

Genetics Exam 2 study guide

Chapter 5 – tetrad analysis

If you can do the homework problems assigned for this section, you will be fine. To test yourself, do more of the homework questions on tetrad analysis.

Understand how to calculate the map distance between two genes, and between a gene and its centromere. Understand what you can do with unordered tetrads and ordered tetrads.

If your data are from ordered tetrads, you can use ascospore order to gain information about the distance between the gene and its centromere. You need to classify asci as first division segregation (AAaa or aaAA) or second division segregation (any other order, such as AaAa or AaaA). You should be able to estimate the distance between a gene and its centromere. You can also use unordered tetrad analysis to determine the distance between two genes.

Unordered tetrad analysis: you need to classify asci as PD, NPD or TT.

If the number of PD=NPD, the two genes you are looking at must be unlinked.

Study figure 5.22 on p. 206 of your book. You should understand each of these rules and why it tells you what it does. You should be able to apply these rules to solve a problem: either to estimate the distance of the gene from the centromere or the distance between two genes. On the test I'll give you the flow chart.

Understand the difference between tetrad analysis and analysis of recombination in organisms without tetrads (like flies or plants or humans).

You won't need to regurgitate the lifecycle of yeast or *Neurospora*, however, you need to understand it sufficiently to know what you are actually doing when you make a yeast or *Neurospora* cross. And you should understand that the ascospores are roughly equivalent to the haploid gametes produced by flies, except that haploid ascospores can grow up into full-fledged fungal bodies. Gametes need to fuse with another gamete.

You should know the terms: ascus, asci ascospore

Chapter 6

Review the Messelson-Stahl experiment. What did they do? And why was it considered evidence for semi-conservative replication?

Remember that DNA replication occurs in 5'-3' direction. Know what this means.

Know the difference between theta replication and rolling circle replication in circular DNA

Terms:

origin of replication

replication fork

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bidirectional replication
elongation
initiation
primer
proofreading
replication bubble

How many origins of replication are there in bacteria? in eukaryotes?

Know the enzymes involved in DNA replication, and their function, or what problem they solve
enzymes (helicase, gyrase, SSB, primosomes/primase/polymerase α , DNA polymerase complex/polymerase III/polymerase δ ,

What is the role of proofreading in DNA replication, how is it carried out? What is the significance of 3'-5' exonuclease activity? How many nucleotides can be excised by the 3'-5' exonuclease activity of the DNA polymerase?

Why is there a lagging strand and leading strand?
Why are there Okazaki fragments?
How are they sealed to create a continuous strand of DNA?
How are the RNA primers removed? What protein carries this out?

Understand how PCR works, what it is used for.

Understand how DNA sequencing works. Understand the role of dideoxynucleotides and colored dyes. You should be able to draw out this process.

What is shotgun sequencing? How is this related to the human genome project?

Understand what a genomic library is and how it is useful in shotgun sequencing.

What is gene conversion? How are recombination and mismatch repair involved in gene conversion?

What is the Holliday model of recombination?

What is the double stranded break model for recombination?

What is heteroduplex DNA? Why is it created?

Chapter 7

Supercoiling:

What enzymes control the level of supercoiling?

What is the advantage of **supercoiling** DNA?

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What is the relative importance of supercoiling in prokaryotes and eukaryotes?
If the DNA is nicked (single-stranded break), how does that influence supercoiling?
Will a nick cause the whole bacterial chromosome to become relaxed?
Why or why not?
What is a plasmid?
What is the difference between **topoisomerase I** and **topoisomerase II**
Which is involved in DNA replication and which is involved in supercoiling?

How is eukaryotic DNA packed?
Remember the different levels of packing
What proteins are important in "beads on a string"?
What are the beads and what is the string?
What are the **nucleosomes** composed of?
Does DNA have a negative or positive charge?
What is the charge of **histones**?
How does this allow the histones and DNA to automatically form beads on a string, even in a test tube?

What is the relationship between DNA packing and gene expression (transcription)?
What is 30nm chromatin fiber?
What is the role of the **scaffold**? Is it made from histones?
What level of chromatin packing is found in most interphase cells (cells not undergoing mitosis or meiosis)?

Know the terms chromatin, euchromatin and heterochromatin
What are the differences between **euchromatin** and **heterochromatin**?
Which is stained more darkly, which is packed more tightly, where are most genes?
Are there certain parts of the chromosome that typically have a lot of heterochromatin?

What are **polytene chromosomes** and why were they useful to early geneticists?

What regions of the chromosomes have a lot of **repetitive DNA**?
Does all repetitive DNA have a necessary role in the cell?
What are some examples of repetitive DNA that is junk repetitive DNA that is necessary?
Does highly repetitive DNA usually code for proteins?

What are some reasons why we might have multiple copies of a gene? Be prepared to give some examples.

What are **telomeres** and **centromeres**?
What is the role of cen sequences in yeast?
What is the effect of having many repeats of the centromere sequence?
Are all centromere sequences located in the same part of the chromosome?

What is the role of repetitive DNA at the ends of chromosomes?
What serious problem does telomerase solve?

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How does telomerase solve that problem?

Why is it difficult to replicate the ends of chromosomes?

Is telomerase active in all cells?

What is the consequence of this?

How does the telomerase know when to stop adding repeats?

Draw out the process by which telomerase prevents telomere shortening. This will help you to understand it.

Chapter 8

Why can humans survive **trisomy** of chromosome 21, but not chromosome 1?

Why can males survive even though they only have 1 X, while females have 2 Xs?

Why can people with 3 copies of the X chromosome survive?

What is **dosage compensation**?

What are the ways in which dosage compensation can occur?

(double expression (transcription) of X in males, suppress expression (transcription) of one X in females)

What is the **Barr body**? How does it relate to dosage compensation and calico cats?

How is the X inactivated in mammals? What gene is involved? How does it function.

What are the different types of chromosomal mutations?

Know the terms: **deletion, duplication, inversion, transversion, trisomy, monosomy, anuploidy, polyploidy**

Why are large deletions more likely to be problematic than small deletions?

Is a deletion likely to be advantageous and spread throughout a population?

How does a deletion effect meiosis?

What is a duplication?

How do duplications and deletions arise?

What happens after a gene is duplicated? Is the result always the same?

How do inversions influence meiosis? How do they prevent us from observing recombination? Do they prevent us from seeing recombination throughout the genome or just in the inverted region?

How do translocations influence meiosis?

What is the fertility level of an individual that is heterozygous for a translocation?

What group of organisms experiences frequent polyploidy events?

Why is polyploidy important in speciation?

What are **allopolyploids** and **autopolyploids**?

How do polyploids arise?

What is a **quantitative trait locus (QTL)**

What is the goal of QTL mapping?

How does it work?

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How does it allow us to find chromosomal regions where quantitative trait genes are located?

What does QTL mapping tell us? What do the results mean?

What is **Hapmap**? Why is it useful? Who was genotyped?

How could hapmap help us find a gene involved in some disease?

What is a haplotype?

What is a SNP?

Epigenetics

What is the difference between epigenetics and genetics

What are the mechanisms of epigenetics?

How can diet cause a heritable epigenetic change in gene expression?