

Molecular Genetics of Human Disease

Bi 410/510

Syllabus - Fall 2013

Date	Topic	Assigned reading	Presenters
Oct 1	Cystic fibrosis - Mendelian recessive disease genes	Rowe et al. 2005. New Eng. J. Med. 352: 1992–2001.	Monte Westerfield
Oct 3	Cystic fibrosis		Monte Westerfield
Oct 8	Muscular dystrophy - X-linked disease genes	Dalkilic and Kunkel. 2003. Curr. Opin. Genet. Dev. 13: 231–238.	Monte Westerfield
Oct 10	Discussion - Cystic fibrosis	Watson et al., 2011. The FASEB Journal. doi:10.1096/fj.11-186080	Group 1
	Discussion - Muscular dystrophy	Lu et al., 2003. Nature Med. 9: 1009–1014.	Group 2
Oct 15	Marfan syndrome - autosomal dominant disease genes	Eldadah et al. 1995. J. Clin. Invest. 95: 874–880.	Monte Westerfield
Oct 17	Genome-wide association studies (GWAS)	Altshuler et al., 2008. Science 322: 881–888.	Albert Edwards
Oct 22	Age-related macular degeneration	Lotery and Trump. 2007. Hum. Genet. (2007) 122:219–236	Albert Edwards
Oct 24	Polycystic kidney disease - somatic mutations	Qian et al. 1996. Cell 87: 979–987.	Monte Westerfield
Oct 29	Fanconi anemia	Taniguchi. 2008. Gene Reviews.	John Postlethwait
Oct 31	Discussion - Polycystic kidney disease	Song et al. 2009. Hum. Mol. Gen. 18: 2328–2343.	Group 3
	Discussion - Macular degeneration	Chen et al. 2010. PNAS 107: 7401–7406.	Group 4
Nov 5	Neurodevelopmental diseases 1: Autism spectrum disorders, Rett Syndrome	Zoghbi, 2003. Science 302: 826-830.	Phil Washbourne
Nov 7	Neurodevelopmental diseases 2: Fragile X mental retardation		Phil Washbourne
Nov 12	Discussion - Fanconi anemia	Ceccaldi et al. 2011. J. Clin. Invest. 121: 184–94.	Group 5
	Discussion - Rett syndrome	Villard et al., 2000. Neurology 55: 1188–1193.	Group 6
Nov 14	Next-gen sequencing & mapping	Lo et al., 2010. Sci. Transl. Med. 2: 61ra91.	Bill Cresko
Nov 19	Usher syndrome - diseases of protein complexes	Saihan et al., 2009. Cur. Opin. Neurol. 22: 19–27.	Monte Westerfield
Nov 21	Alzheimer disease - epistasis in human disease	Combarros et al., 2009. Neurobio. Aging 30: 1333–1349.	Monte Westerfield
Nov 26	Schizophrenia - disease endophenotypes	Thaker 2007. Expert Opin. Ther. Targets 11: 1189–1206.	Monte Westerfield
Nov 28	Thanksgiving - no class		
Dec 3	Discussion - Usher syndrome	Ebermann et al., 2010. J. Clin. Invest. 120: 1812–1823	Group 7
	Discussion - Schizophrenia	Abdolmaleky et al., 2006. Hum. Mol. Genet. 15: 3132–3145.	Group 8
Dec 5	Ethics of genetic testing and gene therapy	Bell et al. 2011. Science Translational Med. 3: 65ra4.	Monte Westerfield