# BIO 100 Biology Today – Exam 3 Study Guide

## Pedigrees and Patterns of Inheritance

Need to Know:
-Autosomes vs sex chromosomes
-Properties of a pedigree
-Modes of inheritance
    -Autosomal Recessive, autosomal dominant, sex-linked recessive, sex-linked dominant
-Important questions to ask to determine inheritance patterns of disease

## Central Dogma

-Layout of the central dogma
-Transcription
    -What is it.
    -What are the key biomolecules involved
    -Relationship between DNA and RNA
    -RNA polymerase is responsible for copying the DNA to RNA
    -Know what it means that the mRNA strand is complementary to the DNA strant
-Translation
    -What is it
    -Key biomolecules involved
    -Relationship between RNA and proteins
-Double-stranded nature of DNA
-Complementary nature of nucleotides in DNA
    -A complements T
    -G complements C
-RNA
    -different sugar group vs DNA
    -How many strands does it have
        -Is this different than DNA?
    -Contains Uracil instead of thymine
    -Know what base uracil is complementary to
    -Role of mRNA
    -Role of tRNA
-DNA vs RNA
    -Table of similarities and differences will be helpful to know
-Gene expression
    -Flow of genetic information from DNA to RNA to Protein
    -Where does transcription and translation fit into this flow
-Role of codons
    -What does the START codon do?
    -What does the STOP codon do?
    -Relationship between codons, tRNA, and amino acids

-***** You DO NOT need to know which codons code for which amino acids!
-*****You DO NOT need to know which codons are the start and stop codons

-mRNA Processing
-Know what introns and exons are
-Splicing
-How alternative splicing can create alternative proteins from the same transcript

-Ribosomes
-Know their role in translation
-Do not need to know APE sites

-Gene regulation
-What is a housekeeping gene?
-How is gene regulation involved in determining the "specialty" of a cell type?
-How is gene expression regulated by heterochromatin vs euchromatin?

Gene layout
-Promoter, coding region, enhancer
-What are the roles of promoters and enhancers in regulating gene expression?
-Role of transcription factors
-Bind to enhancers to speed up transcription

X-inactivation
-Occurs in females
-The inactive X-chromosome is greatly condensed
-What are the effects of condensing this particular copy of the X chromosome on gene expression from that chromosome?

## Myotonic Dystrophy

-Myotonia
-Autosomal Dominant
-Linkage analysis
-What it is used for
-What are "genetic markers" used for
-How does the linkage of genetic markers with mutant alleles allow us to determine chromosomal location?

-Mutation that causes myotonic dystrophy
-Trinucleotide expansion
-Located in a coding region or non-coding region?
-Why was it perplexing to scientists when initially identified?

-Ways to test whether protein levels are involved
-Disease mechanism of myotonic dystrophy
-RNA gain of function means that the RNA that's made from the mutant allele has gained a toxic function
-RNA accumulates and aggregates inside cell nuclei
-Splicing factor MBNL1 sequestered on RNA
-Impact that this has on splicing
-Relationship between mis-splicing and the phenotypes that affect many systems

-Genetic Anticipation

-What is it?

-Why does it occur in myotonic dystrophy?

-All you need to know about antisense morpholinos are that they act in the toxic transcripts in myotonic dystrophy and cause their degradation. This is why they are a potential therapy.