

Molecular Genetics of Human Disease

Bi 427/527

Syllabus Fall 2020

Date	Topic	Assigned reading	Presenter
9/29	Cystic fibrosis - Mendelian recessive disease genes	Rowe et al. 2005. New Eng. J. Med. 352: 1992–2001.	Monte Westerfield
10/1	Cystic fibrosis		Monte Westerfield
10/6	Muscular dystrophy - X-linked disease genes	Dalkilic and Kunkel. 2003. Curr. Opin. Genet. Dev. 13: 231–238.	Monte Westerfield
10/8	Marfan syndrome - autosomal dominant disease genes	Eldadah et al. 1995. J. Clin. Invest. 95: 874–880.	Monte Westerfield
10/13	Polycystic kidney disease - somatic mutations	Qian et al. 1996. Cell 87: 979–987.	Monte Westerfield
10/15	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
10/20	Angelman, Prader-Willi, & Williams syndromes - diseases of copy number variation	Doelken et al., 2013. Dis. Mod. Mech. 6: 358-372.	Monte Westerfield
10/22	Usher syndrome - diseases of protein complexes	Cosgrove & Zallocchi, 2014. Int. J. Biochem. Cell Bio. 46: 80–89.	Monte Westerfield
10/27	Fanconi anemia	Taniguchi. 2008. Gene Reviews.	Monte Westerfield
10/29	Genome-wide association studies (GWAS)	Altshuler et al., 2008. Science 322: 881-888.	Albert Edwards
11/3	Age-related macular degeneration	Lotery and Trump. 2007. Hum Genet (2007) 122:219–236	Albert Edwards
11/5	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
11/10	Neurodevelopmental diseases 1: Autism spectrum disorders	Zoghbi, 2003. Science 302: 826-830.	Phil Washbourne
11/12	Neurodevelopmental diseases 2: Rett Syndrome, Fragile X mental retardation		Phil Washbourne
11/17	Alzheimer disease - epistasis in human disease	Combarros et al., 2009. Neurobio. Aging 30: 1333–1349.	Monte Westerfield
11/19	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
11/24	Schizophrenia - disease endophenotypes	Sakar et al., 2016. Nature 530:177-183.	Monte Westerfield
11/26	Thanksgiving		
12/1	Discussion - Rett syndrome; discussion questions due	Villard et al., 2000. Neurology 55: 1188-1193.	Group 7
	Discussion - Schizophrenia; discussion questions due	Schizophrenia Working Group of the Psychiatric Genomics Consortium, 2014. Nature, 511:421-427.	Group 8
12/3	Ethics of genetic testing and gene therapy	Bell et al. 2011. Science Translational Med. 3: 65ra4.	Monte Westerfield
12/7	Final paper due 8:00 am		