

Karyotyping

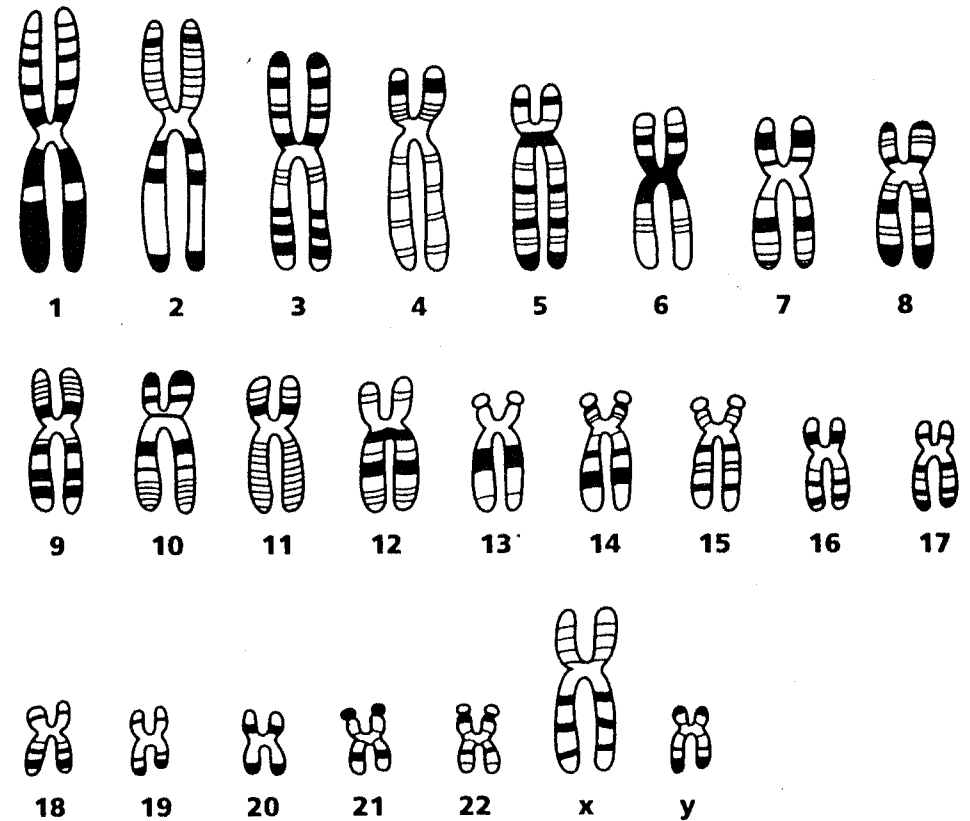
As a medical lab technician, one of your jobs is to assist with prenatal testing. Currently, you are working on the case of Mr. and Mrs. Smith. Mrs. Smith is pregnant, and her doctor has recommended amniocentesis, which is a type of prenatal testing. You have been given photomicrographs of the chromosomes in the unborn baby's cells, which were obtained through amniocentesis. Your job is to complete and analyze a karyotype of these cells to determine the sex of the Smiths' baby and whether the baby is normal or has Down syndrome.


Humans have 46 chromosomes in every diploid ($2n$) body cell. The chromosomes of a diploid cell occur in **homologous pairs**, which are pairs of chromosomes that are similar in size, shape, and the position of their centromere. In humans, 22 homologous pairs of chromosomes are called **autosomes**. The twenty-third pair, which determines the individual's sex, make up the **sex chromosomes**. Females have only one type of sex chromosome, which is called an **X chromosome**. Males have two types of sex chromosomes, an X chromosome and a much smaller **Y chromosome**. The diagram at the top of the next page shows each of the 22 types of autosomes and the 2 types of sex chromosomes.

A **karyotype** is a diagram that shows a cell's chromosomes arranged in order from largest to smallest. A karyotype is made from a photomicrograph (photo taken through a microscope) of the chromosomes from a cell in metaphase. The photographic images of the chromosomes are cut out and arranged in homologous pairs by their size and shape. The karyotype can be analyzed to determine the sex of the individual and whether there are any chromosomal abnormalities. For example, the karyotype of a female shows two X chromosomes, and the karyotype of a male shows an X chromosome and a Y chromosome.

Chromosomal abnormalities often result from **nondisjunction**, the failure of chromosomes to separate properly during meiosis. Nondisjunction results in cells that have too many or too few chromosomes. **Trisomy** is an abnormality in which a cell has an extra chromosome, or section of a chromosome. This means that the cell contains 47 chromosomes instead of 46. **Down syndrome**, or **Trisomy 21**, is a chromosomal abnormality that results from having an extra number 21 chromosome.

Human Chromosomes



 Carefully cut each chromosome from the chromosome spread. Be sure to leave a slight margin around each chromosome.

Arrange the chromosomes in homologous pairs. The members of each pair will be the same length and will have the centromere in the same location. Use the ruler to measure the length of the chromosome and the position of the centromere. Arrange the pairs according to their length, from largest to smallest. The banding patterns of the chromosomes may also help you to pair up the homologous chromosomes.

Tape each homologous pair to a Human Karyotyping Form, positioning the centromeres on the lines. Place the pairs in order, with the longest pair at position 1, the shortest pair at position 22, and the sex chromosomes at position 23.

The diagram you have made is a karyotype. Analyze the karyotype to determine the sex of the individual and whether or not the karyotype is normal.

CHROMOSOMAL ABNORMALITIES

Zygotes with abnormal numbers of chromosomes may undergo spontaneous abortion (about 50%), be stillborn, or lead "normal" lives. One study showed that 5% of all gametes contain chromosomal abnormalities.

The type of chromosomal abnormality seen in this activity is called **aneuploidy** which is the addition or deletion of one chromosome.

The loss of one autosome is always lethal and aborts early.

Aneuploidy Within the Sex Chromosomes

Monosomy X (Turner's Syndrome)

XO

one X chromosome, no Y

female appearance

under 5 feet tall, webbed neck, and a

broad "shieldlike" chest

sterile

may show mild retardation

1/4000 ♀

Trisomy X

XXX

generally, physically normal

may occur in 1/1000 females

may be fertile

Double Y Syndrome

XYY

may occur in 1/1000 males

physically normal, tall, over 6 feet tall

was at one time thought to be connected to criminal behavior (Speck Syndrome)

(newspapers sensationalized this)

Klinefelter's Syndrome

XXY

may occur in 1/1000 males

sterile, some breast development, mild retardation

Aneuploidy Among Autosomes

Trisomy 21 (Down's Syndrome)

47,+21

1/800 live births (incidence increases with age of mother)

mongolian fold in eyelid, retardation, simian increase in

palm, poorly developed circulatory and immune systems

Trisomy 18 (Edward's Syndrome)

47,+18

1/8000 births

severe mental and physical handicaps

die in infancy

Trisomy 13 (Patau's Syndrome)

47,+13

1/20,000 births

severe mental and physical handicaps

die in infancy

Chromosome Spread # _____

Name _____

1. Examine your karyotype. Is the baby male or female? _____

What is the $2n$ chromosome number? _____

Is this normal or abnormal? _____

If abnormal:

name condition _____

describe condition _____

Incidence of condition _____

2. Why are karyotypes important tools for geneticists?

3. Define:

monosomy

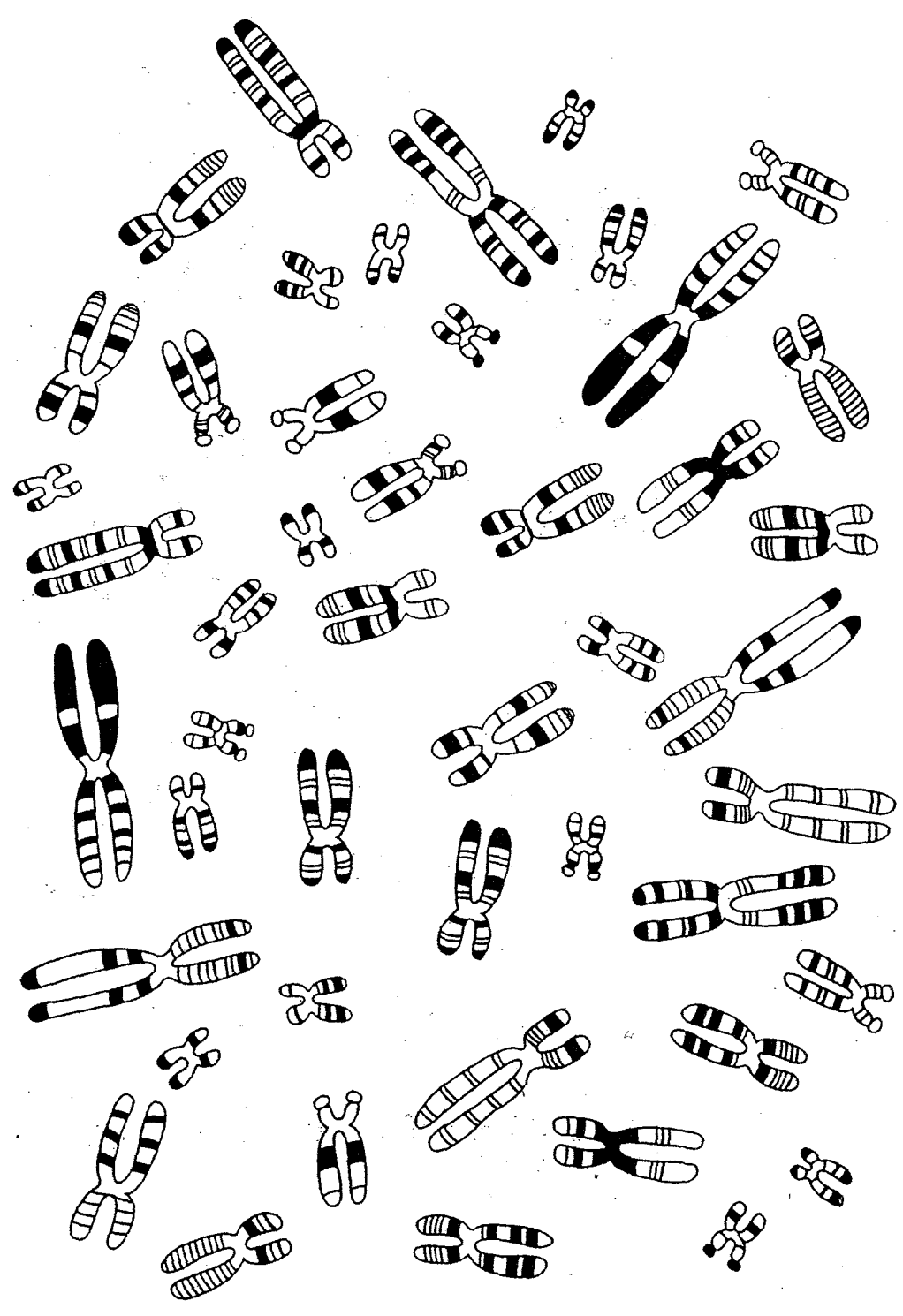
trisomy

nondisjunction

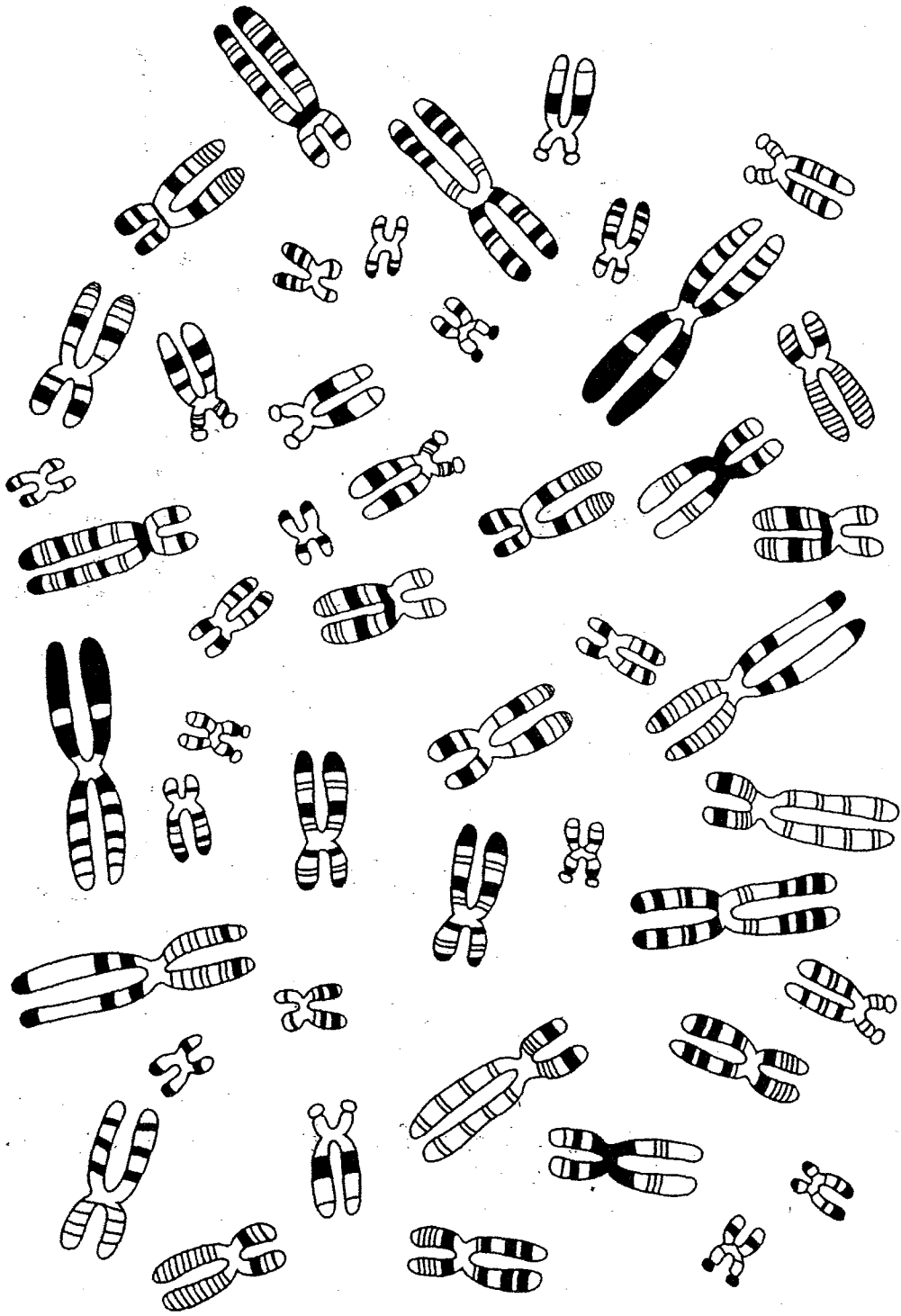
amniocentesis

aneuploidy

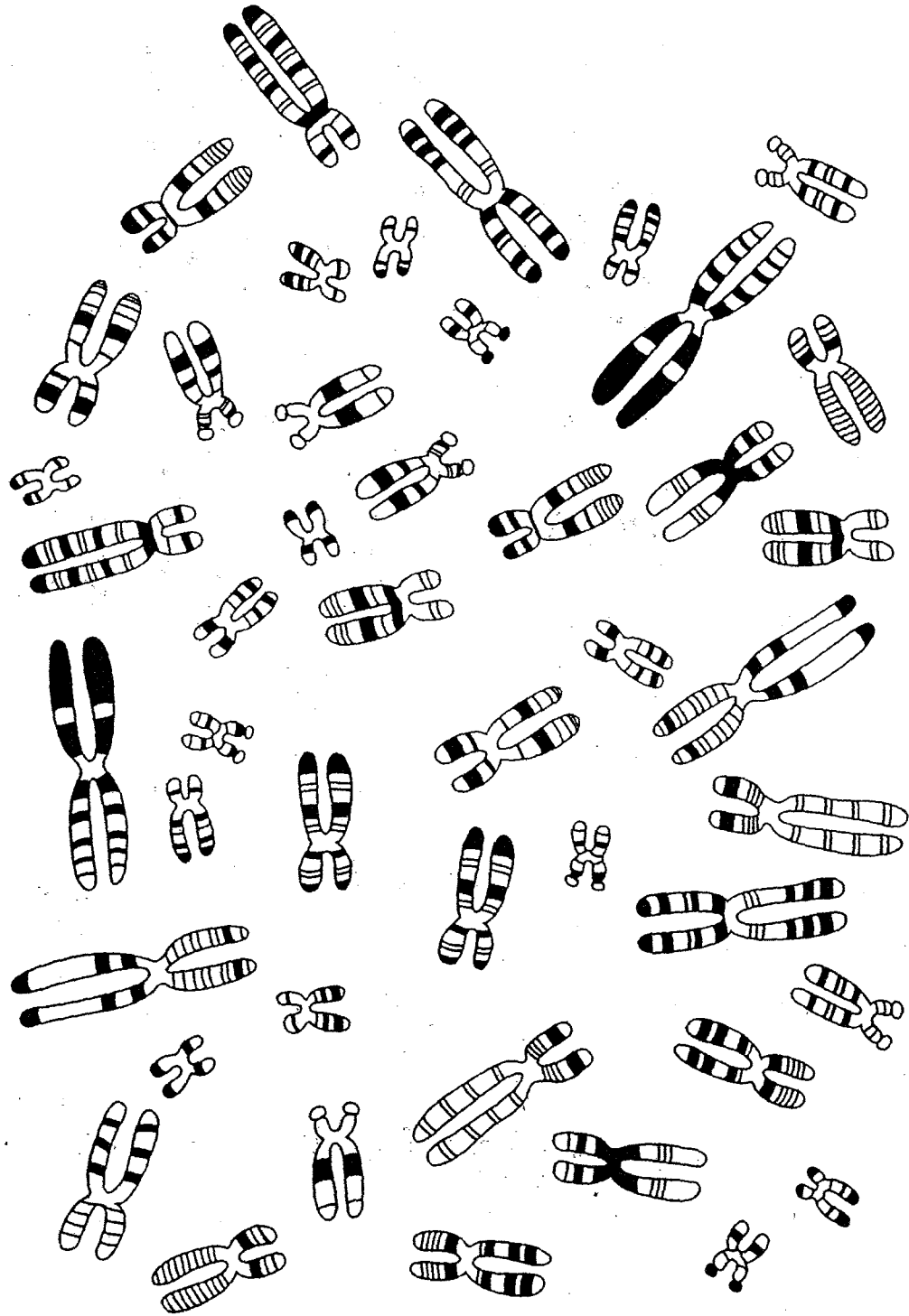
Chromosome Spread 1



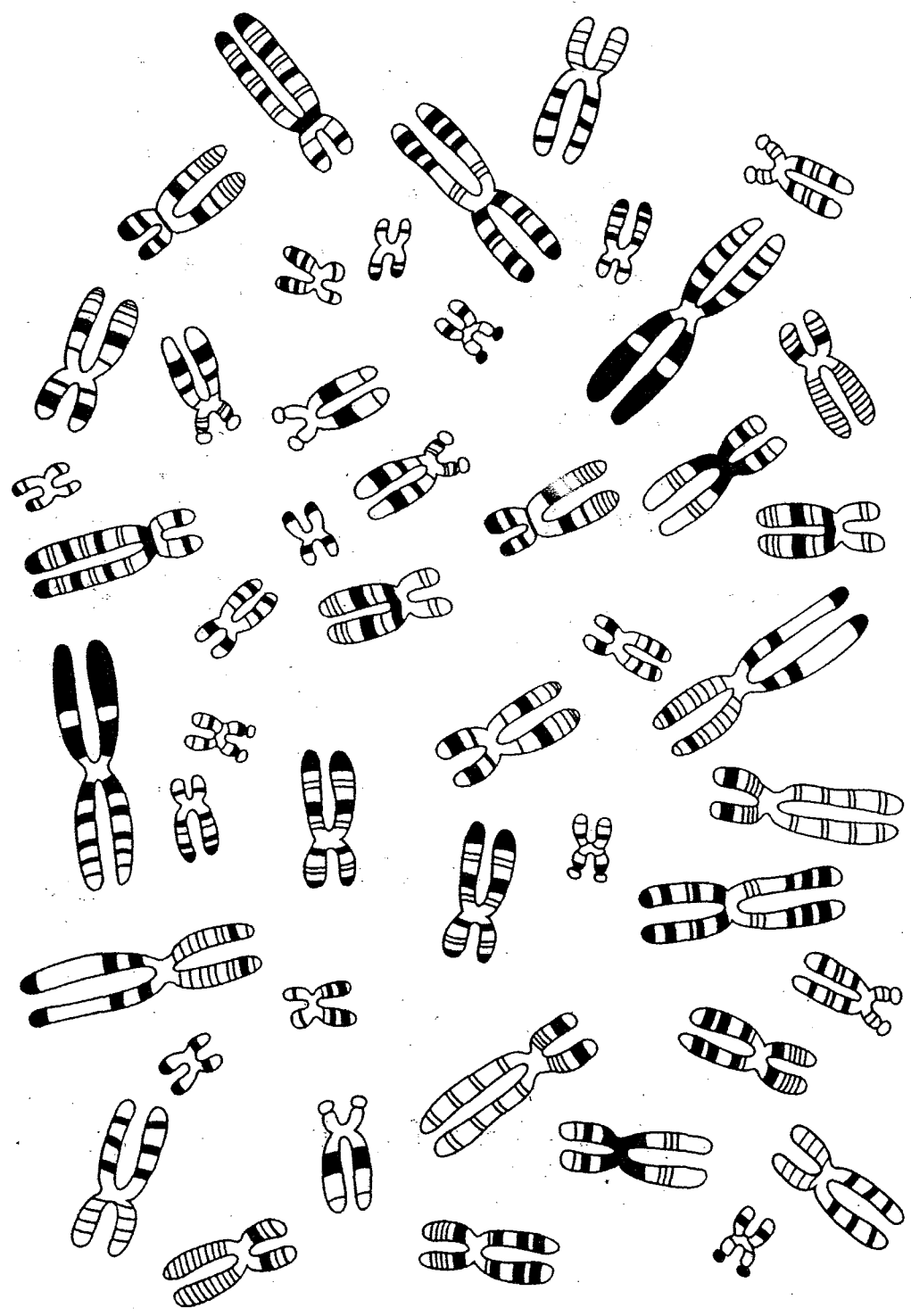
Chromosome Spread 2



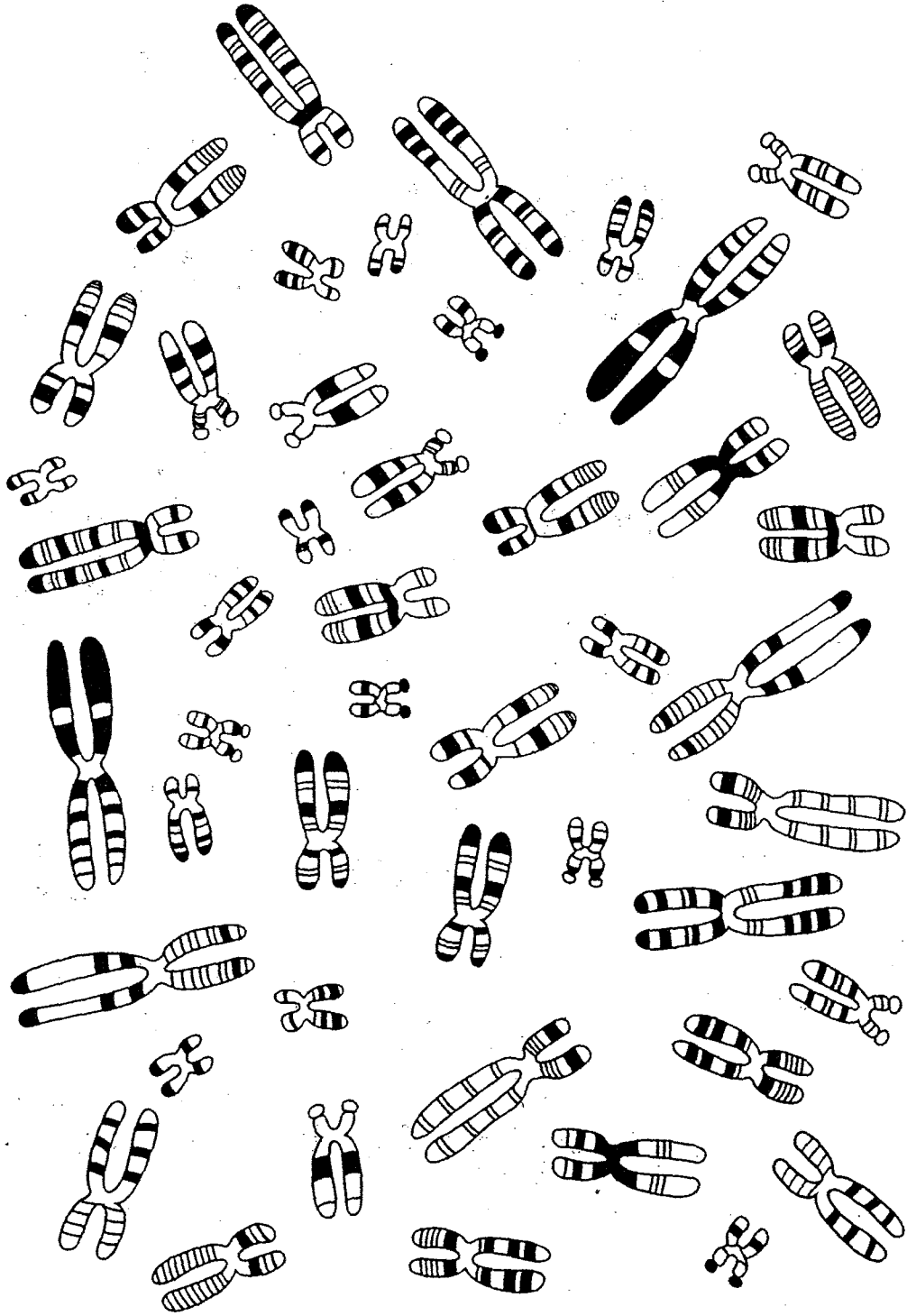
Chromosome Spread 3



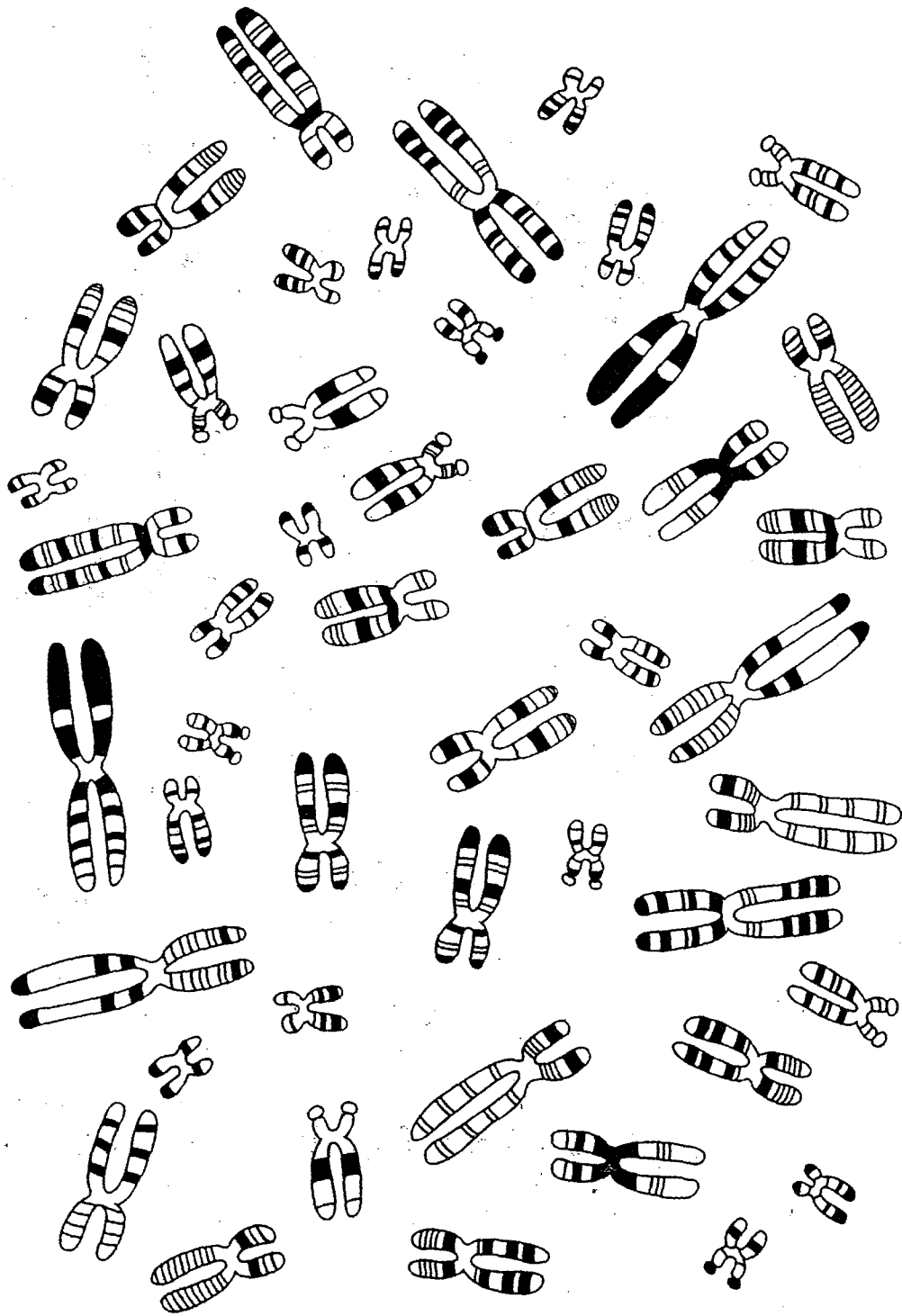
Chromosome Spread 4



Chromosome Spread 5

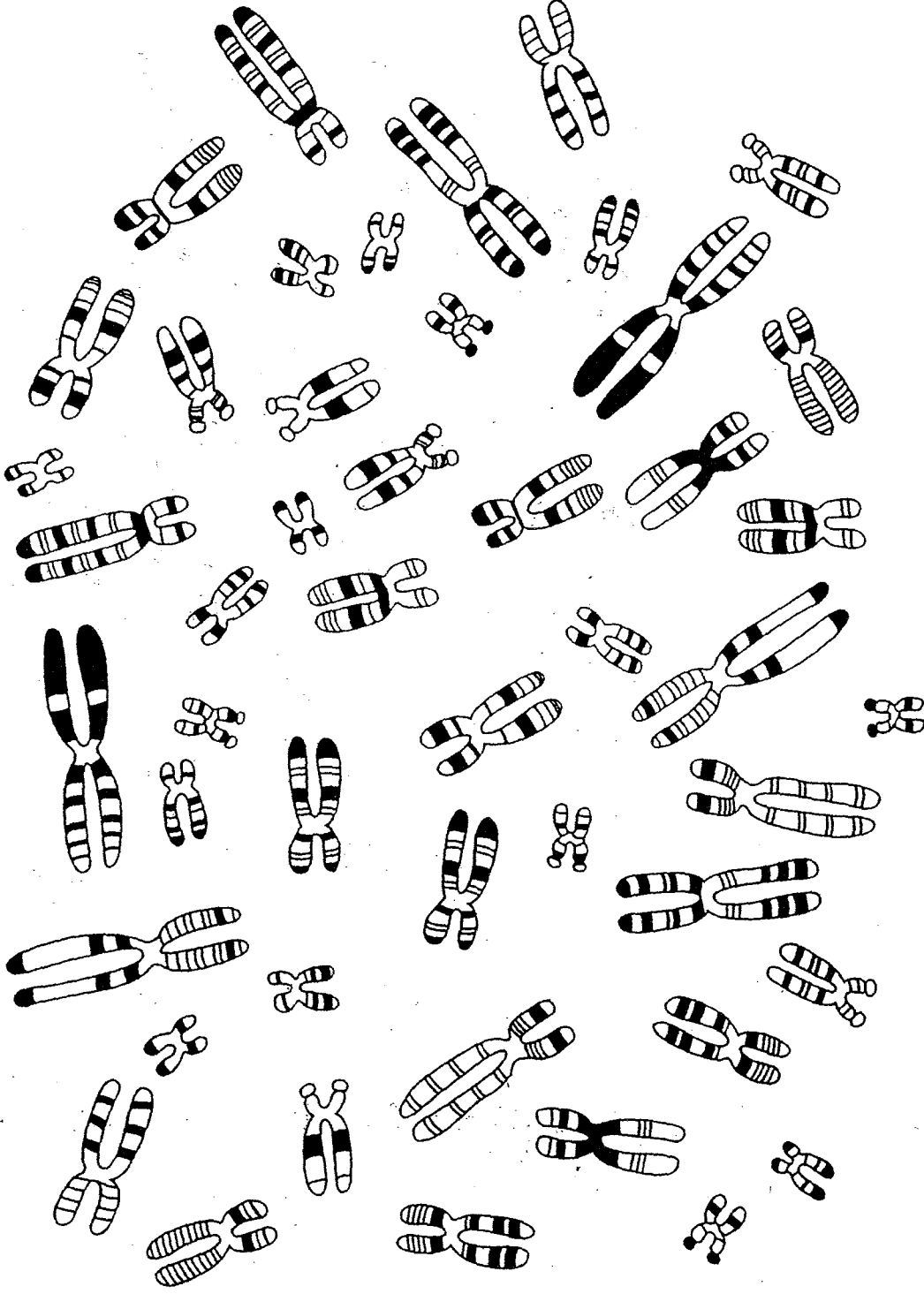


Chromosome Spread 6



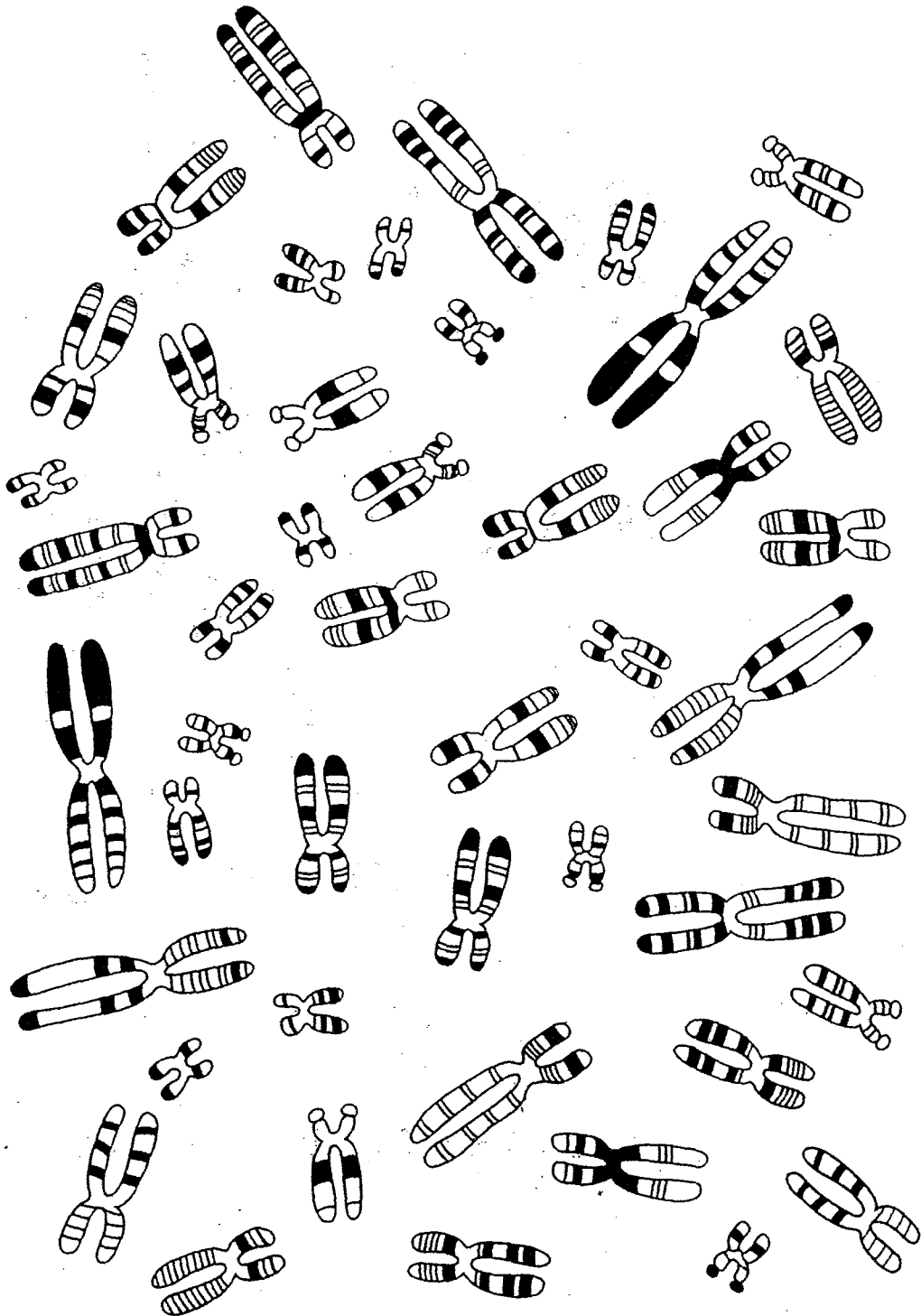
Chromosome Spread

7

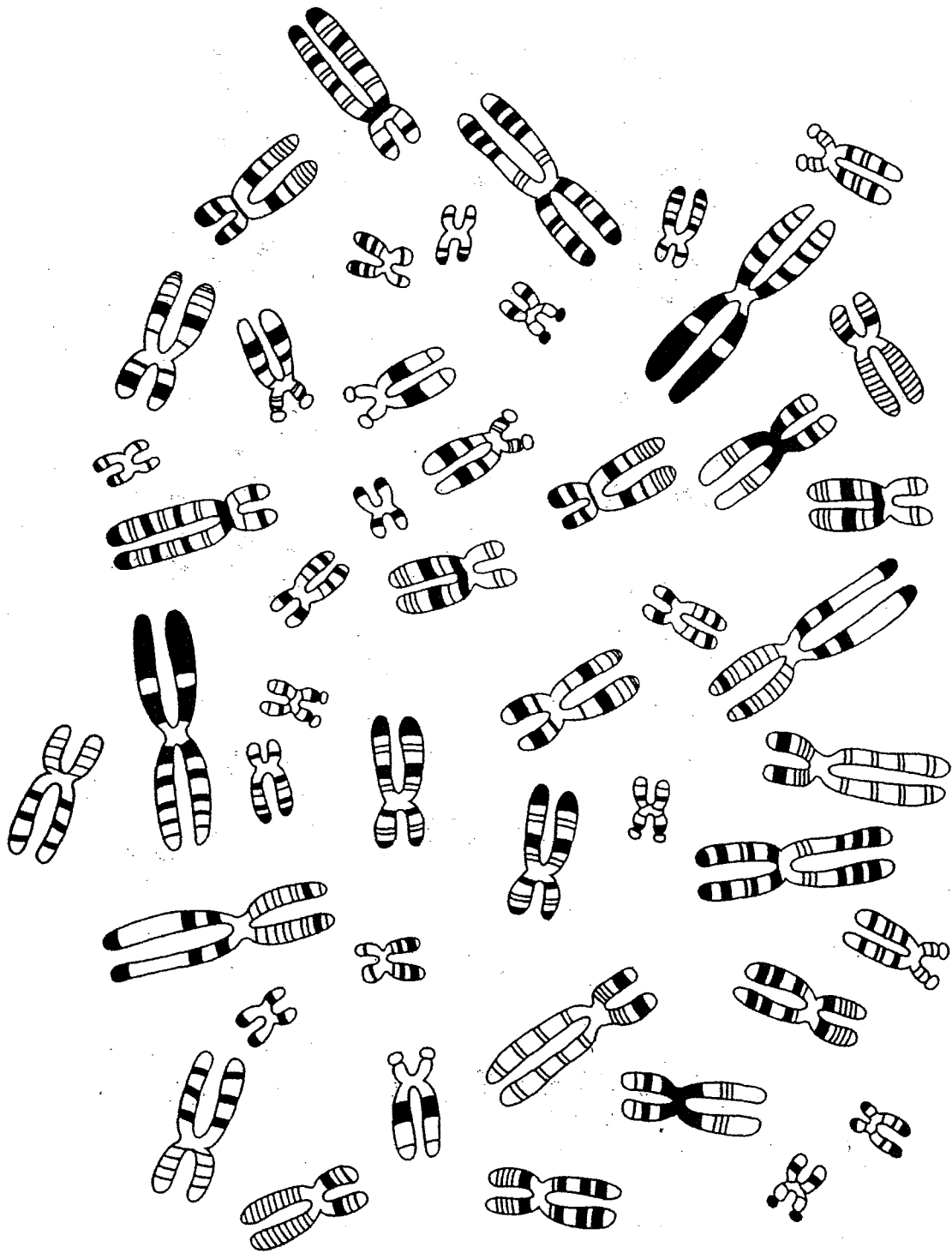


Chromosome Spread

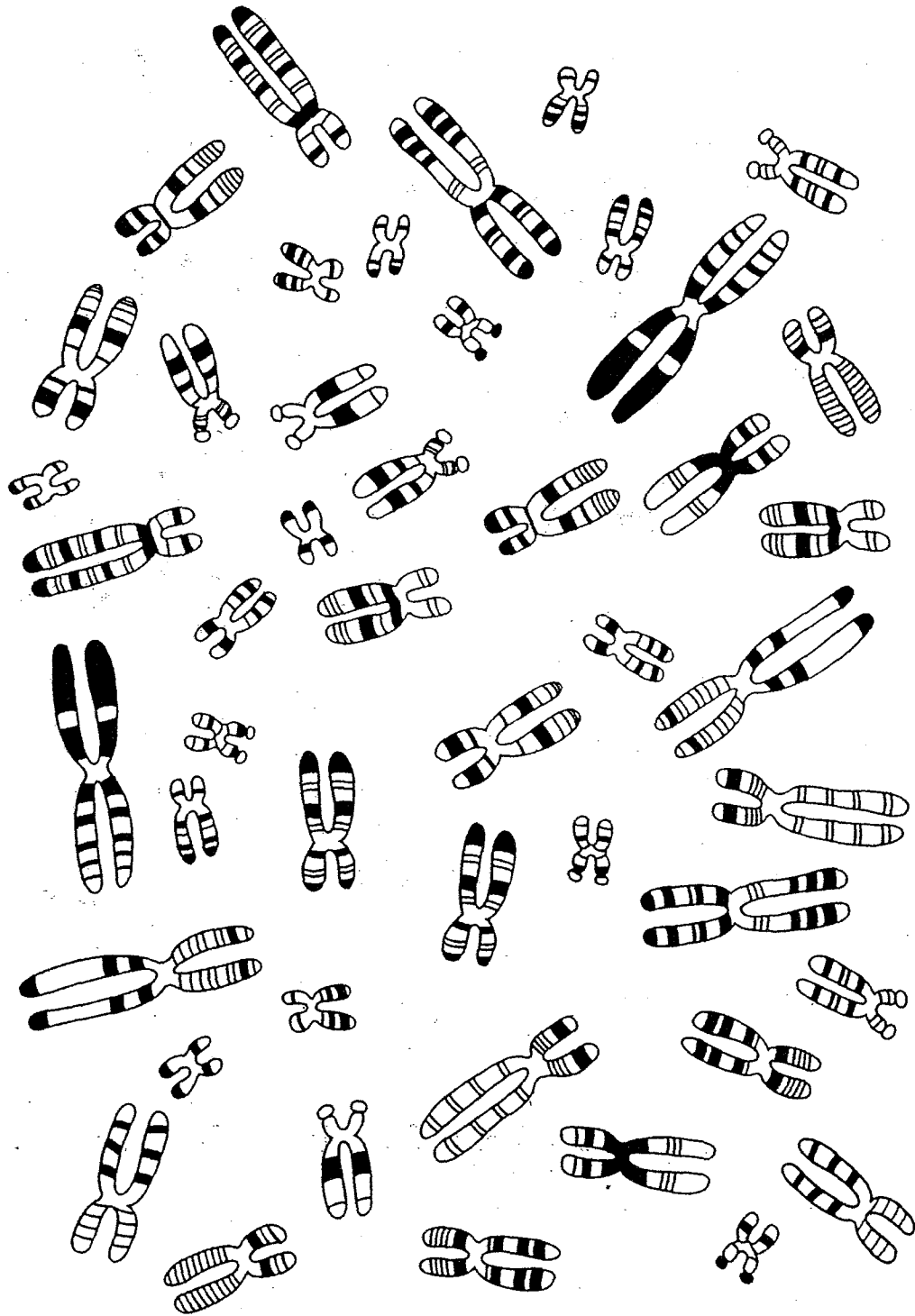
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Chromosome Spread 9



Chromosome Spread 10



Answers to Karyotyping Activity

Chromosome Spread #1 XYY

Speck's

Chromosome Spread #2 male with Trisomy 13

PATAU

Chromosome Spread #3 normal male

Chromosome Spread #4 XXX

Trisomy X

Chromosome Spread #5 female with Trisomy 21

Down Syndrome

Chromosome Spread #6 male with Trisomy 18

Edwards

Chromosome Spread #7 XXY male (Klinefelter's) with Trisomy 21



Chromosome Spread #8 normal female

Chromosome Spread #9 XXY (Klinefelter's)

Chromosome Spread #10 ~~male with Trisomy 13~~

Trisomy 18 Female - Edwards