Tefferi et al (2002). Primer on medical genomics I. Part II: Background principles and methods in molecular genetics. *Mayo Clin Proc 77: 785-*

1. What is a gene in the Mendel sense? What is a gene in the modern sense?
2. What is DNA made of?
3. What is the structure of a gene?
4. How does the orientation of a strand play a role?
5. Name 3 categories of DNA. How do mini- and macrosatellites differ from general tandem repeats?
6. How does RNA differ from DNA
7. What is the diploid number of humans?
8. What is mitochondrial DNA? Does it reside in the cell nucleus?
9. Where does “homologous” in a homologous pair of chromosomes refer to?
10. In the context of gene expression, what is transcription, What is translation? How do these processes fit into the dogma of molecular biology?

11. What is reverse transcription PCR?
12. What is real-time PCR?

Refer to this paper as well, when you need additional information about

- Polymorphisms
- DNA sequencing
- Gene mapping

as supporting document to subsequent chapters.

Pompanon et al (2005). Genotyping errors: causes, consequences and solutions. *Nature Reviews Genetics*, 6:847-

1. What is non-evasive genotyping?
2. How do AFLPs differ from SNPs?
3. How can genotyping errors be spotted?
4. Which genotyping rates can one expect these days? Is the notion of a “small” genotyping error dependent on the context of the subsequent data analysis?
5. Why are there “unverified” SNPs in public data bases?
6. Although too restrictive, broadly speaking, genotyping errors can be categorized in 4 groups. Which ones?
7. What is size homoplasy? Where does it play a role? Link other consequences of the error for the genotype to appropriate causes as well.
8. What are allelic dropouts? Is this the same as “inactivation” (like in X-inactivation)? Why? Why not?

9. Link genetic variants as captured by microsatellites, SNPs and AFLPs to common contexts for those molecular markers.
10. Assign causes of genotyping error to one of four appropriate classes in 6.
11. What is a replicated genotype? What does it involve?
12. Name some common metrics for quantifying genotyping errors.
13. Why is there need for a common metric that is applicable to a wide variety of settings?
14. What is perhaps the most universal metric? Why?
15. When family-designs are considered, Mendelian inheritance checks can be performed. What do these checks involve? Can they exclusively pinpoint genotyping errors? Why? Why not?
16. How can an investigator distinguish between genotyping errors, mutations or the existence of some rare alleles?

17. What are some of the consequences of genotyping error in the context of linkage analysis? Note that recombination is a concept that is particularly relevant for linkage studies…
18. What are some of the consequences of genotyping errors in the context of a genetic association analysis? Note that linkage disequilibrium is one of the key concepts in a genetic association study.
19. What are the important steps in a genotyping process for limiting the occurrence and effect of genotyping errors? Name 5
20. What are F_{ST} estimates? Where do they play a role? Are they prone to genotyping errors?
21. What is a population bottleneck?
22. Name 3 false results that emerge from incorrect genotyping in the context of population genetic studies.
23. In terms of analysis, name 3 strategies to acknowledge the presence of genotyping errors.

Schlötterer (2004). The evolution of molecular markers – just a matter of fashion? *Nature Reviews Genetics*, 5:63-

1. What type of information does a genetic marker provide? How is it used to address one of the key questions in genetics?
2. What are allozymes? Are they useful in the context of genetic association studies? Why or why not?
3. In contrast to allozymes, what are DNA-based markers? What are the advantages of using DNA-based markers?
4. What are RFLPs? What downplayed their popularity?
5. What are minisatellites? How are they similar/dissimilar to RFLPs? What downplayed their popularity?
6. What is DNA fingerprinting?
7. What does PCR stand for and how did it change the evolution on molecular markers?
8. What are micro-satellites? How are they similar/dissimilar to minisatellites? How large is

a typical repeat region? What is the key reason for them to have gained popularity in mapping endeavors? What is a major drawback of microsatellites, hampering their use in population genetics studies?

9. Are AFLPs PCR-based? Do AFLPs require a priori knowledge about primer sequences in the target species?
10. What is a shotgun genome sequence?
11. SNPs have become one of the most important genetic markers in genetic (association) studies. Despite their success, they suffer from some shortcomings. Name 4.
12. Which technique offers the most finegrained genetic information? Hence, Is DNA sequence analysis or comparison an old-fashioned business or will it revive again.
13. Link advantages and weaknesses to the appropriate markers.
14. Which markers or variation capturing technique is the most optimal in the context of making inferences of demographic processes?

Can you make the link with the use of DNA sequences and phylogenetics?

15. Which markers or variation capturing technique is the most optimal in the context of paternity testing and forensics?
16. Which markers or variation capturing technique is the most optimal in the context of linkage analysis?
17. Which markers or variation capturing technique is the most optimal in the context of association analysis?