Appendix B

Karyotyping and Genetic Disorder

Karyotyping

Introduction

You are a genetic counselor whose job is to discuss with expecting parents any genetic disorder that may affect their child. Currently, you are working with two couples. The two expectant mothers are over thirty-five and are concerned that their unborn children may have chromosomal abnormalities. You have been given a chromosome spread of each of the children. For each couple, you must construct and analyze a karyotype. In addition, you will be expected to give each couple a brief explanation of the test results, including the characteristics of any genetic disorder that may affect their unborn child.

Purpose

To connect and analyze several karyotypes.

Materials

Glue (1)
Ruler (1)
Scissors (1 pair)
Chromosome Spread Sheets (2)
Karyotype Templates #'s 1 and 2

Procedure

1. Obtain two different chromosome spread sheets from your teacher.
2. Cut out the chromosomes of one of the chromosome spread sheets.
3. Arrange the chromosomes into 22 pairs on the karyotype template. The chromosomes of each pair should be the same length (use a ruler!) and have the same centromere position. They should also have similar banding patterns. The two remaining chromosomes are the sex chromosomes. Since the X and Y chromosomes are nonhomologous, they will not have similar lengths, centromere positions or banding patterns. (Refer to the normal human karyotype.)
4. Now, place the chromosomes in order, with the longest pair at position 1, the shortest at position 22, and the sex chromosomes at position 23.
5. Finally, glue each chromosome into position. Be sure to label your karyotype according to the chromosome spread you were given.
6. Use your constructed karyotype # 1 to answer the analysis question 1.
7. Repeat steps 1 to 5 for your second chromosome spread sheet.
8. Use your constructed karyotype #2 to answer the analysis question 2.

Analysis

1. Analyze karyotype #1 to determine if a chromosomal abnormality exists.

   a) Will the child have a genetic disorder?
   b) Explain the reason for your answer.
   c) Using the student chart, determine which genetic complication will affect the child.

2. Analyze karyotype #2 to determine if a chromosomal abnormality exists.

   a) Will the child have a genetic disorder?
   b) Explain the reason for your answer.
   c) Using the student chart, determine which genetic complication will affect the child.

Further Analysis

3. Describe two procedures used by genetic counselors to obtain a DNA sample used for karyotyping.

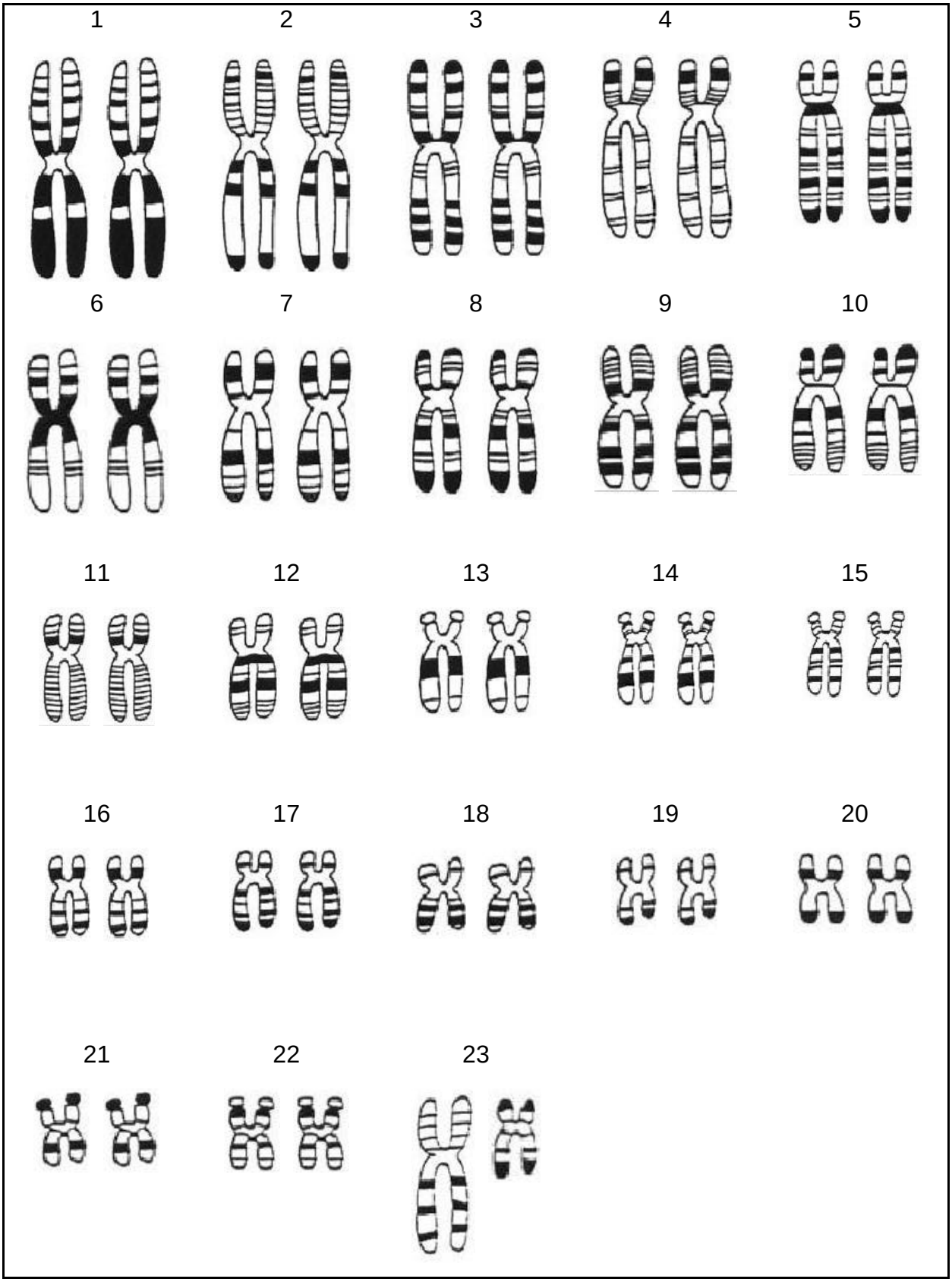
4. Describe two other techniques used to examine the developing fetus for abnormalities.

5. Write a letter to one of the expectant couples, informing them of the results of the genetic tests. The letter must include the name of the genetic disorder that affects their child and the characteristics of the disorder. Since you are a professional, be sure that you use clear language to communicate the test results.

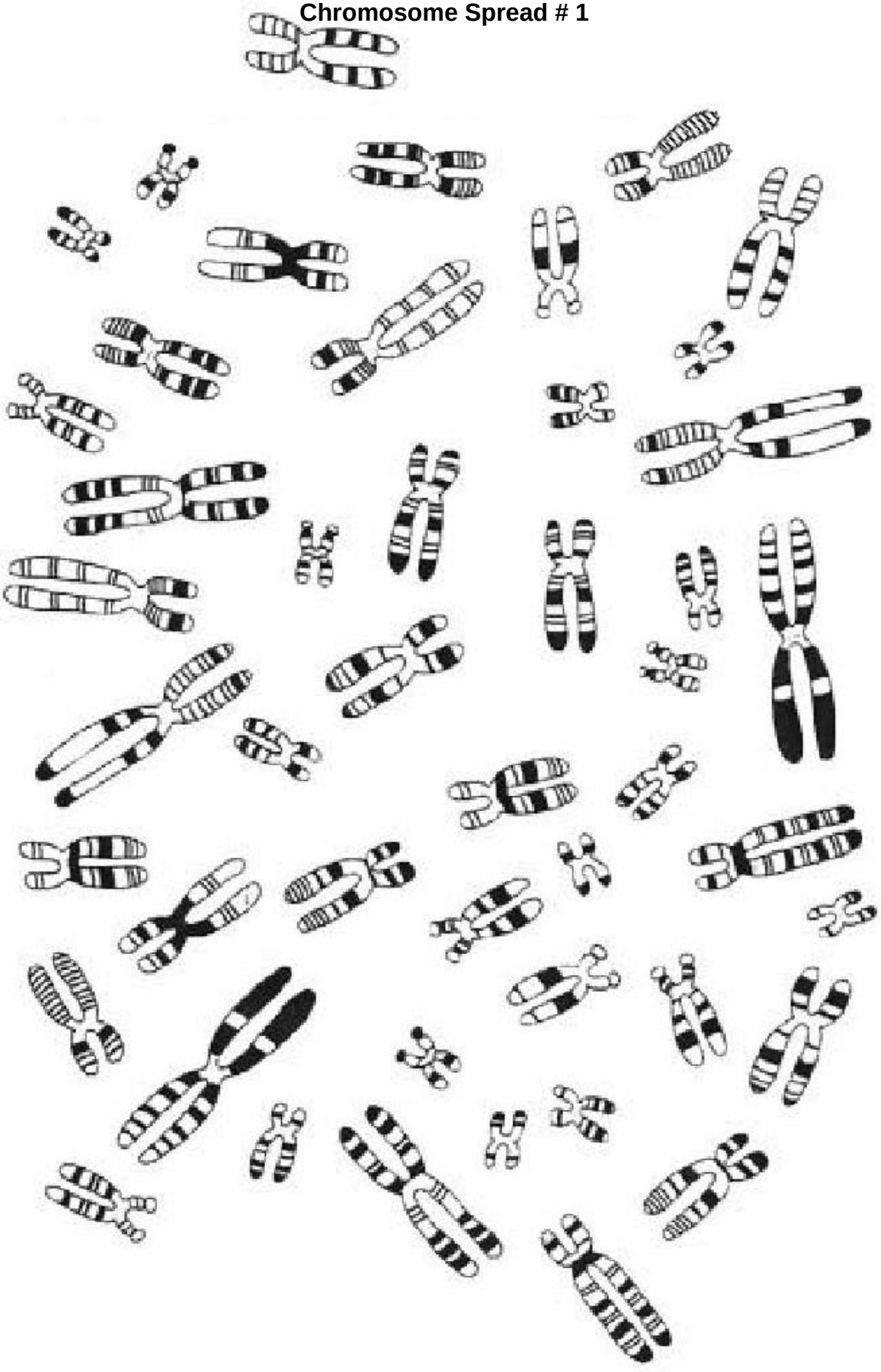
Genetic Disorders

Genetic Disorder	Chromosome Affected	Description of Disorder
Down Syndrome	# 21	47 chromosomes, mild to severe developmental disabilities, almond - shaped eyes, large tongue, prone to heart defects and respiratory problems.
Turner Syndrome	Single X in female (XO)	45 chromosomes, female lacking an X chromosome, normal in childhood, normal intelligence, fails to develop secondary sex characteristics and remains infertile.
Klinefelter Syndrome	Extra X in Male (XXY)	47 chromosomes, male with an additional X chromosome, usually normal in appearance, normal intelligence, tall, underdeveloped testes, sterile, may also cause female characteristics (breast development, feminine body shape).
Jacobs Syndrome	Extra Y in Male (XYY)	47 chromosomes, male with an additional Y chromosome, low mental ability, normal in appearance.
Triple X Syndrome	Extra X in Female (XXX)	47 chromosomes, female with an extra X chromosome, normal intelligence, normal in appearance, may be sterile.

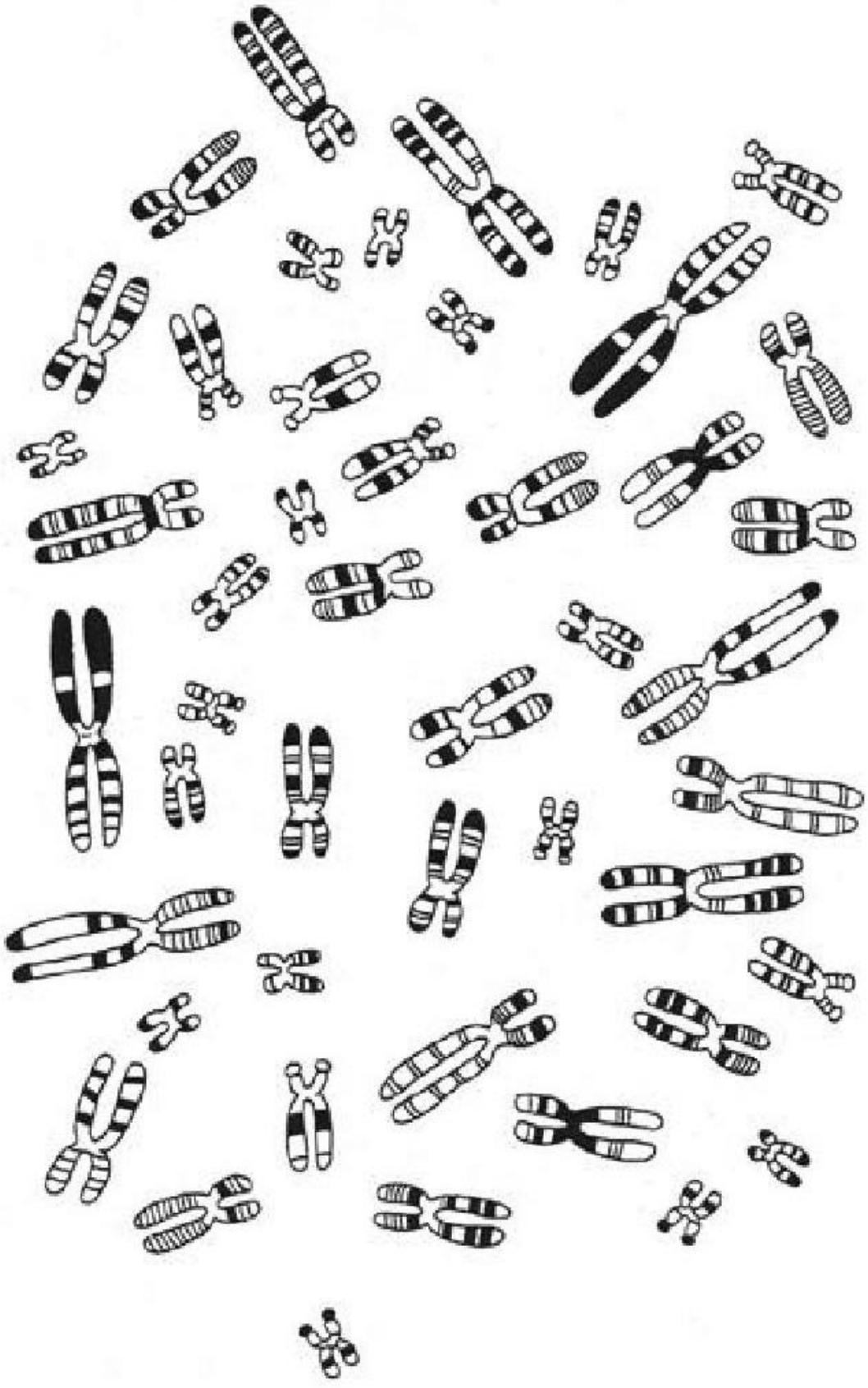
Normal Human Karyotype



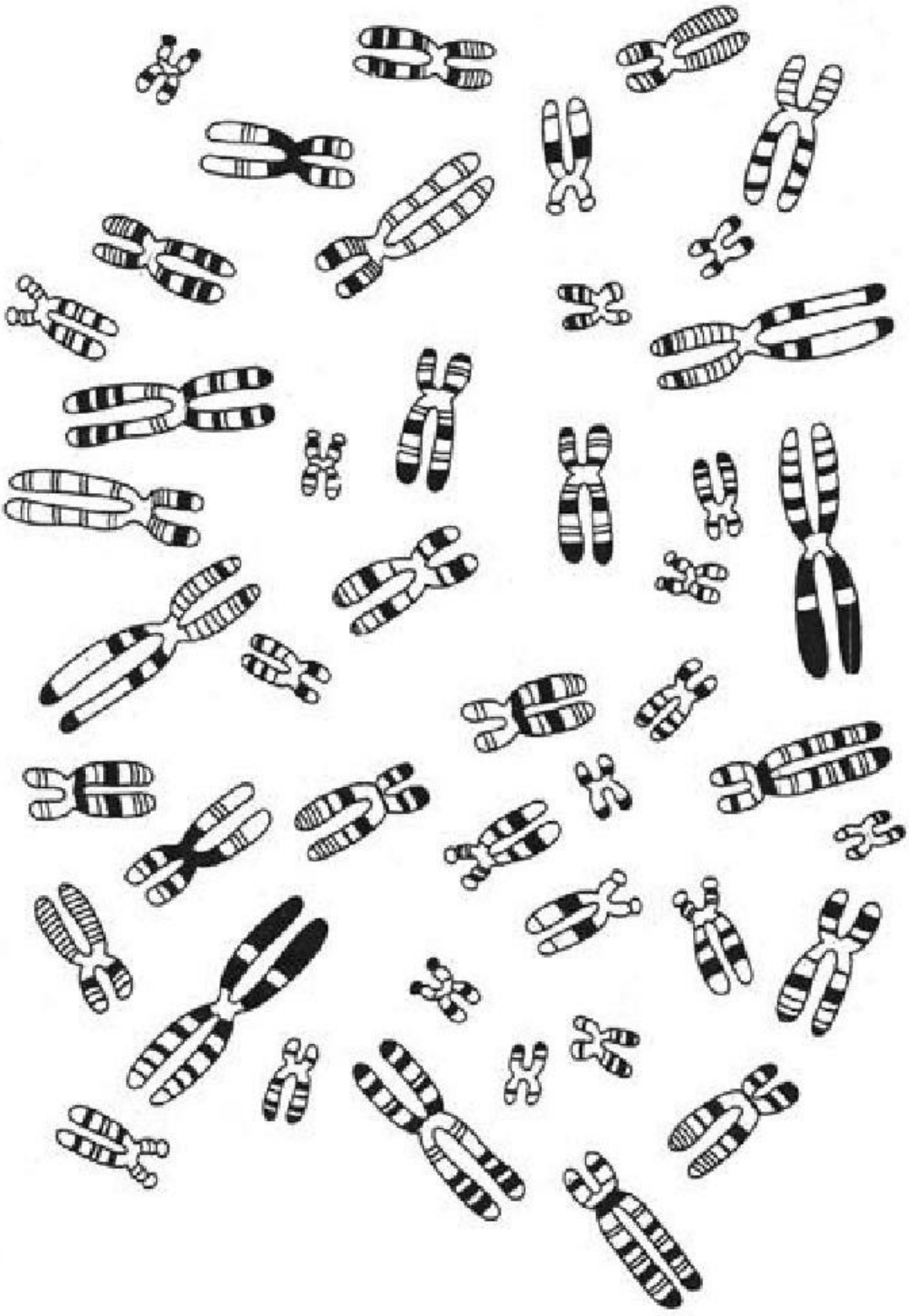
Chromosome Spread # 1



Chromosome Spread # 2



Chromosome Spread # 3



Karyotype # 1

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

Karyotype # 2

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

Karyotype Lab
(Answers)

1. a) Yes or no, according to whether or not the completed Karyotype matches the normal human Karyotype.

   b) Answers would probably include that there is an extra chromosome or a missing chromosome.

   c) This will depend on the chromosome spread given to the student.
   Chromosome spread # 1 - Klinefelter's Symdrome
   Chromosome spread # 2 - Down Syndrome
   Chromosome spread # 3 - Turner's Syndrome

2. Refer to answer in question 1.

3. Two processes: 1. Amniocentesis → needle to withdraw amniotic fluid

   2. Chorionic villus sampling → remove a small section of chorion (villi)

4. Fetoscopy → fibre optic camera

   Ultrasound → ultrasound imaging

5. Use, for example, trisomy 21 (Down Syndrome) which is caused by non-disjunction of the 21st tetrad. The child will have a mild to moderate mental impairment, large thick tongue causing speech defects, underdeveloped skeleton, and be more susceptible to infections. However, the individual will be quite functional in society, generally happy and outgoing.