Study Guide for Midterm Exam #1 – ECOL 453/553

1. What is the structure of a typical eukaryotic gene? A typical prokaryotic gene?

2. Describe the general differences between eukaryotic and prokaryotic genome structure.

3. What is the C value paradox? What is the major explanation for variation in genome size?

4. Describe the general kinds of repetitive DNA that are common in eukaryotic genomes.

5. What proportion of the human genome codes for proteins? What constitutes the remainder of the genome? (i.e. what are the major classes of sequences?)

6. Are mutations random with respect to fitness? What is the evidence for this?

7. What are the different ways that DNA sequences can mutate?

8. What causes mutations?

9. How can mutation rates be estimated directly? Indirectly? Do these approaches give similar answers? What is the average mutation rate in humans per bp? Per gene? Per genome? Does this rate differ among sites?

10. Does the mutation rate in humans differ among chromosomes? Why?

11. Define mutation. Define substitution. How are they different? Be very precise.

12. What is genetic drift? What is the consequence of genetic drift for genetic variation in natural populations?

13. What is effective population size? How is the effective population size different from the census population size?

14. What is the probability of fixation of a neutral mutation? Of a new neutral mutation?

15. Describe three mutational models. Which one is most commonly used to analyze DNA sequences?

16. Nucleotide diversity (π) and the proportion of segregating sites (θ) are two common estimators of genetic variation that use DNA sequence data. Make up a fake dataset of DNA sequences and then calculate π and θ from your dataset. What population parameter can be estimated with this information?

17. What is the neutral theory of molecular evolution? Outline the major ideas and predictions of the neutral theory. Give explicit mathematical predictions, and then say, in words, what these relationships mean.

18. Does the rate of evolution depend on population size? If so, why? If not, why not? Consider advantageous, deleterious, and neutral mutations.

19. What are the different kinds of selection and how do they affect patterns of genetic variation in natural populations? Consider the consequences for levels of variation, patterns of LD, sojourn times, and the distribution of allele frequencies.

20. What is genetic hitchhiking?

21. Describe the major classes of statistical tests for selection that utilize data from DNA sequences. What kinds of data are needed for each? What are the assumptions behind each test? What patterns would be consistent with positive directional selection? With balancing selection? With the presence of slightly deleterious mutations? How would changes in population size affect these tests? How would you distinguish between selection and changes in population size in explaining observed patterns?

22. The MK test and HKA test both rely on patterns of polymorphism and divergence. How do these tests differ? What kinds of conclusions can be drawn from each? What is the effect of selection on linked sites in generating deviations from the null model using each test?

23. What is a genetic map and what is a physical map of a genome? What are they used for? Describe different kinds of physical maps and how they are constructed.

24. What is linkage mapping? What are the various strategies that can be used to map the genes underlying traits of interest using linkage mapping?

25. How do recombination rates vary across mammalian genomes? Outline three methods by which recombination rates can be measured or inferred. In what ways do these different approaches give consistent results?

26. What is LD? What is the relationship between levels of LD and recombination rate?

27. What is linkage disequilibrium mapping? How does it differ from linkage mapping? What are the advantages and disadvantages of each approach and when would you use them? What is the human HapMap?

28. What is meant by “forward genetics” and by “reverse genetics”?

29. Describe the commonly used forward genetic and reverse genetic approaches. When are they used and what kinds of information do these different techniques provide?