Name: _____

Date: _____ Per: _____

Karyotype Lab

PURPOSE:
This lab will give students the opportunity to study amniocentesis and karyotype procedures.

MATERIALS:
Activity sheet
Glue or tape
Pen or pencil
Scissors

PROCEDURES

1. In this lab activity, you will pretend you have been hired by a genetics testing laboratory. A lab report comes to you with amniotic fluid sample taken from a pregnant woman. The physician suspects the fetus may have a birth defect for the following reasons:
    a. The father's family has a history of children born with defects
    b. The mother is 39 years old.
    c. The mother is known to have used the medication Accutane during this pregnancy.

    If the karyotype analysis from the amniotic fluid is normal, then the physician may prescribe a sonogram to detect any physical deformities in the fetus. While it may not be possible to detect physical defects caused by Accutane, the mother's use of this drug puts the child at risk for birth disorder. This is the first time you have performed the amniocentesis.

2. You now have received the chromosome scatter sheet and you prepare to make the karyotype. Record the letter on the scatter sheet for future reference.

3. Carefully cut out each of the chromosomes in the scatter worksheet you have been given. Save the large capital letter in the bottom right-hand corner. Rearrange the chromosomes into matching pairs using your karyotype reference sheet as a guide. When searching for a mate for each chromosome, remember to consider the length of the "arms" and the placement of the CENTEROMERE, the point where the two CHROMATIDS (halves) of the chromosome are joined.

4. Copy the large letter that you found on the lower right-hand corner of your scatter sheet. Write it at the top of your Karyotype Layout Sheet

5. After all the chromosomes pairs are matched, glue the matched pairs onto the Karyotype Layout Worksheet, arranging the pairs in order from longest to shortest.

LABORATORY QUESTIONS:
Answer the following questions, using complete sentences.

1. What is the name of the fluid in which a developing embryo or fetus floats?

2. What is the name of the procedure that removes and tests amniotic fluid?

3. How many pairs of chromosomes were present in the karyotype you completed?

4. What was the sex of the child who donated these chromosomes?

5. Did you find the fetal chromosomes to be (apparently) normal or abnormal?

6. If the karyotype has an abnormal number of chromosomes, use the chart "Human Birth Defects Resulting in Chromosomal Disorders," to identify the condition. List the genetic characteristics of this condition. If the karyotype has a normal number of chromosomes, answer this "normal baby"

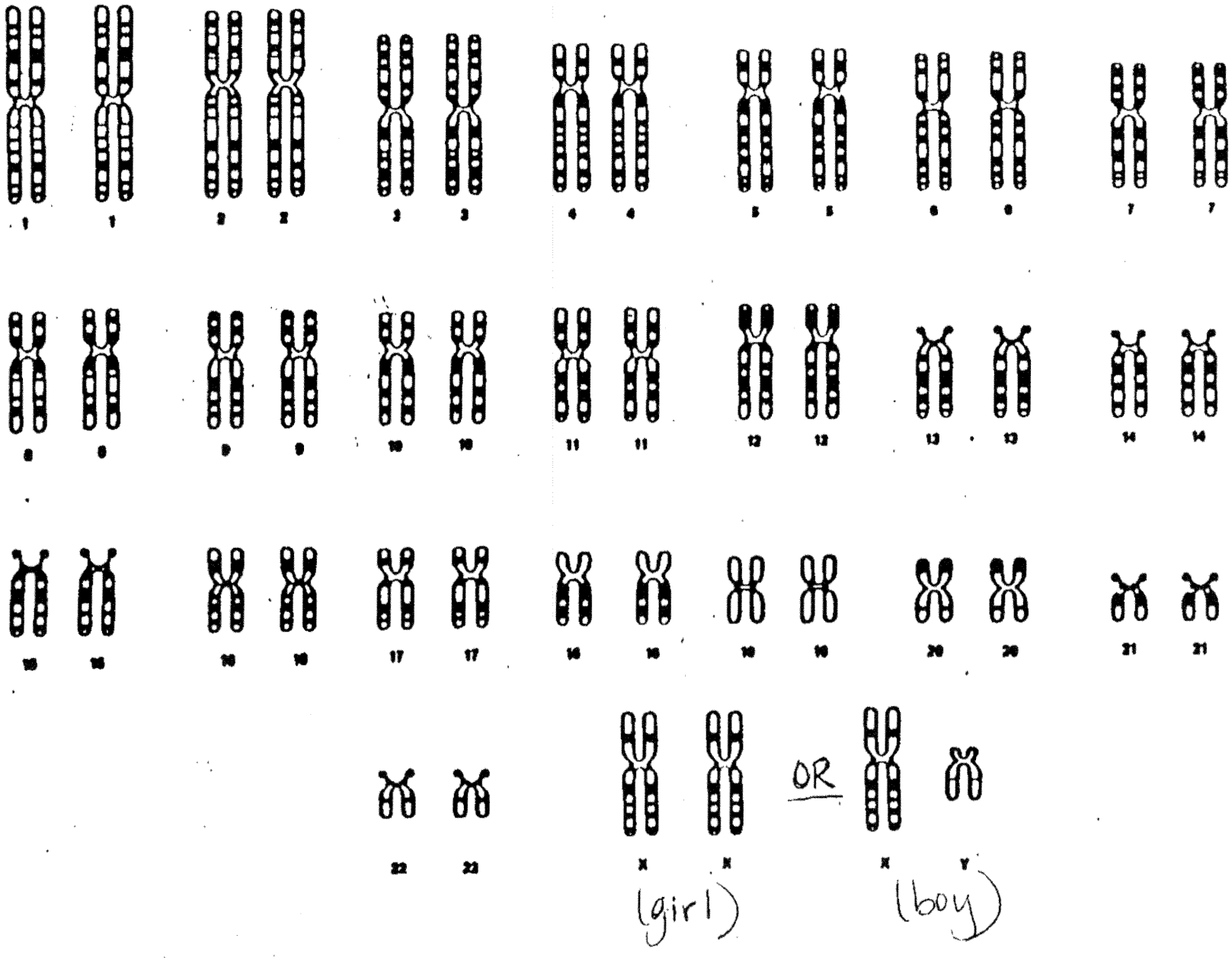
Karyotype Scatter Sheet reference letter _____ Names _____

_____

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 X Y	

Kayotype Reference Sheet



Background Information (cont.)

Chromosome Deletions and Their Resulting Characteristics

Abnormal Chromosome Number	Number of Cases per 1000 Live Births	Main Characteristics
4	Several dozen	Microcephaly, severe mental retardation seizures, characteristic wide nose bridge, frequent cleft lip and palate, early death but may survive several decades.
5	100	*Cri-du-chat* syndrome: "moon" face, microcephaly, hypertelorism, small mandible, severe mental retardation, distinctive infant cry like kitten, good survival.
9	10	Trigonocephaly, frequent cardiac malformations, IQ 30–60, good survival.
11	6	Trigonocephaly, mental retardation.
12	6	Microcephaly, moderate mental retardation.
13	Several dozen	Microcephaly; moderate to severe mental retardation, bridge of nose broad and high; frequent hypertelorism; ocular anomalies, bone anomalies, retinoblastoma.
18	100	Small head and body size; moderate to severe mental retardation; some have severe malformations of head; good survival.
18	100	Hypotonia, moderate microcephaly, depressed midface, ocular malformations, mild to severe mental retardation; high childhood death rate with some survival to adulthood.

Background Information (cont.)

Human Birth Defects Resulting from Chromosome Disorders

Total Number of Chromosomes	Affected Chromosome Pair	Birth Defect	Estimated Frequency at Birth	Main Characteristics
47	21	Down syndrome	1 / 700	Short broad hands with simian palmar crease, short stature, hyperflexibility of joints, mental retardation, broad head with round face, open mouth with large tongue, slanting eyes.
47	13	Patau syndrome	1 / 5000	Mental deficiency and deafness, minor muscle seizures, cleft lip and/or palate, polydactyly, cardiac anomalies, posterior heel prominence.
47	18	Edwards trisomy	1 / 4000 to 1 / 18000	Multiple congenital malformation of many organs; low-set, malformed ears; receding mandible, small mouth and nose with general elfin appearance; mental deficiency; horseshoe or double kidney; short sternum. 90 percent die in the first six months.
45	23 (-X)	Turner (XO) syndrome	1 / 5000	Female with retarded sexual development, usually sterile; short stature, webbed skin in neck region, cardiovascular abnormalities, hearing impairment.
47 48 48 49 50	23 (+X) 23 (+XX) 23 (+XY) 23 (+XXY) 23 (+XXXY)	Klinefelter (XXY) syndrome (XXXY) (XXYY) (XXXYY) (XXXXYY)	1 / 500	Male, subfertile with small testes, developed breasts, feminine pitched voice, mental deficiency, long limbs, knock knees, rambling talkativeness, frequent early death.
47	23 (+X)	Triple X	1 / 700	Female with underdeveloped genitalia and limited fertility; frequent neuromotor delay.

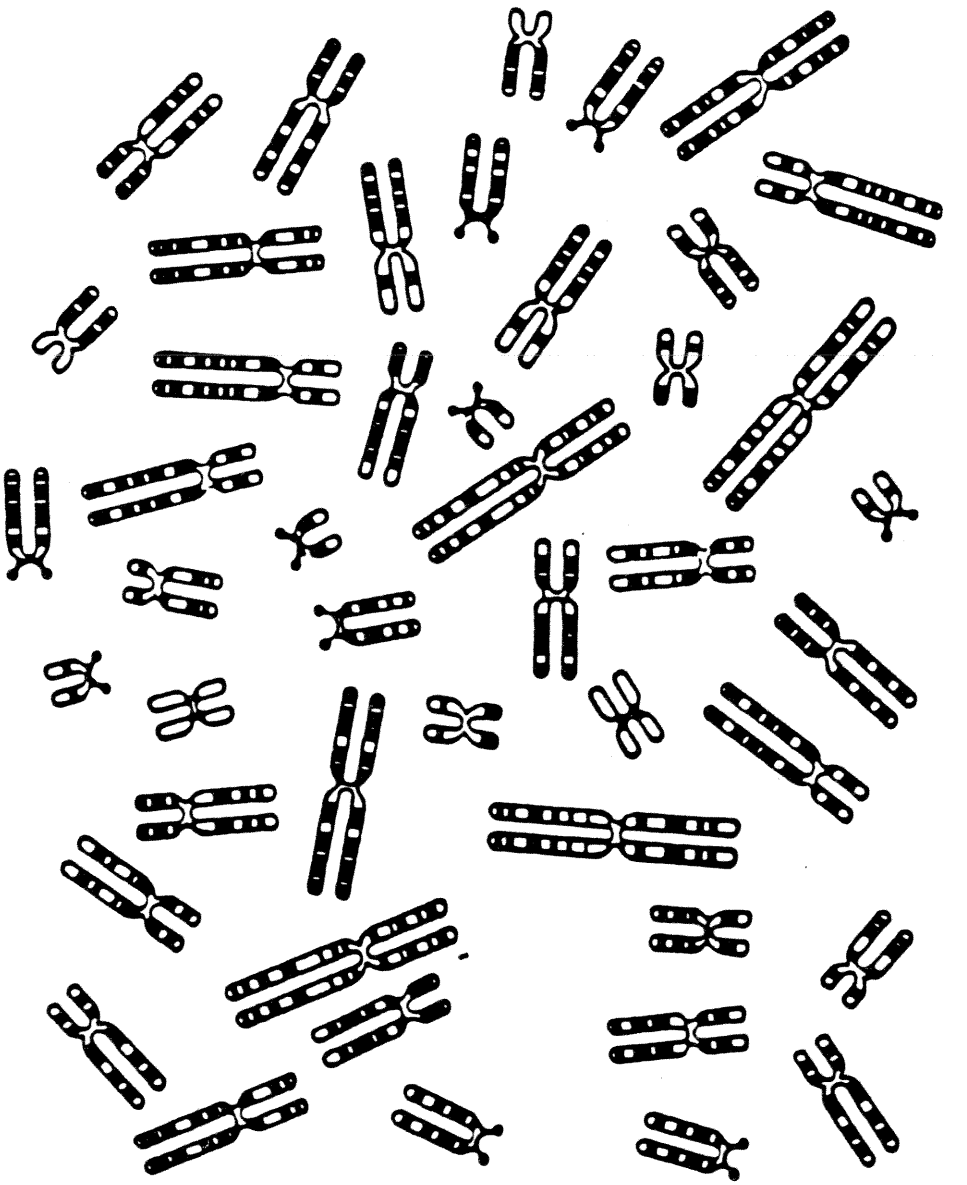
\+ = additional chromosome

\- = deleted chromosome

X and Y = sex chromosomes

"A"
100
White

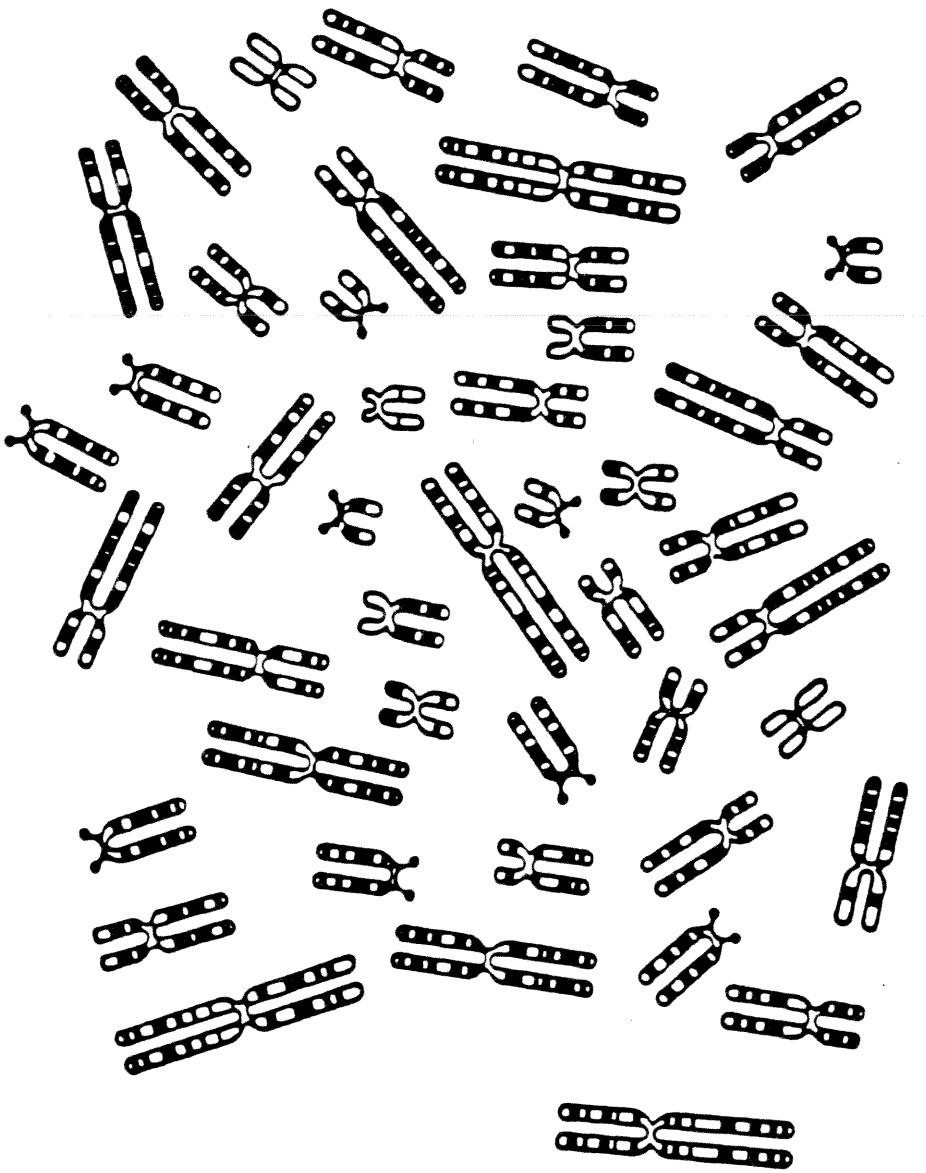
Chromosome Scatter Sheet



A

Green "B"
~~75~~ 100
Copies

Chromosome Scatter Sheet

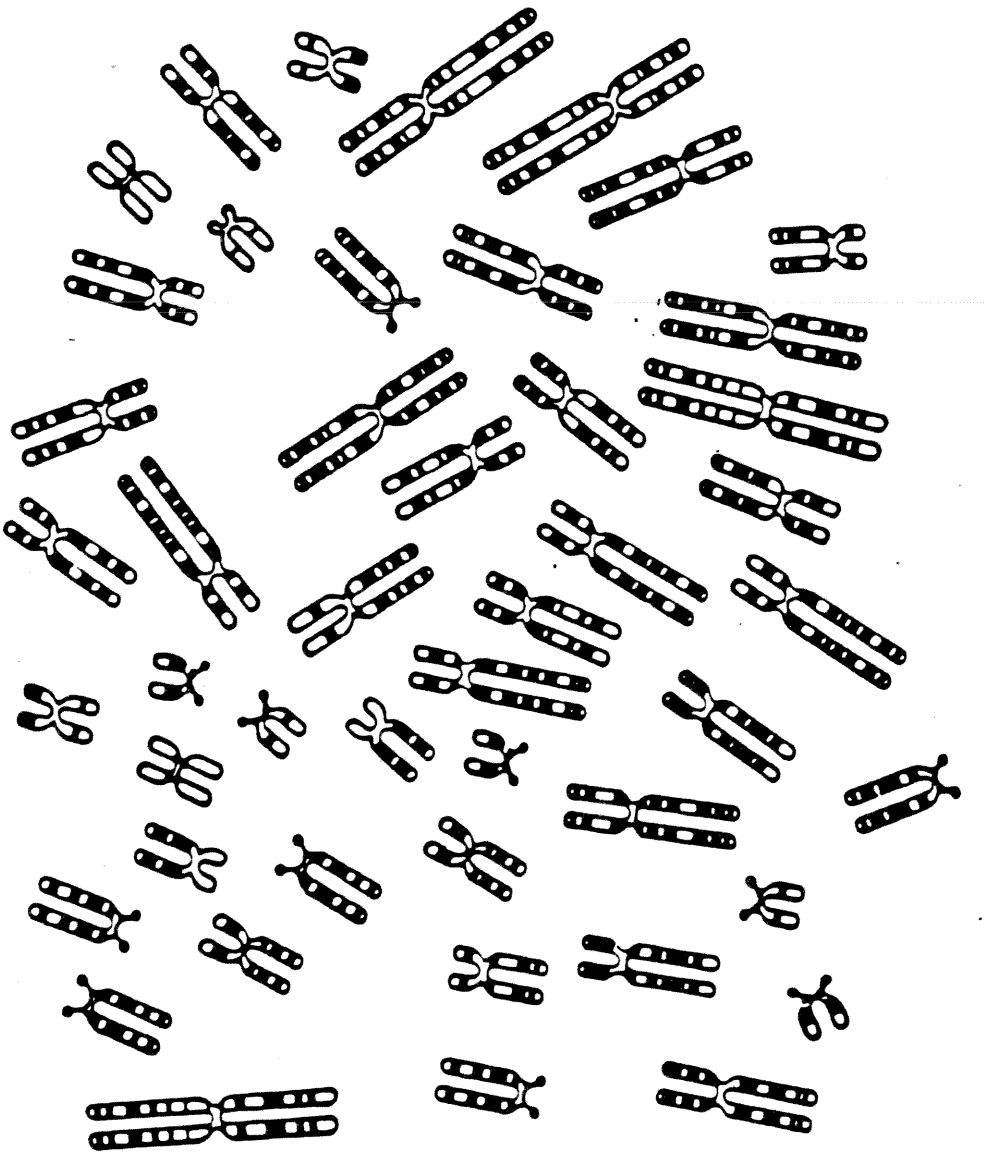


B

pink 'D'

75 copies

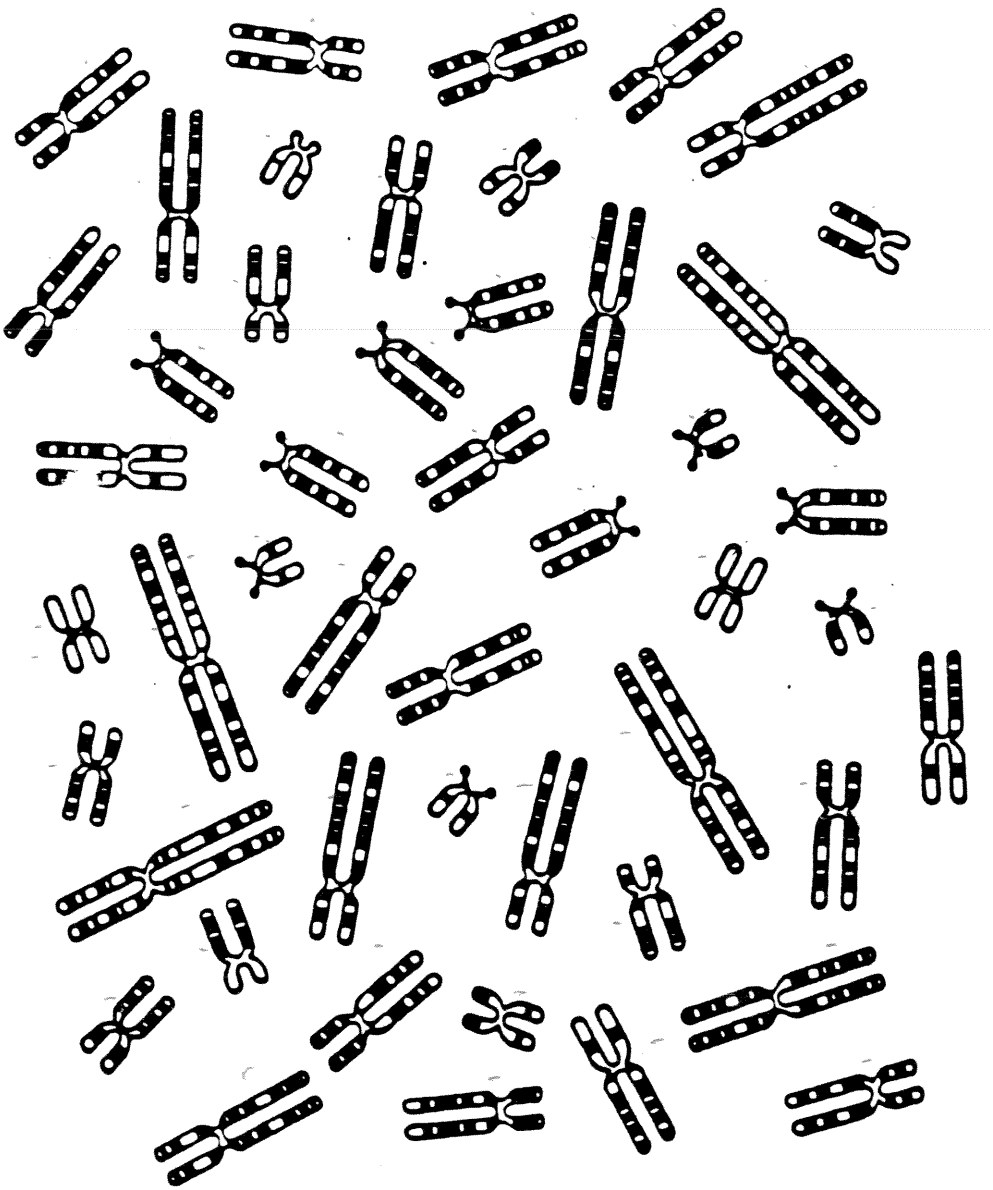
Chromosome Scatter Sheet



D

75 Copies "E"
blue

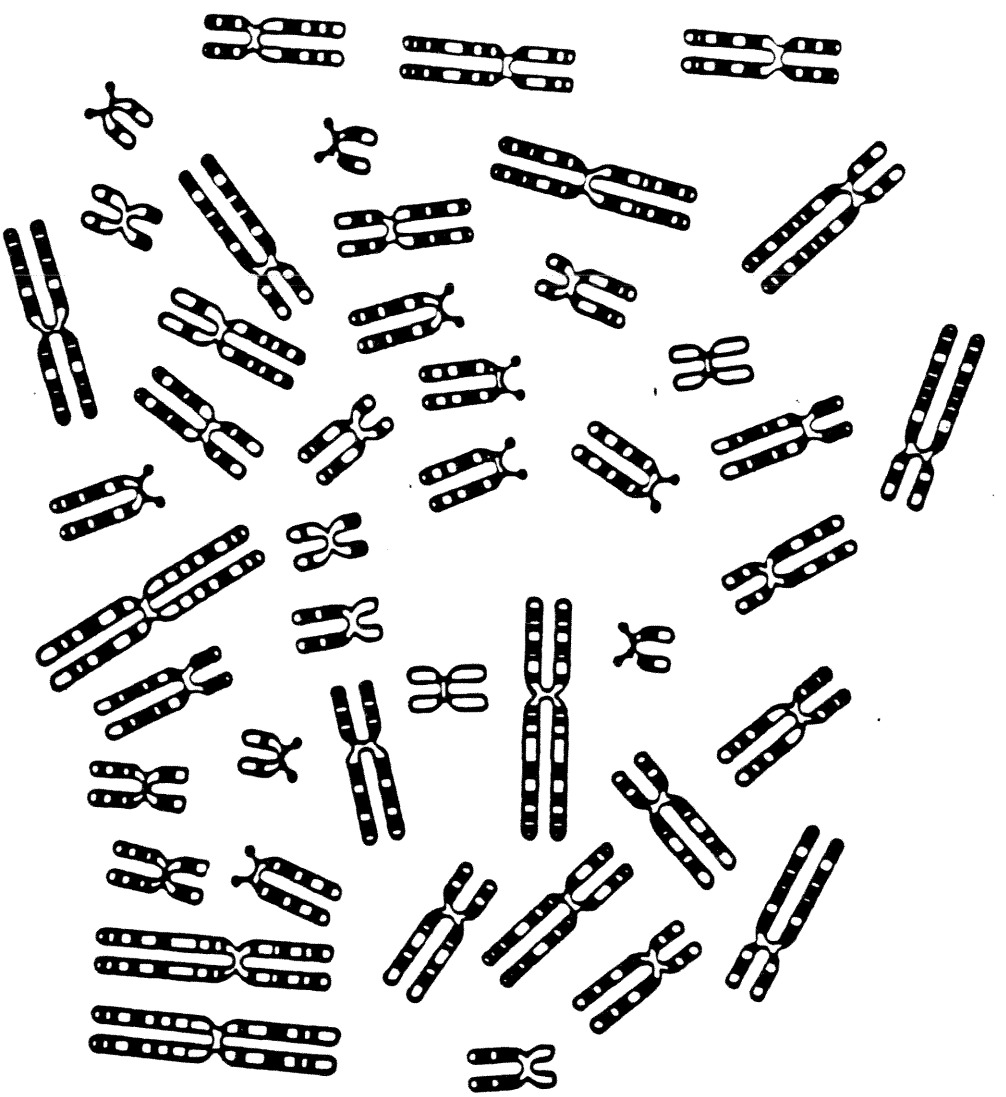
Chromosome Scatter Sheet



E

75 Copies "F"
Yellow

Chromosome Scatter Sheet



F

F = Turners = yellow

E = Kleinf blue

D = Down - pink

B = ♂ green

A = ♀ white