GRADE 12 BIOLOGY
CHAPTER 14 STUDY GUIDE
HUMAN GENETICS

Name: MrE

Date: _____

Answer open ended questions in grammatically proper form; three or four questions may require external research

SUMMARY

Human Heredity

Biologists can analyze human chromosomes by looking at a karyotype. A karyotype is a picture of the chromosomes from a cell arranged in homologous pairs.

Humans have 46 chromosomes. Two of these chromosomes, X and Y, are the sex chromosomes. Females have two X chromosomes (XX). Males have one X and one Y chromosome (XY). The other 44 chromosomes are called autosomes.

Human genes are inherited according to the same principles of genetics described by Mendel. To study the inheritance of human traits, biologists use a pedigree chart.

A pedigree shows the relationships within a family. The inheritance of a certain trait in a family can be traced using a pedigree. From this, biologists can infer the genotypes of family members.

It is difficult to associate an observed human trait with a specific gene. Many human traits are polygenic, meaning that they are controlled by many genes. The environment also influences many traits.

Some of the first human genes to be identified were those that control blood type. Red blood cells can carry two different antigens, called A and B. Antigens are molecules that can be recognized by the immune system. The presence or absence of the A and B antigens produces four possible blood types: A, B, AB, and O.

The ABO blood types are determined by a single gene with three alleles. In addition to the ABO antigens, there is another antigen on red blood cells called the Rh antigen. People who have the Rh antigen are Rh positive. People without it are Rh negative. A single gene with two alleles determines the Rh blood group.

There are several human genetic disorders, including phenylketonuria (PKU), Huntington's disease, and sickle cell disease.

PKU is caused by a recessive allele. It is expressed only in individuals who have inherited a recessive allele from each parent.

Huntington's disease is caused by a dominant allele. It is expressed in any person who has that allele.

Sickle cell disease is caused by a codominant allele. Scientists are beginning to understand which changes in the DNA sequence cause certain genetic disorders.

Cystic fibrosis is caused by the deletion of three bases in the middle of the sequence for a protein. This deletion inactivates the protein, which causes the symptoms of this disorder.

14–2 Human Chromosomes

The two smallest human chromosomes, chromosomes 21 and 22, were the first chromosomes to have their DNA sequences identified. Both have many genes important for health. Both have regions of DNA that do not code for proteins. Genes located on the X and Y chromosomes, the sex chromosomes, are said to be sex-linked. They are inherited in a different pattern than genes located on autosomes.

For example, all alleles linked to the X chromosome, including those responsible for colour blindness, hemophilia, and Duchenne muscular dystrophy, are expressed in males even if they are recessive alleles. However, in order for these recessive alleles to be expressed in females, there must be two copies of them.

Females have two X chromosomes. Males have only one. To account for this difference, one X chromosome in females is randomly turned off. The turned-off chromosome forms a dense region in the nucleus known as a Barr body. Barr bodies are not found in males because their single X chromosome must be active.

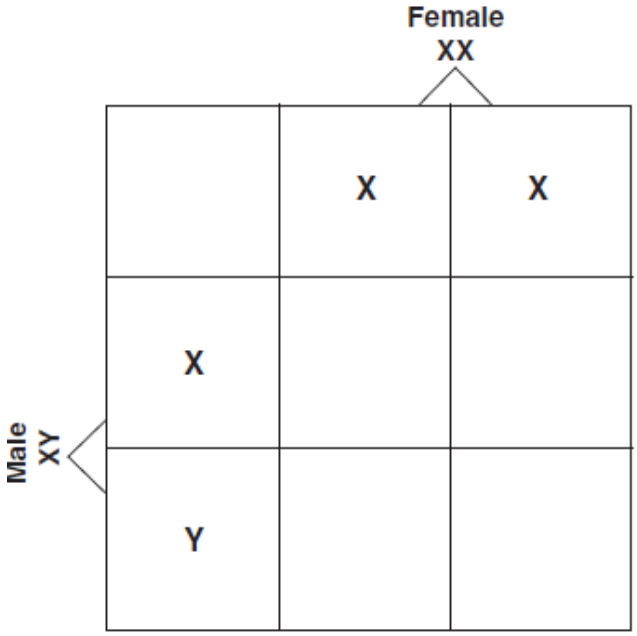
Human Chromosomes

1. How do biologists make a karyotype?

2. Circle the letter of each sentence that is true about human chromosomes:

a. The X and Y chromosomes are known as sex chromosomes because they determine an individual's sex.
b. Males have two X chromosomes.
c. Autosomes are all the chromosomes, except the sex chromosomes.
d. Biologists would write 46,XY to indicate a human female

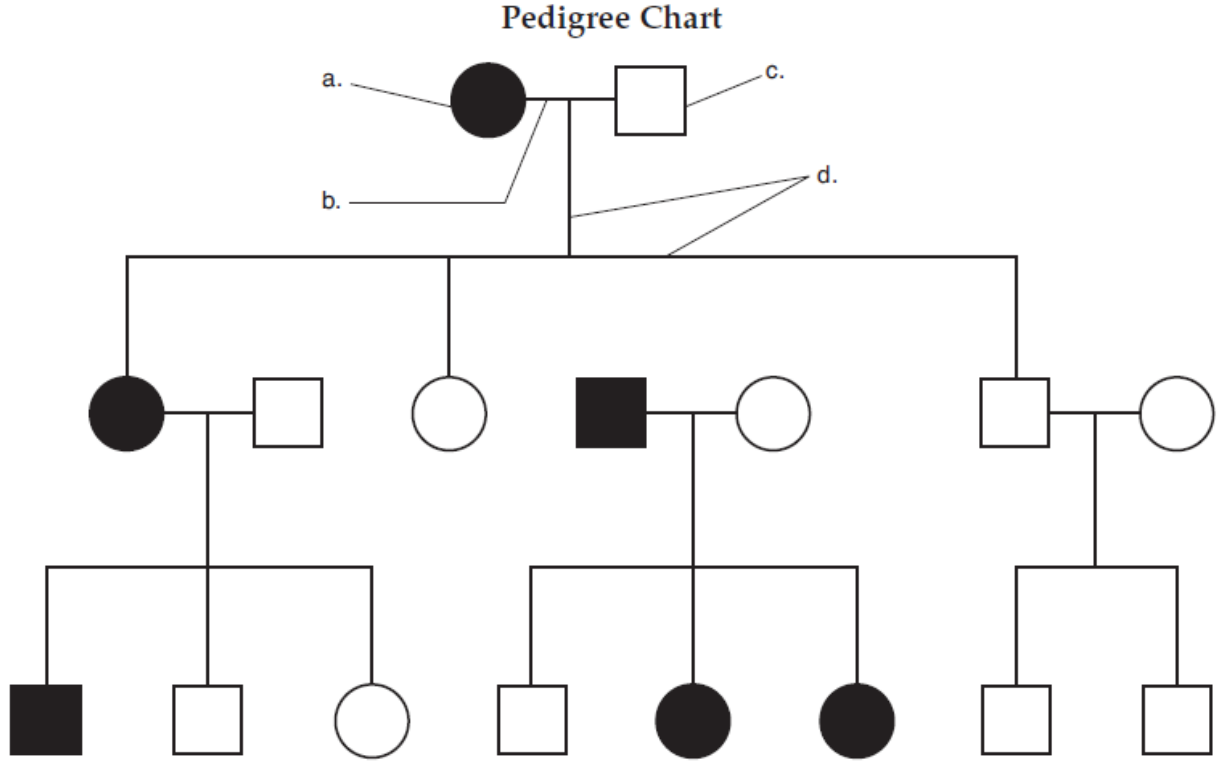
3. Complete the Punnett square below to show how the sex chromosomes segregate during meiosis.



4. Why is there the chance that half of the zygotes will be 46,XX and half will be 46,XY?

5. What does a pedigree chart show?

Match the labels to the parts of the pedigree chart shown below. Some of the parts of the pedigree chart may be used more than once.



_____ **6.** A person that expresses the trait
_____ **7.** Amale
_____ **8.** Aperson who does not express the trait
_____ **9.** Represents a marriage
_____ **10.** Afemale
_____ **11.** Connects parents to their children

20. A normal human diploid zygote contains
    a. 23 chromosomes.
    b. 46 chromosomes.
    c. 44 chromosomes.
    d. XXY chromosomes.

21. A chart that traces the inheritance of a trait in a family is called a(n):

a. pedigree. b. karyotype. c. genome. d. autosome.

22. An example of a trait that is determined by multiple alleles is:

a. cystic fibrosis. b. ABO blood groups.
c. Down syndrome. d. colourblindness.

23. What is the difference between autosomes and sex chromosomes?

24. Is it possible for a person with blood type alleles I^A and I^B to have blood type A? Explain your answer.

Human Genes

25. Why is it difficult to study the genetics of humans?

26. Circle the letter of each sentence that is true about human blood group genes.

a. The Rh blood group is determined by a single gene.

b. The negative allele (Rh^-) is the dominant allele.

c. All of the alleles for the ABO blood group gene are codominant.

d. Individuals with type O blood are homozygous for the *i* allele (*ii*) and produce no antigen on the surface of red blood cells.

27. Is the following sentence true or false? Many human genes have become known through the study of genetic disorders. _____

28. *Match the genetic disorder with its description (Will need to investigate this on the internet or another Biology book).*

Description	**Genetic Disorder**
_____ **30.** Nervous system breakdown caused by an autosomal recessive allele	**a.** Phenylketonuria (PKU)
_____ **31.** A form of dwarfism caused by an autosomal dominant allele	**b.** Tay-Sachs disease
_____ **32.** A buildup of phenylalanine caused by an autosomal recessive allele	**c.** Achondroplasia
_____ **33.** A progressive loss of muscle control and mental function caused by an autosomal dominant allele	**d.** Huntington's disease

Human Genes and Chromosomes

1. Circle the letter of each sentence that is true about human genes and chromosomes.

a. Chromosomes 21 and 22 are the largest human chromosomes.
b. Chromosome 22 contains long stretches of repetitive DNA that do not code for proteins.
c. Biologists know everything about how the arrangements of genes on chromosomes affect gene expression.
d. Human genes located close together on the same chromosome tend to be inherited together.

Sex-Linked Genes

2. What are sex-linked genes?

3. Is the following sentence true or false? _____

" The Y chromosome does not contain any genes at all "

6. Complete the Punnett square to show how colour blindness is inherited. (It is a recessive trait)

		X^cY	
		X^c	Y
X^cX^c	X^c		
	X^c		

8. A mutation involving a change in a single DNA base pair

a. will definitely result in a genetic disease.
b. will have no effect on the organism's phenotype.
c. will produce a positive change.
d. may have an effect on the organism's phenotype.

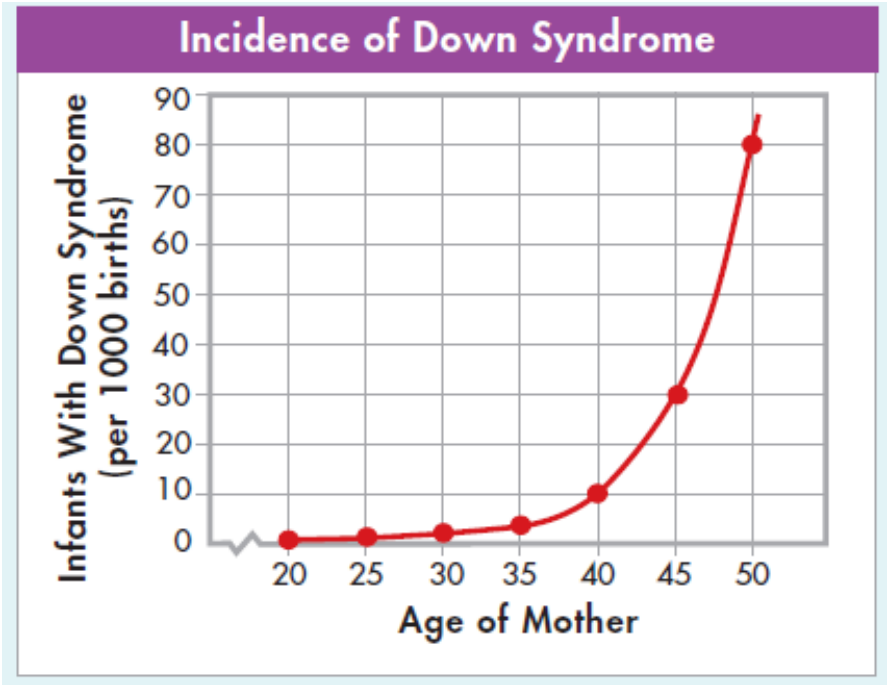
9. Cystic fibrosis is caused by:

a. nondisjunction of an autosome.
b. a change of three base pairs in DNA.
c. nondisjunction of a sex chromosome.
d. deletion of an entire gene from a chromosome.

10. Malaria is a disease caused by a:
a. gene mutation.
b. defect in red blood cells.
c. bacterium found in water.
d. parasite carried by mosquitoes.

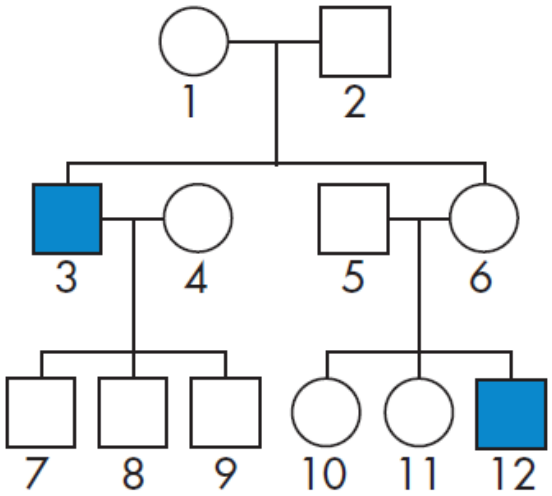
12. What is a chromosomal disorder?

15. **Interpret Graphs**. What can you infer about the relationship between the age of the mother and the incidence of Down syndrome?



Answer:

16. Hemophilia is an example of a sex-linked disorder. Two genes carried on the **X** chromosome help control blood clotting. A recessive allele in either of these two genes may produce hemophilia. The pedigree shows the transmission of hemophilia through three generations of a family.



31. **Interpret Diagrams** Which mothers are definite carriers of the gene?
32. **Apply Concepts** Why did the sons of Person 3 not inherit the trait?
33. **Apply Concepts** How could Person 12 have hemophilia if neither of his parents had hemophilia?

Standardized Test MUTIPLE CHOICE

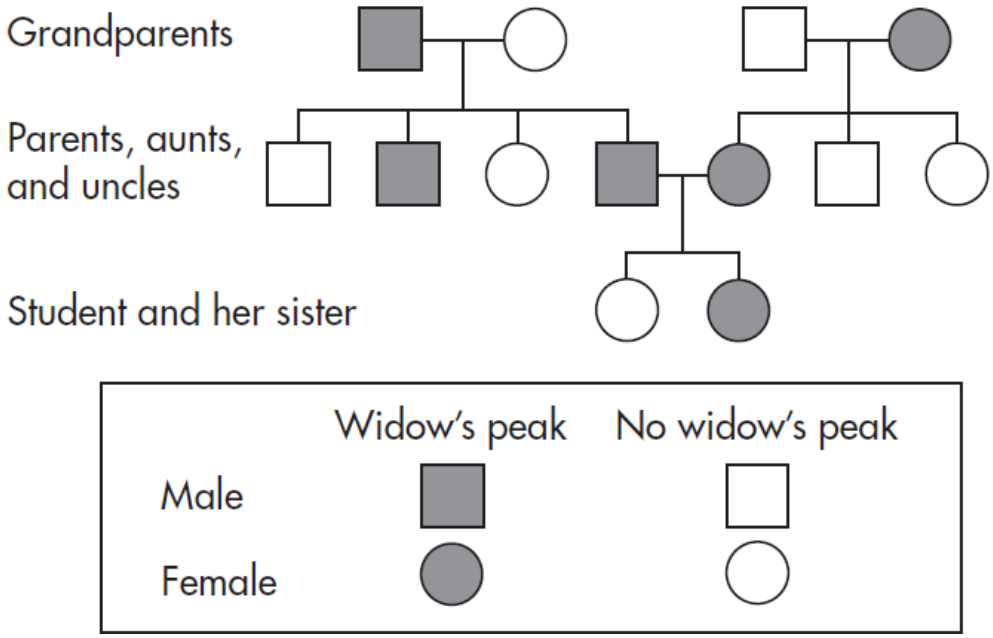
1. Which of the following disorders can be observed in a human karyotype?:
   A colourblindness
   B trisomy 21
   C cystic fibrosis
   D sickle cell disease

2. Which of the following disorders is a direct result of nondisjunction?:
   A sickle cell disease
   B Turner's syndrome
   C Huntington's disease
   D cystic fibrosis

3. A woman is homozygous for A^- blood type. A man has AB^- blood type. What is the probability that the couple's child will have type B^- blood?

   A 0% **C** 75% **B** 50% **D** 100%

4. A student traced the recurrence of a widow's peak hairline in her family. Based on her interviews and observations, she drew the pedigree shown below.



5. Which pattern of inheritance is consistent with the pedigree?

A sex-linked inheritance
B complete dominance
C codominance
D multiple alleles

6. What are the probable genotypes of the student's parents?

A Mother—*Ww*; Father—*ww*
B Mother—*ww*; Father—*ww*
C Mother—*WW*; Father—*Ww*
D Mother—*Ww*; Father—*Ww*