

KARYOTYPING LAB

Background Information:

Scientists have developed several different tools and techniques for studying chromosomes, genes and base pairs in humans and other organisms. One of the most useful techniques is karyotyping.

A karyotype is a photograph of all of an organism's chromosomes. The chromosomes in the karyotype are arranged in homologous pairs according to size (largest to smallest). Homologous pairs can be determined by centromere placement, equal length of top and bottom arms as well as similar band placement on each arm.

Karyotyping helps doctors diagnose and treat genetic disorders. Doctors use a normal human karyotype and compare it to the karyotype of a patient to determine if there are abnormalities. Some of the characteristics the physician will compare are:

- A. Total number of chromosomes - normal humans have 46 chromosomes (23 pairs), so if the number is higher or lower then an abnormality exists
- B. Homologous pairs for the first 22 pairs of chromosomes (autosomes) - once centromeres are aligned, top and bottom arms are of equal length and if not then an abnormality exists
- C. Sex Chromosomes (23rd pair) - if female, then 2 homologous X chromosomes (XX) will be present and if male, an X chromosome and a Y chromosome (XY) will be present, so if there are additional or fewer sex chromosomes then an abnormality exists.

Procedure:

1. Arrange the chromosomes into homologous pairs on the Layout Sheet, using your Karyotype Reference Sheet as a guide.
2. Once all chromosomes are laid down on the Layout Worksheet, answer the Discussion Questions using the background information, your karyotype, and Explanation of Chromosome Disorders Handout..
Hint: You may have abnormalities in your karyotype (more or less than 46 chromosomes), so maintain the number of chromosomes you were given.

EXPLANATION OF CHROMOSOME DISORDERS

Cri-du-chat

Babies with the "cry of the cat" syndrome have a cry which sounds like that of a cat in distress because the infant's larynx is improperly developed. The cause of this condition is a deletion of about half of the short arm of chromosome number five. Cri-du-chat babies are severely mentally disabled, have a small cranium, a small jaw and a moon-shaped face. The incidence of this syndrome is 1/100,000 live births.

****Karyotype:** 46XX or 46XY with one chromosome #5 upper arm deletion

Down Syndrome

Trisomy 21, one of the most common causes of mental retardation is due to an extra chromosome 21. This results in a number of characteristic features, such as short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult and mental disabilities. Individuals with this syndrome have a high incidence of respiratory infections, heart defects and leukemia. The average risk of having a child with trisomy 21 is 1/750 live births. Mothers in their early twenties have a risk of 1/1,500 and women over 35 have a risk factor of 1/70, which jumps to 1/25 for women 45 or older.

****Karyotype:** 47XX or 47XY with 3 of the chromosome #21

Edward's Syndrome

This syndrome, trisomy 18 (an extra chromosome 18), produces severe mental disabilities and a highly characteristic pattern of malformations such as elongated skull, a very narrow pelvis, rocker bottom feet, malformed heart and a grasping of the two central fingers by the thumb and little finger. In addition, the ears are often low set and the mouth and teeth are small. Nearly all babies born with this condition die in early infancy. The frequency of this syndrome is 1/5,000 live births.

****Karyotype:** 47XX or 47XY with 3 of the chromosome #18

Patau Syndrome

This syndrome (trisomy 13 - extra chromosome 13) causes severely abnormal cerebral functions and virtually leads to death in early infancy. The baby has very pronounced clefts of the lip and palate, broad nose, polydactyly (extra fingers and toes), small cranium and nonfunctional eyes. Heart defects and severe mental disabilities are also part of the clinical picture. The frequency is 1/15,000 live births.

****Karyotype:** 47XX or 47XY with 3 of the chromosome 13

Discussion Questions:

Write the answers to these questions on the back of your Karyotype Layout Worksheet.

Use Background Information to answer the following questions.

1. Define Karyotype.
2. What can karyotypes be used to determine?
3. How many chromosomes do normal humans have?
4. How did you know if 2 chromosomes were a homologous pair (What made you pair 2 chromosomes together)?
5. What does the 23rd pair of chromosomes determine?
6. The first 22 pairs of chromosomes are homologous in a normal person. Is that "always true for the 23rd pair of chromosomes? Explain your answer.

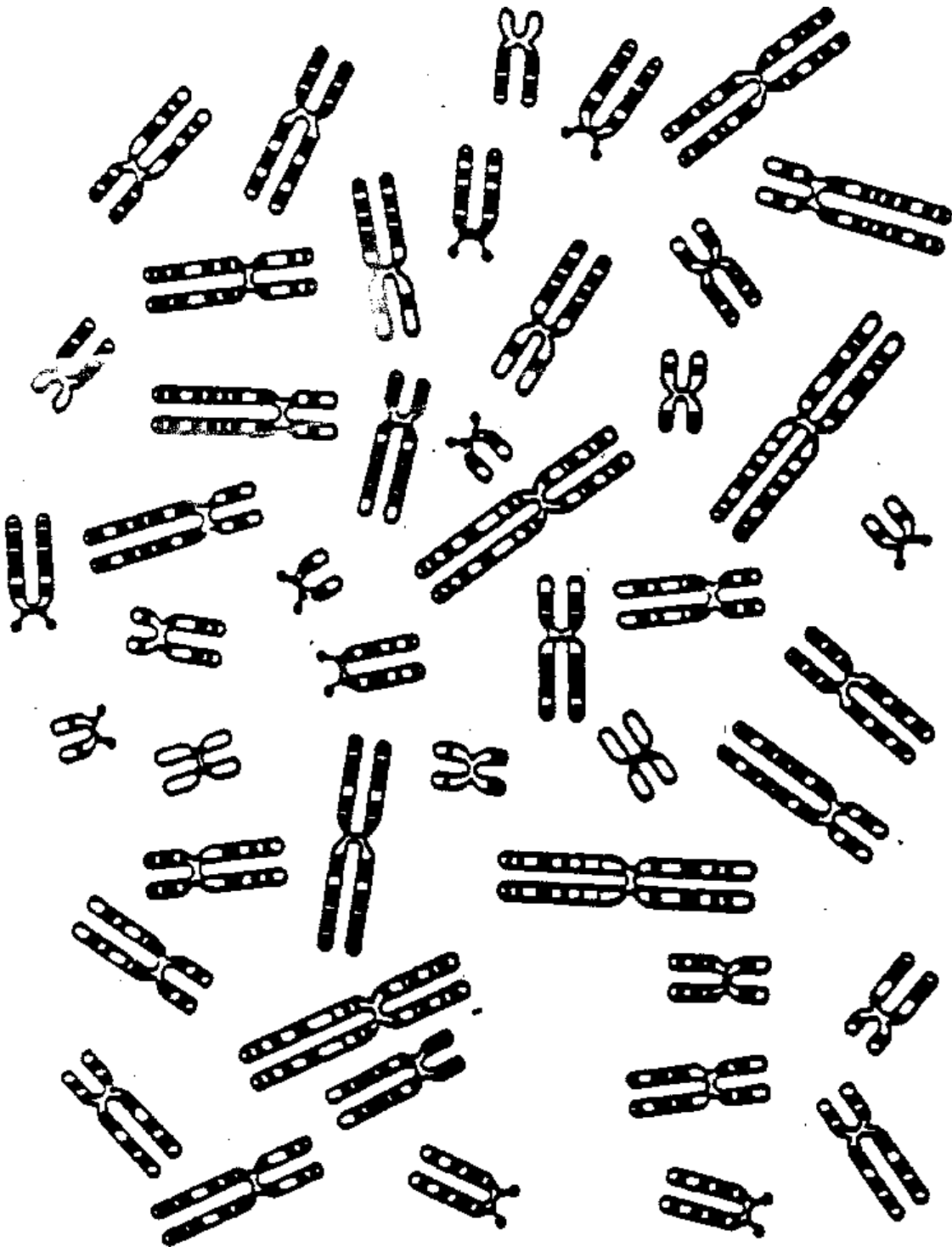
Use your karyotype to answer the following questions.

7. What is the gender (sex) of your baby?
8. Was your baby's karyotype normal (yes/no)?
9. If the karyotype was abnormal, what is the disorder your baby has (use the Explanation of Chromosome Disorders Handout)?
10. Describe the disorder your baby has. If your baby is normal write "Normal Baby".

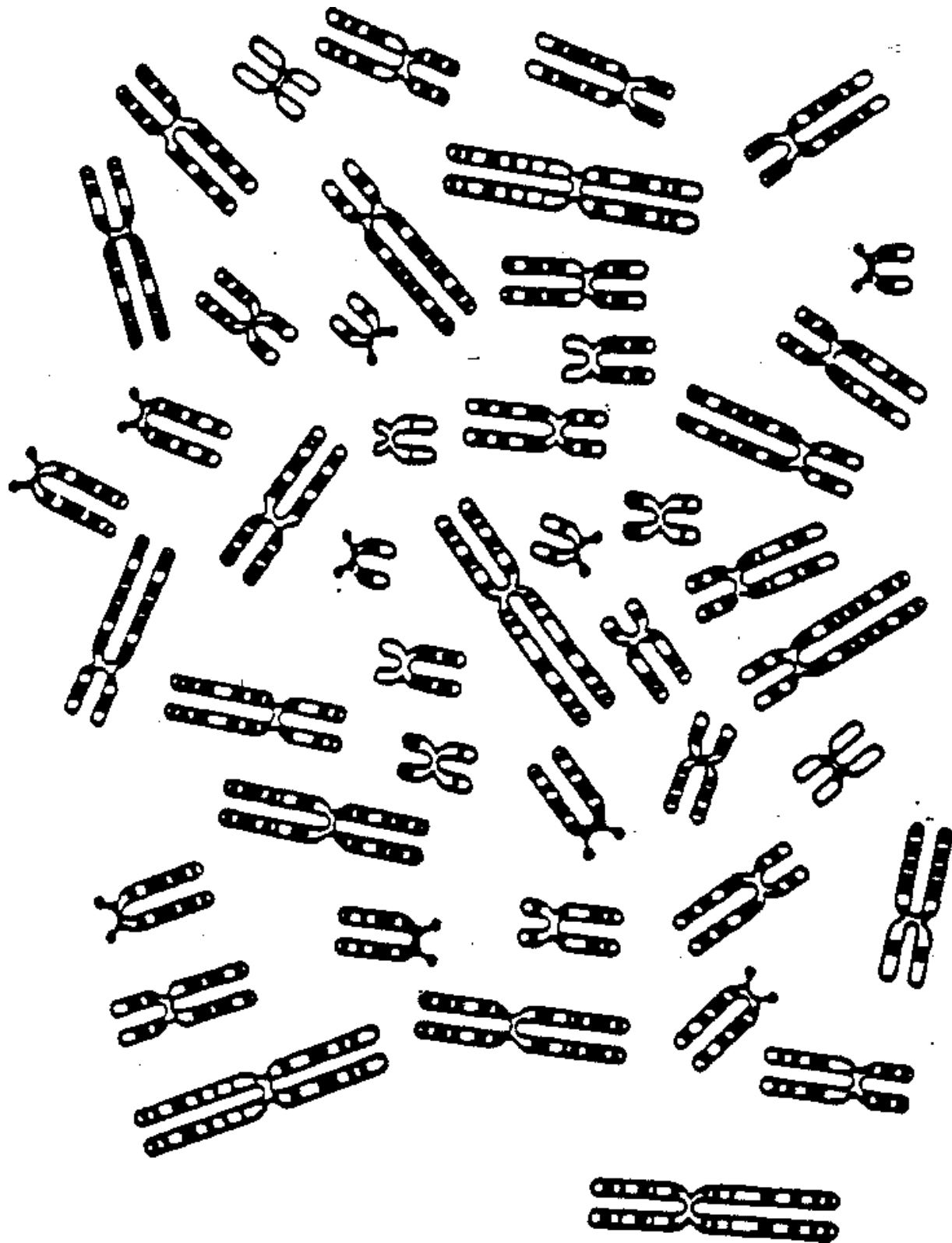
KARYOTYPE LAYOUT WORKSHEET

1	2	3	4	5	6
7	8	9	10	11	12
13	14	15	16	17	18
19	20	21	22	23 XX	XY

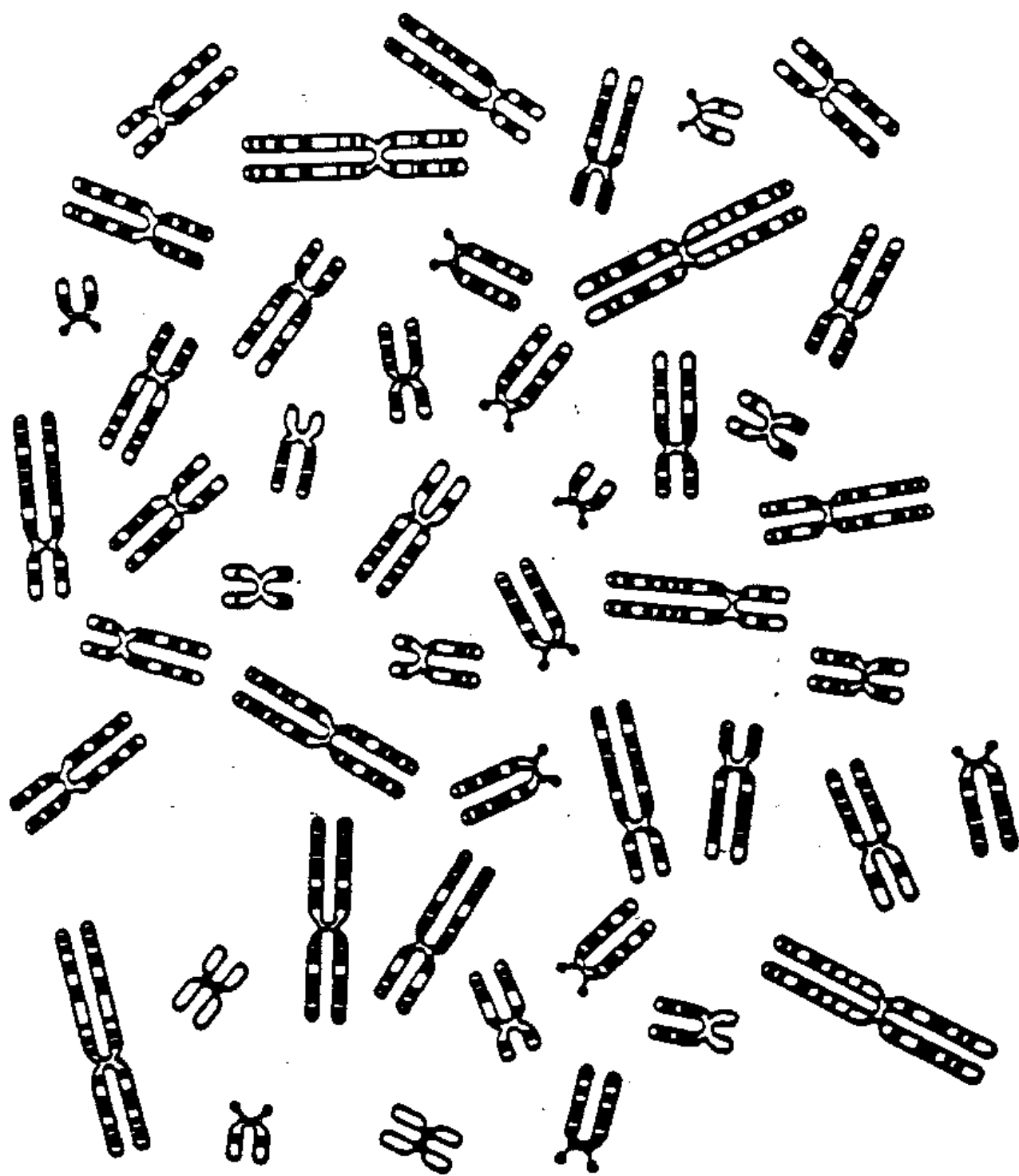
Chromosome Scatter Sheet-A



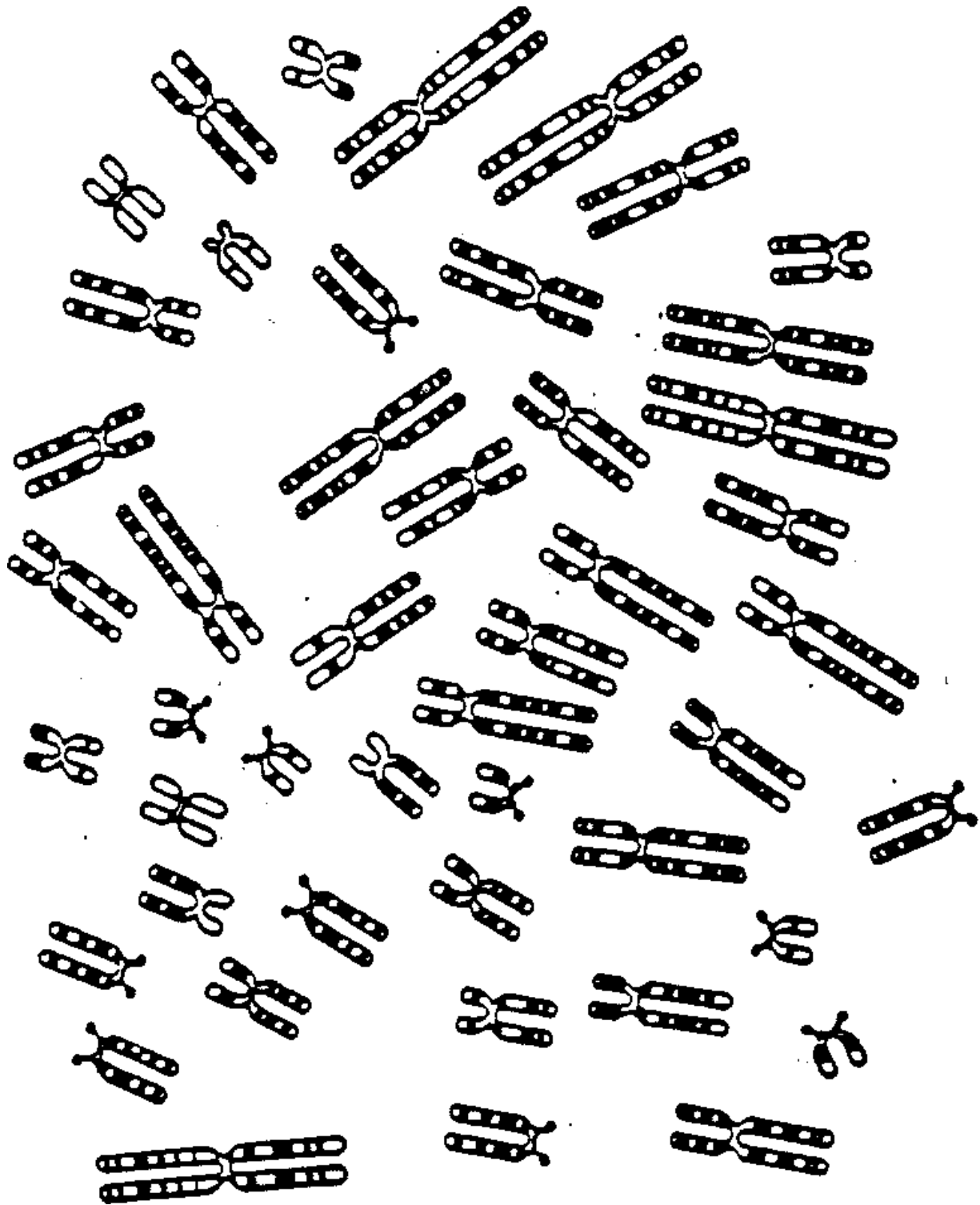
Chromosome Scatter Sheet B



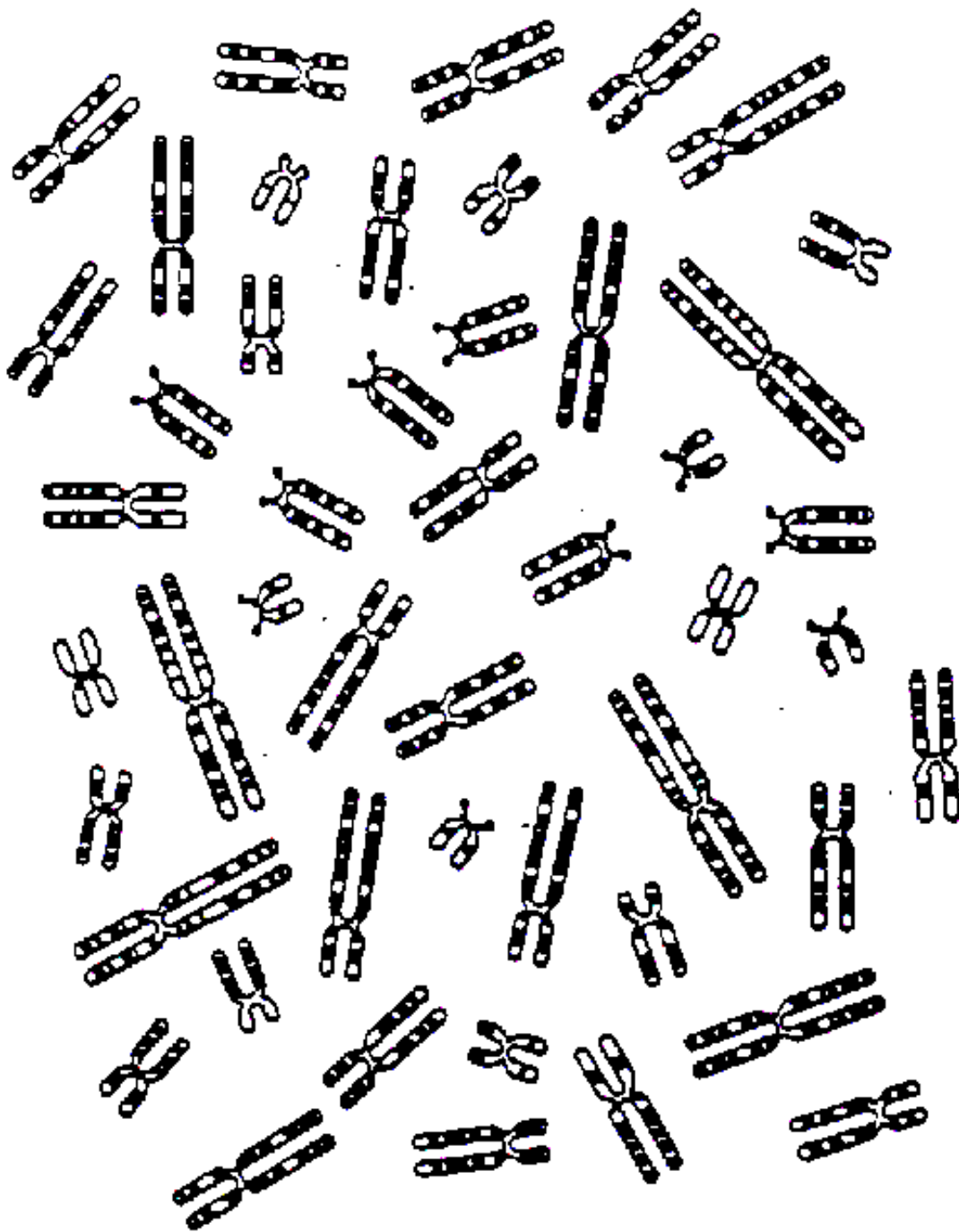
Chromosome Scatter Sheet-C



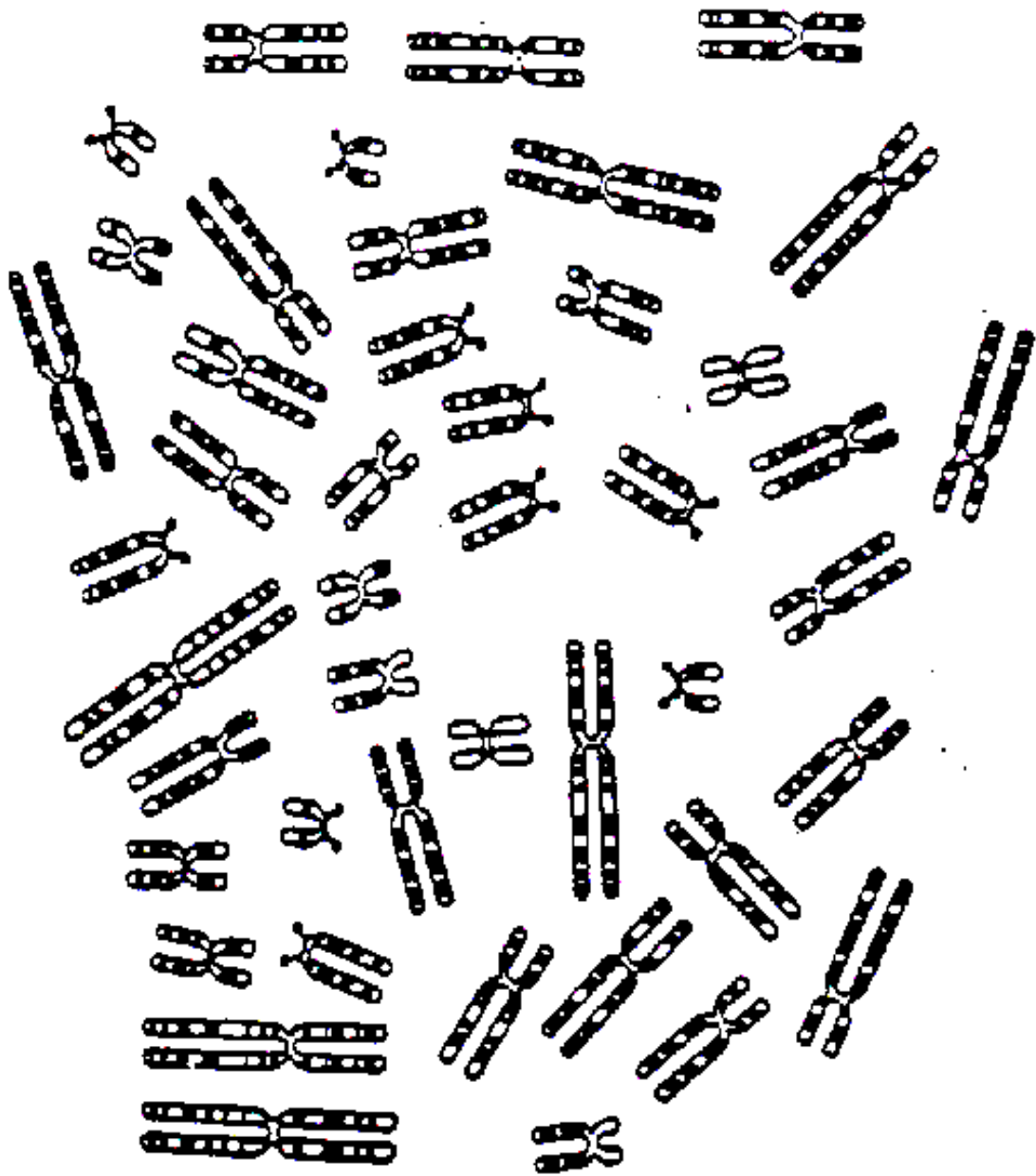
Chromosome Scatter Sheet-D



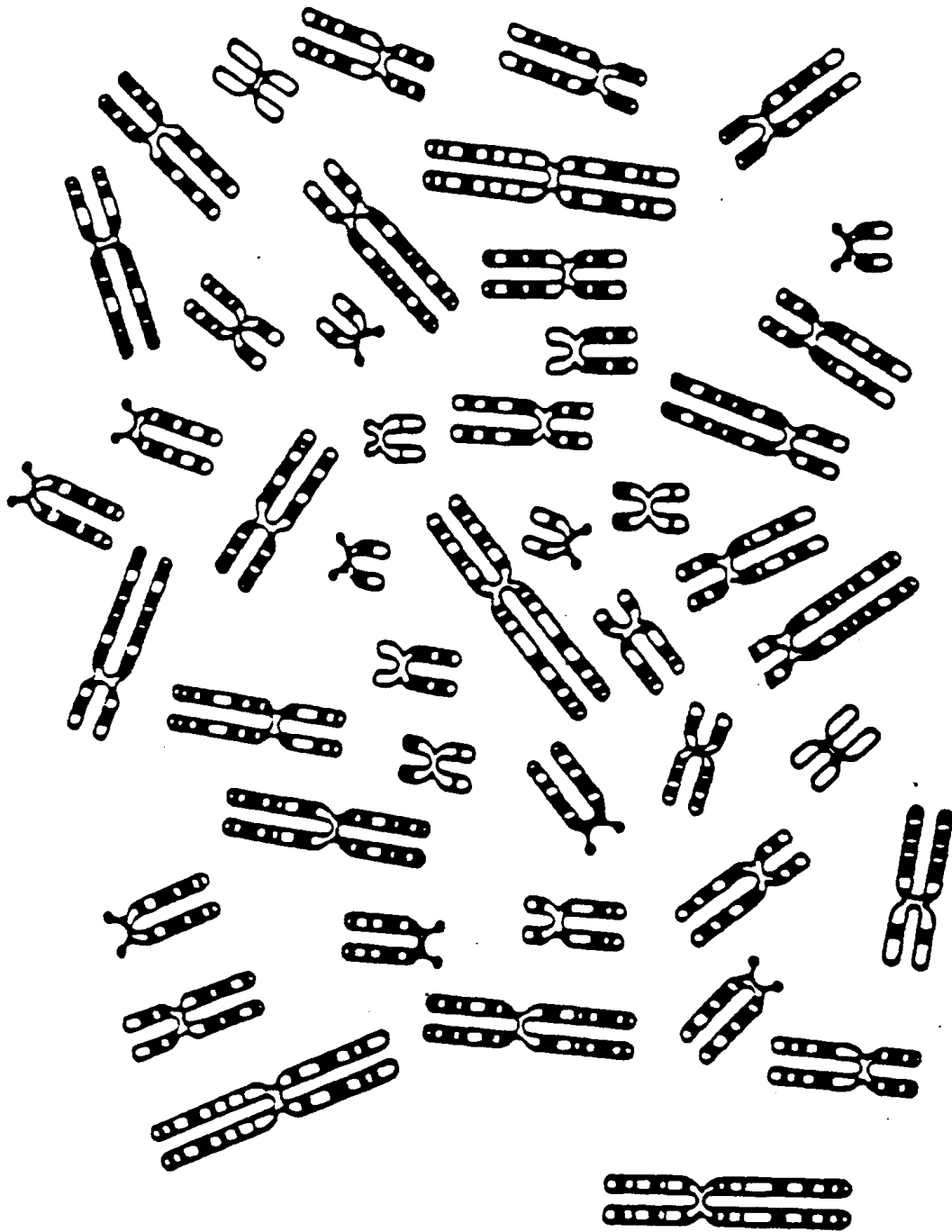
Chromosome Scatter Sheet-E



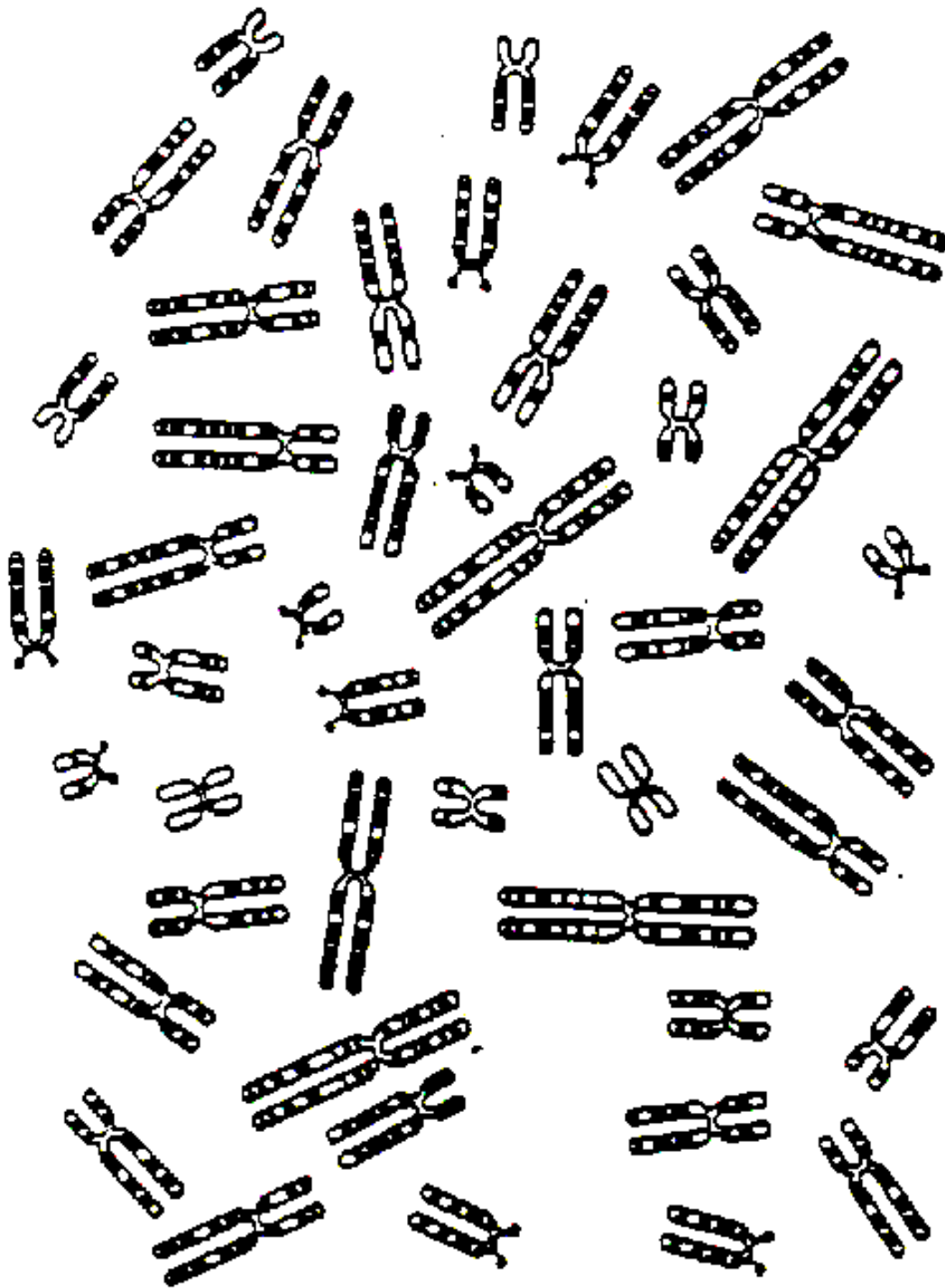
Chromosome Scatter Sheet-F



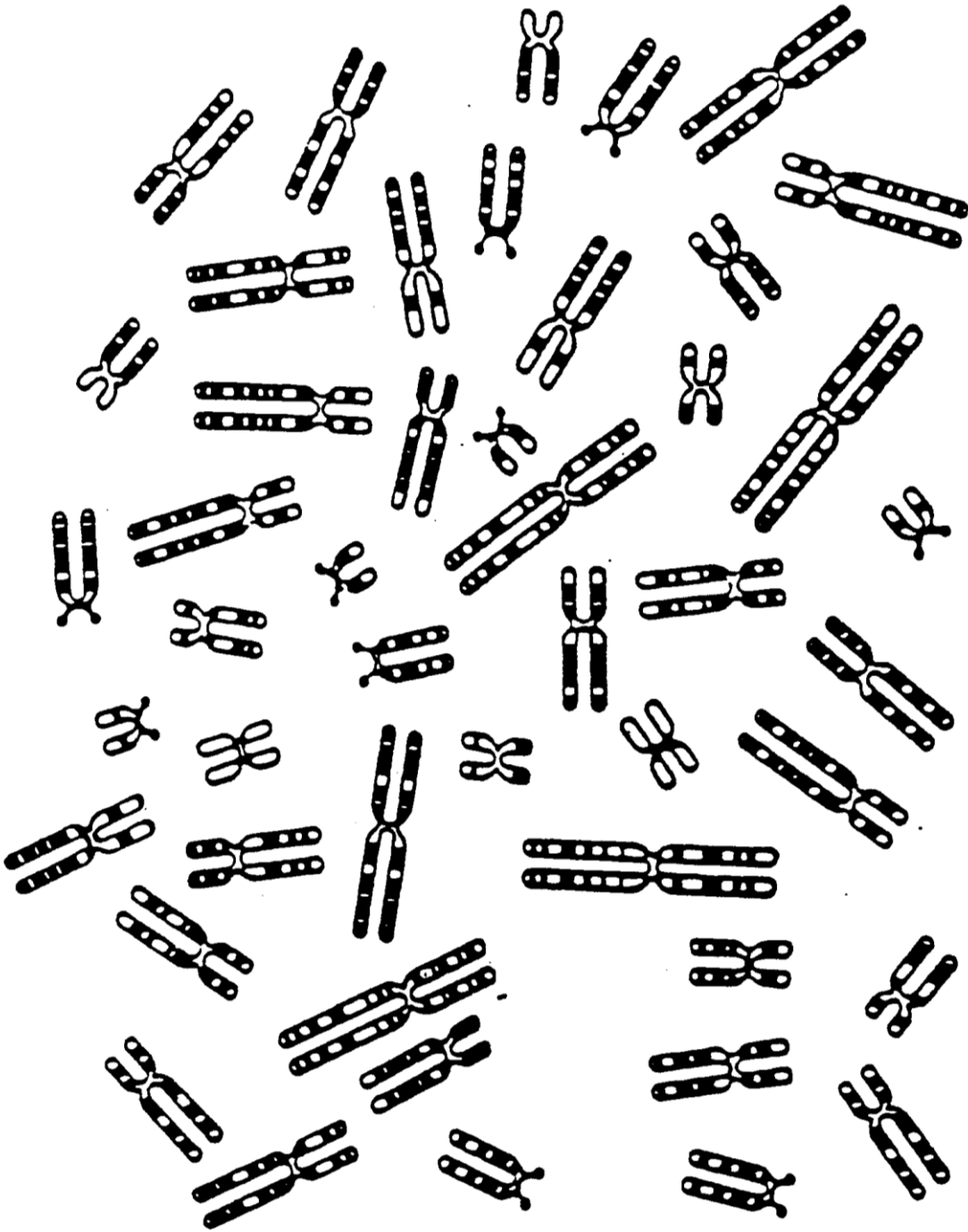
Chromosome Scatter Sheet-G



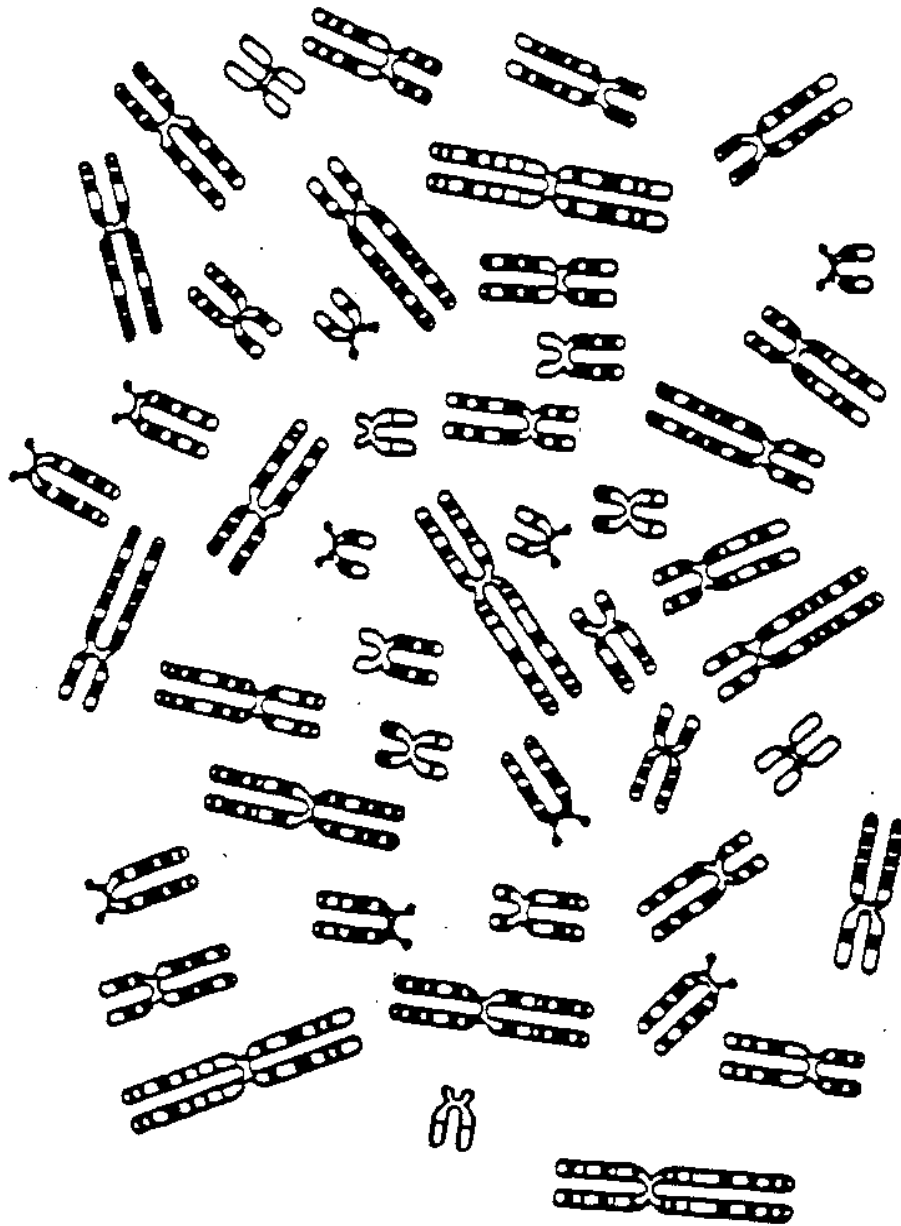
Chromosome Scatter Sheet-H



Chromosome Scatter Sheet-I



Chromosome Scatter Sheet-J



Karyotype Reference Sheet

