Name: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ U5T3 Meiosis

Period: \_\_\_\_\_\_ Page: \_\_\_\_\_

**Practice Using Karyotypes**

# Objectives

* Define and explain the vocabulary terms listed below.
* Analyze karyotypes to determine the sex.
* Infer chromosomal abnormalities from a karyotype.
* Observe similarities and differences among human chromosomes.

# Vocabulary

* Karyotype
* Autosome
* Sex chromosome
* Nondisjunction
* Monosomy
* Trisomy

# Introduction

* *Karyotype Formation*

Karyotyping is the way geneticists identify, organize, and study human chromosomes. Cells are taken from tissue and made to reproduce in a culture. After cells have reproduced a few times, the cells are then treated with a chemical that stops mitosis at metaphase. At this stage of cell division, the chromosomes are easiest to observe. This is because during metaphase the chromosomes are at the best length for identification. The chromosomes in the cells are then observed under a microscope where they are counted, checked for abnormalities, and photographed. The photo is enlarged and the individual chromosomes are cut out. The chromosomes are then matched with their homologous pairs. Recall that homologous chromosomes are identical or matching chromosomes, one chromosome is from the mother and the other chromosome is from the father. The arrangement of homologous pairs of chromosomes is called a **karyotype.**

The geneticists then look at the distinctive banding patterns created after the chromosomes have been stained. They use these patterns to help them organize the chromosomes into homologous, or matching, pairs. This arrangement of chromosomes is called a karyotype. Geneticists use karyotypes to determine the sex of a person. They also can use them to see whether a person has a genetic disorder.

* *Naming Chromosomes*

Humans have 46 chromosomes (23 homologous pairs) in every single one of their diploid body (somatic) cells. Out of these 23 homologous pairs, the first 22 pairs of chromosomes are called the **autosomes**, and the 23rd pair of chromosomes are called the **sex chromosomes** because they are the ones that determine the individual’s sex. A female’s sex chromosomes are XX, where a male’s sex chromosomes are XY. The twenty-three pairs of chromosomes are divided into seven groups according to their length and position of their centromeres.

* *Chromosomal Abnormalities*

Several human genetic disorders are caused by the addition of a chromosome, the deletion of a chromosome, and/or damaging chromosomes. One way of studying genetic disorders is to observe a karyotype of an individual’s chromosomes. Chromosomal abnormalities often result from **nondisjunction**. Nondisjunction is the failure of chromosomes to separate properly during meiosis, specifically during anaphase I. Nondisjunction results in cells having too many or too few chromosomes. One type of nondisjunction is **trisomy**. Trisomy is an abnormality in which a cell has an extra chromosome or section of a chromosome. This means the cell will contain 47 chromosomes instead of 46 chromosomes. Down syndrome, also known as Trisomy 21, is an example of a chromosomal abnormality that results from having an extra copy of chromosome 21.

**Karyotype Activity**

## **Part A: Analysis of a “Normal” Karyotype**

Observe the karyotype below. Notice the following:

1. The banding patterns, size, and general shape are identical for each of the homologous pairs of chromosomes.
2. The homologous pairs of chromosomes are arranged by sized and then numbered from 1-23. This is to make the analysis easier.

****

Sex is Explain why you know this is the sex.

# Part B: “What’s Wrong with my Karyotype?”

For each of the given karyotypes complete the following steps:

1. Identify: Next to each karyotype, identify it as either normal or abnormal. If the karyotype is normal skip to step #3.
2. Abnormality: If the karyotype is abnormal, circle the abnormality in the karyotype. Then explain what is wrong with the karyotype, and where the abnormality is located specifically on the chromosome. You will be looking for missing chromosomes and/or parts of chromosomes, extra chromosomes and/or parts, or anything that makes the karyotype abnormal or different from the normal karyotype above with the exception of the 23rd pair of chromosomes.
3. Gender: Identify the karyotype as male or female.

### **Karyotype #1**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

### **Karyotype #2**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

### **Karyotype #3**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

### **Karyotype #4**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

### **Karyotype #5**



Is the karyotype normal or abnormal? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

### **Karyotype #6**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the

karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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The gender of the karyotype is .

### **Karyotype #7**



Is the karyotype normal or abnormal?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

If abnormal, what is wrong in this karyotype? And where specifically is the abnormality located in the karyotype?

The gender of the karyotype is .

# Part C: Analysis Questions

1. How many autosomes are in a normal human karyotype?
2. Explain the following statement: The human male determines the sex of all of his offspring.
3. Explain how genetic counselors might use karyotypes with their patients.

## **Part D: Identifying Genetic Disorders**

The chart below describes several genetic chromosomal abnormalities and their symptoms. All of the abnormalities below result from nondisjunction.

|  |  |  |
| --- | --- | --- |
| **Name of Abnormality** | **Affected Chromosome** | **Brief Description of the Resulting Phenotypic Abnormality** |
| Down Syndrome  | 21 | 47 chromosomes, mental retardation with specific characteristics or physical features, may have heart defects and respiratory problems. |
| Edwards Syndrome | 18 | 47 chromosomes, severe mental retardation very characteristic malformations of the skull, pelvis, and feet among other things. Die in early infancy. |
| Patau Syndrome | 13 | 47 chromosomes, abnormal brain function that is very severe, many facial malformations, usually die in early infancy. |
| Turner’s Syndrome | Female XOonly 1 X chromosomes | 45 chromosomes, in females only, missing an X chromosome, secondary sex characteristics do not develop, infertile. |
| Klinefelter’s Syndrome | Male XXY2 XX chromosomes | 47 chromosomes, in males only, tall, sterile, small testicles, otherwise normal in appearance. |
| XYY Syndrome | Male XYY2 YY chromosomes | 47 chromosomes, in males only, low mental ability, otherwise normal in appearance. |
| Triple X Syndrome | Female XXXextra X chromosomes | 47 chromosomes, sterility sometimes occurs, normal mental ability. |

# USE THE CHART

# Now go back to karyotypes #1-7 in Part B and identify the genetic abnormality by name. If there are NO abnormalities in the karyotype write “normal” on the line.

# Karyotype #1 Karyotype #5

# Karyotype #2 Karyotype #6

# Karyotype #3 Karyotype #7

Karyotype #4