

**Study material.**

This study guide covers material for the third hour exam: Regulation of transcription; RNA processing, stability, localization and translation; Physical and genetics maps; LOD scores and allelic association; Complex traits and types of allele. The exam is Tuesday, Nov. 22.

**Be able to define, discuss, and explain the following:**

polycistronic	monocistronic	antitermination
helix-turn-helix motif	recognition helix	CTD
core promoter	enhancer	silencer
general transcription factors	preinitiation complex	zinc finger
homeodomain	bZIP domain	bHLH domain
nucleosome	euchromatin	heterochromatin
PEV (position effect variegation)	consensus sequence	RNA polymerases I, II & III
DNAase hypersensitive site	LCR (locus control region)	<i>de novo</i> methylation
DNA methylation	histone acetylation	epigenetic
imprinting	maintenance methylase	spliceosome
exon	intron	lariat
SR protein	snRNP	ESE
cap	polyadenylation signal	nonsense-mediated decay
RNA editing	trans-splicing	aminoacyl-tRNA
aminoacyl-tRNA synthetase	initiator methionyl tRNA	peptidyl-tRNA
Shine-Dalgarno sequence	scanning model of eukaryotic initiation	
A site	P site	E site
nonsense mutations	missense mutations	Ran GTPase
small ribosomal subunit	large ribosomal subunit	EJC (exon junction complex)
RNAi	siRNA	miRNA
dicer	RISC	ubiquitin
FISH		
physical map	genetic map	cytological map
chromosome painting	pulsed gel electrophoresis	contig
radiation hybrid	YAC	BAC
STS	EST	microsatellites
CEPH kindred	lod score	SNP
autozygosity	linkage disequilibrium	allelic association
identity by descent	informative meioses	pseudogenes
haplotype	incomplete penetrance	phenocopy
genetic heterogeneity	polygenic determination	allelic series
hypermorph	neomorph	hypomorph
antimorph	dominant negative	haploinsufficient

**Thought-provoking questions:**

Why is it advantageous in a complex genome for genes to have multiple independently regulated enhancers?

What differences in gene expression and genome structure between prokaryotes and eukaryotes can be attributed, directly or indirectly, to differences in the mechanism of translation initiation?

Which tRNA is able to enter the P site of the ribosome without passing through the A site?

**Review questions.**

Know the properties of active and inactive chromatin.

Review sex determination in *Drosophila* (pp. 605-609) as an example of regulation by transcription and alternative splicing. Know and be able to compare the mechanisms of dosage compensation in mammals and in *Drosophila*.

Review pattern formation in *Drosophila* (pp. 826-838) as an example of transcriptional and post-transcriptional regulation of gene expression including RNA localization.

Be sure that you understand the yeast two-hybrid assay for protein-protein interactions, both as a technique and as an illustration of principles behind transcriptional activation.

Sometimes, two wild-type alleles at a locus are inherited, but only one is expressed. Be able to give at least two examples of this phenomenon (which is known as monoallelic expression).

Be able to explain the transmission disequilibrium test and sib pair analysis.

You are studying a disease known as Mount's syndrome that you believe is genetic. You have 100 patients, and you determine their genotypes at each of four biallelic marker loci A, B, C and D. As a control, you examine 100 unaffected people from the same region of the country. The following is a table of gene frequencies for each locus:

	locus A	locus B	locus C	locus D
Unaffected	85 A <sub>1</sub> , 15 A <sub>2</sub>	51 B <sub>1</sub> , 49 B <sub>2</sub>	78 C <sub>1</sub> , 22 C <sub>2</sub>	5 D <sub>1</sub> , 95 D <sub>2</sub>
Affected	83 A <sub>1</sub> , 17 A <sub>2</sub>	48 B <sub>1</sub> , 52 B <sub>2</sub>	37 C <sub>1</sub> , 63 C <sub>2</sub>	30 D <sub>1</sub> , 70 D <sub>2</sub>

Which alleles are associated with Mount's disease?

Can you conclude that marker locus C is linked to a gene affecting Mount's disease? If not, suggest alternative explanations.