

Human Heredity Exam 2 Study Guide

Chapter 10 - From Proteins to Phenotypes

- Characteristics, regulation of transcription for proteins
- Ways of cloning regulatory sequences - proto-oncogenes, enhancers, and enhancers and insulators - what did you call regulatory elements?
- Essential vs. nonessential amino acids
- The structure of primary, secondary - alpha helix, beta sheet and other types
- Structure of hemoglobin
- Hemoglobin variants - what have they taught us?
- Sickle cell anemia - what type of mutation causes it?
- Beta thalassemia - what genetic mechanism is the cause?
- Regulation
- Phenotypic variation
- Drug resistance as genetic trait

Chapter 11 - Mutation: The Source of Genetic Variation

- Measuring mutation rates
- Spontaneous mutations vs. genetic mutations - what are the different results - silent mutations vs. disease?
- How mutations can be identified in populations
- How are mutation rates in humans measured? Number of silent vs. silent per silent?
- What is the mutation rate in a particular gene (beta-globin)?
- Environmental sources of mutations (ex. mutagens)
- Background mutation
- How much mutation is the average person exposed to?
- Spontaneous substitutions (ex. point mutations) vs. chromosomal mutations
- Mitosomes, cancer and mutagenic mutations
- What is the cause of Down's disease?
- Chromosomal repeats and deletions: expansion
- Gene amplification
- DNA repair and proofreading mechanisms and efficiency for DNA repair systems
- Genetic recombination

Chapter 12 - An Overview of the Cloning and Recombinant DNA (RMA)

- Cloning procedure: transfer and embryo splitting - what are the uses for cloning and RMA?
- Recombinant DNA technology and the use of recombinant proteins
- Use of vectors in recombinant DNA technology
- Advantages of using MHCs for cloning and human genes
- Cloning efficiency

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VM Jensen



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