Human Inheritance

Ch. 11.1

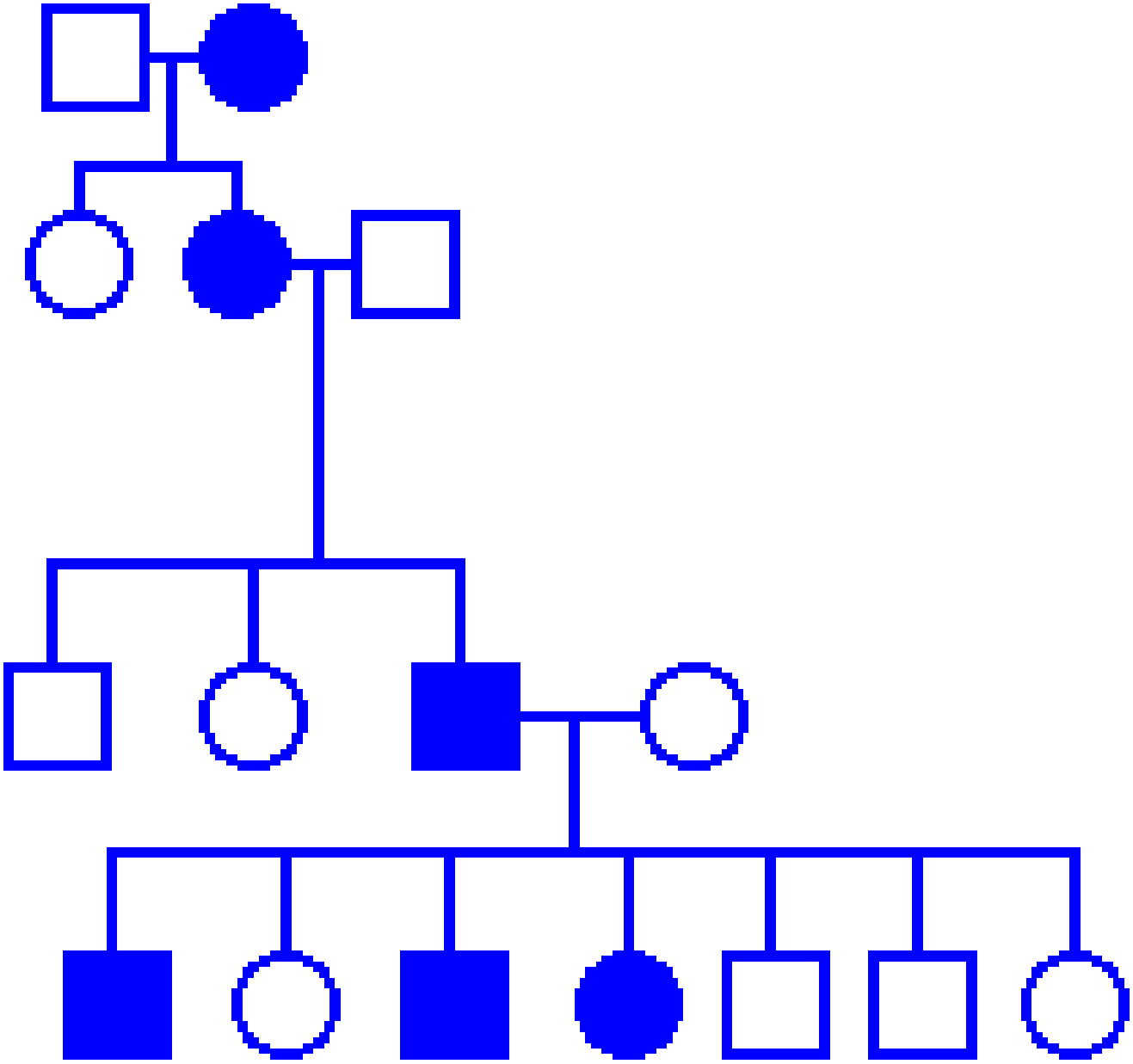
A. Mutations

1. Recessive

2. If lethal

B. Pedigrees

1. Squares vs. circle
2. Horizontal line
3. Vertical line

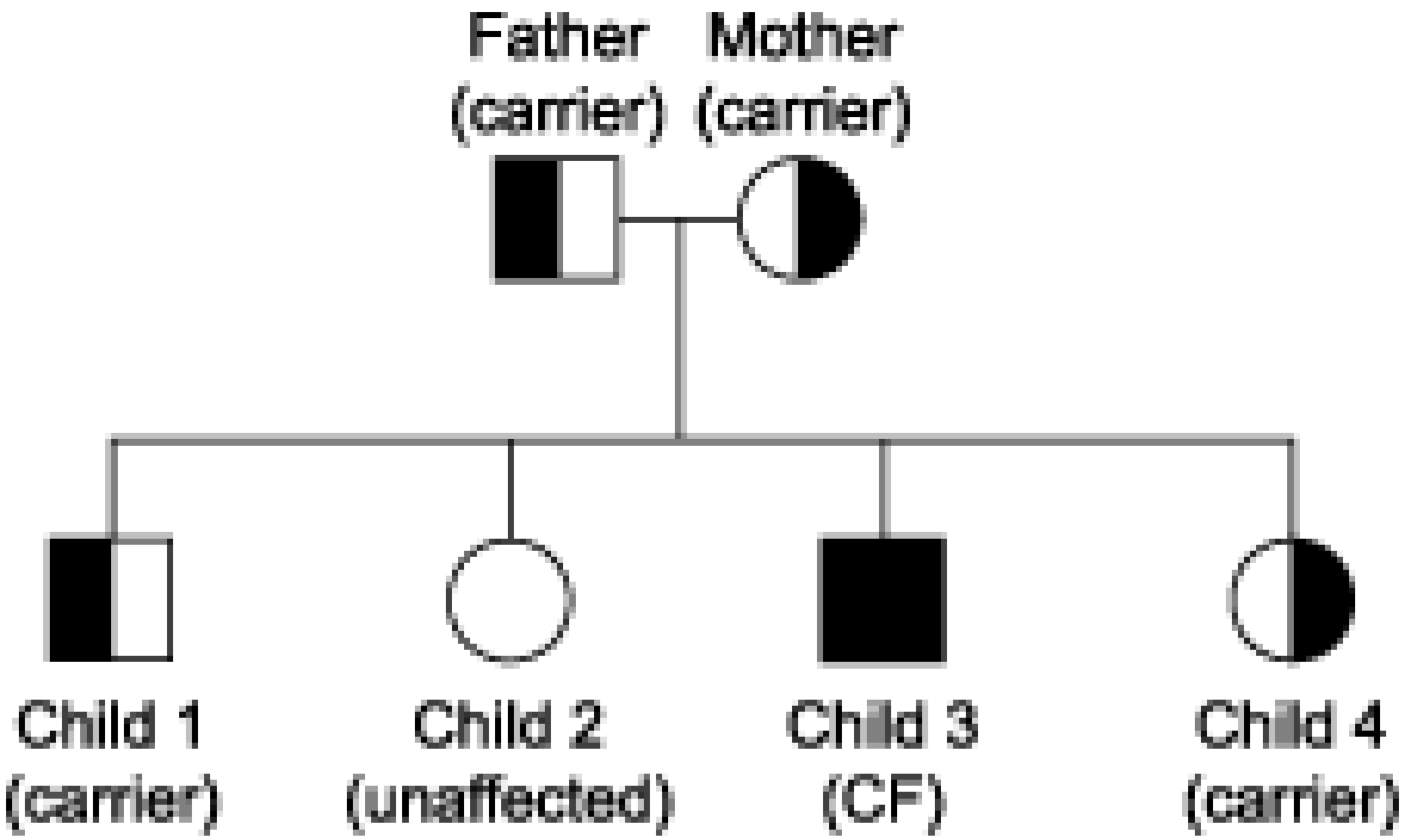


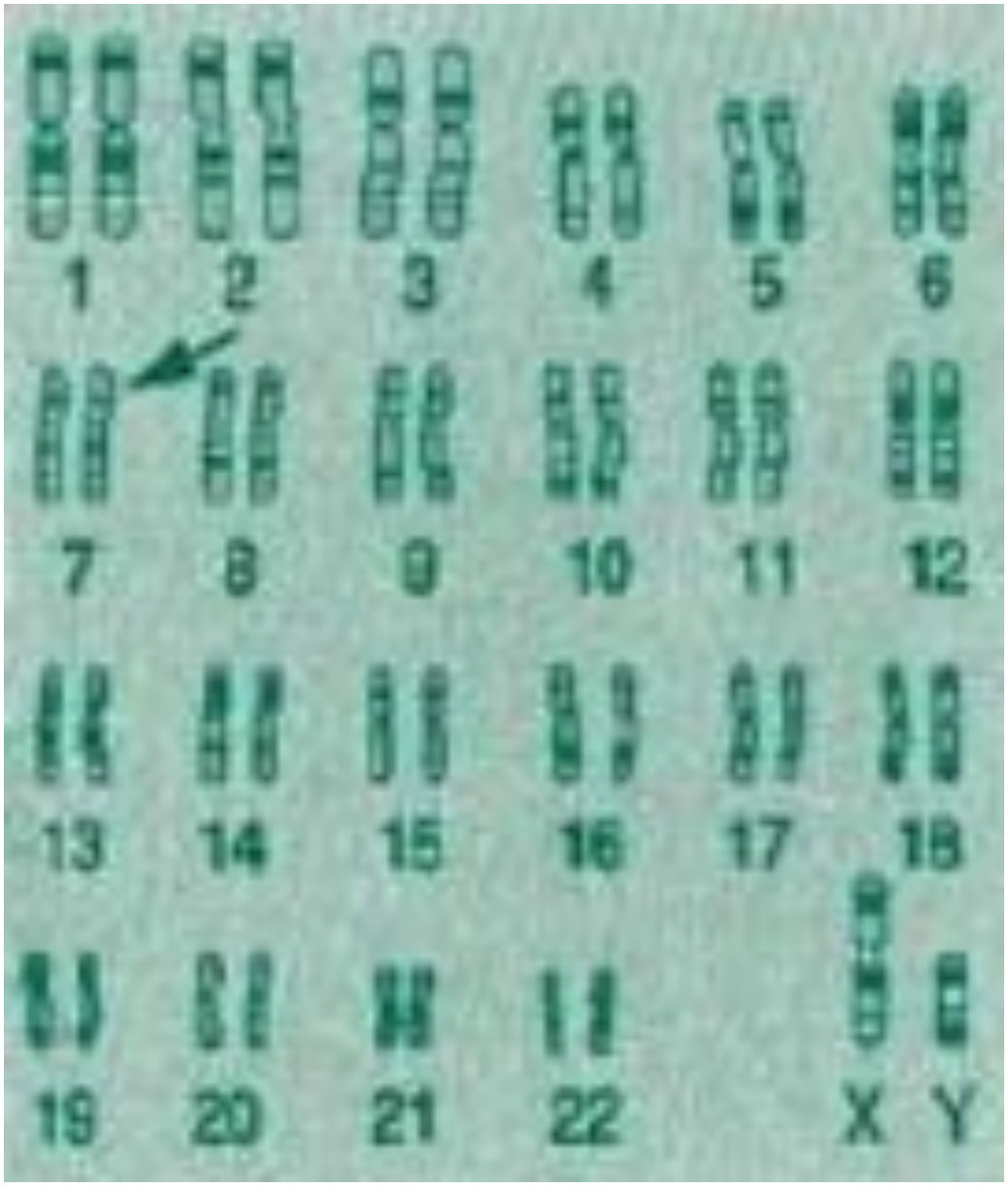
C. Examples

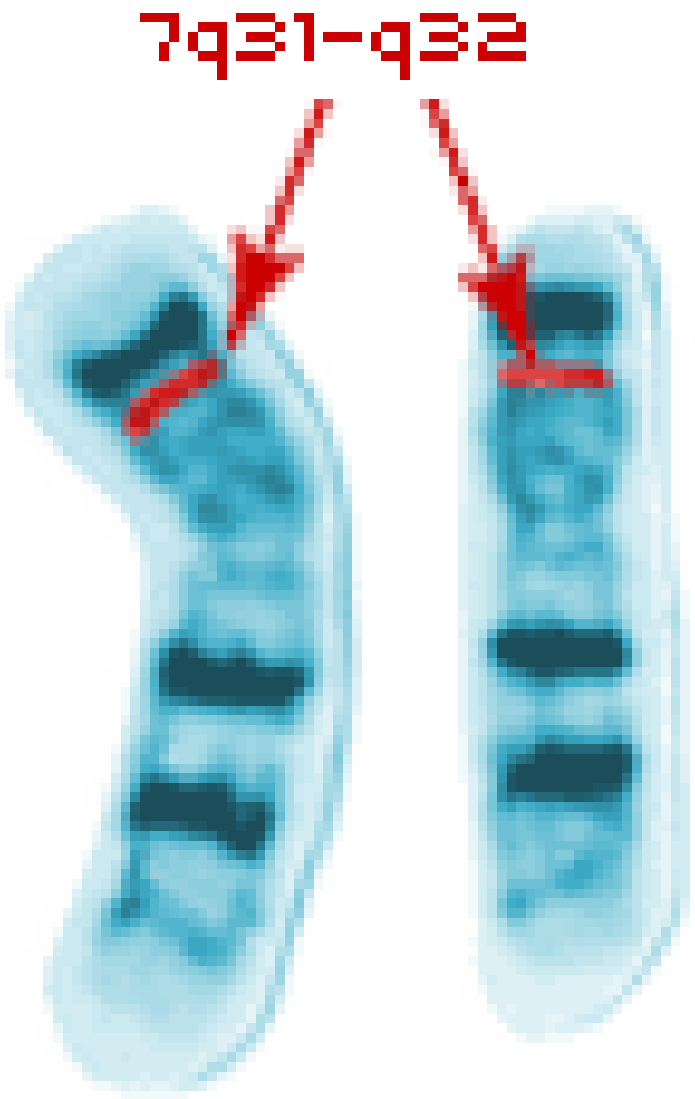
1. CF-#7

    a. Recessive

    b. Membrane protein


Father
(carrier)
Mother
(carrier)
Child 1
(carrier)
Child 2
(unaffected)
Child 3
(CF)
Child 4
(carrier)


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


7q31-q32

c. Die by 40

d. European

2. Sickle cell anemia-#11

    a. Recessive

    b. Hemoglobin

    c. African

    d. Heterozygous adv.

HBB Sequence in Normal Adult Hemoglobin (Hb A):

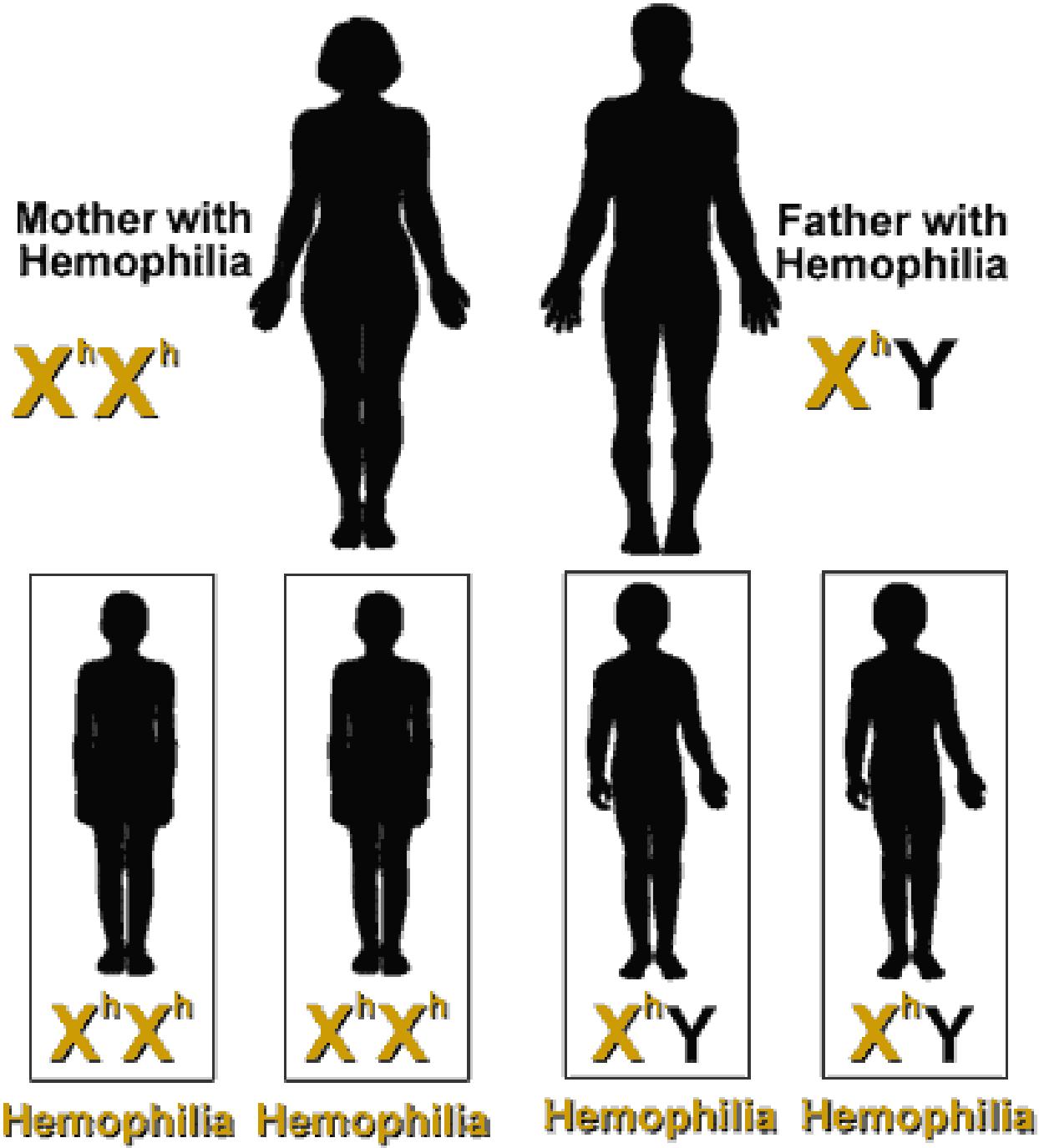
Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Glu	Glu	Lys	Ser
	3			6			9

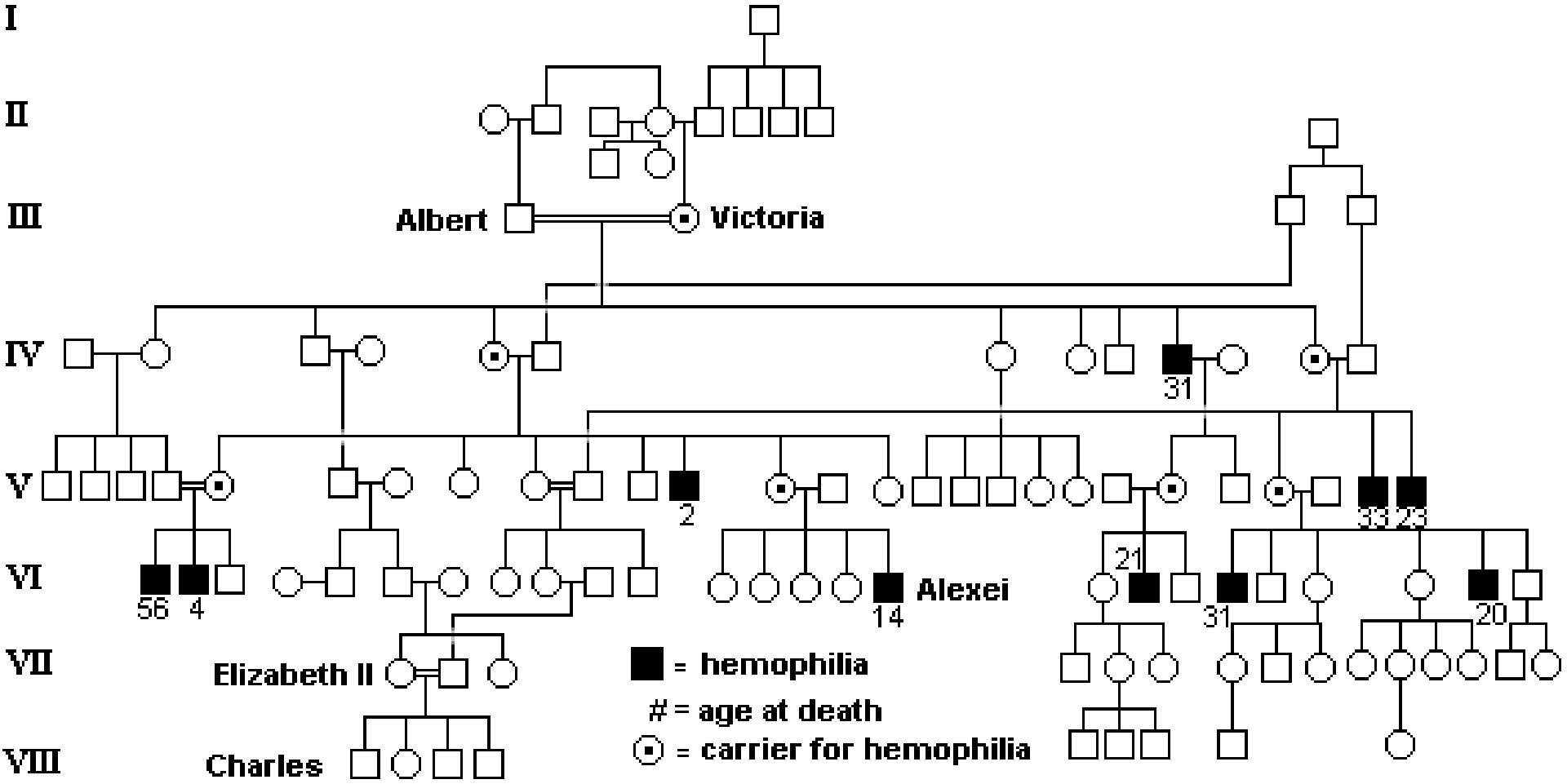
HBB Sequence in Mutant Adult Hemoglobin (Hb S):

Nucleotide	CTG	ACT	CCT	GTG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Val	Glu	Lys	Ser
	3			6			9

*Replace “T” with “U” for the RNA transcript

3. Hemophilia-#23X
    a. Sex linked trait
    b. Mainly males


Mother with Hemophilia
X^hX^h
Father with Hemophilia
X^hY
X^hX^h
X^hX^h
X^hY
X^hY
Hemophilia
Hemophilia
Hemophilia
Hemophilia

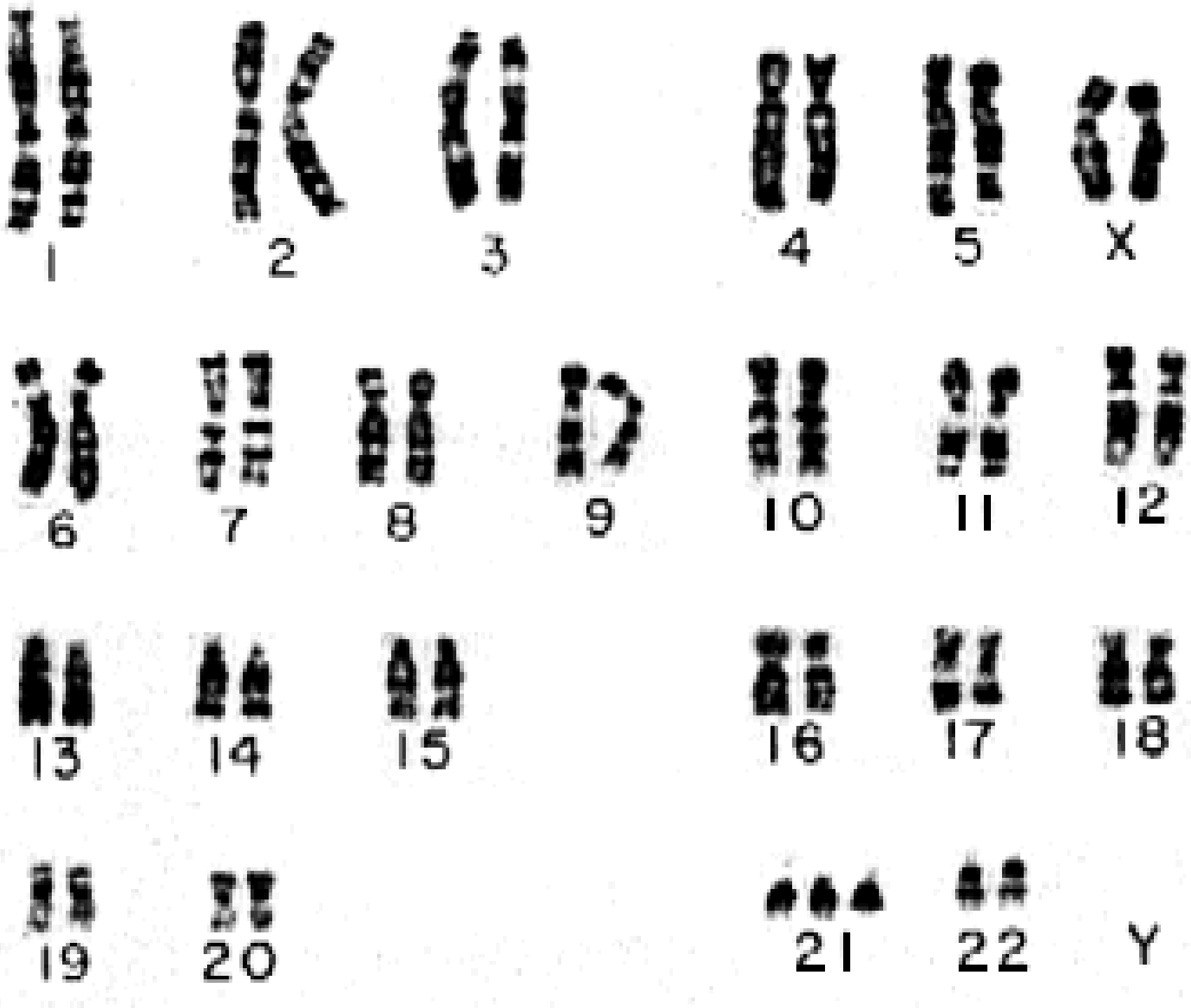

I
II
III
IV
V
VI
VII
VIII
Albert
Victoria
31
2
33
23
21
56
4
14
Alexei
31
20
Elizabeth II
Charles
= hemophilia
= age at death
= carrier for hemophilia

4. Down's syndrome- #21

a. Trisomy 21

b. Mental/physical difficulty

c. Mom over 40


1
2
3
4
5
X
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7
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14
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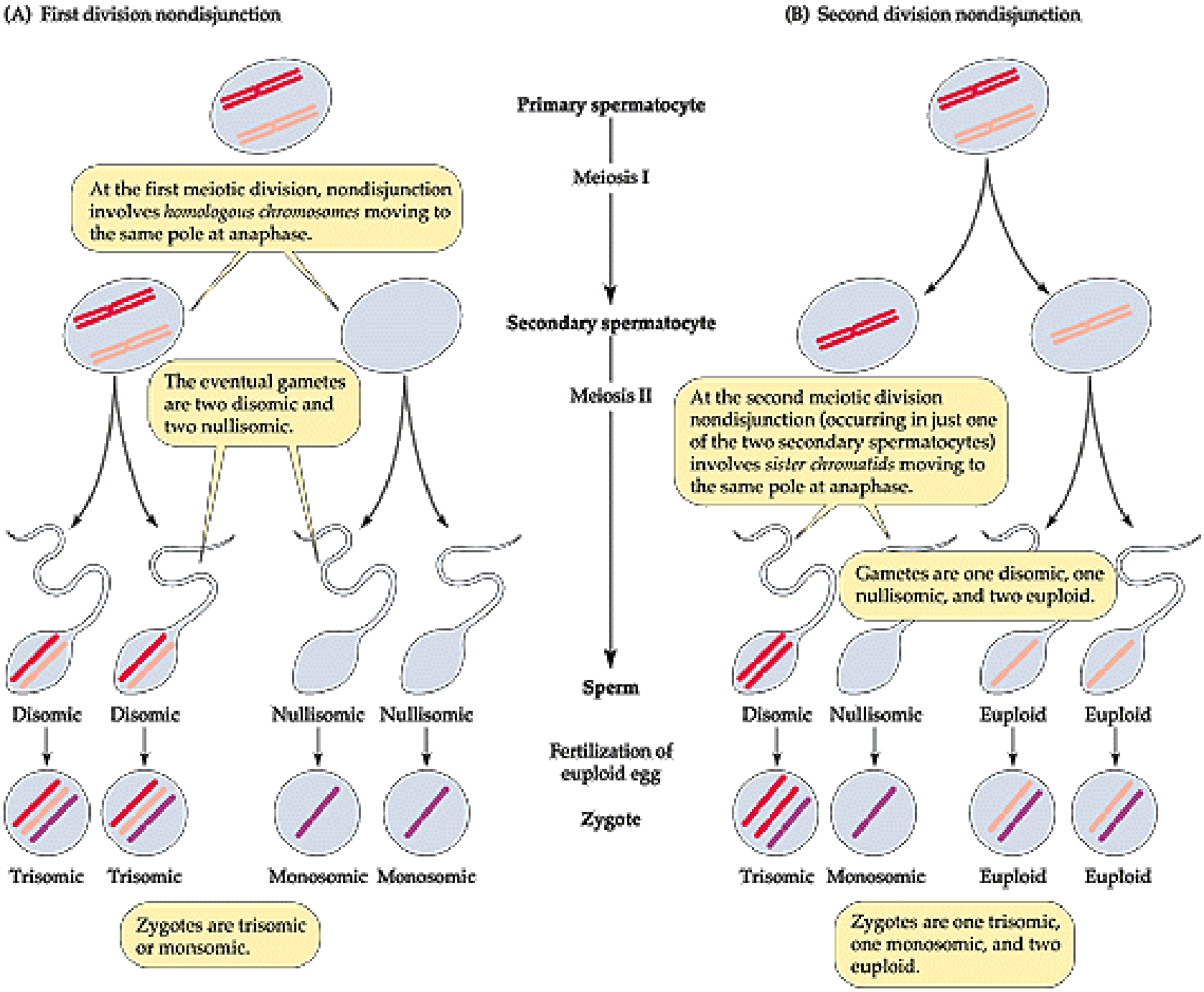
16
17
18
19
20
21
22
Y


(A) First division nondisjunction
(B) Second division nondisjunction
Primary spermatocyte
Meiosis I
Secondary spermatocyte
Meiosis II
Sperm
Fertilization of euploid egg
Zygote
At the first meiotic division, nondisjunction involves homologous chromosomes moving to the same pole at anaphase.
The eventual gametes are two disomic and two nullisomic.
Disomic
Disomic
Nullisomic
Nullisomic
Trisomic
Trisomic
Monosomic
Monosomic
Zygotes are trisomic or monosomic.
At the second meiotic division nondisjunction (occurring in just one of the two secondary spermatocytes) involves sister chromatids moving to the same pole at anaphase.
Gametes are one disomic, one nullisomic, and two euploid.
Disomic
Nullisomic
Euploid
Euploid
Trisomic
Monosomic
Euploid
Euploid
Zygotes are one trisomic, one monosomic, and two euploid.

Nondisjunction: chromosomes do not separate evenly into the developing gametes.

Gamete has too many (47): Down's
Kleinfelters

Gametes have too few (45): Turner

D. Genetic counseling

1. Tools
2. Huntington's correa

A new study from Nancy Wexler, in Venezuela in the 1990s with a boy with Huntington's disease, suggests there may be ways to delay the onset of the disease.