midterm Exam**
3/5	Session 6:Prenatal Genetics I (Quiz 6)- **Kimberly Anderson, MS, CGC** • Advanced maternal age • Family history
3/12	Session 7:Prenatal Genetics II (Quiz 7)- **Kimberly Anderson, MS, CGC** • Fetal abnormalities • Pregnancy loss
3/19	Session 8:Constitutional Genetics I (Quiz 8)- **Kimberly Anderson, MS, CGC** • Infertility • Intellectual and developmental disabilities • Multiple congenital anomalies • Autism spectrum disorders Organization of Group Projects
3/26	Session 9: Constitutional Genetics II (Quiz 9)- **Chris Vlangos, PhD** • Risk for inherited disease (including cancer predisposition syndromes) • Pharmacogenetics
4/2	**Spring Break**
4/9	Session 10: Oncology/Acquired I (Quiz 10)- **Chris Vlangos, PhD** • Pharmacogenetics • Leukemia
4/16	Session 11: Oncology/Acquired II (Quiz 11)- **Chris Vlangos, PhD** • Lymphoma • Solid tumors
4/23	**Group Projects**
4/30	**Group Projects**
5/7	**Group Projects/Review**
5/11	**10:05-12:05 FINAL EXAM**