

1. What is aneuploidy?

2. If a *female* is not completely colorblind, but sees fewer colors than a typical person, what must her genotype be?

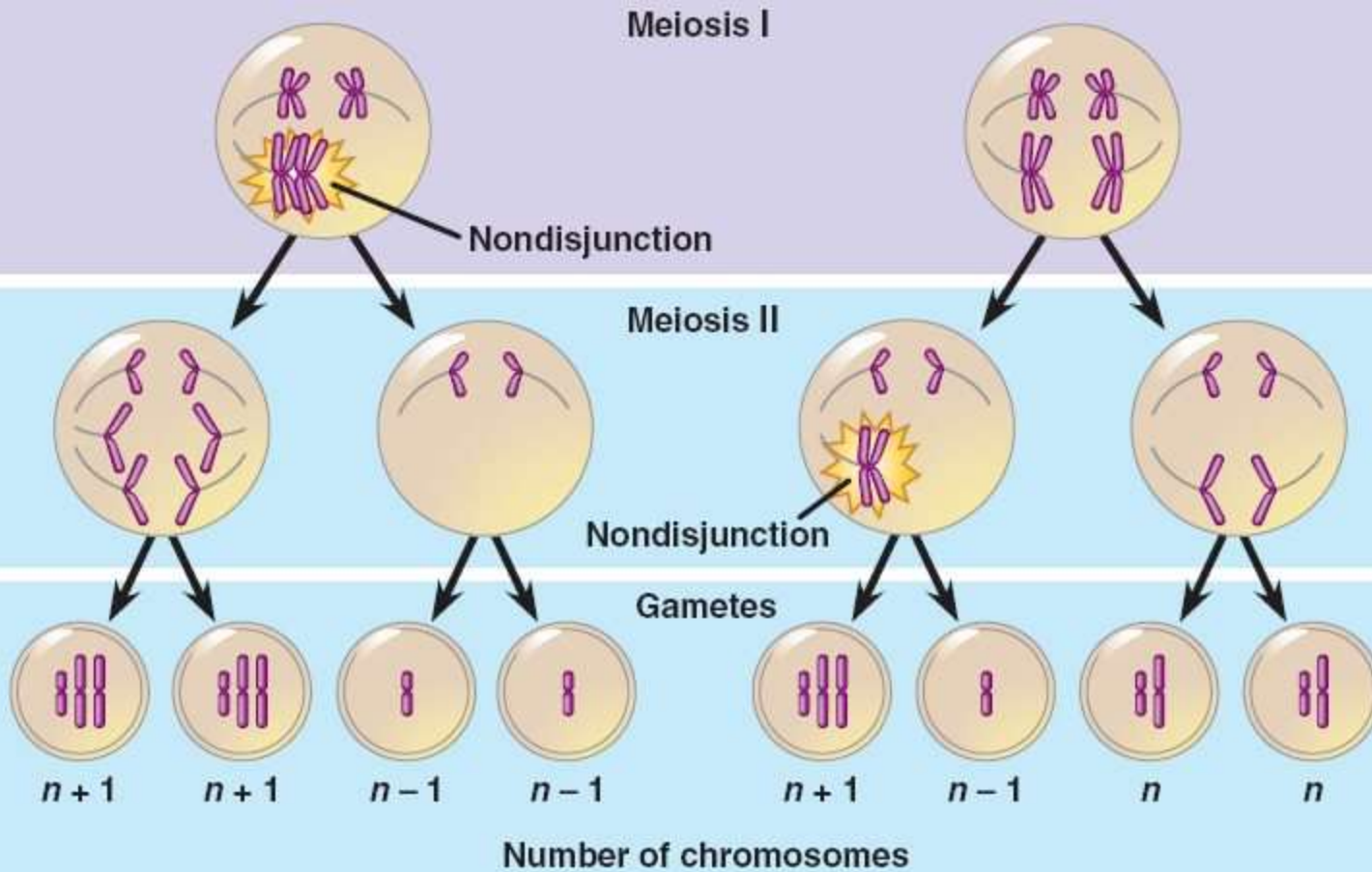
Hemophilia A is a sex-linked recessive trait:

3. What is the genotype of a female carrier?

4. If her husband is unaffected, what is the probability that their sons will be affected?



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.

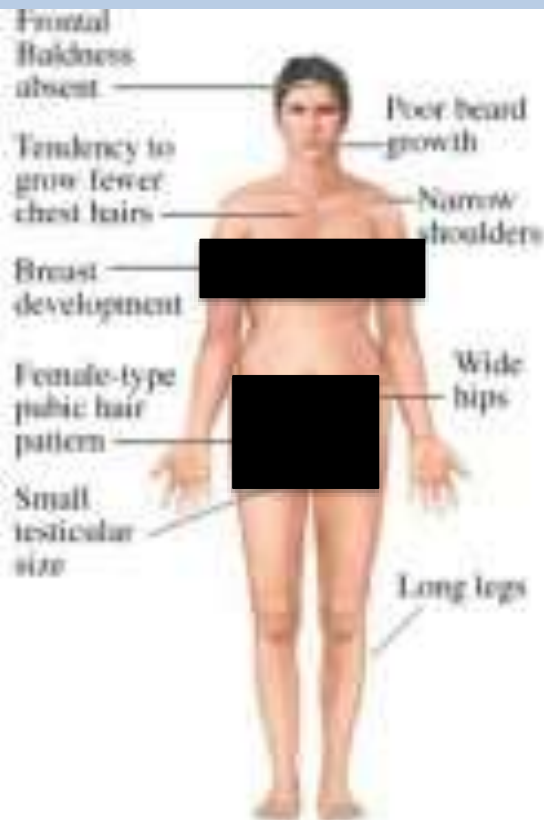
single palmar crease, short fifth finger that curves inward

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

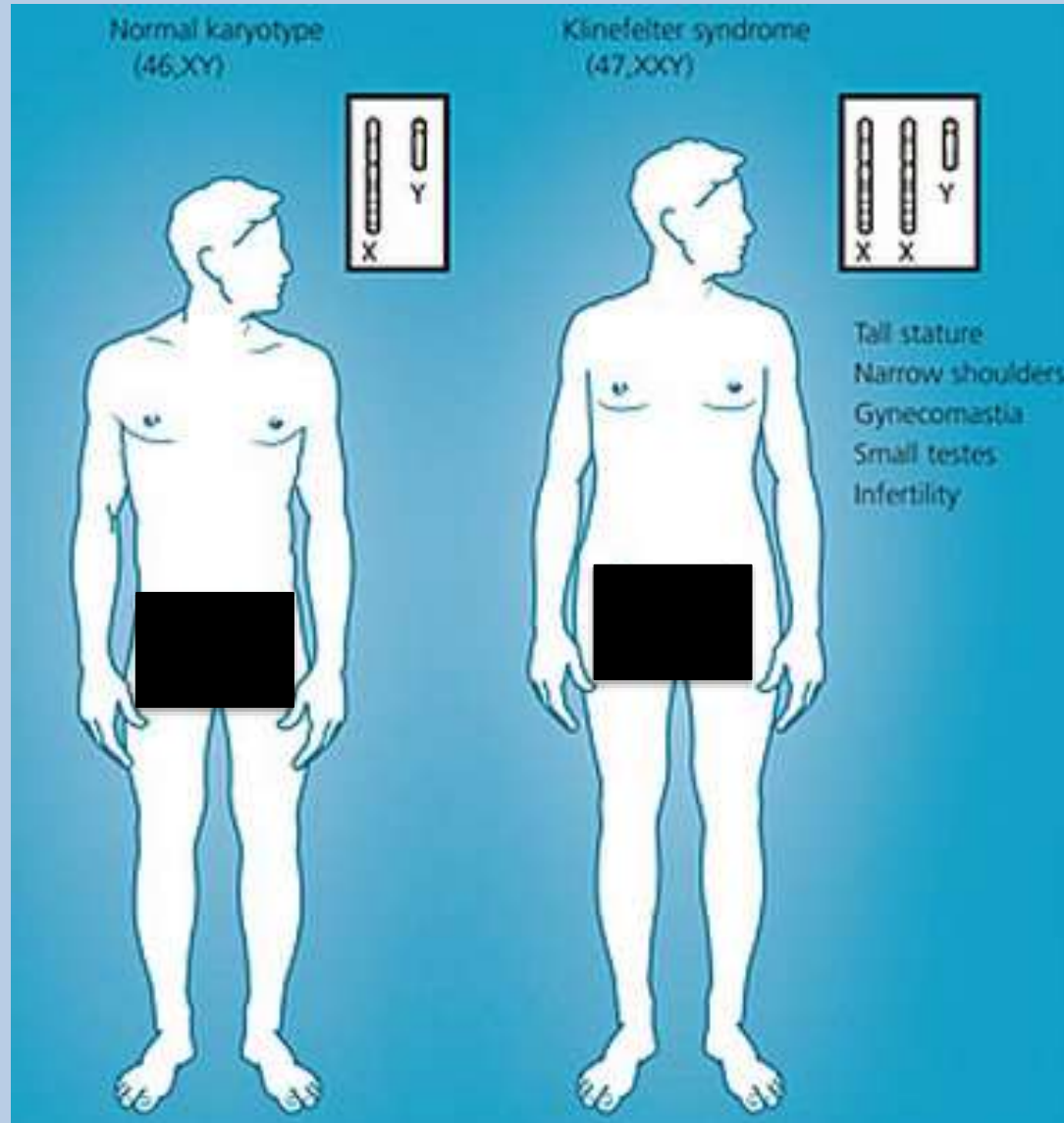


- **Lower IQ than sibs**
- **Tall stature**
- **Poor muscle tone**
- **Reduced secondary sexual characteristics**
- **Gynaecomastia (male breasts)**
- **Small testes/infertility**

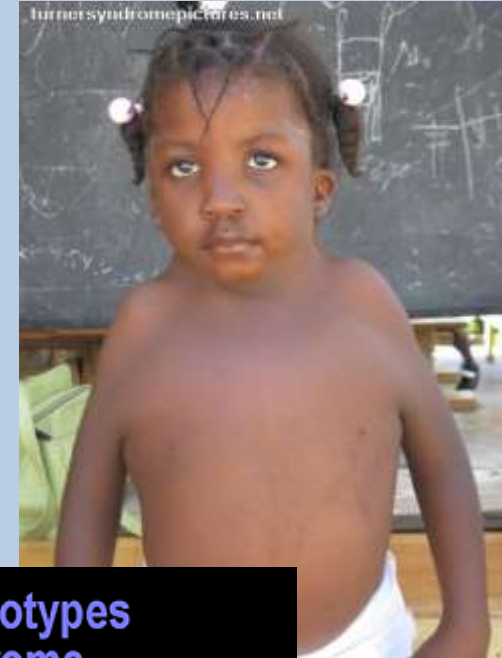
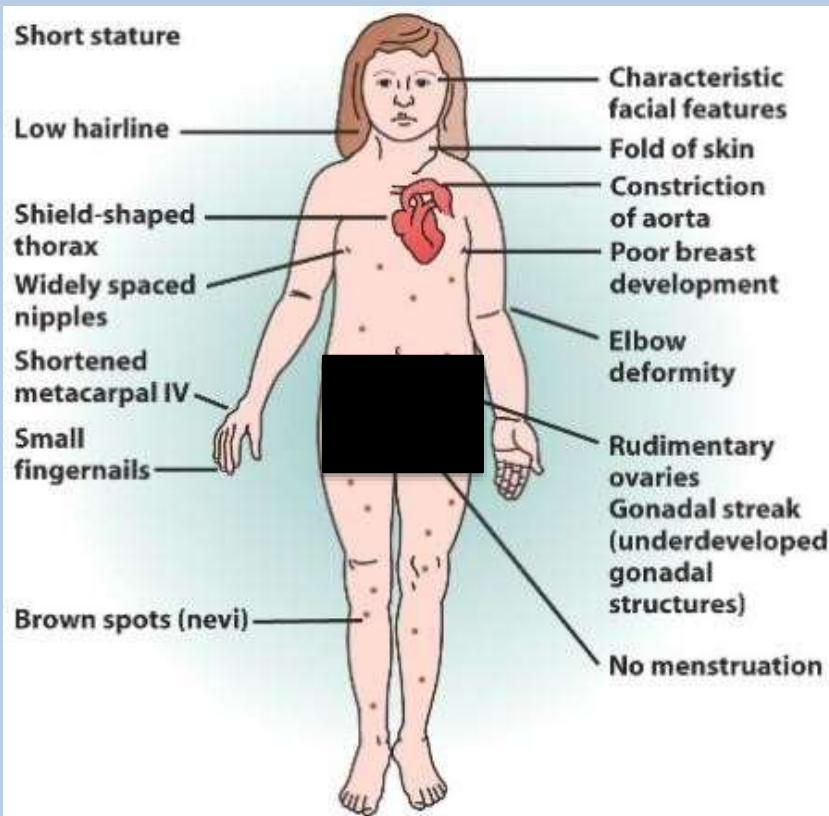


www.angelfire.com/wy/XXY/

Klinefelter Syndrome



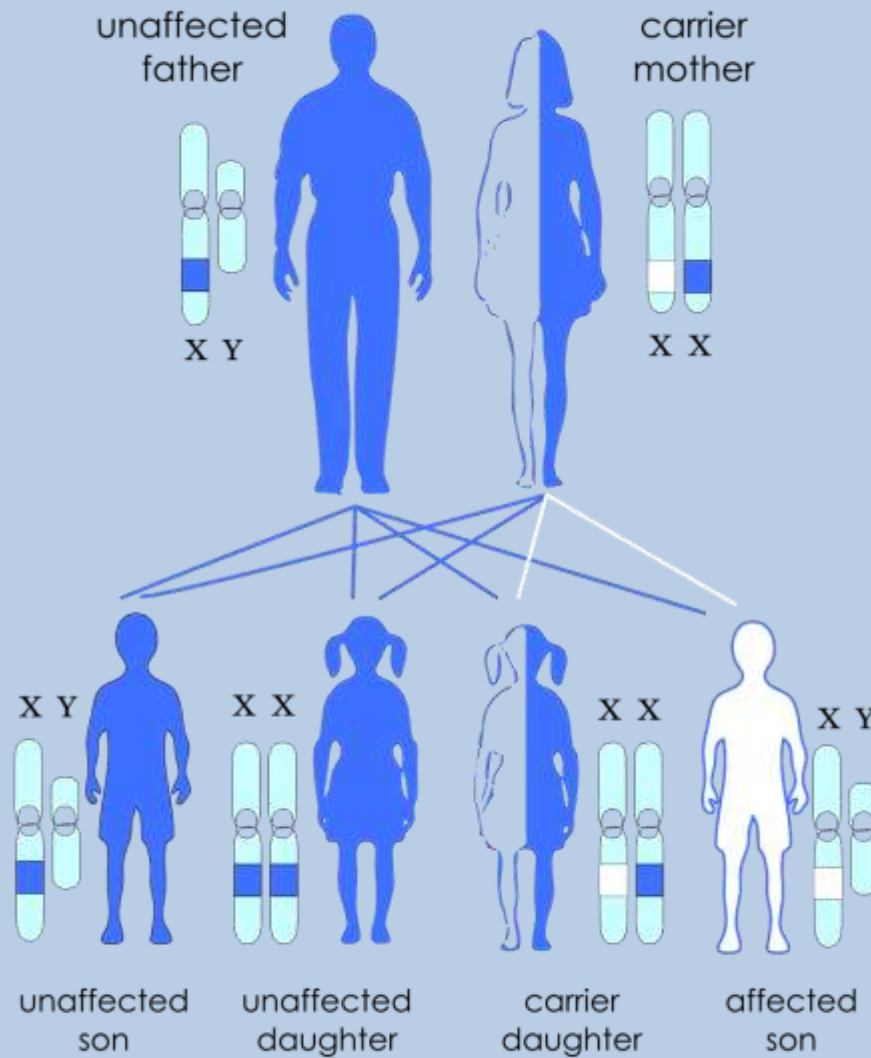
Turner Syndrome




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%

X-linked recessive inheritance



 *unaffected*

 *carrier*

 *affected*

Activity

- **At your groups there are two beakers**
 - **Male and Female**
- **These are your parents!**

Activity

- **When you are told to do so you will draw 1 card from the male and one from the female**
- **We will discover what genes when everyone has drawn**

Activity

- **Based on your genotype, are you affected by any of these genetic disorders?**

Activity

- **Genotypes:**
 - **D/d= Duchenne muscular dystrophy, sex-linked recessive**
 - **H/h= Hemophilia A, sex-linked recessive**
 - **R/r= Rett syndrome, sex-linked dominant**
 - **V/v= Vitamin D resistant Rickets, sex-linked dominant**

Pedigree Activity

- Turn to **page 62** in your notebook

Pedigree Activity

- Use the beakers to pull out your genotype
- Write your genotype on the paper

**DO NOT DISCUSS YOUR
GENOTYPE**

Activity

- Under your genotype write your phenotype (what disorder(s) do you have?)

**DO NOT DISCUSS YOUR
PHENOTYPE**

Activity

- **Genotypes:**
 - **D/d= Duchenne muscular dystrophy, sex-linked recessive**
 - **H/h= Hemophilia A, sex-linked recessive**
 - **R/r= Rett syndrome, sex-linked dominant**
 - **V/v= Vitamin D resistant Rickets, sex-linked dominant**

Activity

- **FRONT ROW:** For now focus on D/d =
Duchenne muscular dystrophy,
sex-linked recessive
- **BACK ROW:** For now focus on H/h =
Hemophilia A, sex-linked recessive

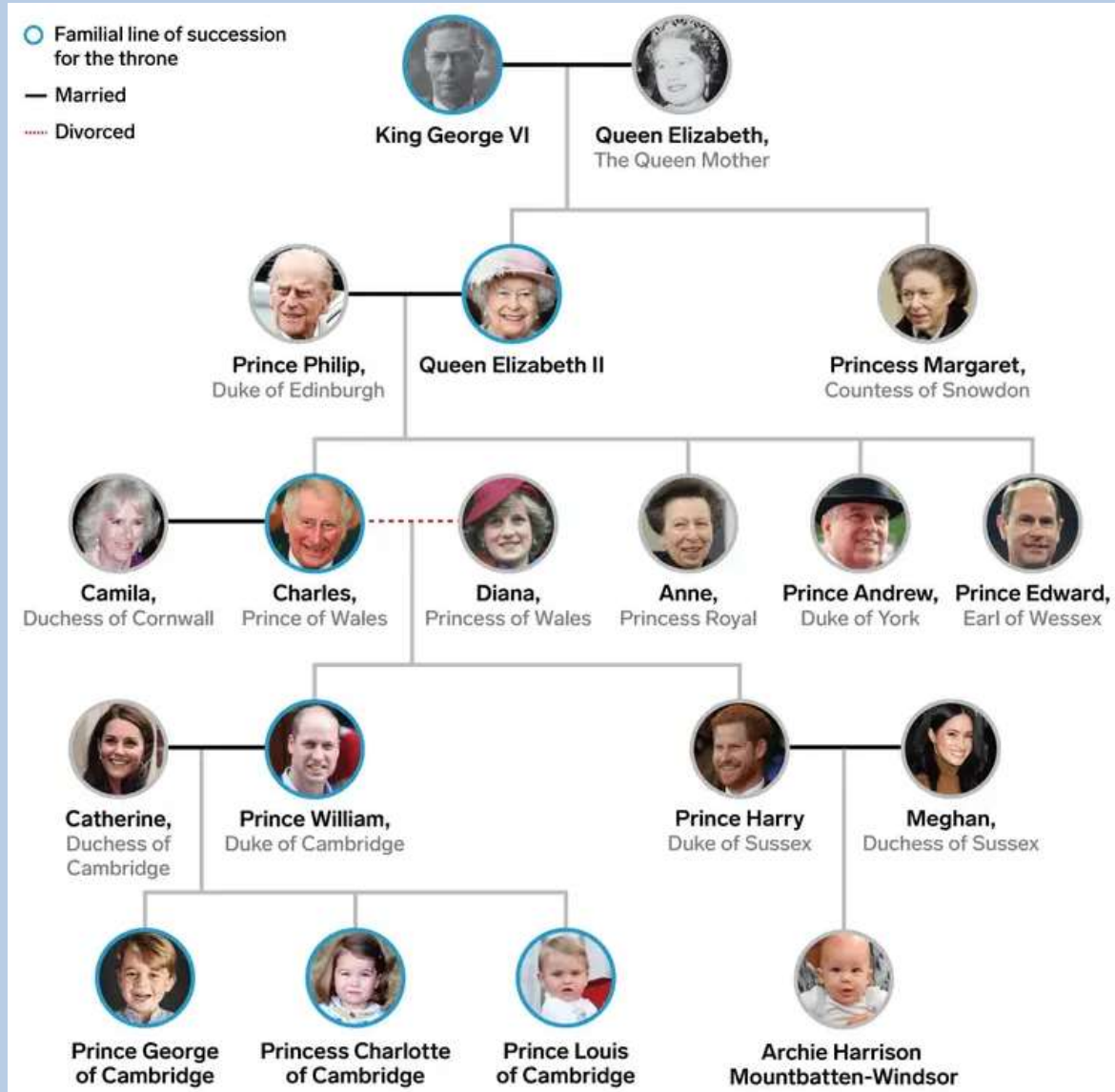
Pedigrees

DUQUESA DEL CIELO dun. M, PERUVIAN PASO, 1998

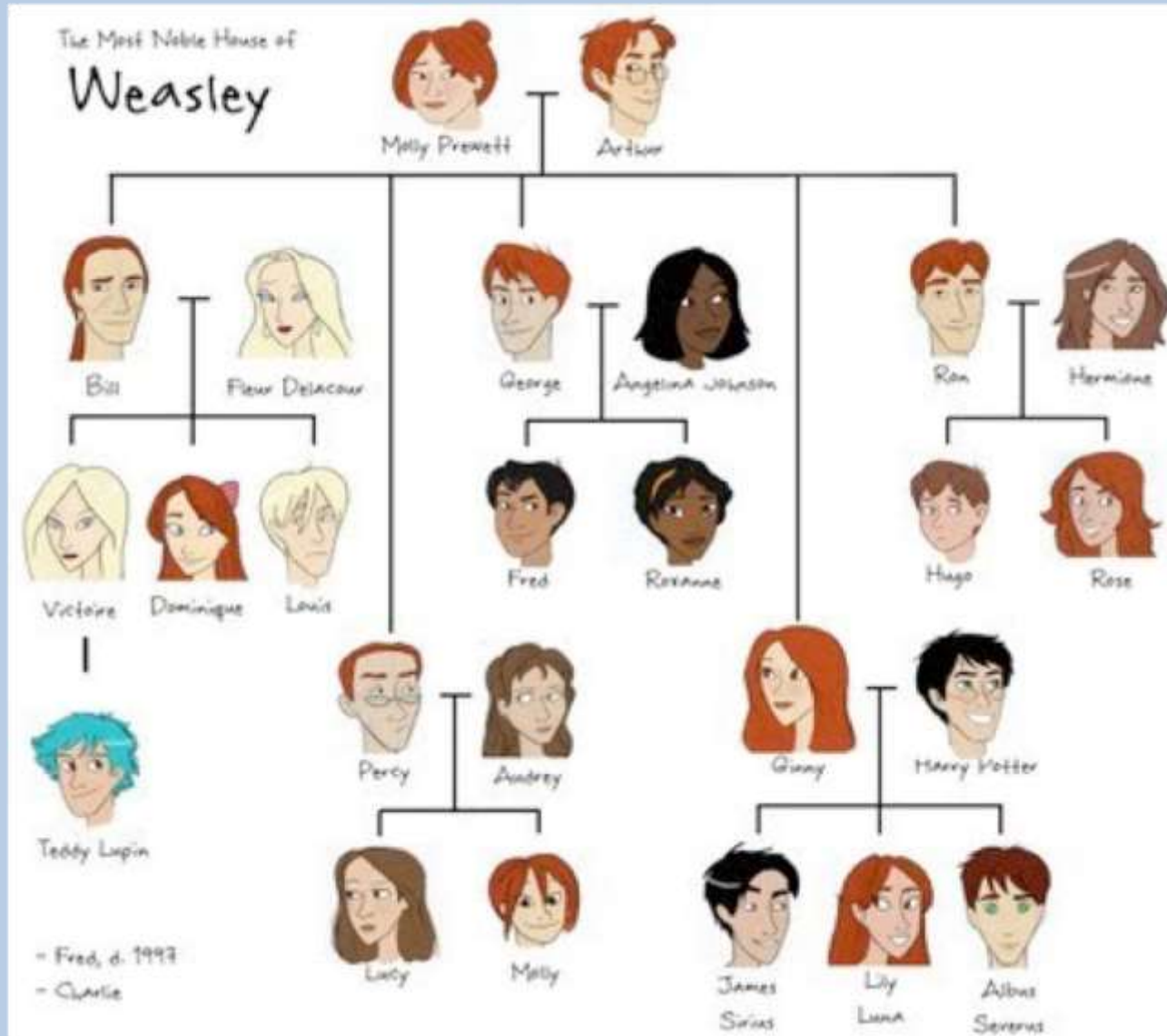
DUQUESA DEL CIELO* dun 1998 PERUVIAN PASO	CIELITO LINDO* dun 1977 PERUVIAN PASO	VEA BUCEFALO* ch 1970 PERUVIAN PASO	LAUREL* ch 1964 PERUVIAN PASO	AEV CARAMELO* ch 1958	SOL DE ORO V* ch 1945	
				CENTELLA* ch 1946	SULTANA* ch	
				SENORITA ch 1958 PERUVIAN PASO	SENORON* 1947	ELEGANTE
					MARUJA	RAIMUNDA
		HNS LIRA* b 1972 PERUVIAN PASO	HNS PALADIN 1962 PERUVIAN PASO	SOL DE ORO V* ch 1945	POTRO DE CALAPALLA	
				HNS PASCUALA 1954	YEGUA DE FRANCISCO DEGREGORI	
				PCD SENORON 1947	DARTMAN	
				HNS TORMENTA II 1951	HNS TORMENTA II 1951	
					ELEGANTE	
					RAYMUNDA	
	HE POR FIN 1980 PERUVIAN PASO	DON FREDERICO* blk 1970 PERUVIAN PASO	MANDINGA* blk 1959 PERUVIAN PASO	PIURANO ~1949		
				ZAPATILLA PUCALA		
		SR VENTISCA* b 1969 PERUVIAN PASO	DONA MELINDRES PERUVIAN PASO	SOL DE ORO V* ch 1945	POTRO DE CALAPALLA	
				PRIMOROSA DE CEPEDA 1958	YEGUA DE FRANCISCO DEGREGORI	
				FRONTINO* ch 1953	LUNANCO	
				FLORIDA V	DIAS	
				VENTARRON* b 1959 PERUVIAN PASO	FRONTINO* ch 1953	EMPERATRIZ* 1947
					FLORIDA V	FIGARO
	REINA DE MUSANTE ch 1957 PERUVIAN PASO	DICTADO* 1950	PRIMAVERA PARODI			
		EMPERATRIZ* 1947	NORBO* Dun			
			PERCAL			
			NO			
			GOYITA* 1943			

* - Photo Available

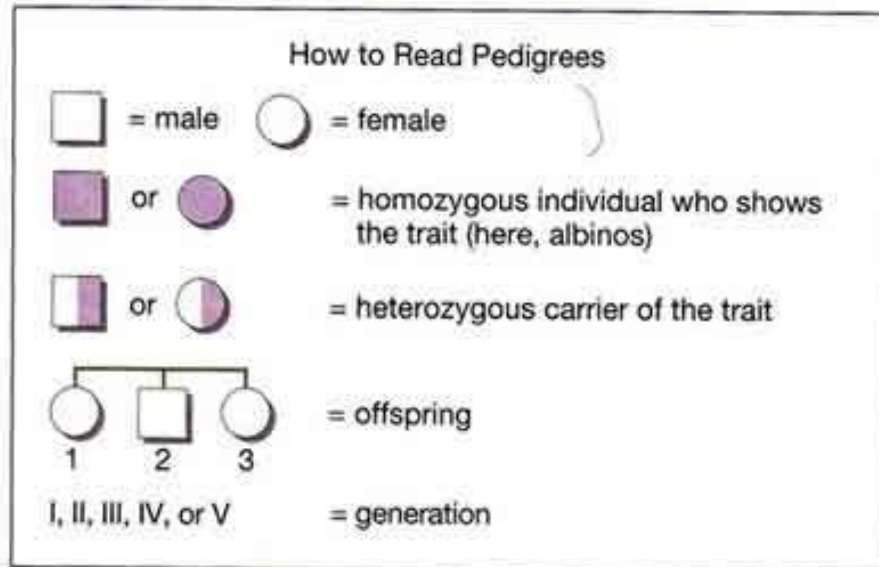
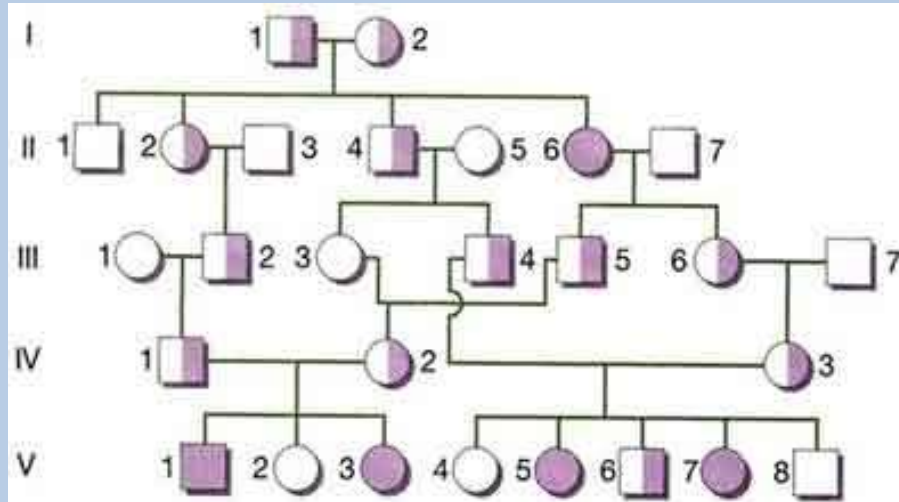
Pedigrees



Pedigrees



Pedigrees

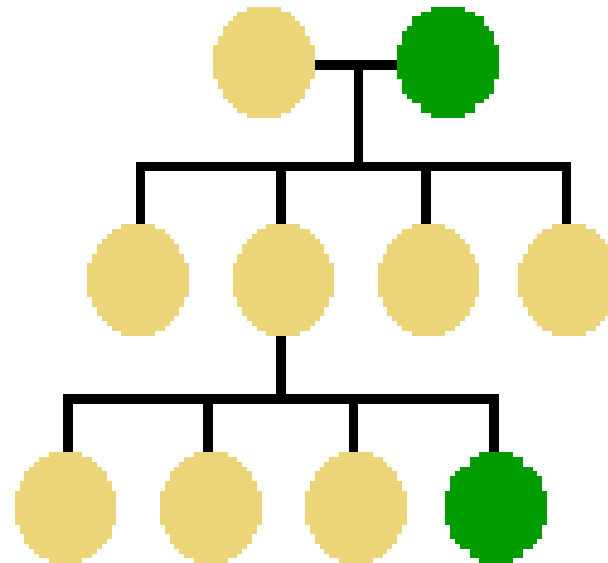


Pedigrees

 = yellow pea seed

 = green pea seed

3:1 ratio



parent generation

f1 generation

f2 generation

Activity

- **On your paper, under the genotype, draw the shape that represents you based on your new genotype**

Activity

- You will have **ONE MINUTE TO COMPLETE THIS STEP**

**DO NOT DISCUSS WHETHER
YOU ARE AFFECTED OR NOT**

Activity

- Find a mate, **DO NOT DISCUSS WHETHER YOU ARE AFFECTED OR NOT**
- **FRONT ROW STICK WITH FRONT ROW, BACK ROW WITH BACK ROW**
- Find a seat with your papers

Activity

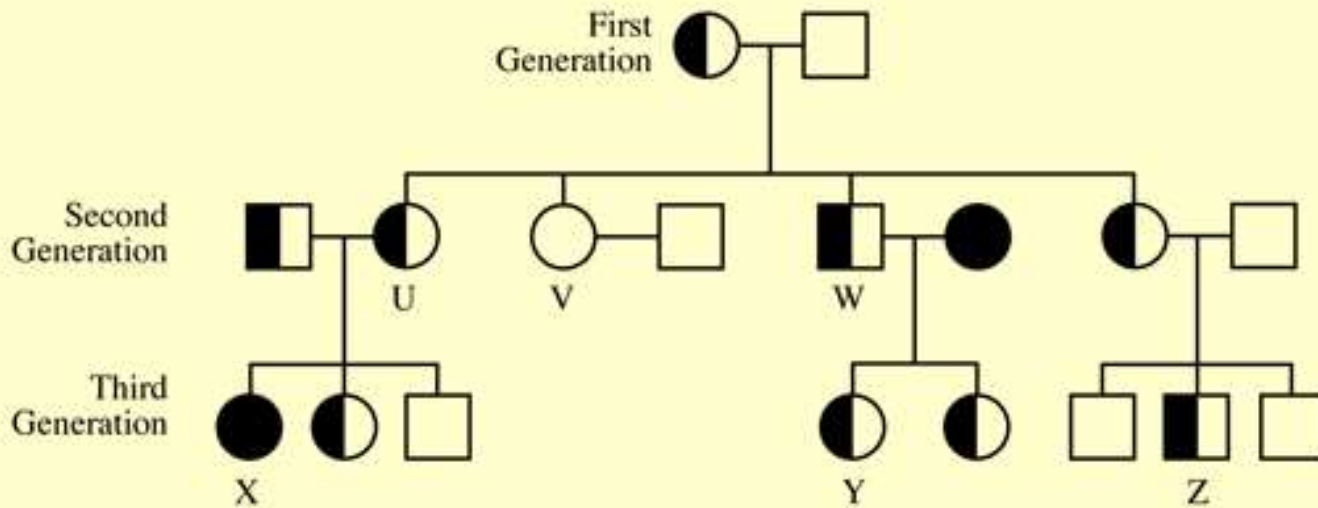
- **Reveal your genotypes**
- **On your own paper EACH PARTNER should use a Punnett square to show the probable genotypes of your offspring**

Activity

- **Make a pedigree:**
- **Show you and your partner, as well as your 4 offspring**

Pedigrees

Genetic Pedigree



Key

- | | |
|---------------------|-------------------|
| ○ = normal female | □ = normal male |
| ● = affected female | ■ = affected male |
| ◐ = carrier female | ◑ = carrier male |

Activity

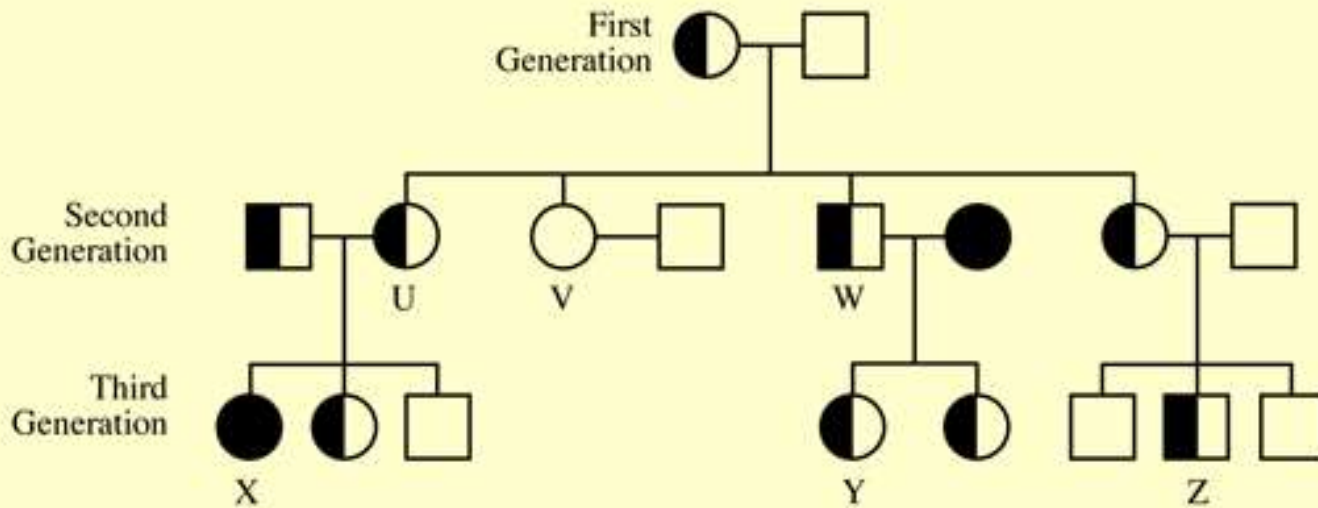
- **With your partner select a mate for one of your offspring from the children of another partner group**
- **Complete a Punnett square to show their possible offspring genotypes**

Activity

- **On your pedigree show their offspring**

Pedigrees

Genetic Pedigree



Key

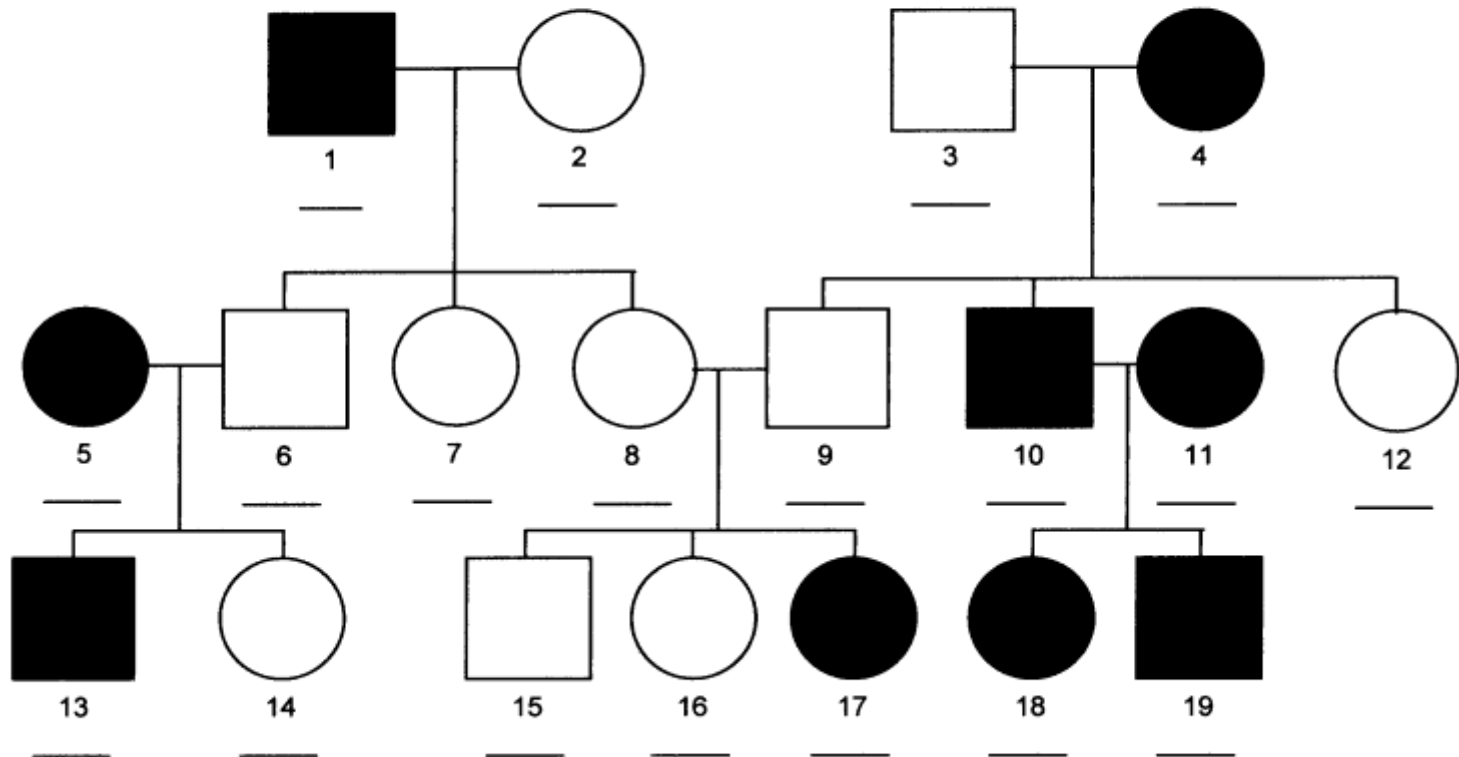
- | | |
|---------------------|-------------------|
| ○ = normal female | □ = normal male |
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Deciphering Pedigrees

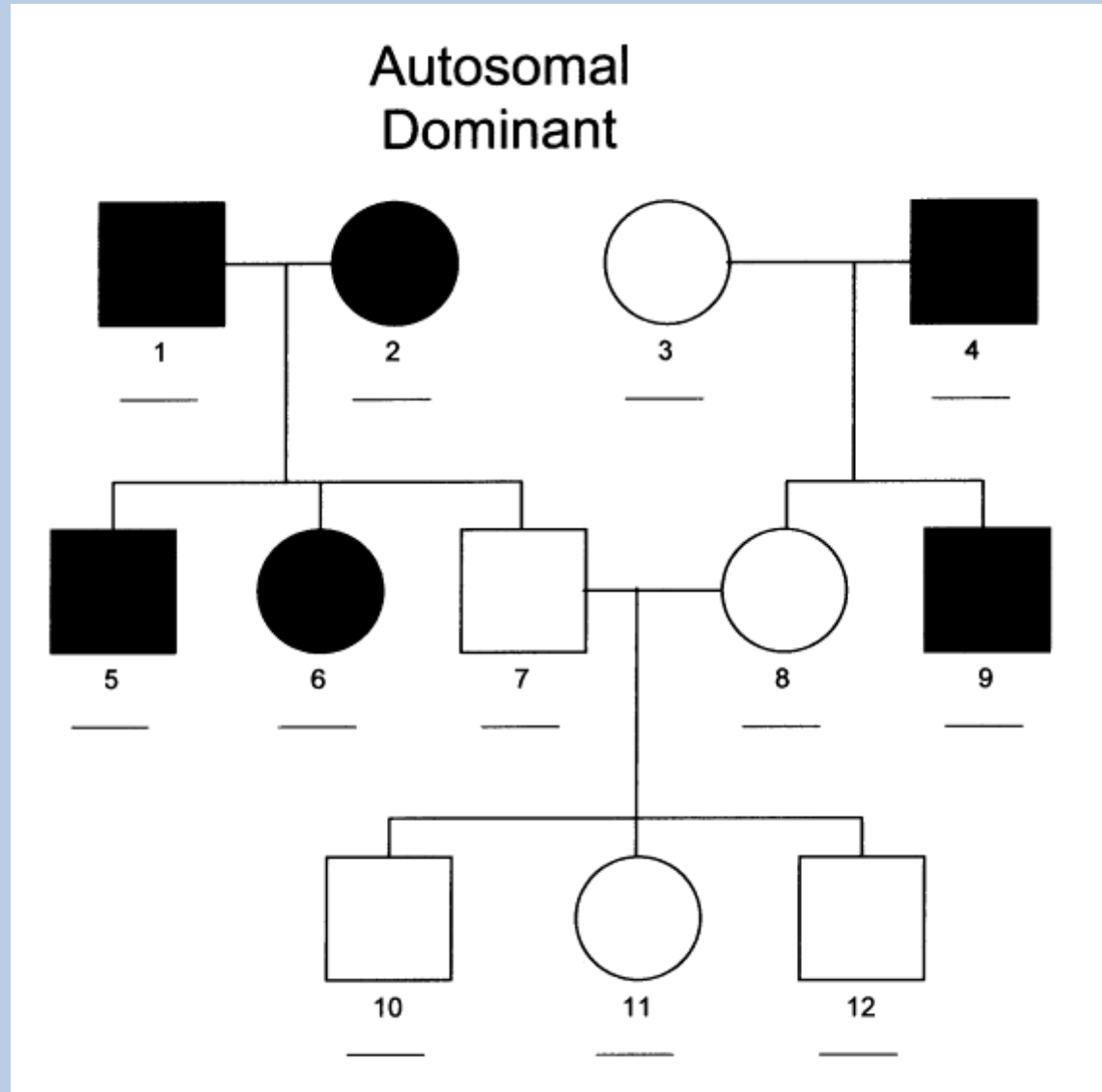
- **Two general hints:**
 - **If the trait SKIPS a generation it is recessive**
 - **If the trait is EQUAL in males and females it is autosomal**

Deciphering Pedigrees

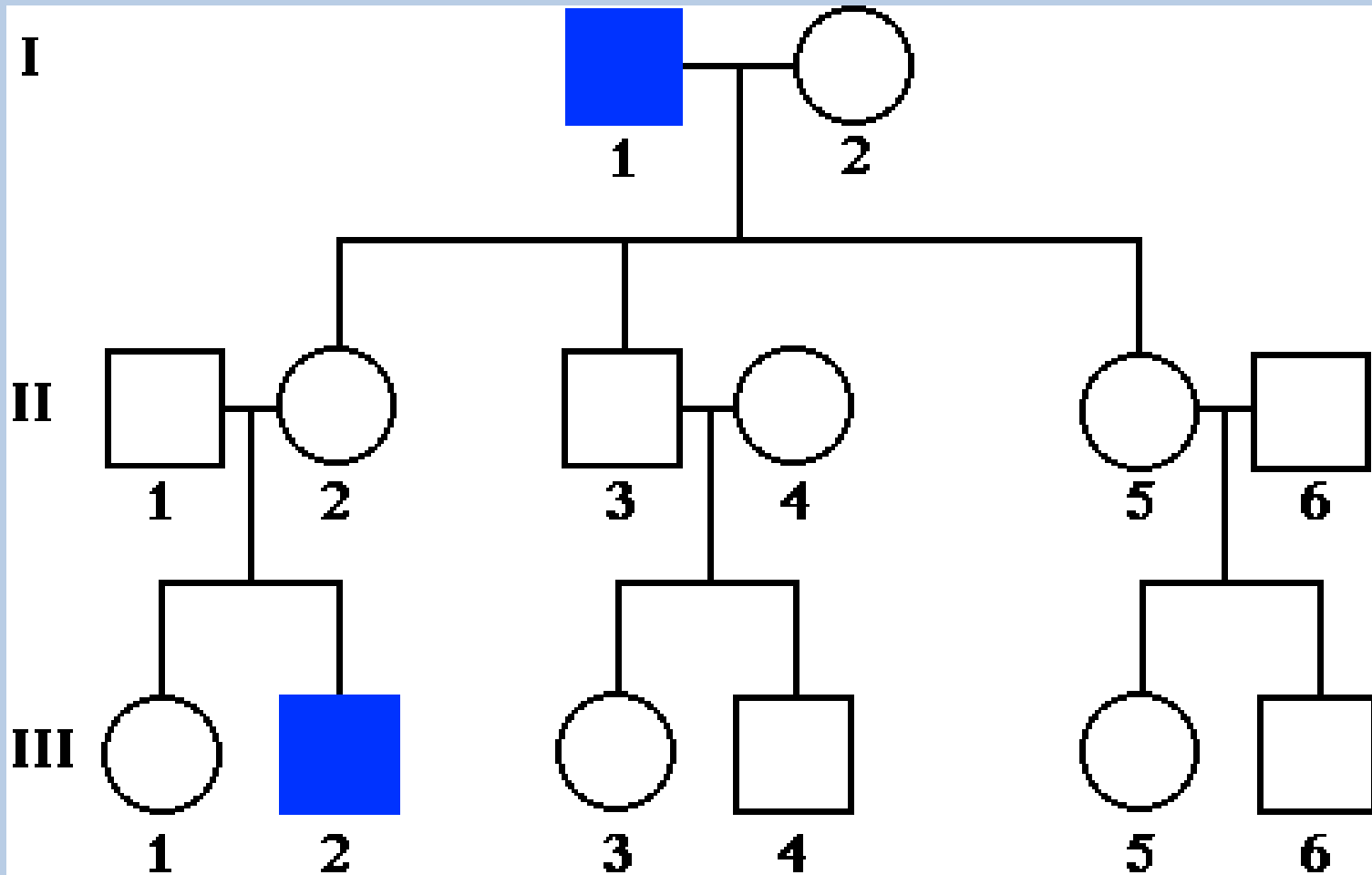
AUTOSOMAL RECESSIVE



Deciphering Pedigrees

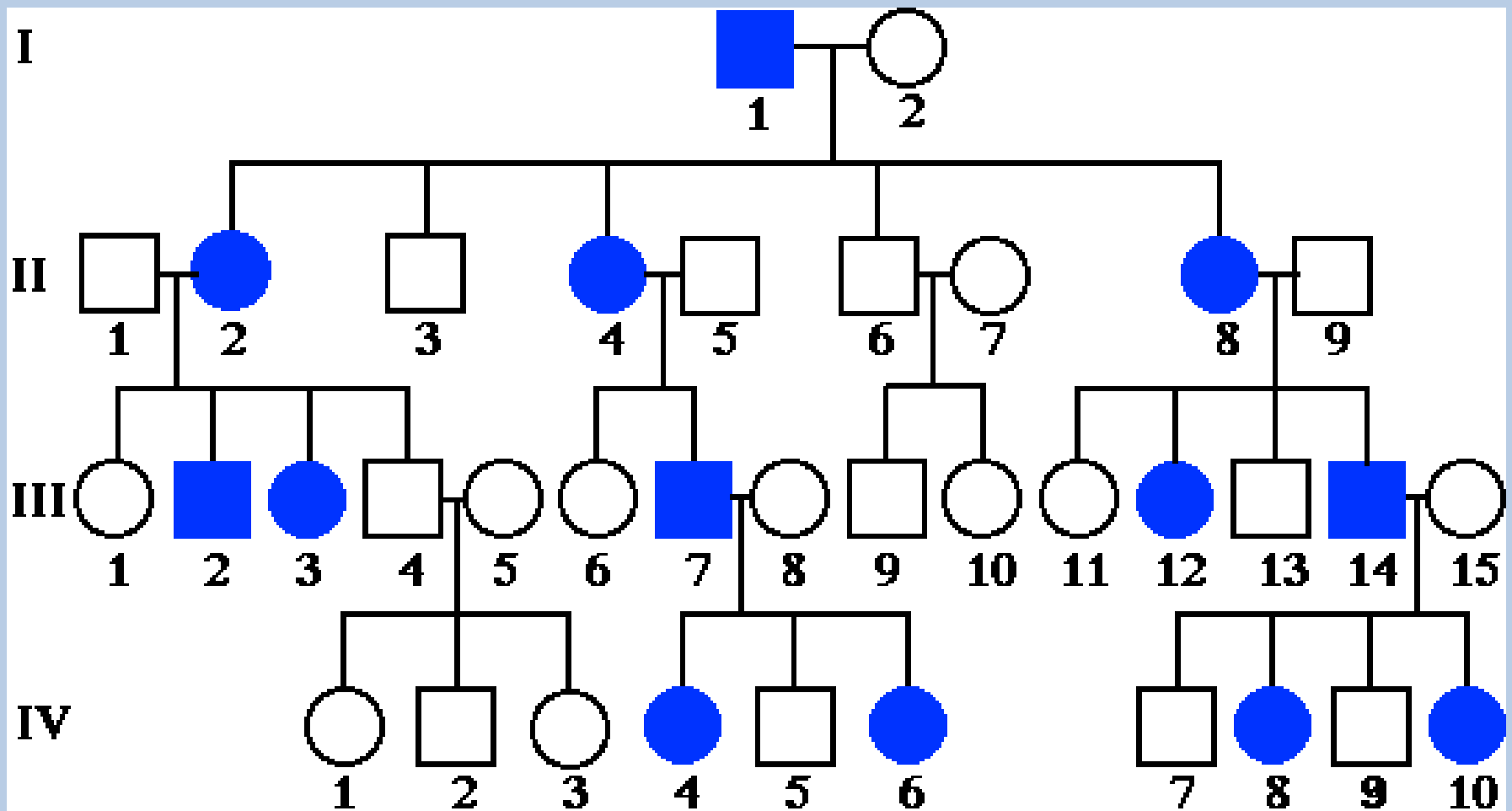


Deciphering Pedigrees



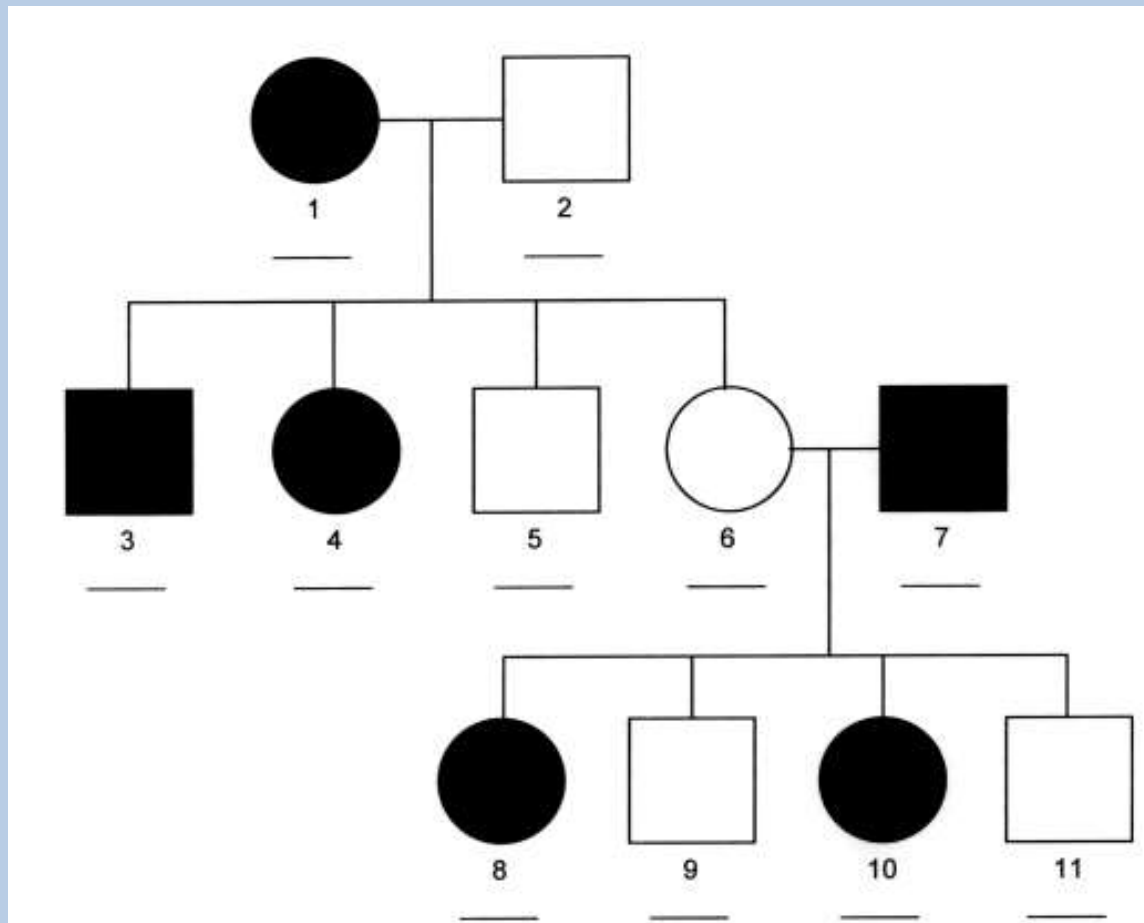
Pedigree 7. X-linked recessive inheritance.

Deciphering Pedigrees

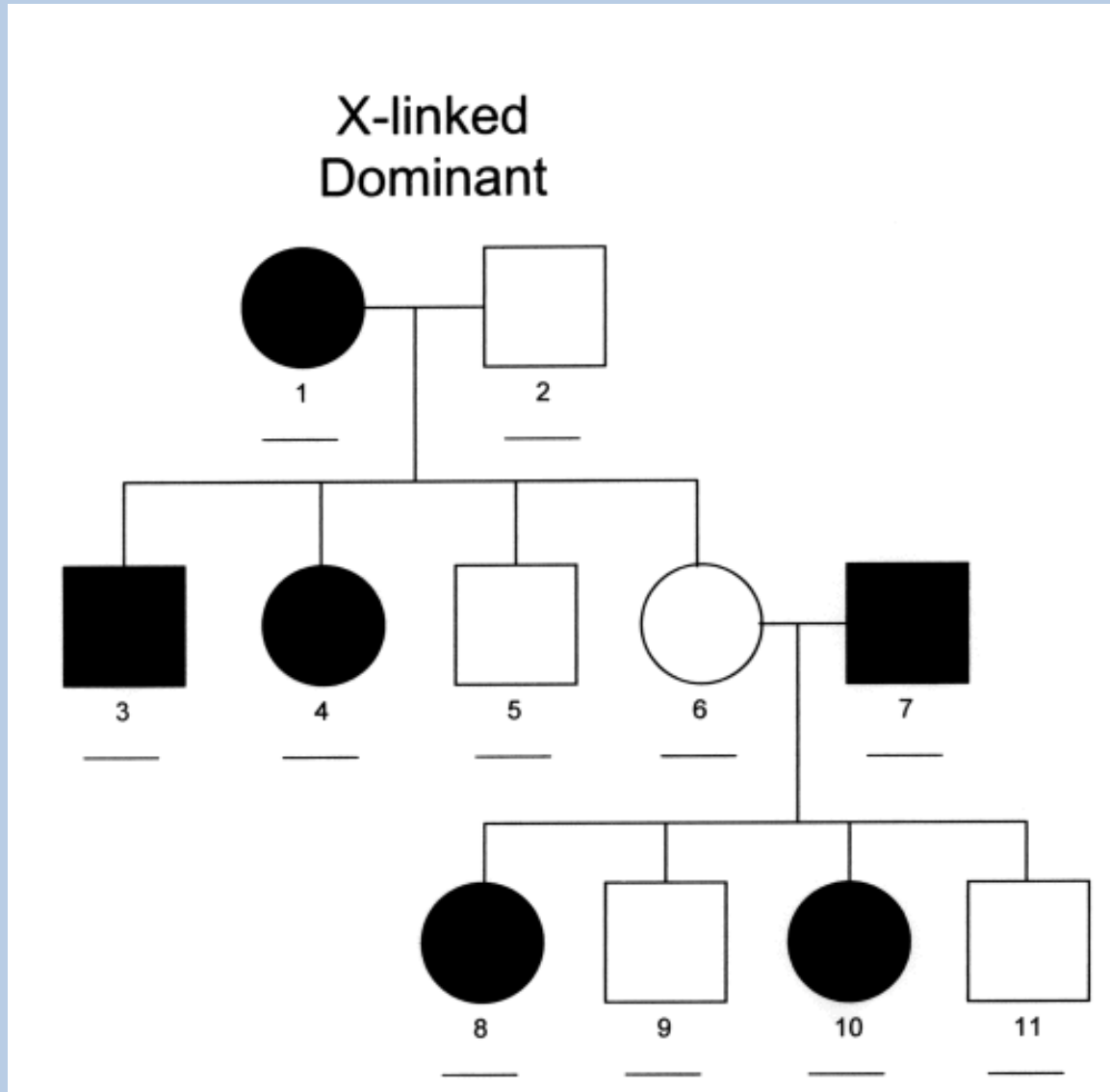


Pedigree 5. X-linked dominant inheritance.

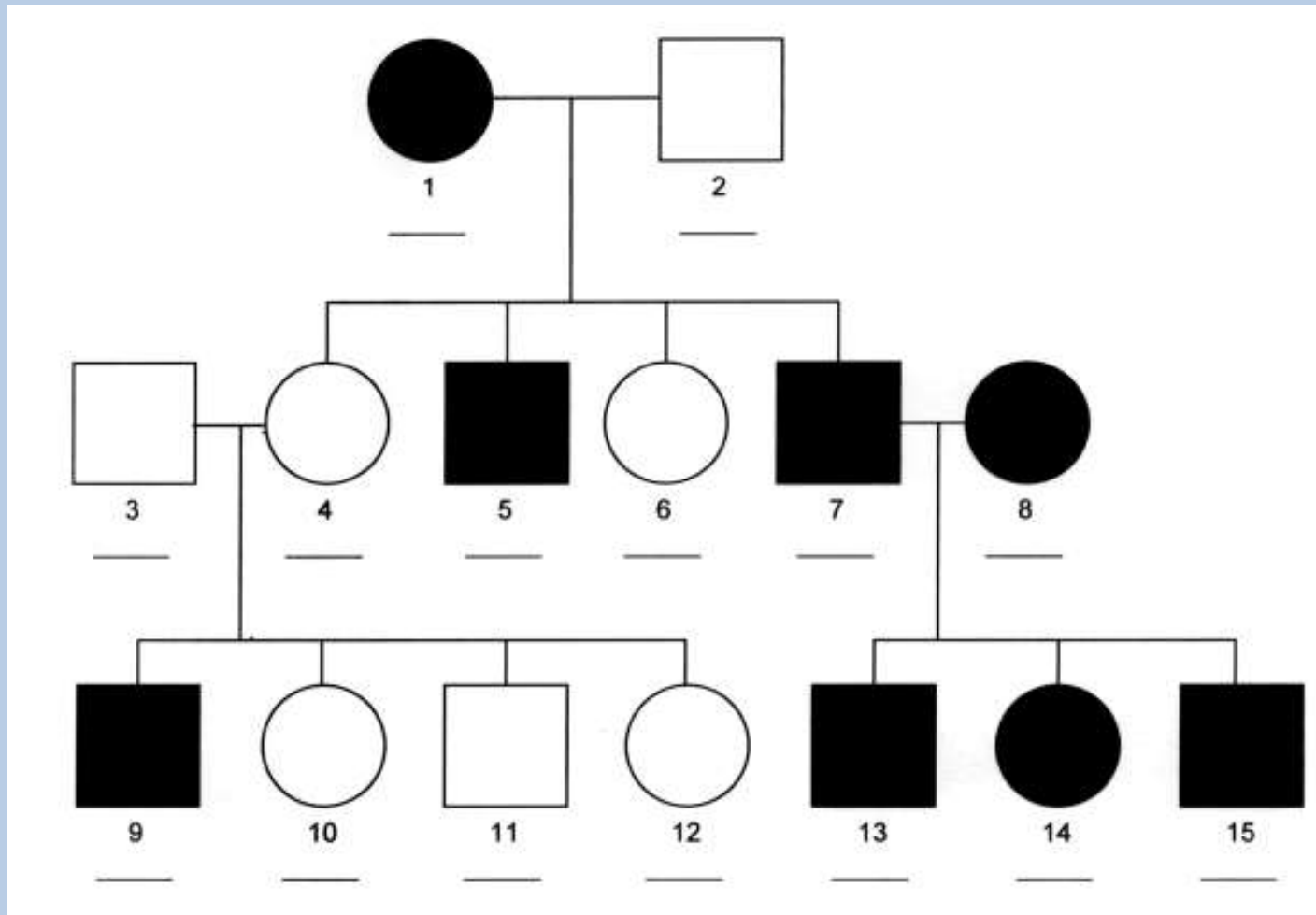
What is the pattern of inheritance?



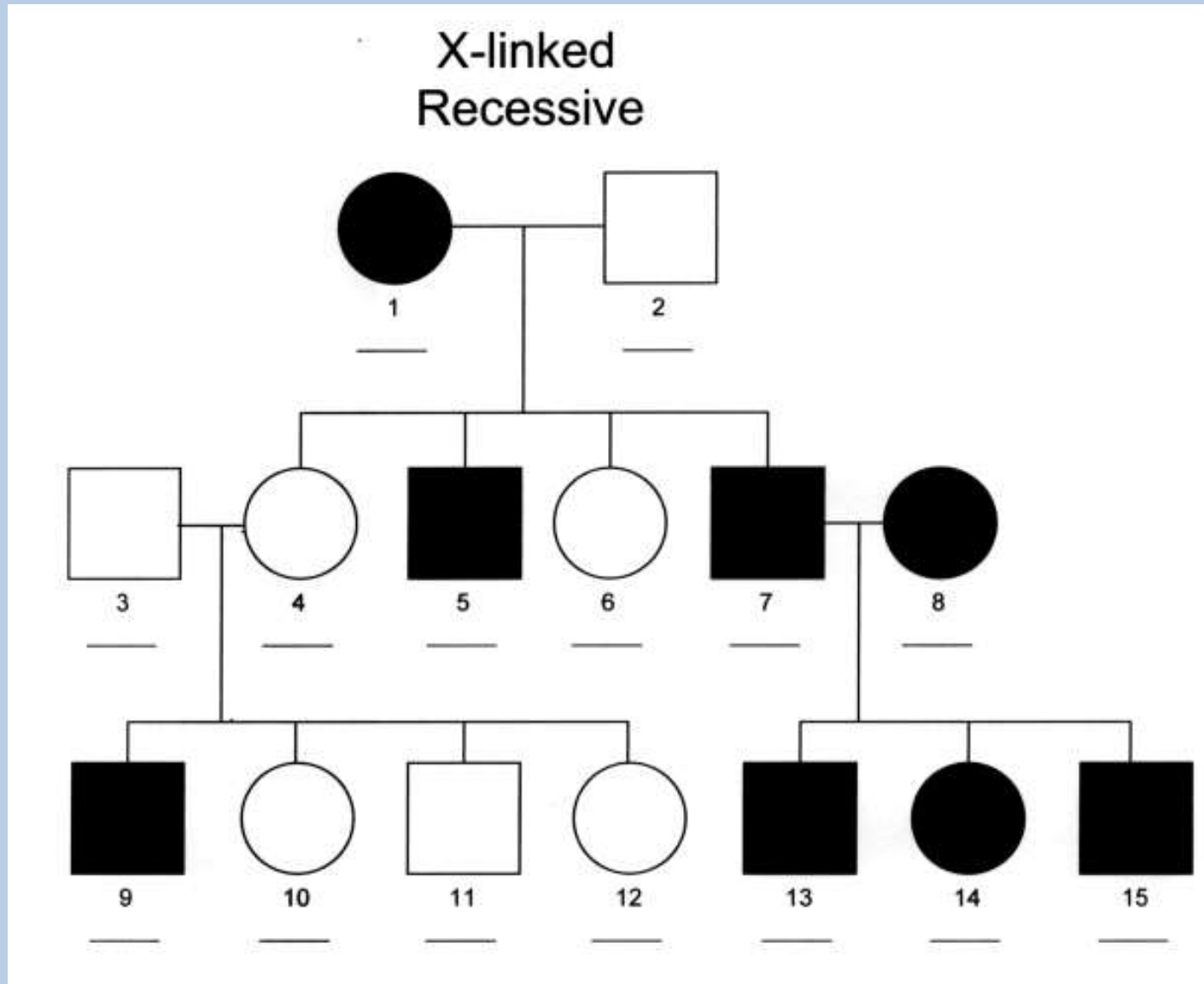
What is the pattern of inheritance?



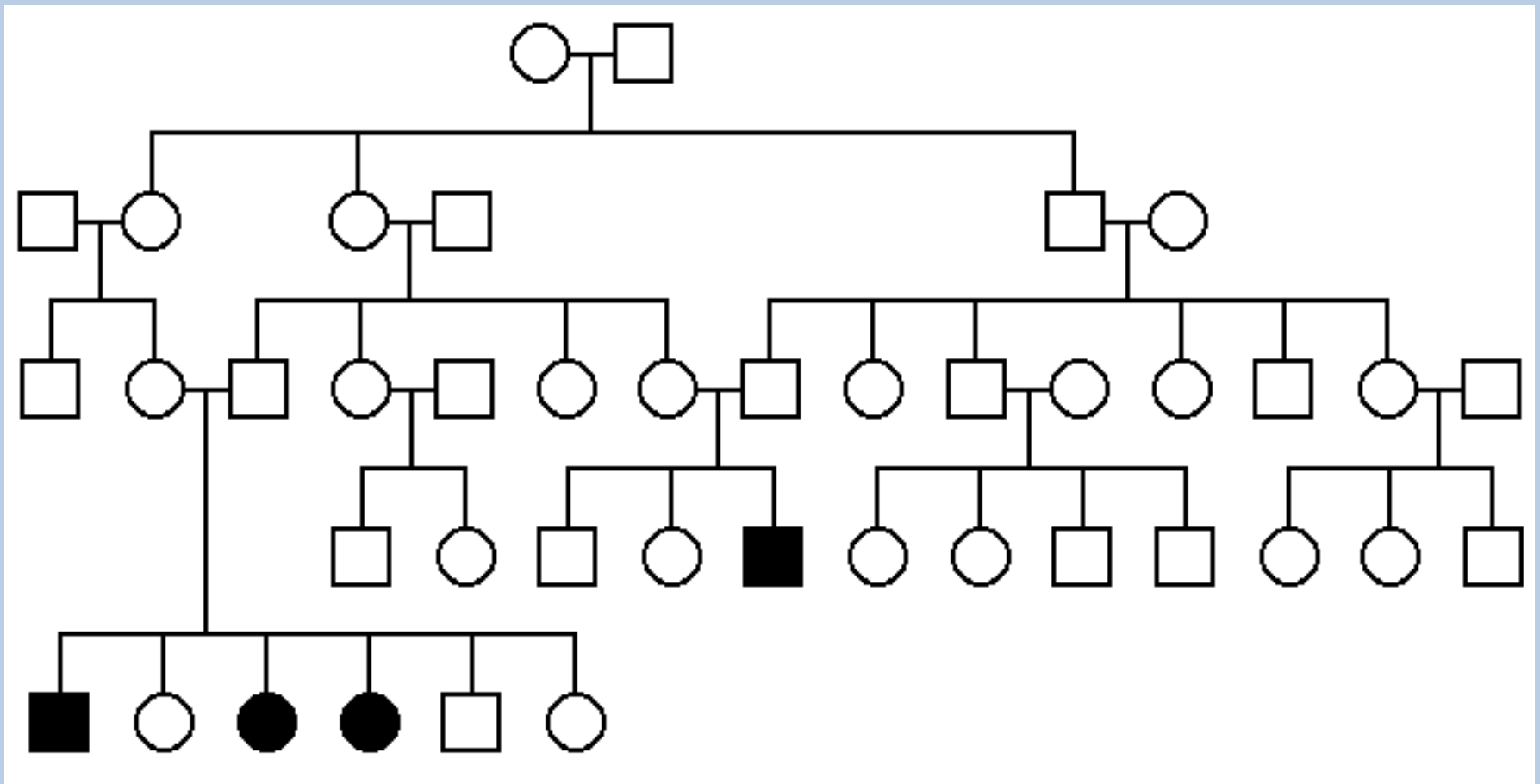
What is the pattern of inheritance?



What is the pattern of inheritance?

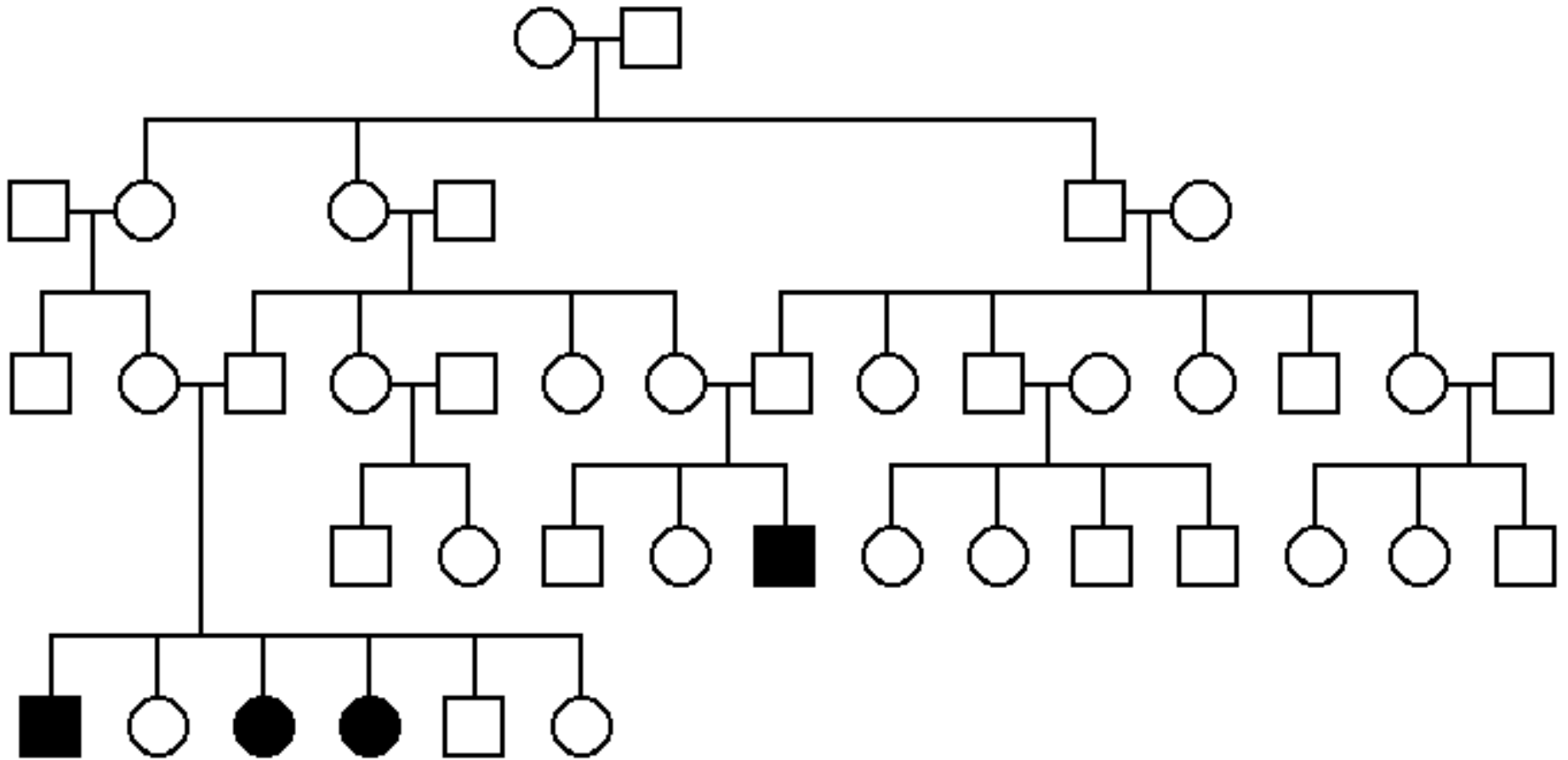


What is the pattern of inheritance?

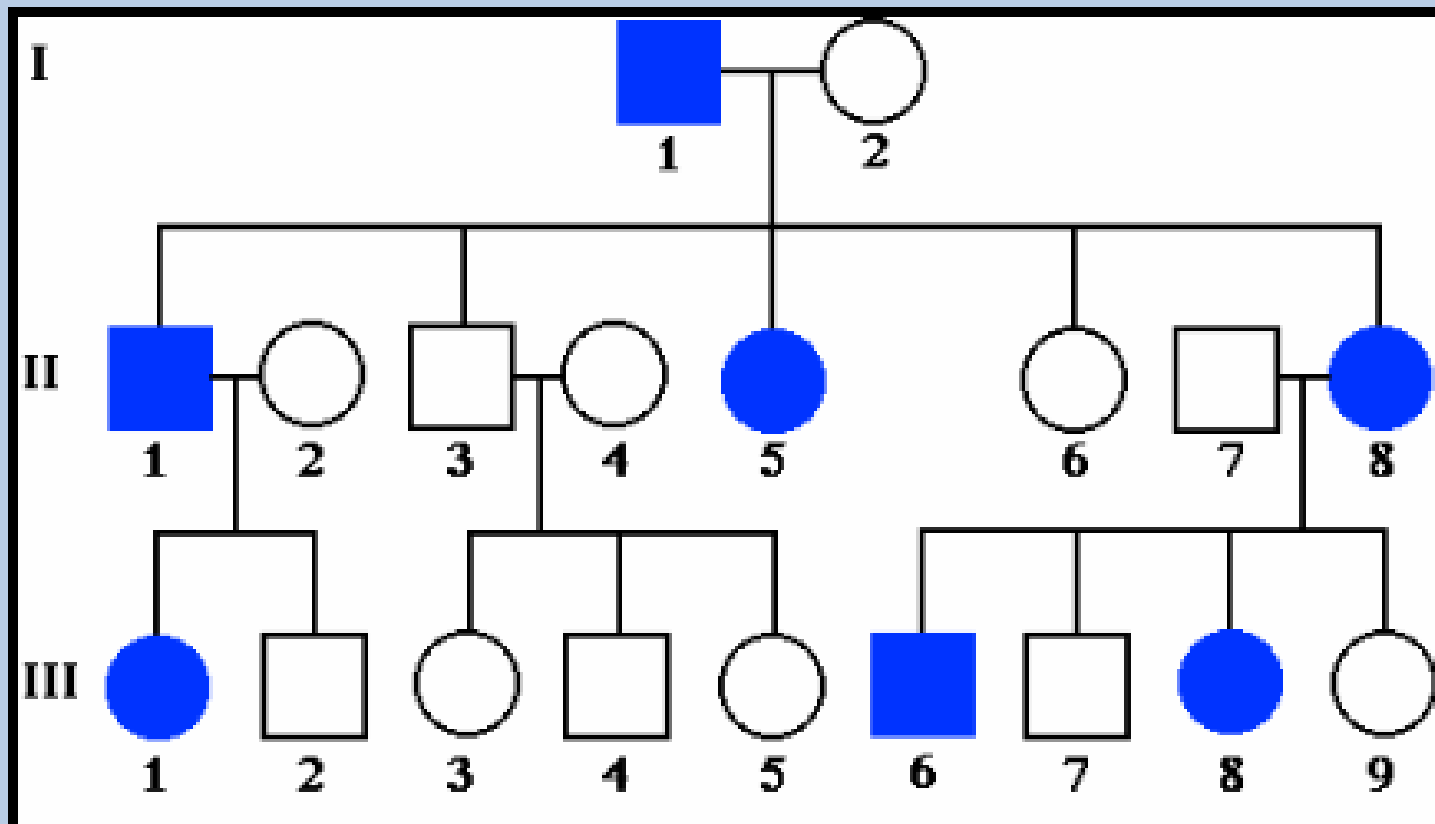


What is the pattern of inheritance?

Autosomal Recessive



What is the pattern of inheritance?



What is the pattern of inheritance?

Autosomal Dominant

