INVESTIGATION 11

Human Karyotypes

Purpose

To construct a karyotype, detect any abnormalities, and identify the sex of the person.

Science Concepts

- Homologous chromosomes
- Karyotyping
- Chromosomal abnormalities

Background

Recall that each person has two copies of every chromosome: one of the pair comes from the mother and the other comes from the father. Each cell that has a nucleus has the same genetic material as every other cell in the body (except for the gametes, which contain half the information). A karyotype is a picture of the chromosomes which is examined for any visual abnormalities such as missing, extra or broken chromosomes. To perform a karyotype, cells are removed from the patient and are given chemicals to start mitosis. When they reach metaphase another chemical is given to stop mitosis because the chromosomes are most visible at this stage. The chromosomes are then put on a microscope slide, photographed, and the pictures of each chromosome are cut out. They are then arranged in pairs by looking at the pattern of the bands, the placement of the centromere, or pinched portion, and the length of the chromosome arms. They are then inspected for abnormalities.

Each chromosome of a pair has somewhere between 50,000 and 100,000 genes depending on the size. There are between a dozen and a few hundred genes in between each band on a chromosome. Each gene on a chromosome has an " address" so that scientists can tell each other where to find them. The part of a chromosome that lies above the centromere is called the "p arm" and the part that lies below the centromere is called the "q arm". For example, the gene address 9q3.1 means that the gene is on the q arm of chromosome 9 in region 3. (See Figure 1.)

9	q	3.	1
chromosome	arm	section	subsection

Figure 1. Earlobe shape "gene address"

Materials

- Chromosome smear patterns
- Scissors
- Karyotyping worksheet
- Tape or glue

Safety

Be careful when using scissors and follow all other normal laboratory safety rules.

Procedure

1. Count the total number of chromosomes on the Chromosome Smear Sheet.
2. Carefully cut out each chromosome from the Chromosome Smear Sheet.
3. Arrange the chromosomes by size from largest to smallest.
4. Use the banding pattern and centromere placement to arrange the chromosomes into pairs.
5. Keeping the chromosomes in pairs, arrange them on the Karyotyping Worksheet. Arrange them neatly and in order with the centromeres on the line. Use the shape, size, banding patterns and group descriptions to arrange the karyotype.
6. When the chromosomes have been classified and you are confident of their identity, tape them to your worksheet and answer the following questions:

   a. Look at the karyotype. What is the relationship between chromosome number and size of the chromosomes?

   b. How many autosomes are present in your karyotype?

   c. How many sex chromosomes are present in your karyotype?

   d. Are there any abnormalities? Explain.

   e. What is the sex of the person?

   f. What is the genetic disorder of the person in the karyotype?

   g. What is the name of the mechanism by which this disorder occurs?

Name: _____

Karyotyping Worksheet

Group A: Long chromosomes; centromere near center. 1 2 3

Group B: Long chromosomes; centromere not in center. 4 5

Group C: Medium chromosomes; centromeres slightly off center.

6 7 8 9 10 11 12

Group D: Medium chromosomes; centromeres near one end of chromosome. 13 14 15

Group E: Short chromosomes; centromeres slightly off center. 16 17 18

Group F: Short chromosomes; centromeres near center. 19 20

Group G: Very short chromosomes; centromere near end. 21 22

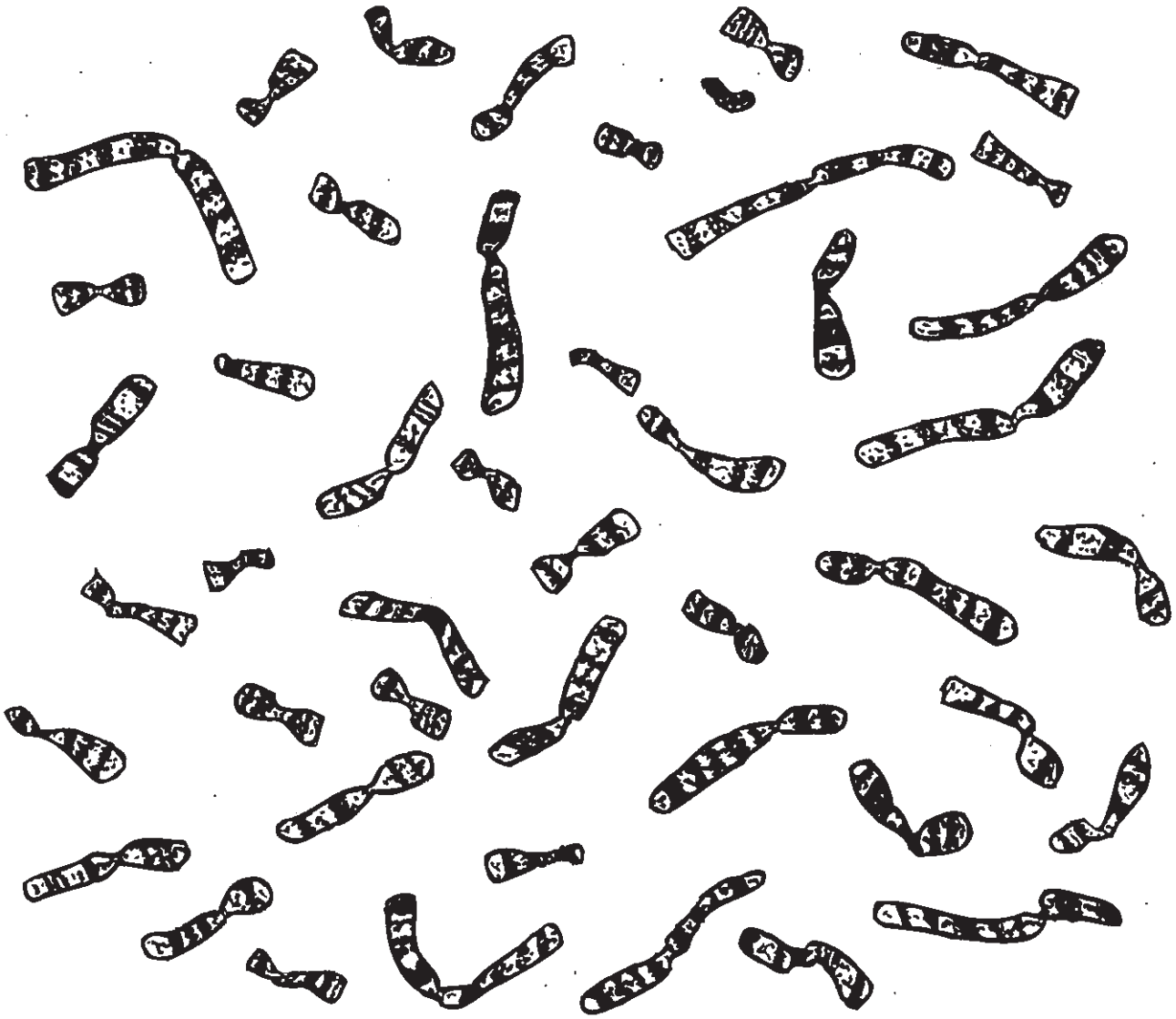
Sex Chromosomes _____

Smear #_____ How many chromosomes does this individual possess? _____

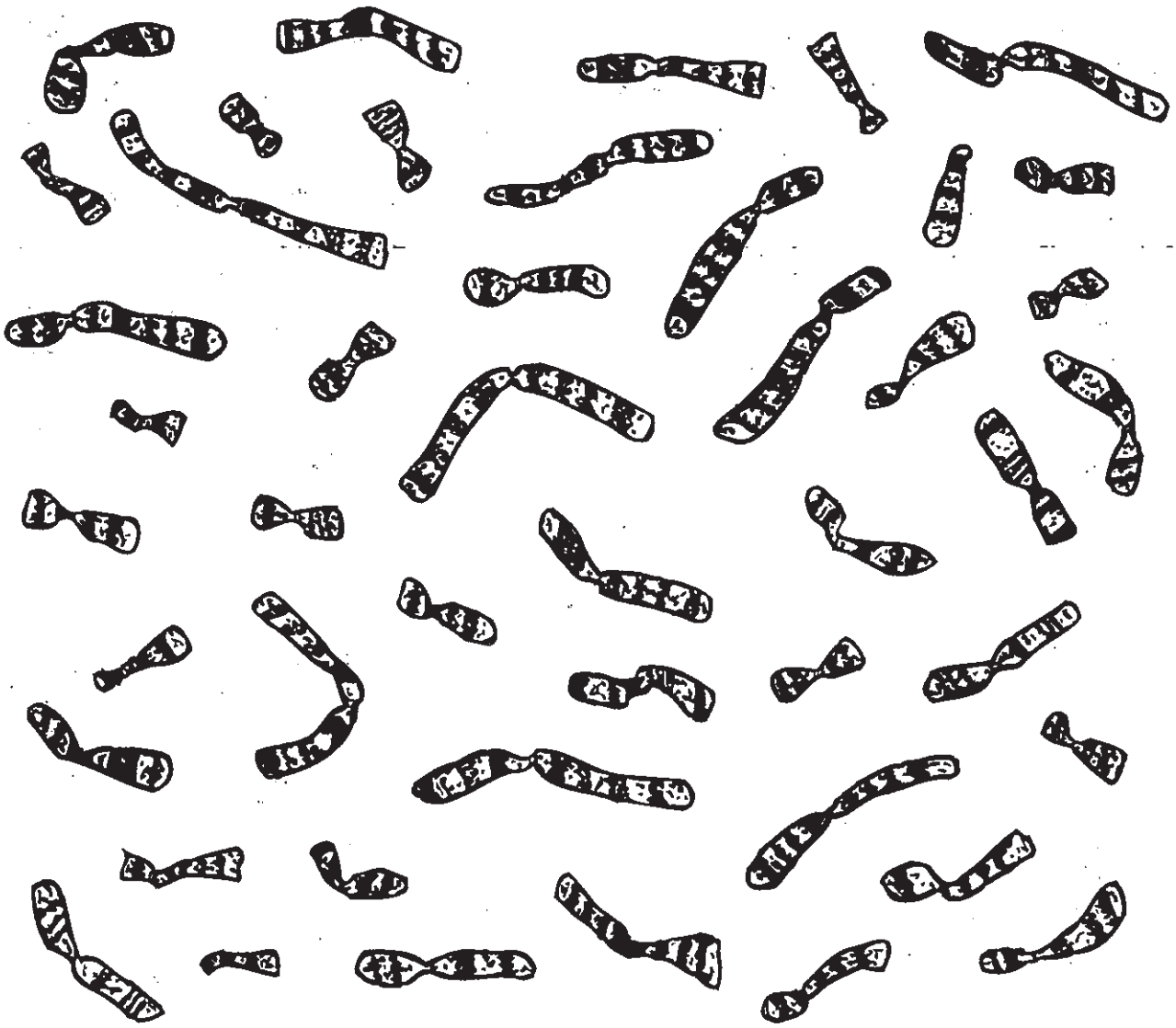
What is the sex of the individual? _____

Describe any possible chromosome abnormalties. _____

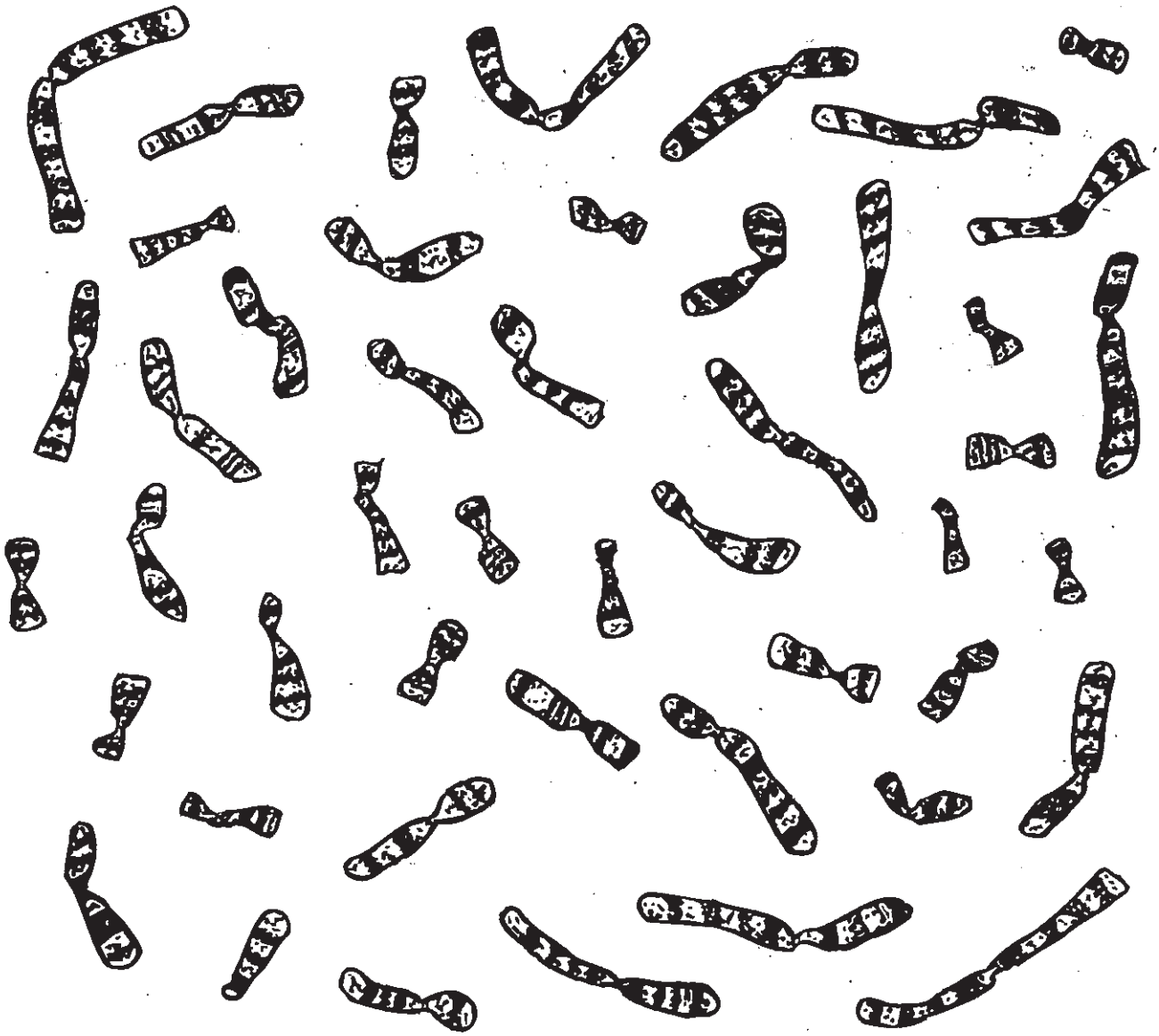
Chromosome Smear #1



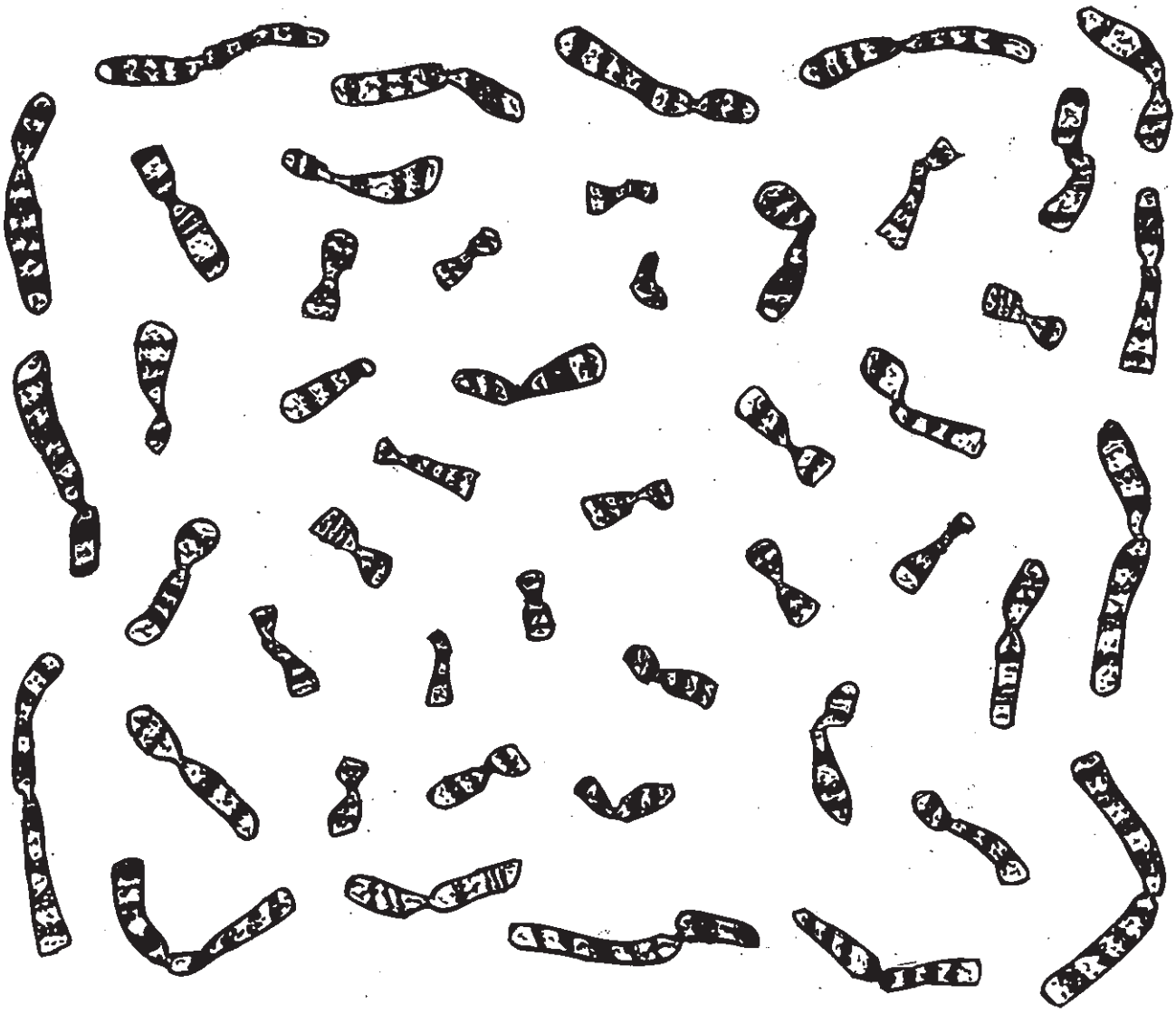
Chromosome Smear #2



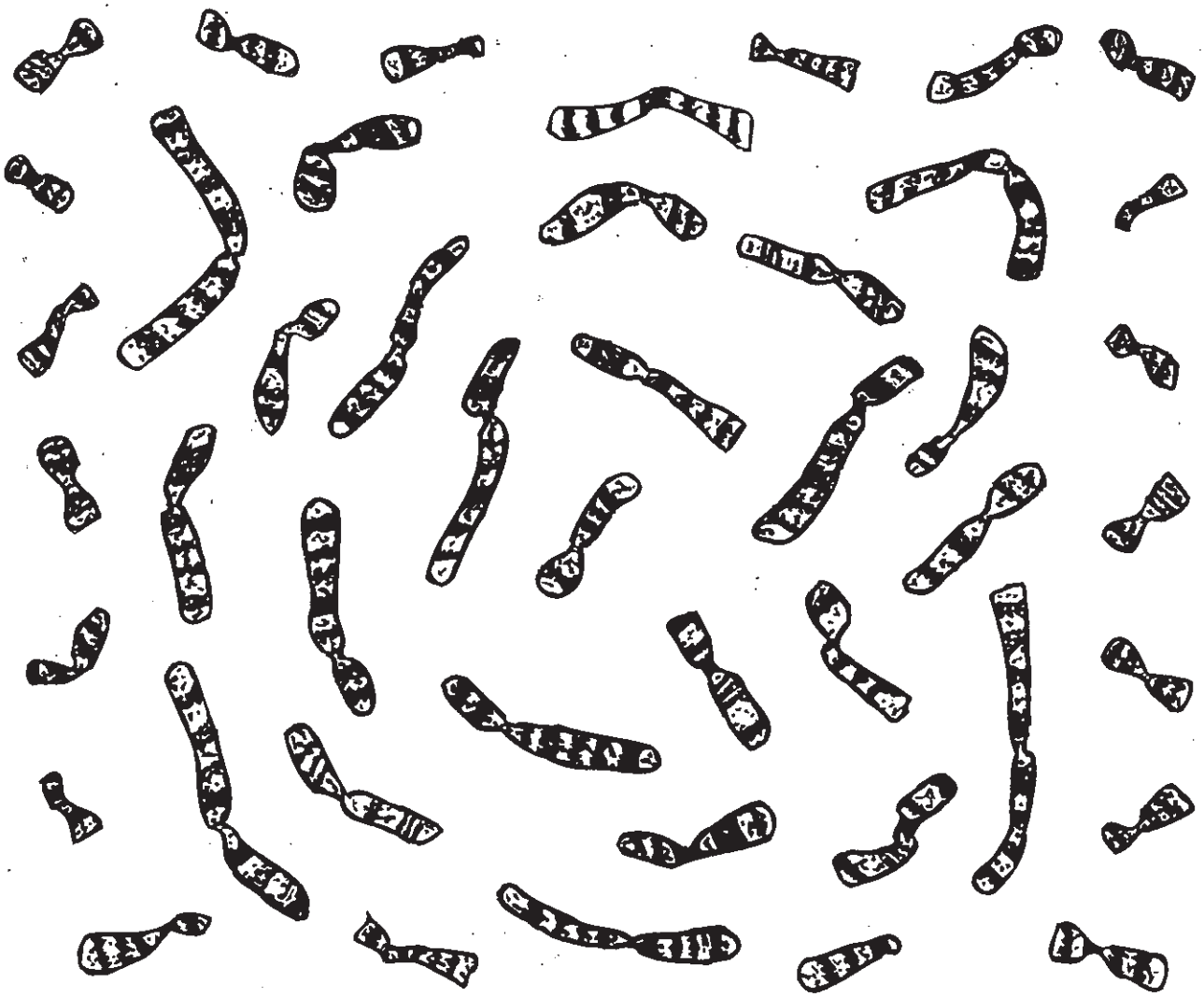
Chromosome Smear #3



Chromosome Smear #4



Chromosome Smear #5



Chromosome Smear #6