

**Molecular Genetics of Human Disease**

**Bi 427/527**

**Syllabus Fall 2019**

| <b>Date</b> | <b>Topic</b>   | <b>Assigned reading</b>   | <b>Presenters</b> |
|-------------|--|---|-------------------|
| 10/1        | Cystic fibrosis - Mendelian recessive disease genes                              | Rowe et al. 2005. New Eng. J. Med. 352: 1992–2001.                | Monte Westerfield |
| 10/3        | Cystic fibrosis  |   | Monte Westerfield |
| 10/8        | Muscular dystrophy - X-linked disease genes                                      | Dalkilic and Kunkel. 2003. Curr. Opin. Genet. Dev. 13: 231–238.   | Monte Westerfield |
| 10/10       | Marfan syndrome - autosomal dominant disease genes                               | Eldadah et al. 1995. J. Clin. Invest. 95: 874–880.                | Monte Westerfield |
| 10/15       | Polycystic kidney disease - somatic mutations                                    | Qian et al. 1996. Cell 87: 979–987.                               | Monte Westerfield |
| 10/17       | Discussion - Cystic fibrosis; discussion questions due                           | Watson et al., 2011. The FASEB Journal. doi:10.1096/fj.11-186080  | Group 1           |
|             | Discussion - Muscular dystrophy; discussion questions due                        | Lu et al., 2003. Nature Med. 9: 1009–1014.                        | Group 2           |
| 10/22       | Angelman, Prader-Willi, & Williams syndromes - diseases of copy number variation | Doelken et al., 2013. Dis. Mod. Mech. 6: 358-372.                 | Monte Westerfield |
| 10/24       | Usher syndrome - diseases of protein complexes                                   | Cosgrove & Zallocchi, 2014. Int. J. Biochem. Cell Bio. 46: 80–89. | Monte Westerfield |
| 10/29       | Fanconi anemia   | Taniguchi. 2008. Gene Reviews.                                    | John Postlethwait |
| 10/31       | Genome-wide association studies (GWAS)   | Altshuler et al., 2008. Science 322: 881-888.                     | Albert Edwards    |
| 11/5        | Age-related macular degeneration   | Lotery and Trump. 2007. Hum Genet (2007) 122:219–236              | Albert Edwards    |
| 11/7        | Discussion - Polycystic kidney disease; discussion questions due                 | Song et al. 2009. Hum. Mol. Gen. 18: 2328–2343.                   | Group 3           |
|             | Discussion - Usher syndrome; discussion questions due                            | Ebermann et al., 2010. J. Clin. Invest. 120: 1812–1823            | Group 4           |
| 11/12       | Neurodevelopmental diseases 1: Autism spectrum disorders                         | Zoghbi, 2003. Science 302: 826-830.                               | Phil Washbourne   |
| 11/14       | Neurodevelopmental diseases 2: Rett Syndrome, Fragile X mental retardation       |   | Phil Washbourne   |
| 11/19       | Alzheimer disease - epistasis in human disease                                   | Combarros et al., 2009. Neurobio. Aging 30: 1333–1349.            | Monte Westerfield |
| 11/21       | Discussion - Fanconi anemia; discussion questions due                            | Ceccaldi et al. 2011. J. Clin. Invest. 121: 184-94.               | Group 5           |
|             | Discussion - Macular degeneration; discussion questions due                      | Chen et al. 2010. PNAS 107: 7401–7406.                            | Group 6           |
| 11/26       | Schizophrenia - disease endophenotypes   | Sakar et al., 2016. Nature 530:177-183.                           | Monte Westerfield |
| 11/28       | Thanksgiving   |   |                   |
| 12/3        | Discussion - Rett syndrome; discussion questions due                             | Villard et al., 2000. Neurology 55: 1188-1193.                    | Group 7           |
|             | Discussion - Schizophrenia; discussion questions due                             | Abdolmaleky et al., 2006. Hum. Mol. Genet. 15: 3132–3145.         | Group 8           |
| 12/5        | Ethics of genetic testing and gene therapy                                       | Bell et al. 2011. Science Translational Med. 3: 65ra4.            | Monte Westerfield |
| 12/13       | Final paper due 8:00 am  |   |                   |