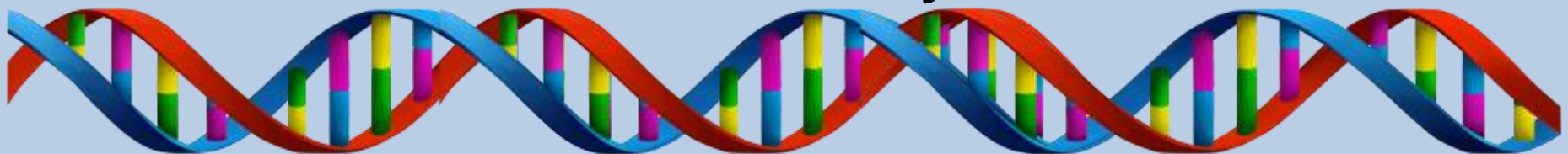
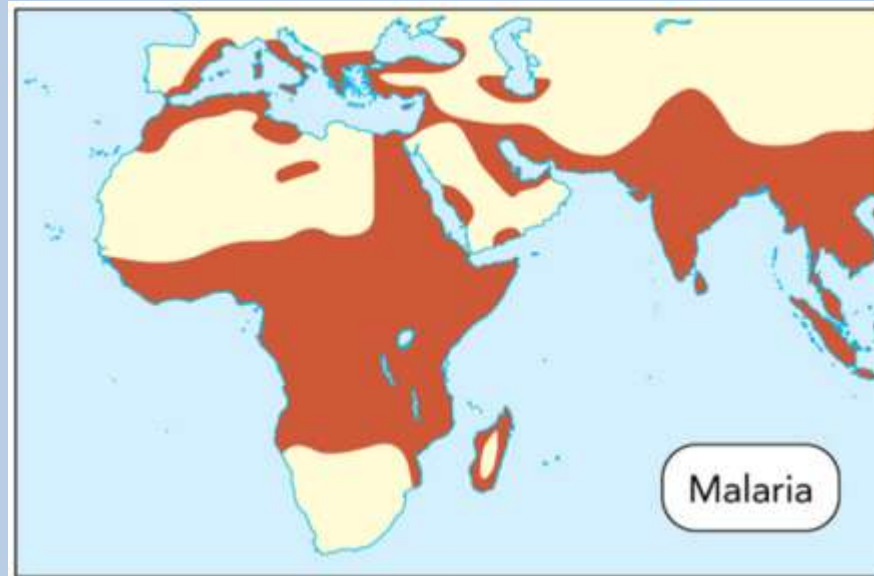


1. Use the maps to answer the questions:
 - a. In 1805 a group of European geographers were surveying in Africa. Most of them died of malaria, but their native guides survived. Why might the native Africans have survived while the Europeans died? (think about genotypes)
 - b. Why do southern African populations not have sickle cell anemia?
 - c. Why might the disease sickle cell anemia still exist if it is deadly?



Sickle Cell Anemia



Sickle Cell Anemia

- Heterozygous Advantage:

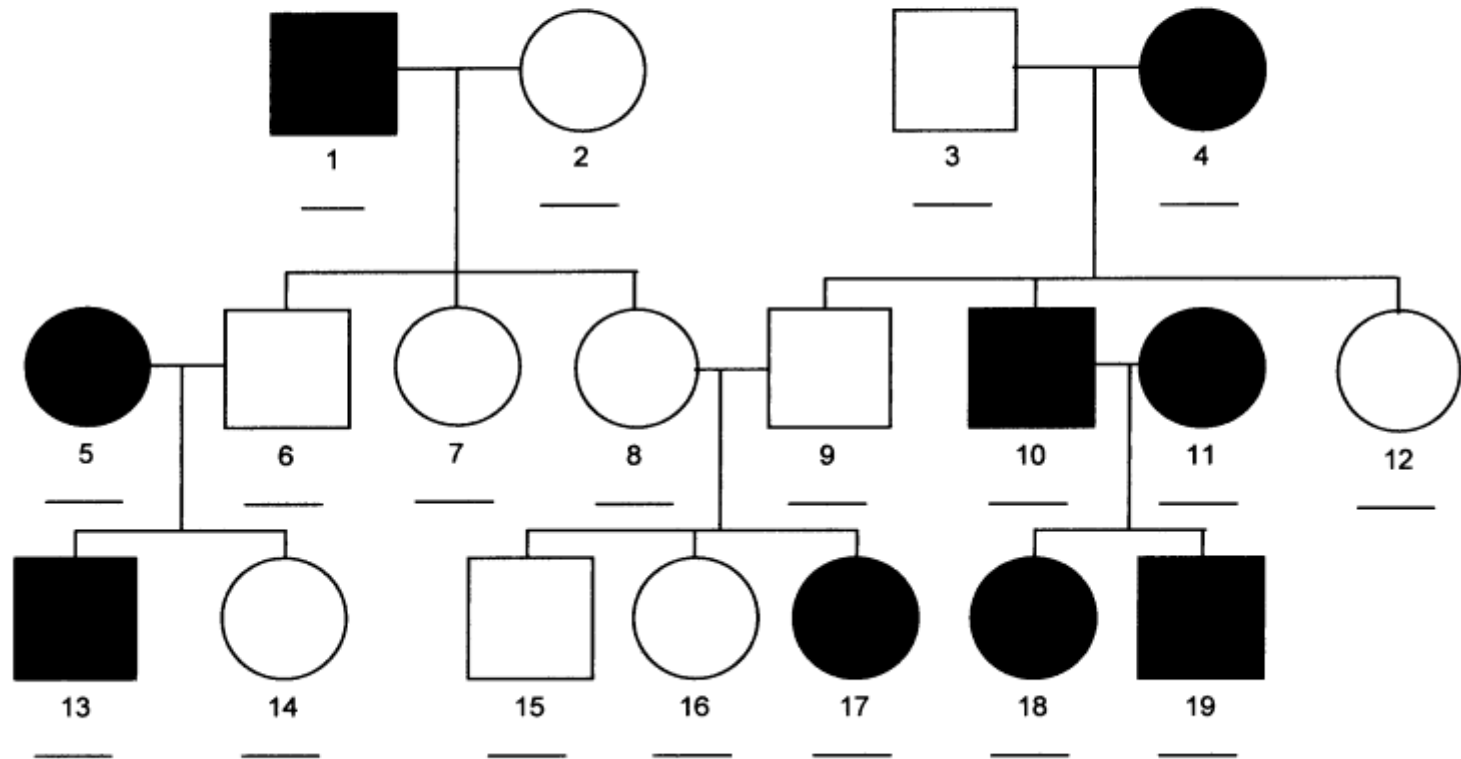
<https://www.youtube.com/watch?v=1fN7rOwDyMQ>

Deciphering Pedigrees

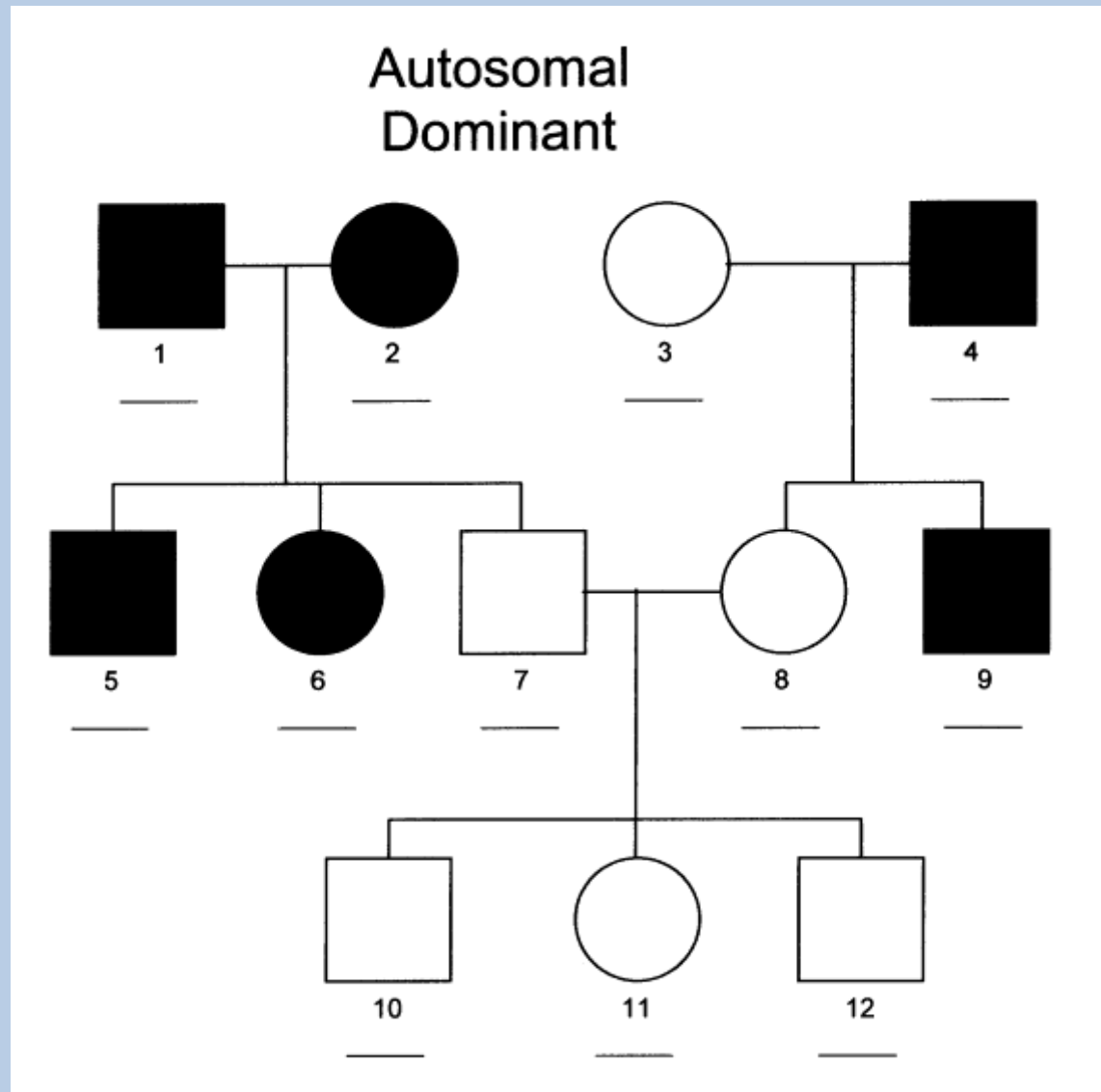
- **Two general hints:**
 - **If the trait SKIPS a generation it is recessive**
 - **If the trait is close to EQUAL in males and females it is autosomal**
 - **You can also look for an affected male and observe his daughters**

Deciphering Pedigrees

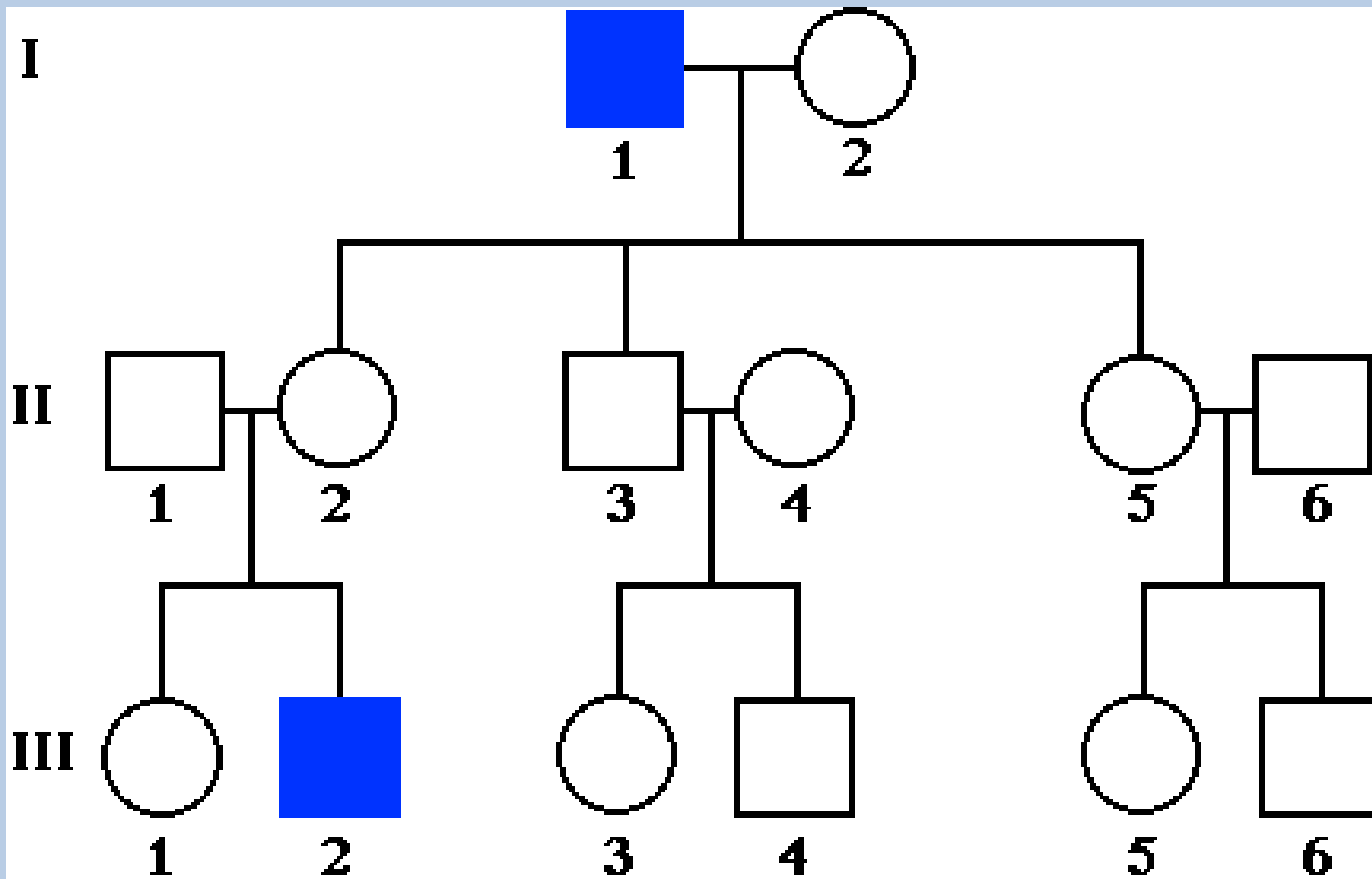
AUTOSOMAL RECESSIVE



Deciphering Pedigrees

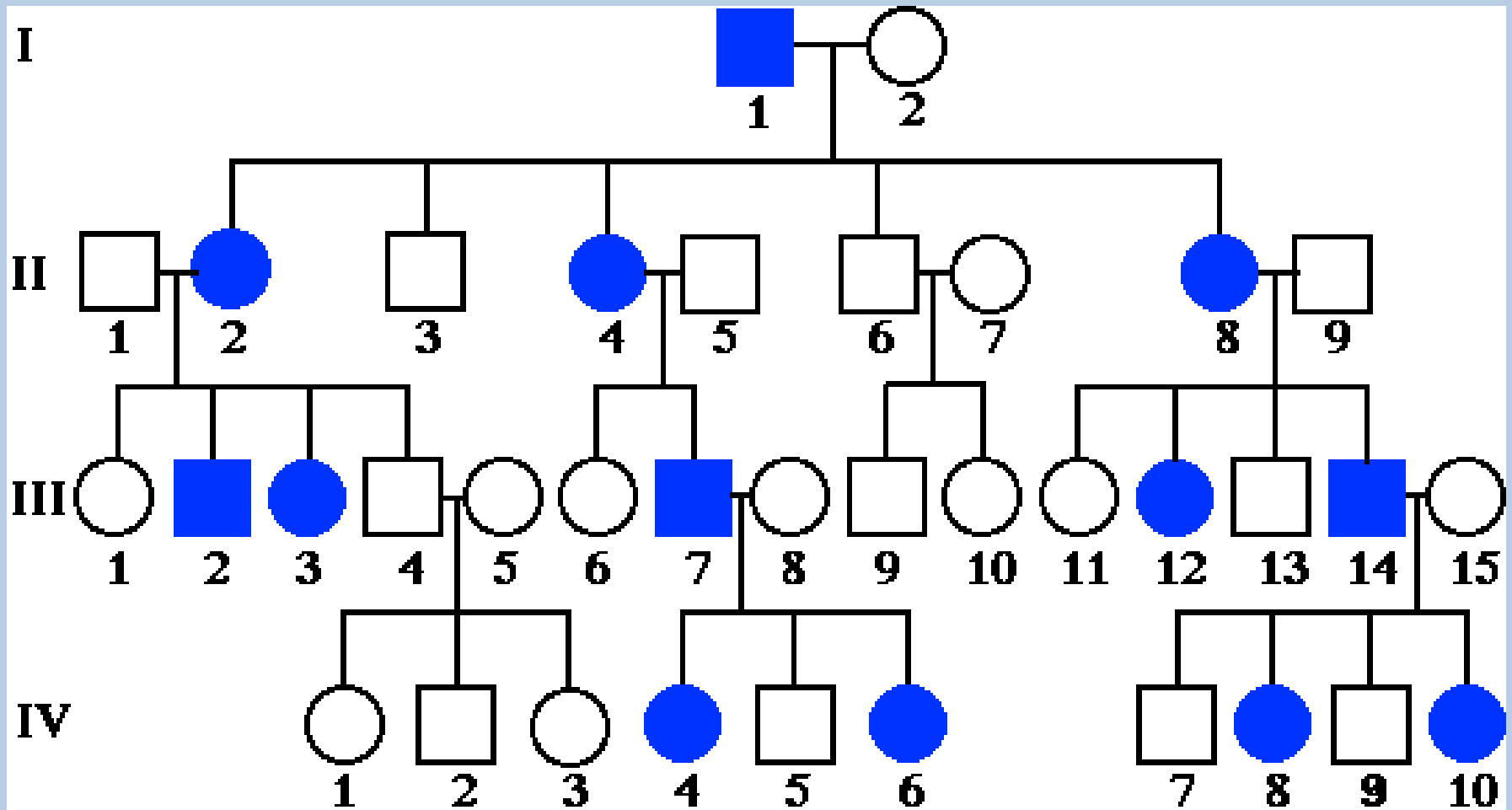


Deciphering Pedigrees



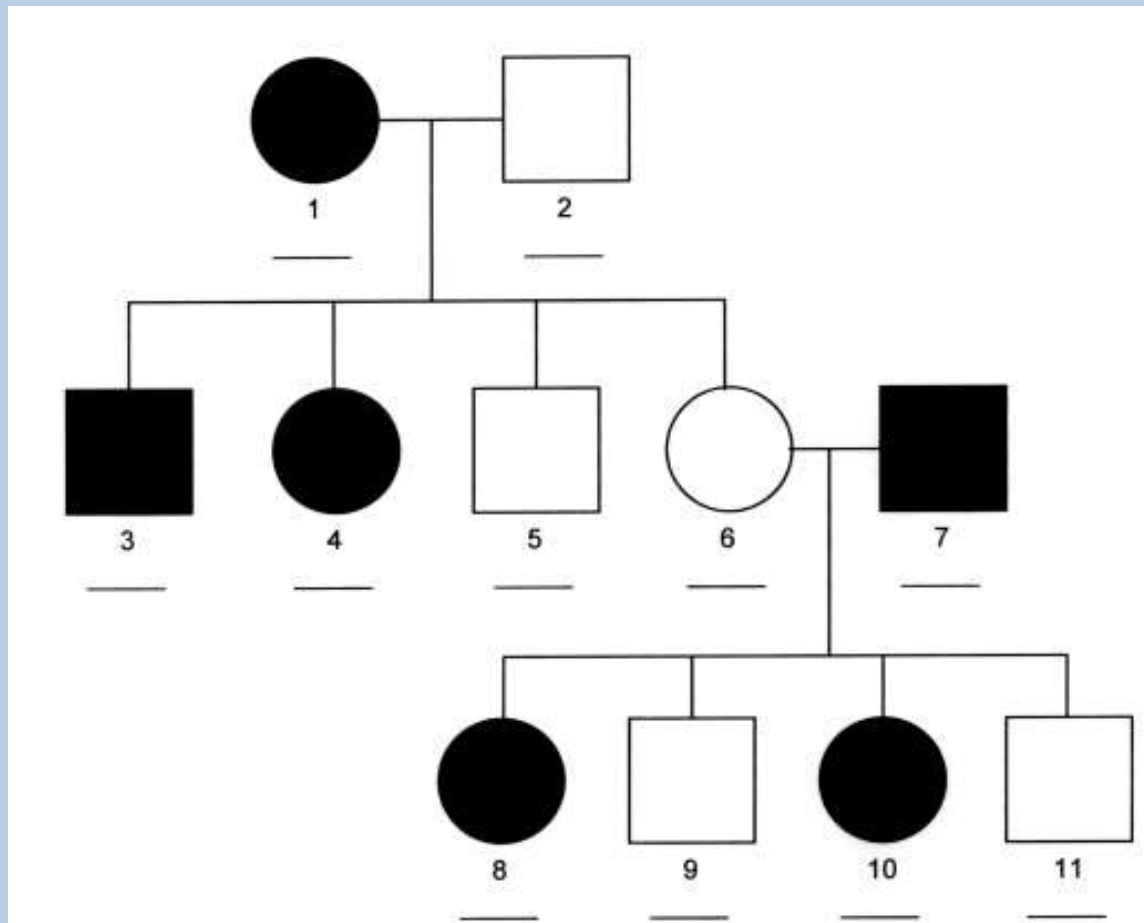
Pedigree 7. X-linked recessive inheritance.

Deciphering Pedigrees

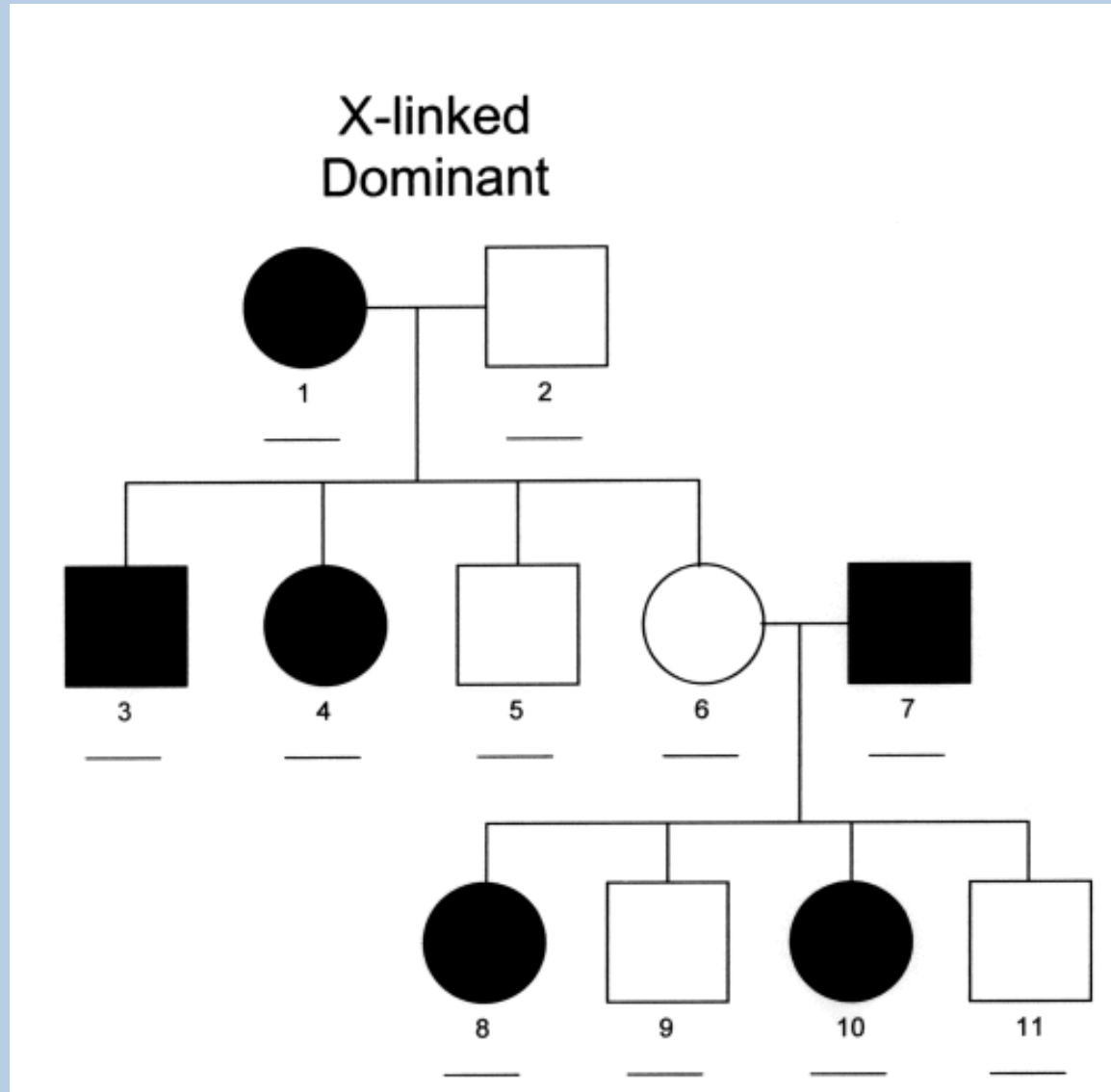


Pedigree 5. X-linked dominant inheritance.

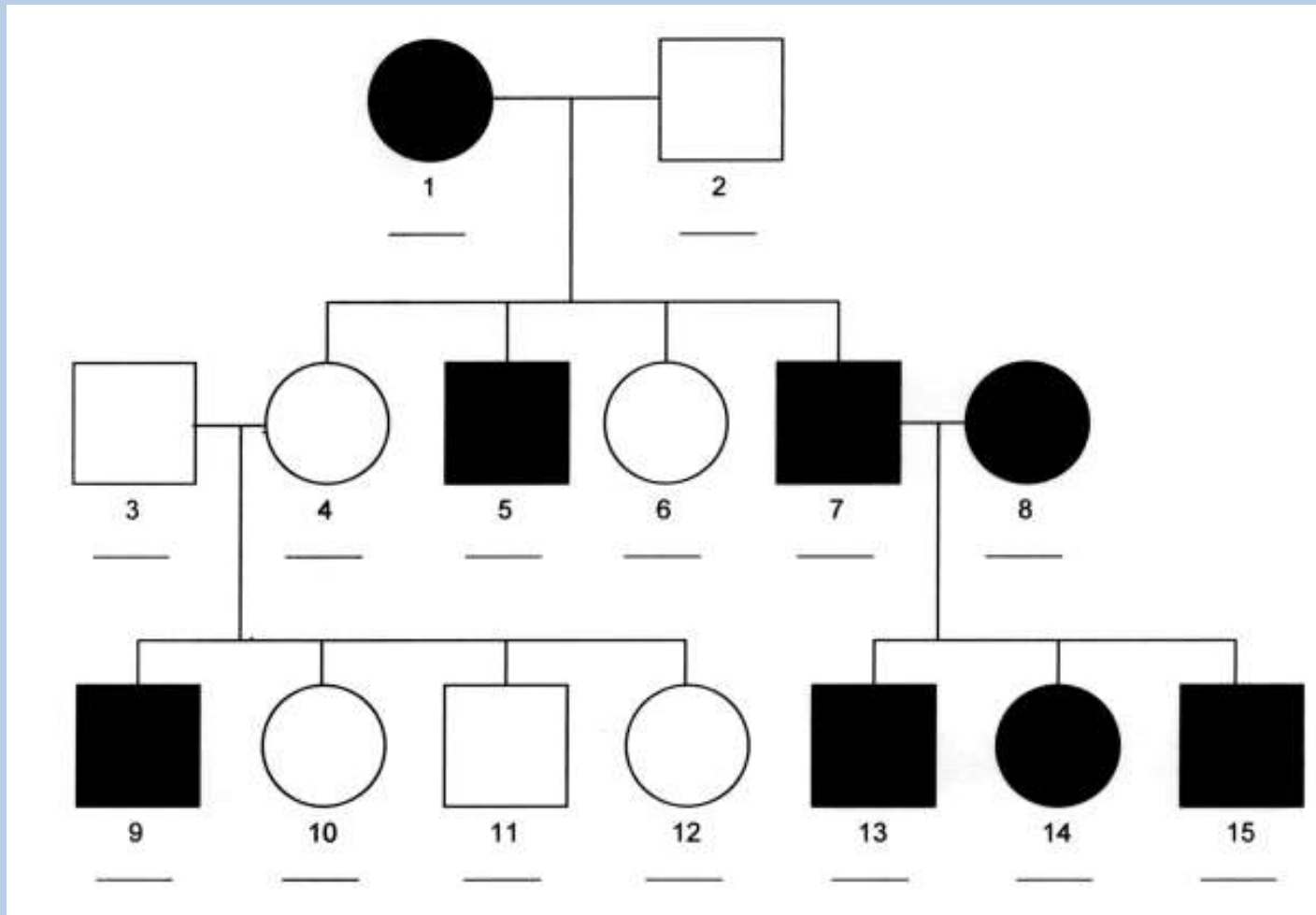
What is the pattern of inheritance?



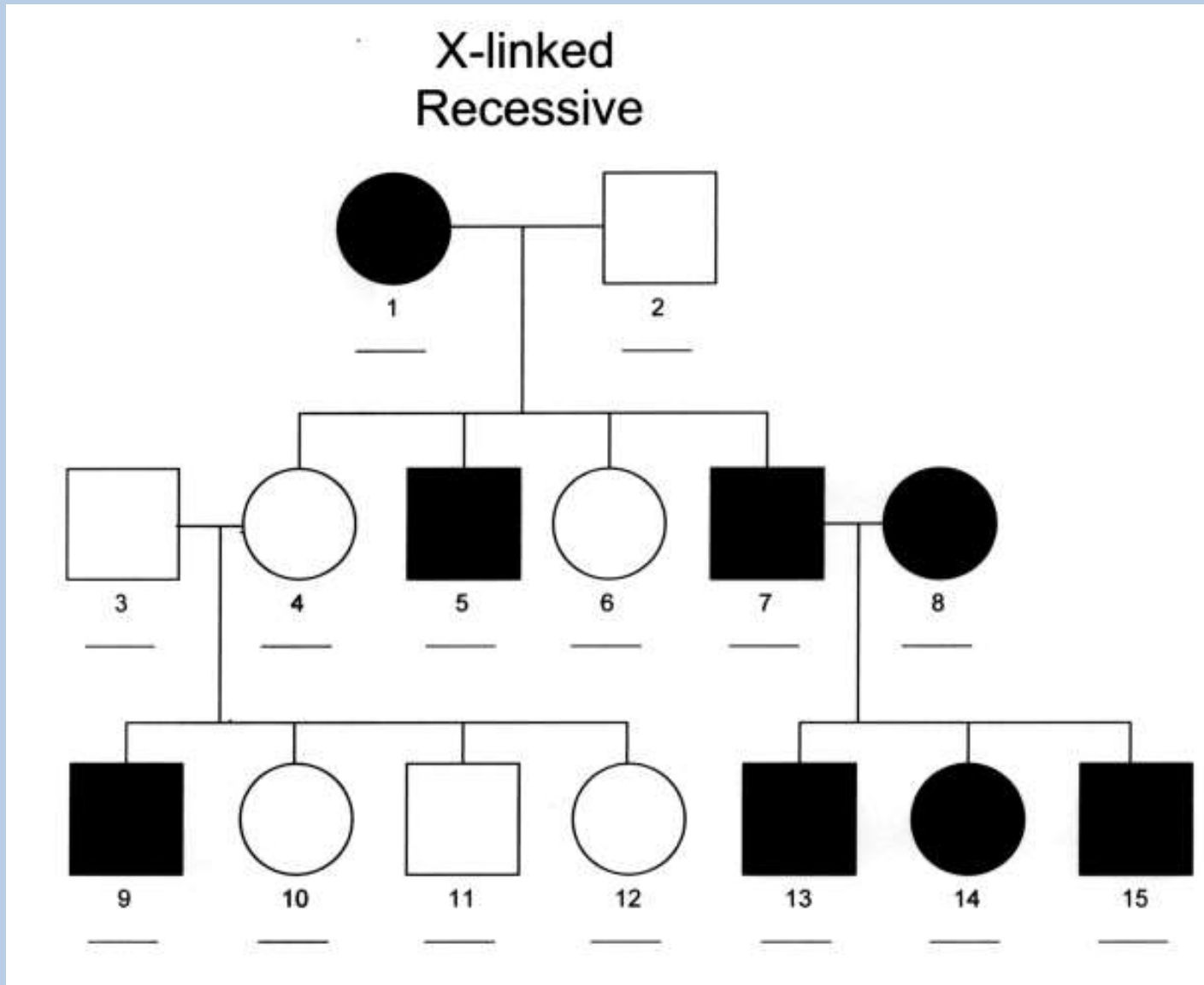
What is the pattern of inheritance?



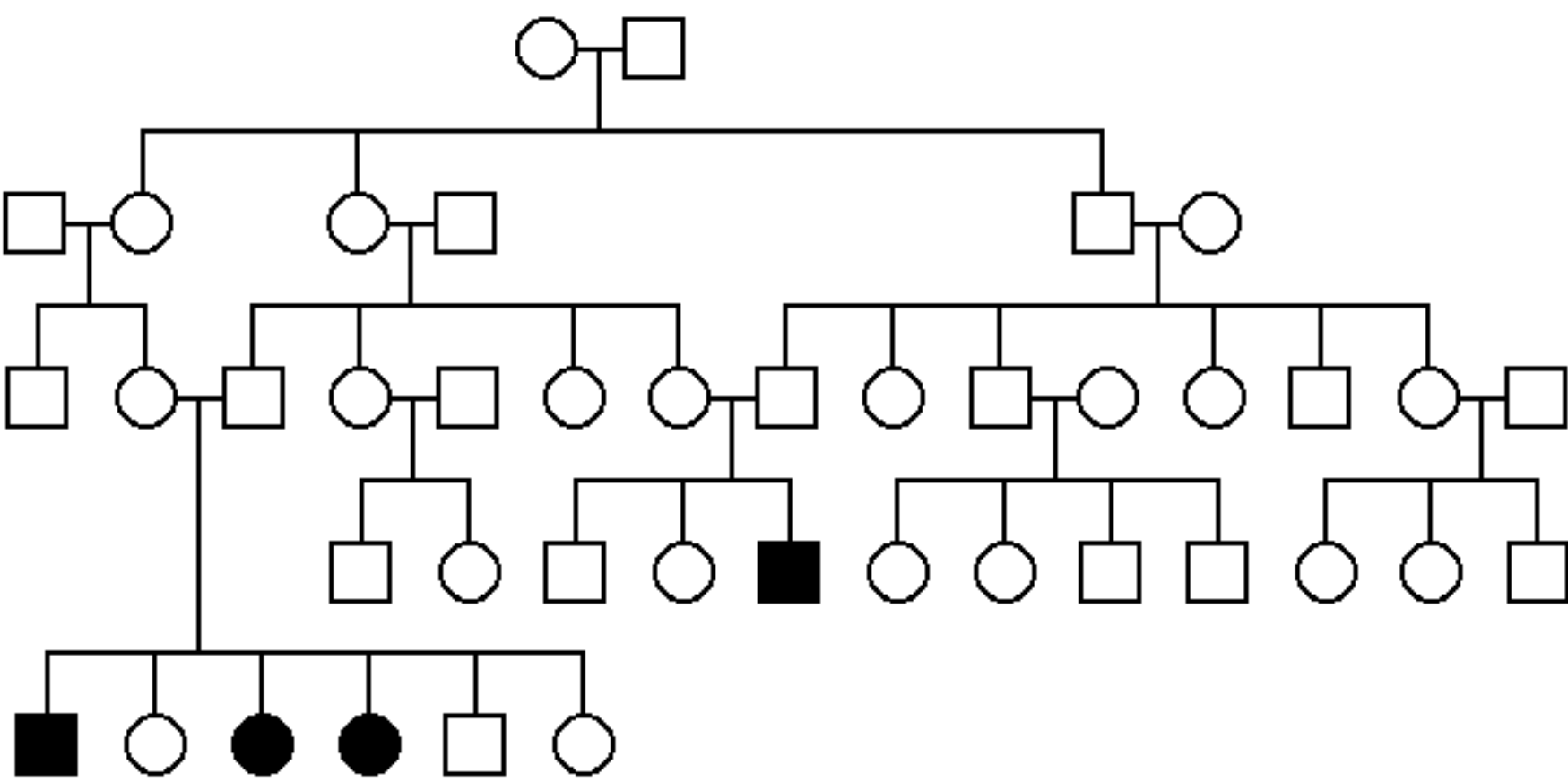
What is the pattern of inheritance?



What is the pattern of inheritance?

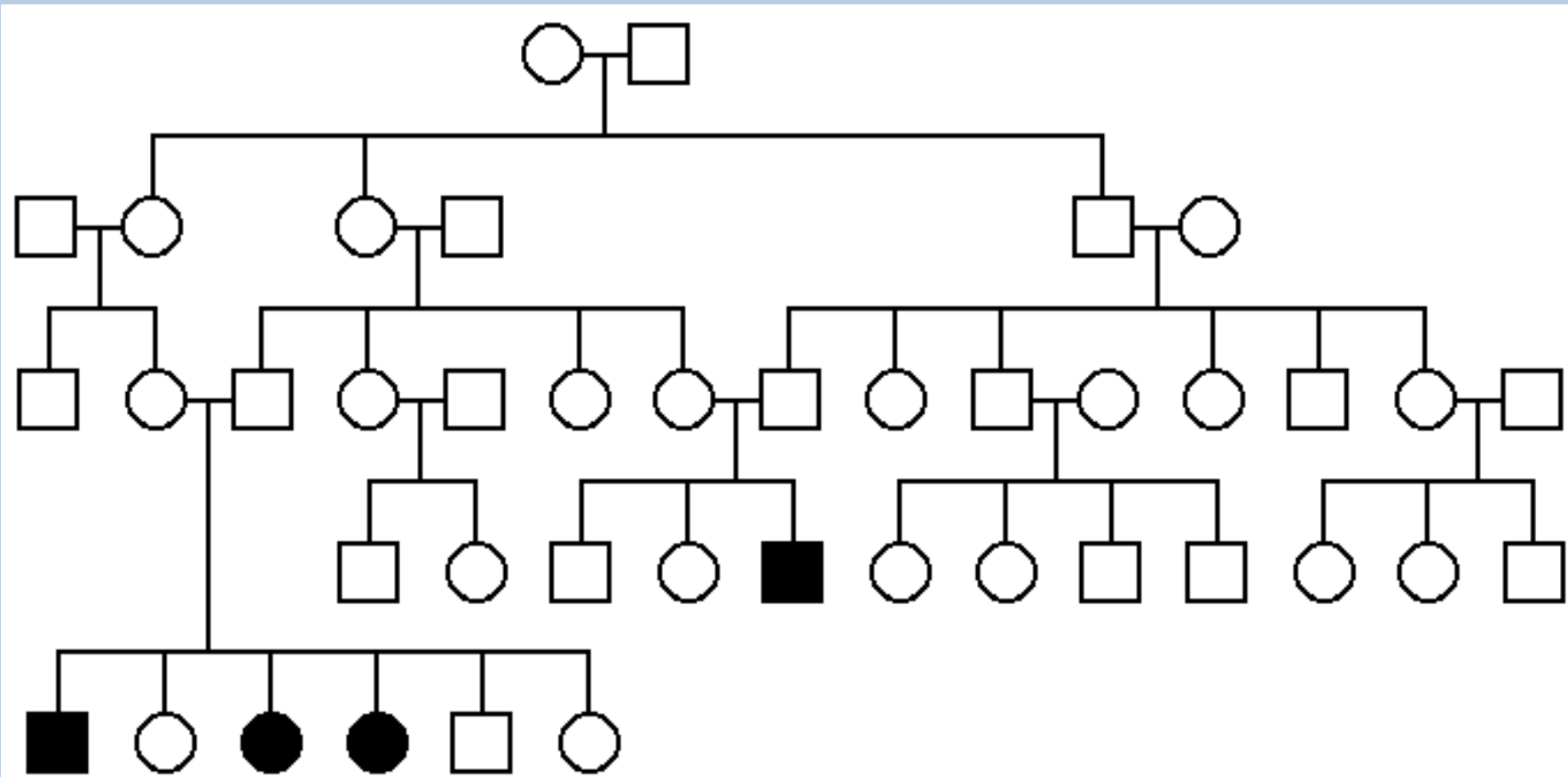


What is the pattern of inheritance?

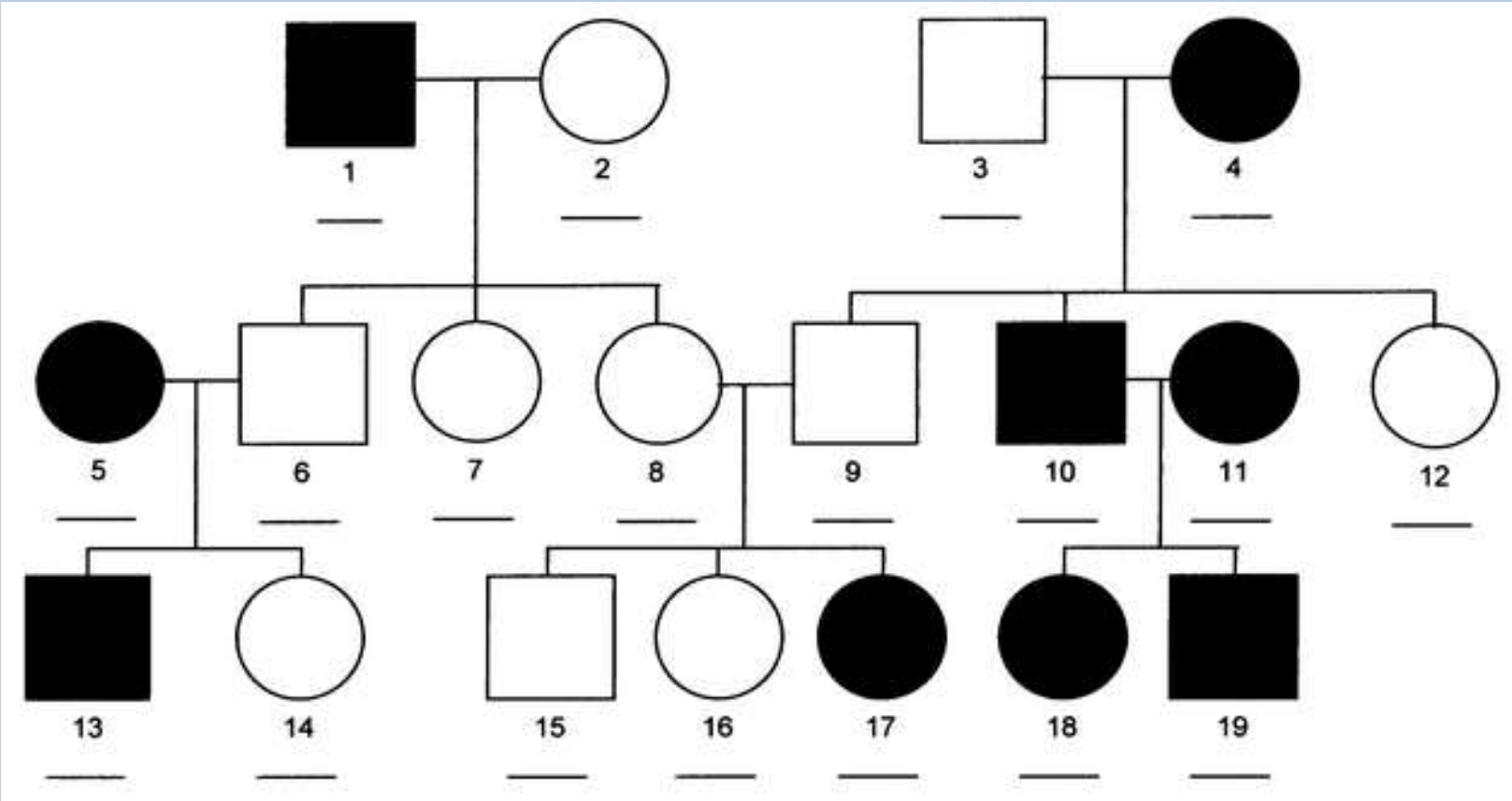


What is the pattern of inheritance?

Autosomal Recessive

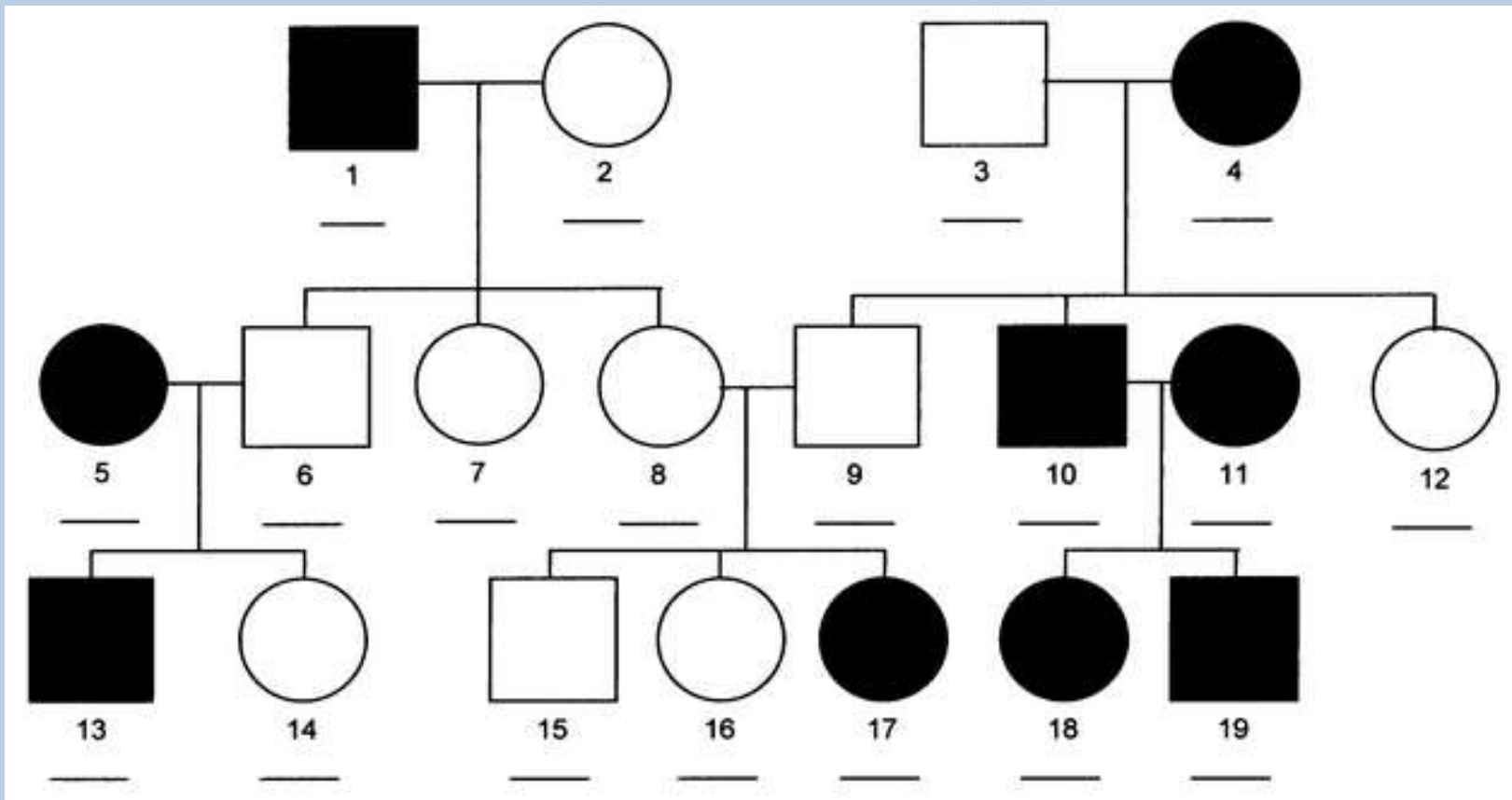


What is the pattern of inheritance?

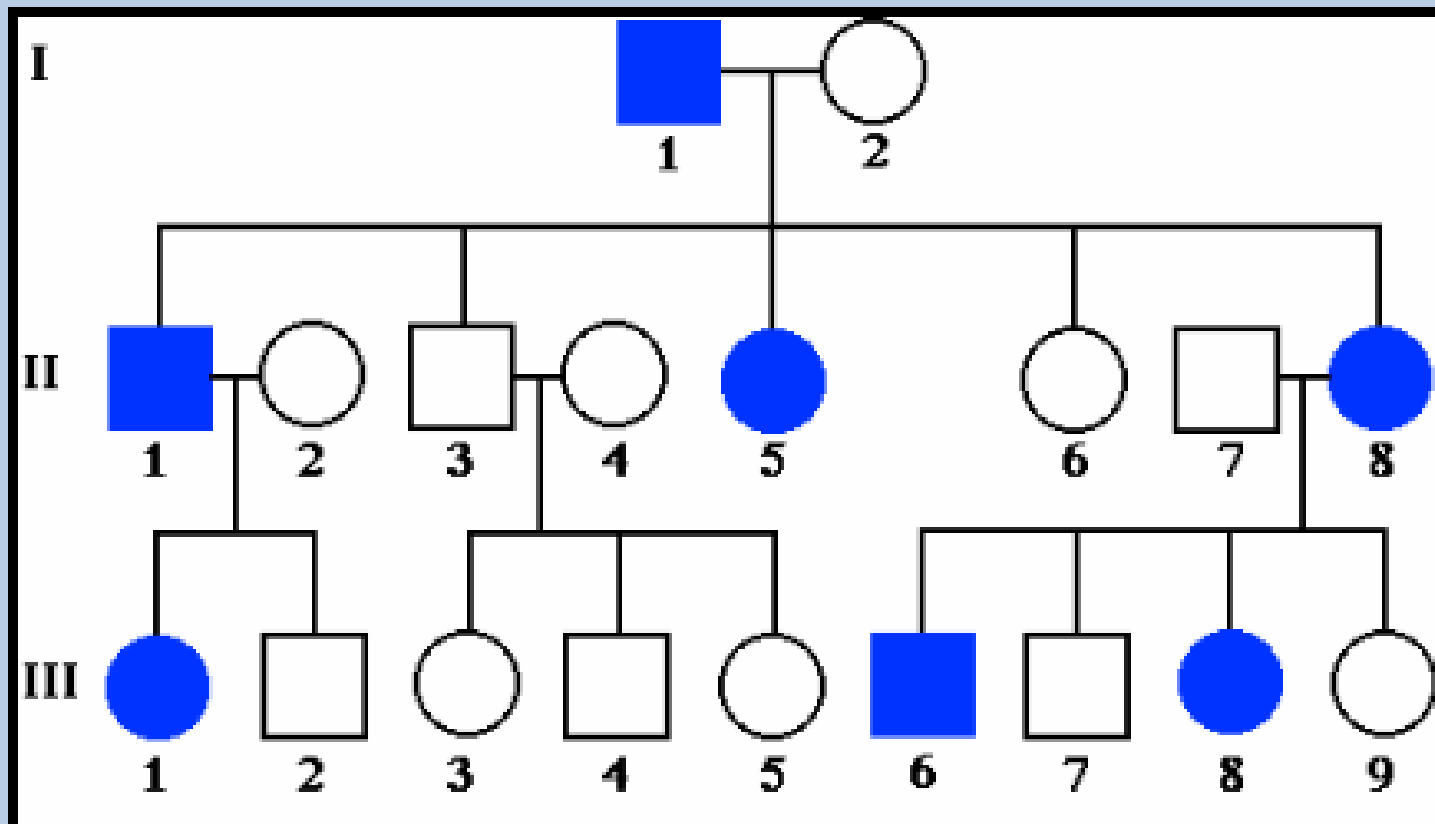


What is the pattern of inheritance?

Autosomal Recessive

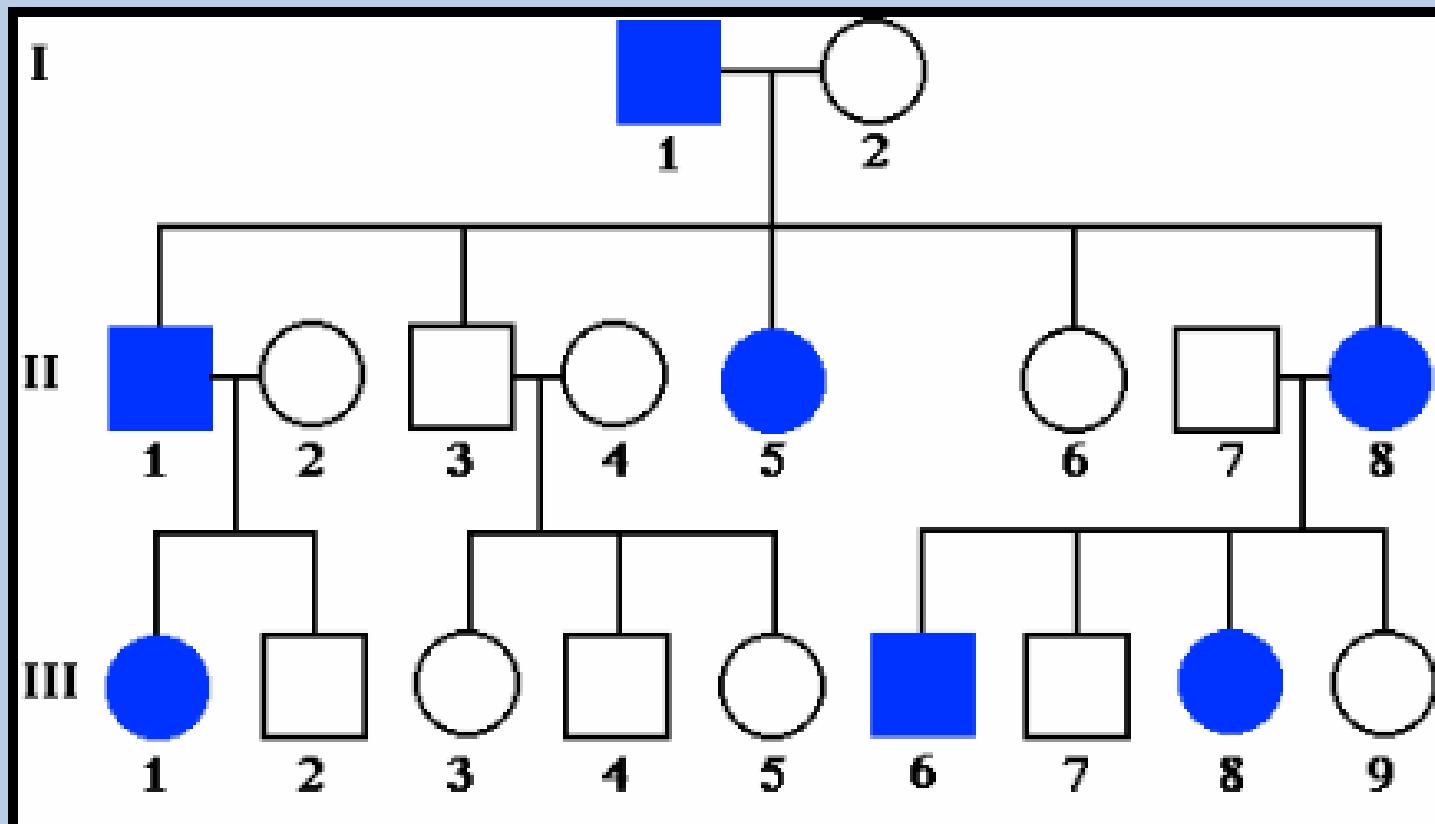


What is the pattern of inheritance?



What is the pattern of inheritance?

Autosomal Dominant



Deciphering Karyotypes

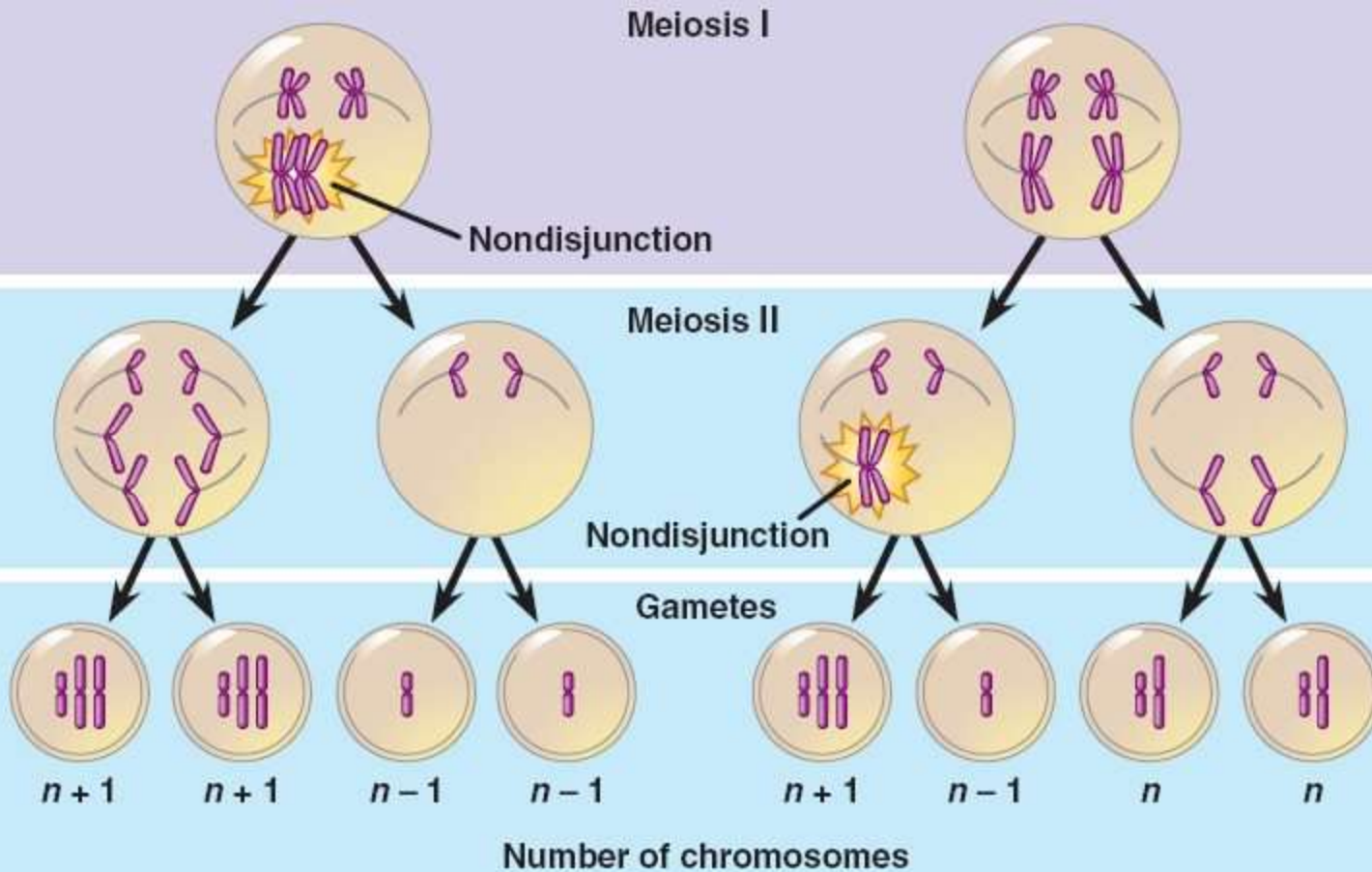
- Karyotypes are used to diagnose **NONDISJUNCTION**

Deciphering Karyotypes

- Karyotypes are used to diagnose **NONDISJUNCTION**



Nondisjunction



(a) Nondisjunction of homologous chromosomes in meiosis I

(b) Nondisjunction of sister chromatids in meiosis II

Trisomy 21 (Down Syndrome)



flattened nose and face, upward slanting eyes.

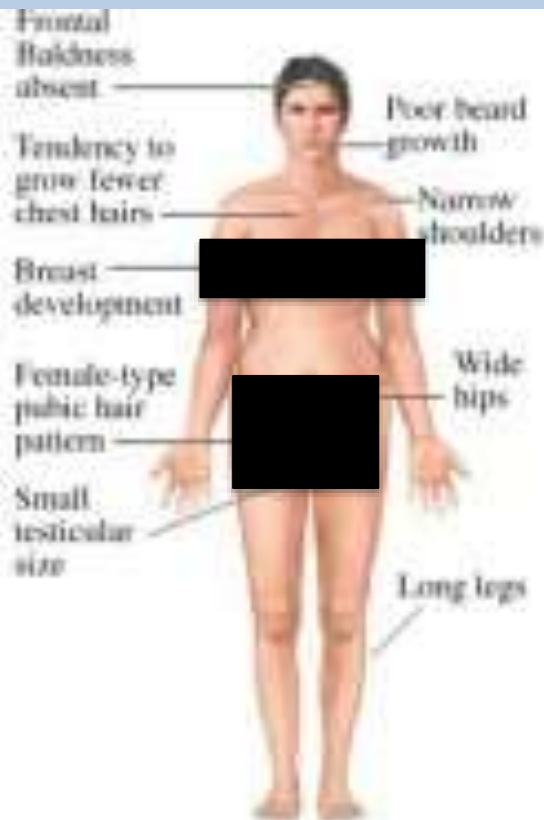
single palmar crease, short fifth finger that curves inward

widely separated first and second toes and increased skin creases

- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Upward slanting eyes
- Small ears
- Small mouth
- Wide, short hands with short fingers
- Separated joints between the bones of the skull
- Single crease in the palm of the hand
- White spots on the colored part of the eye



Klinefelter Syndrome

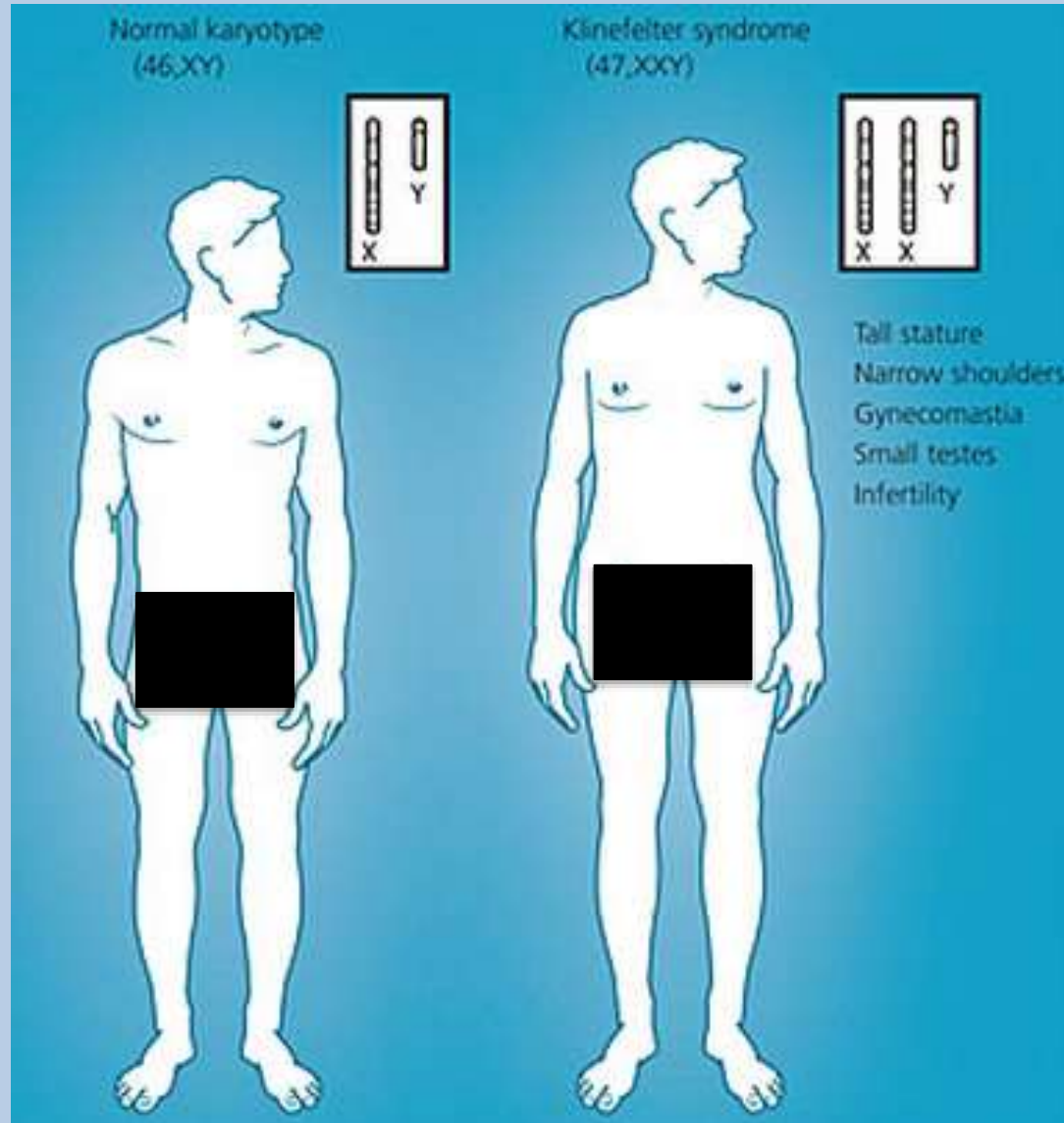


- **Lower IQ than sibs**
- **Tall stature**
- **Poor muscle tone**
- **Reduced secondary sexual characteristics**
- **Gynaecomastia (male breasts)**
- **Small testes/infertility**

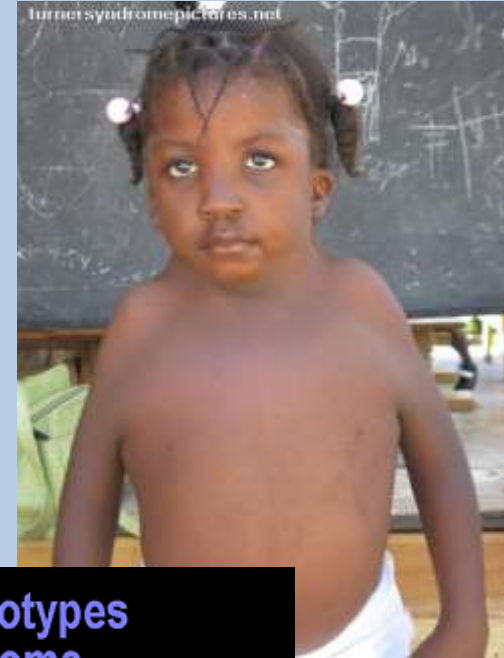
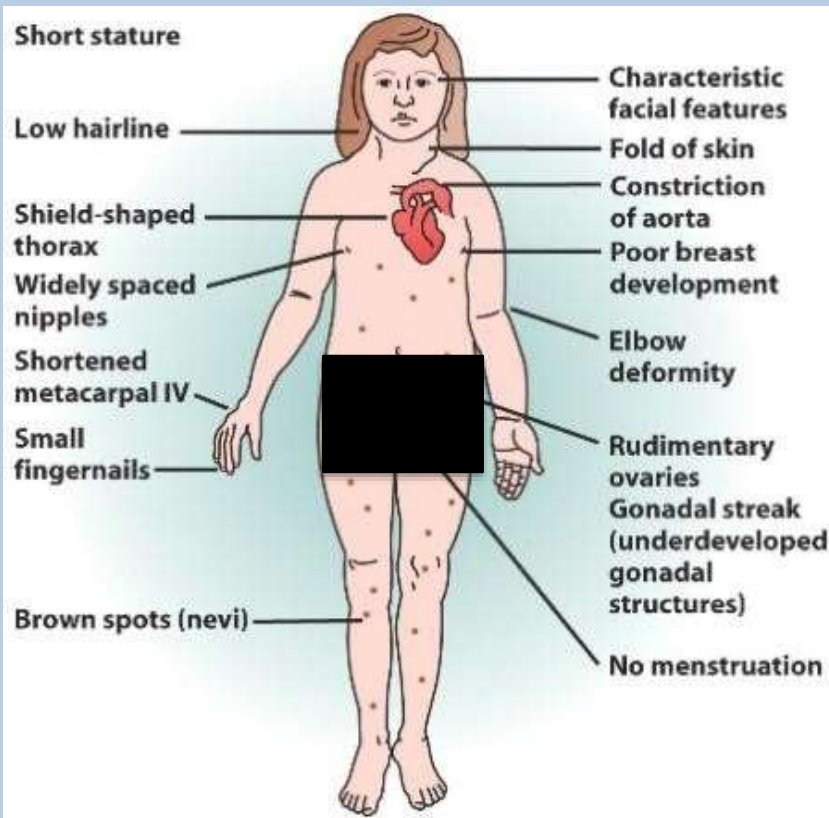


www.angelfire.com/wy/XXY/

Klinefelter Syndrome



Turner Syndrome



Incidence of Phenotypes in Turner Syndrome

♦ Short stature	100%
♦ Infertility	98%
♦ Primary gonadal failure	95%
♦ Osteoporosis	50%
♦ Cubitus valgus	45%
♦ Low posterior hairline	40%
♦ Carbohydrate intolerance	30-40%
♦ High blood pressure	25-40%
♦ Short metacarpals	35%
♦ High arched palate	35%
♦ Structural abnormalities in kidney	35%
♦ Hypothyroidism (Hashimoto thyroiditis)	35%

Deciphering Karyotypes

- Autosomal Nondisjunction in humans

#	monosomy	trisomy
1	1p36 deletion syndrome 1q21.1 deletion syndrome	Trisomy 1
2	2q37 deletion syndrome	Trisomy 2
3		Trisomy 3
4	Wolf-Hirschhorn syndrome	Trisomy 4
5	Cri du chat 5q deletion syndrome	Trisomy 5
6		Trisomy 6
7	Williams syndrome	Trisomy 7
8	Monosomy 8p Monosomy 8q	Trisomy 8
9	Alfi's syndrome Kleefstra syndrome	Trisomy 9
10	Monosomy 10p Monosomy 10q	Trisomy 10
11	Jacobsen syndrome	Trisomy 11

12		Trisomy 12
13		Patau syndrome
14		Trisomy 14
15	Angelman syndrome Prader-Willi syndrome	Trisomy 15
16		Trisomy 16
17	Miller-Dieker syndrome Smith-Magenis syndrome	Trisomy 17
18	Distal 18q- Proximal 18q-	Edwards syndrome
19		Trisomy 19
20		Trisomy 20
21		Down syndrome
22	DiGeorge syndrome Phelan-McDermid syndrome 22q11.2 distal deletion syndrome	Cat eye syndrome Trisomy 22

Deciphering Karyotypes

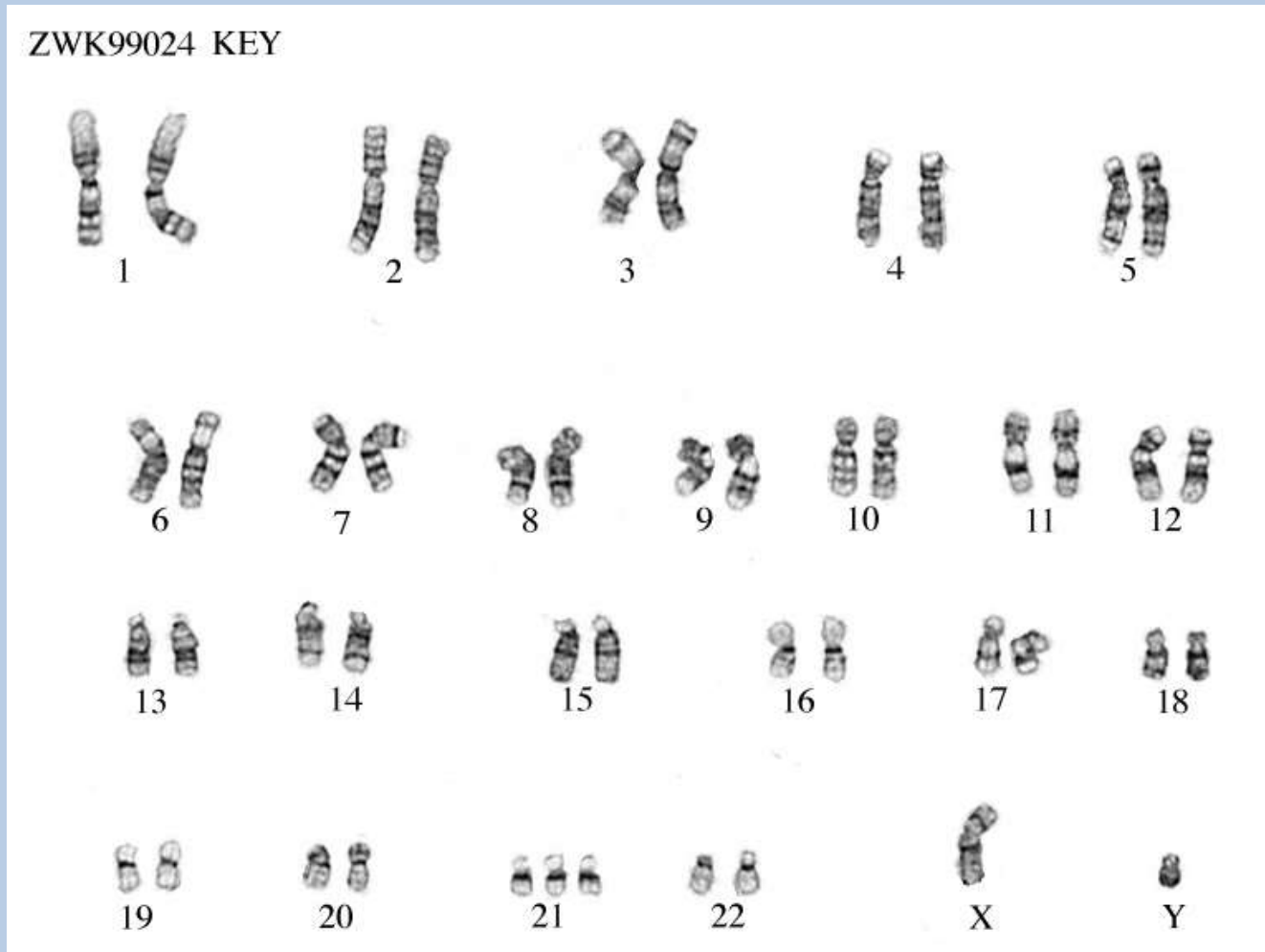
- Sex-linked nondisjunction in humans

key	
color	significance
	lethal
	normal male phenotype
	Klinefelter syndrome (abnormal male)
	polysomy X and/or Y, (abnormal male)
	normal female phenotype
	Turner's syndrome (abnormal female)
	tetrasomy X, pentasomy X, (abnormal female)

Non-autosomal						
	0	X	XX	XXX	XXXX	XXXXX
0	00	X0	XX	XXX	XXXX	XXXXX
Y	Y0	XY	XXY	XXXY	XXXXY	XXXXXY
YY	YY	XYY	XXYY	XXXYY	XXXXYY	XXXXXY
YYY	YYY	XYYY	XXYYY	XXXYYY	XXXXYYY	XXXXXY
YYYY	YYYY	XYYYY	XXYYYY	XXXYYYY	XXXXYYYY	XXXXXY
YYYYY	YYYYY	XYYYYY	XXYYYYY	XXXYYYYY	XXXXYYYYY	XXXXXY

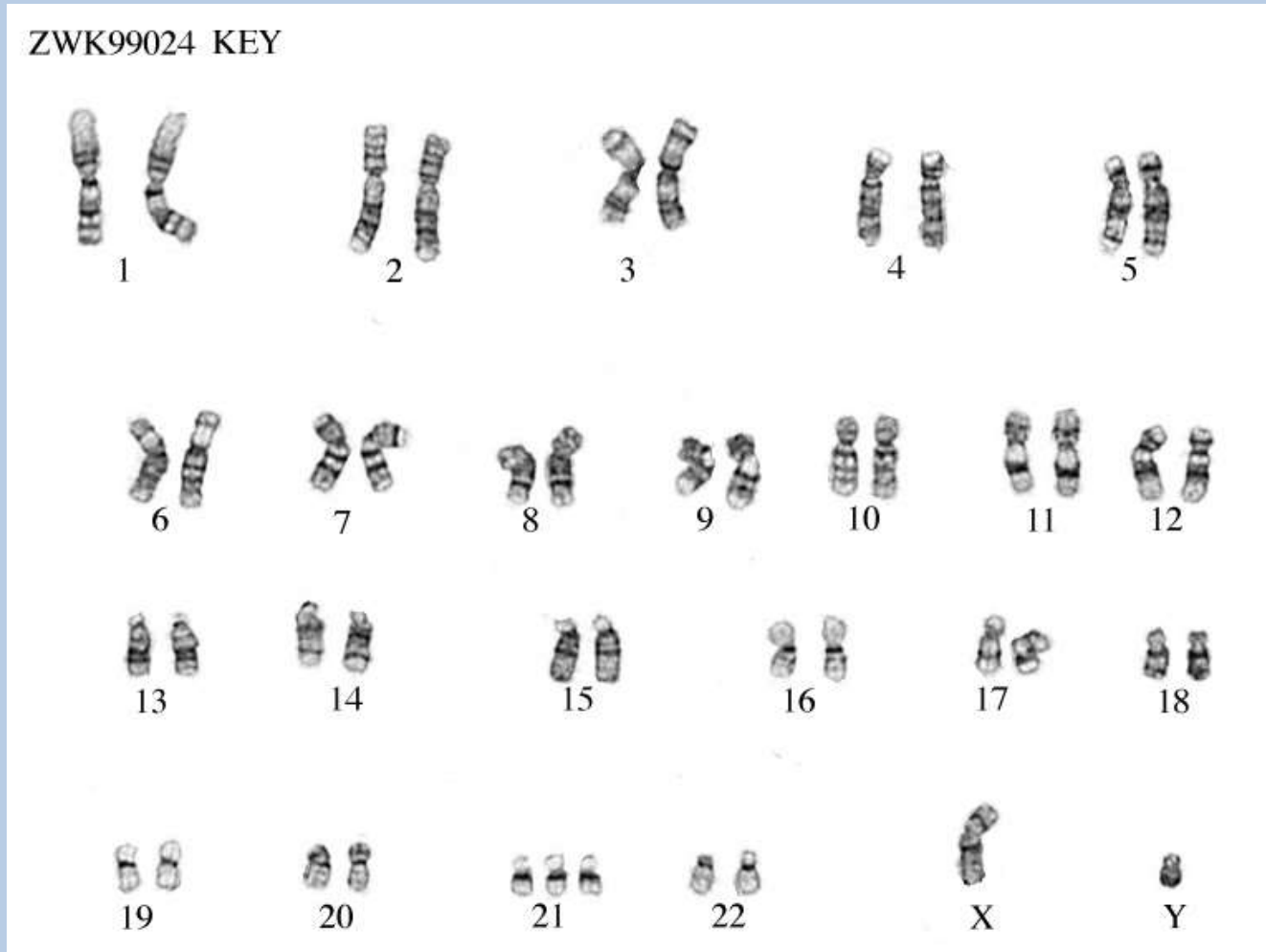
Deciphering Karyotypes

- What is wrong here?



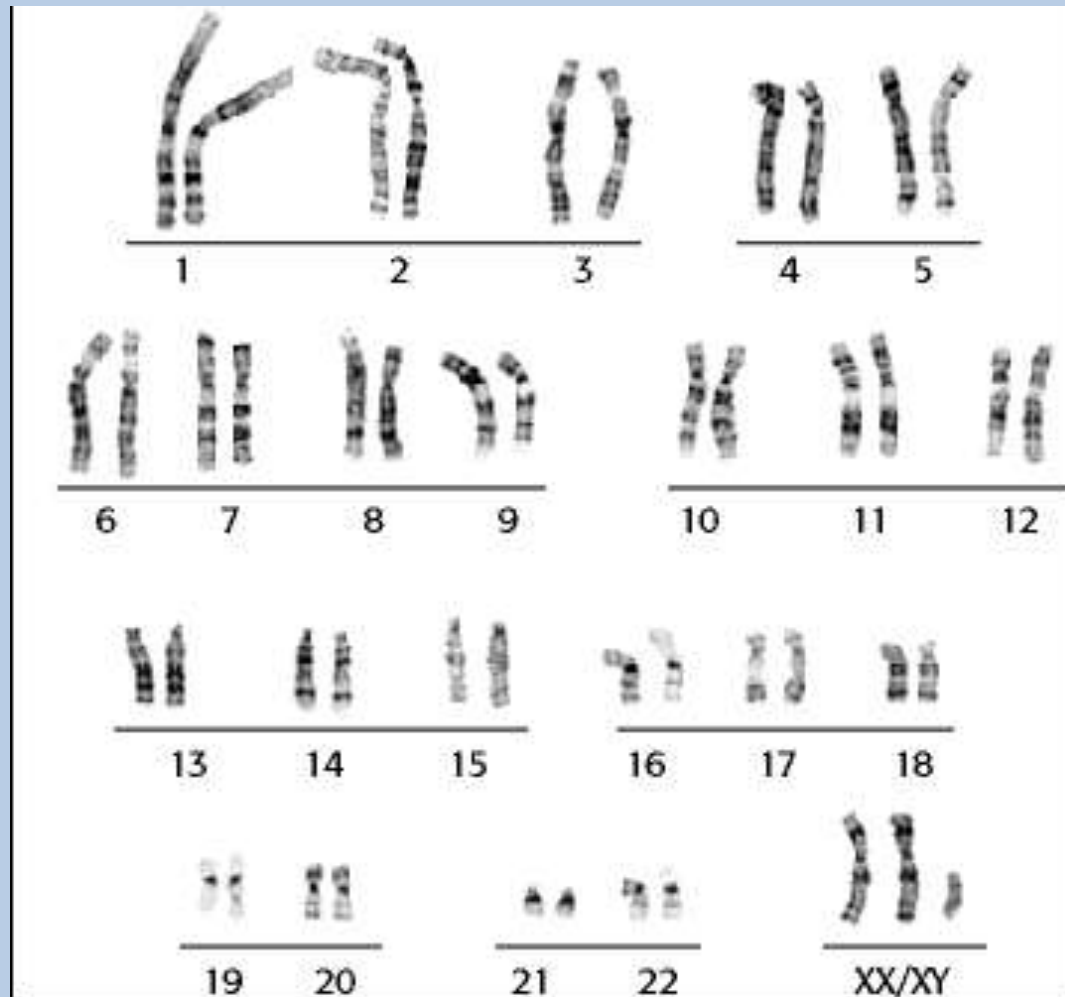
Deciphering Karyotypes

- What is wrong here?



Deciphering Karyotypes

- What is wrong here?



Deciphering Karyotypes

- What is wrong here?



Deciphering Karyotypes

- **To understand how karyotypes are used you and a partner will be diagnosing people using their karyotypes**

Deciphering Karyotypes

- PLEASE DO NOT LOSE ANY CHROMOSOMES, BE VERY CAREFUL
- If you do, tell Mrs. Moberly *IMMEDIATELY*

Deciphering Karyotypes

- **When dismissed:**
 - **Go to a lab station**
 - **Complete the activity**
 - **Put chromosomes away**
 - **Move to a new station**
- **DO NOT LOSE
CHROMOSOMES**