Chapter 14.1 & 14.2 Study Guide

Know what a Karyotype is, and how a cytogeneticist prepares a karyotype.

Know that humans have 23 pairs of chromosomes for a total of 46. Chromosomes are a combination of histone proteins and DNA (deoxyribonucleic acid).

22 pairs of chromosomes are autosomes; they do not determine sex. The 23rd pair is the sex chromosomes. Human females (and mammals in general) have two X chromosomes, while males have one X chromosome and one Y chromosome.

The sex of a baby is determined by the sex chromosome in a father's sperm. If the sperm carries an X chromosome and fertilizes an ovum (which also has an X chromosome) the offspring is female (XX). If the sperm carries a Y chromosome and fertilizes an ovum the offspring is male (XY).

The Y chromosome is both necessary and sufficient for being male. No one who lacks a Y chromosome is a biological male, and the mere presence of the Y chromosome is enough to make someone a biological male.

Some human genetic disorders arise from having the wrong number of sex chromosomes (caused by non-disjunction in meiosis).

XXY = Kleinfelter syndrome (above average height, cognitive disabilities, decreased muscle tone)

X = Turner syndrome (cognitive disabilities, neck webbing, swollen digits, short stature)

XXX = Triple X syndrome (mild symptoms, sometimes have learning disabilities)

XYY = Jacob's syndrome (mild symptoms, sometimes have learning disabilities)

Know how to do a Punnett square for a sex-linked (X-linked) trait. Know that sex-linked recessive traits show up in males far more often than females (and know why!).

Know the difference between codominance, incomplete dominance, polygenic inheritance, and multiple alleles. Be able to give an example for each one. Describe the distribution of a human trait for polygenic inheritance.

For an extra challenge, be able to analyze the inheritance of epistatic traits (one gene controls the expression of another gene, 9:3:4 ratio with a dihybrid cross) and lethal alleles (2:1 ratio with a dihybrid cross).

Know the difference between a chromosome based disorder versus a disease caused by a gene mutation. For example, Down syndrome is caused by a trisomy in chromosome 21, while sickle cell anemia is caused by a nucleotide substitution.

Describe the cause, symptoms, and pattern of inheritance for Down syndrome, sickle cell anemia, cystic fibrosis, Huntington's disease, and color blindness.

Analyze human blood types and their pattern of inheritance. Know that human blood types are examples of both codominance and multiple alleles. Know that type O blood is a universal donor, while type AB blood is a universal recipient. Be able to explain these blood donation patterns, too.

What is heterozygote advantage, and which two disease causing alleles (above) can be advantageous in some circumstances?

14.3 Study Guide

Describe the human genome project. What DNA sequences did researchers look for to locate a gene? Why is there so much variety in human phenotypes if all humans share the same genes?

Why were restriction enzymes useful for the human genome project? Where do restriction enzymes come from, and what is their function in nature?

Be able to show how a restriction enzyme like *EcoR1* cuts DNA along a palindromic sequence, i.e. GAATTC. Why does *EcoR1* create "stick ends?"

Explain the process of *gel electrophoresis*. Know that the smaller fragments travel farther and faster through the gel, and that the larger fragments travel slower and a shorter distance. Describe how a scientist would compare the genetics of two individual with the results of gel electrophoresis.

How were *dideoxyribonucleic acid* nucleotides used to sequence fragments of DNA? Your book refers to these as nucleotides with a colored dye attached to them.

Shotgun sequencing was first used to construct large genomes from DNA fragments. Fragments are sequenced individually, and then a computer algorithm looks for areas of overlap and puts the fragments together.

Human DNA sequences are very similar regardless of race or ethnicity. However, comparing individuals, *single nucleotide polymorphisms* (SNPs) occur once about every 1,200 nucleotides.

The complexity of an organism is not a reliable indication of how much DNA or how many chromosomes it has. *Ophioglossum reticulatum* (a type of fern) has 1260 chromosomes!