Fall 2015

Course Announcement

*CMM 695D Human Genetic Disease Colloquium* (3 units)

Cross-listed with BIOC, GENE, MCB, PSIO, NRSC

**Instructor:** Linda L. Restifo, MD, PhD  
Prof. of Neurology, Neuroscience, and Cellular & Molecular Medicine  
Member, GIDPs in Genetics, Neuroscience, and Insect Science  
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**Time:** Tuesdays and Thursdays, 10:30 am – 12 noon  
**Place:** Life Sciences North 452

**Format:** Introductory lectures by the instructor (classification of human disease, differential diagnosis, types of mutations, and genetic technology), followed by student presentations. We focus on FOUR topics representing a range of genetic mechanisms, disease pathogenesis, and organ system involvement.

**Topics** in Fall 2014 were (alternatives for Fall 2015 include):
- **Fragile X syndrome** (cystic fibrosis, muscular dystrophy)
- **Alzheimer’s disease** (cardiomyopathies, epilepsies)
- **Type 1 diabetes** (multiple sclerosis, asthma, type 2 diabetes)
- **Autism spectrum disorder** (alternative: schizophrenia)

**Readings:** primarily from the biomedical literature.
- Student presenters will select assignments in consultation with the instructor.  
- Highly recommended: *Thompson & Thompson Genetics in Medicine, 7th ed.*  
- Use of a medical dictionary is essential.

**Educational goals:**
- approaching the study of any human disease topic;  
- understanding historical changes in research approaches to a disease;  
- critical analysis of research on disease genetics and pathophysiology;  
- understanding the range of genetic causation of and risk for disease;  
- evaluation of animal models of disease;  
- understanding the challenges in translational medicine for therapeutics;  
- effective, professional presentation skills and peer review;  
- appreciating the challenge of classification of human disease.

**Student assessment:**
Student’s presentations (50%) and participation in class discussion (50%)

**Prerequisites:**
Graduate-level genetics, molecular biology, or biochemistry; some knowledge of mammalian anatomy/physiology is helpful. Rarely, advanced undergraduates may be ready for this course; contact Dr. Restifo to discuss.

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*a This course is complementary to Dr. Vercelli’s *Biology of Complex Diseases* (595H). Many students find it valuable to take both courses (in either order).*