Fall 2016

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 and Neuroscience
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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 pathogeneses, and organ system involvement.

Topics in Fall 2015 were (alternatives for Fall 2016 include):

- **Duchenne/Becker muscular dystrophy** (cystic fibrosis, Fragile X syndrome)
- **Amyotrophic lateral sclerosis** (cardiomyopathies, epilepsies, Alzheimer's disease)
- **Asthma** (multiple sclerosis, diabetes, types 1 or 2)
- **Schizophrenia** (autism spectrum disorder)

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;
- recognizing 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. Please contact Dr. Restifo to discuss.

This course is complementary to Dr. Vercelli's *Biology of Complex Diseases* (CMM595H).
Many students find it valuable to take both courses (in either order).