PEDIGREES

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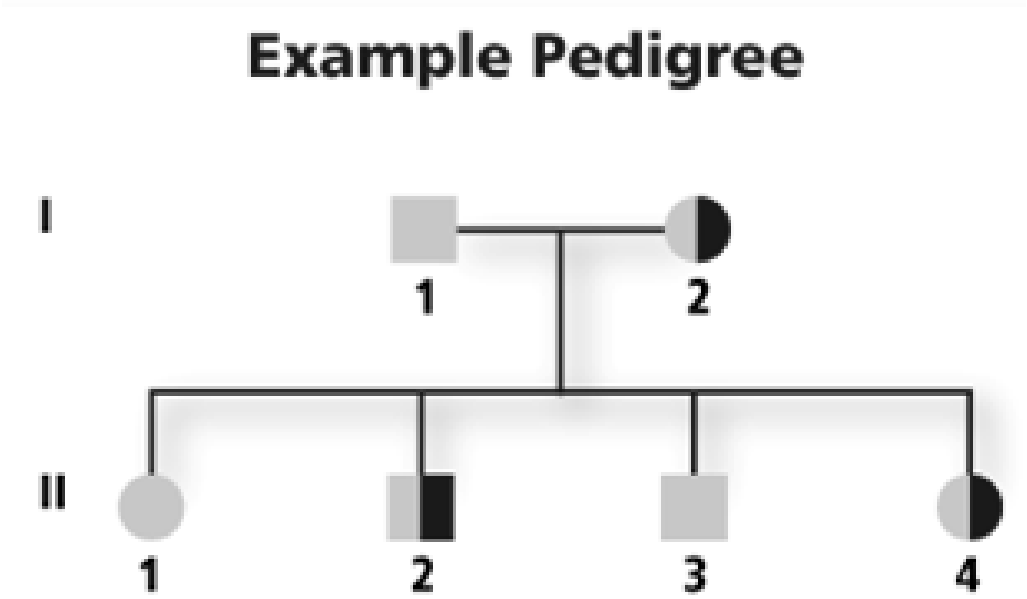
Pedigree: a family tree used to look at the inheritance of certain traits, especially of genetic diseases.



Parents= top row

Children= bottom row

PEDIGREE REVIEW

1. **Draw a male:**

2. **Draw a female:**

3. **Draw a couple:**

4. **Now draw that with three kids (from oldest to youngest)—a boy and two girls:**

GENETIC DISEASES

For any genetic disease there are 2 alleles (*just like there are for a trait*):

- One allele causes you to have the disease.
- Other allele does not cause you to have the disease.

2 types of genetic diseases:

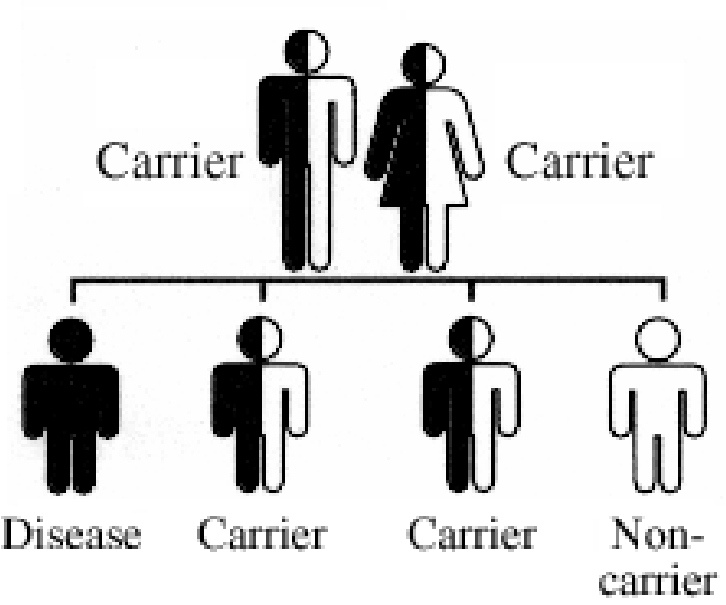
1. Recessive Genetic Diseases
2. Dominant Genetic Diseases

RECESSIVE GENETIC DISORDERS

- The disease is in the recessive allele (a).
- You must be homozygous recessive (aa) to have the disease.
- Therefore, those with a dominant allele (AA or Aa) will not have the disorder.
- An individual who is heterozygous (Aa) for a recessive disorder is called a carrier.

A = _____

a = _____



Recessive Genetic Disorders in Humans

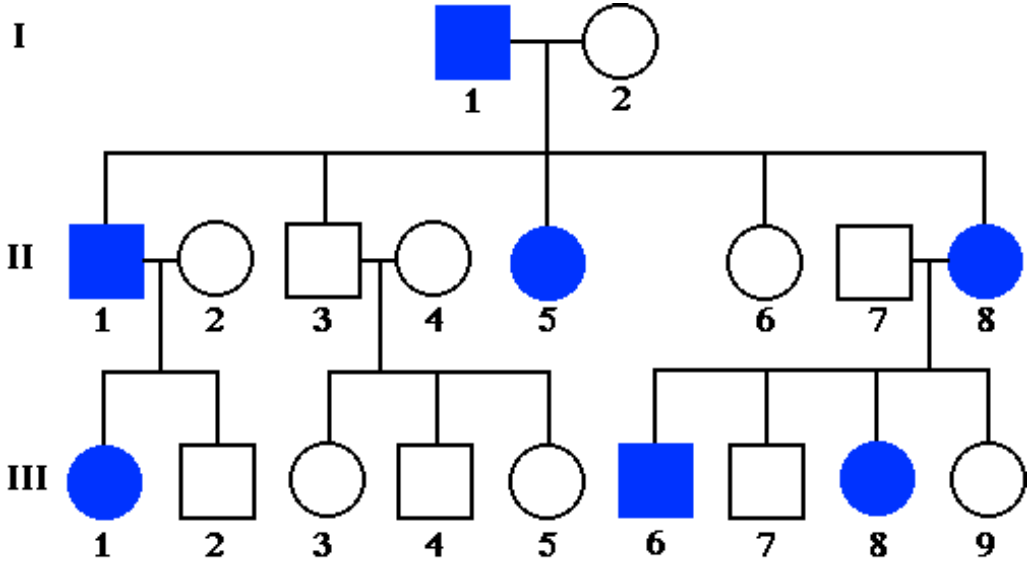
Disorder	Occurence in the U.S.	Cause	Affect	Cure/Treatment
Cystic fibrosis	1 in 3500	The gene that codes for a membrane protein is defective.	• Excessive mucus production • Digestive and respiratory failure	• No cure • Daily cleaning of mucus from the lungs • Mucus-thinning drugs • Pancreatic enzyme supplements
Albinism	1 in 17,000	Genes do not produce normal amounts of the pigment melanin.	• No color in the skin, eyes and hair • Skin susceptible to UV damage • Vision problems	• No cure • Protect skin from the Sun and other environmental factors • Visual rehabilitation
Galactosemia	1 in 50,000 to 70,000	Absence of the gene that codes for the enzyme that breaks down galactose.	• Mental disabilities • Enlarged liver • Kidney failure	• No cure • Restriction of lactose/ galactose in the diet
Tay-Sachs disease	1 in 2500 (affects people of Jewish descent)	Absence of a necessary enzyme that breaks down fatty substances.	• Buildup of fatty deposits in the brain • Mental disabilities	• No cure or treatment • Death by age 5

DOMINANT GENETIC DISORDERS

- Some genetic diseases are caused by dominant alleles (B).
- If you have a dominant allele, you have the disease.
- If you are homozygous dominant (BB) or heterozygous (Bb), you have the disease.
- Those who do not have the disorder are homozygous recessive (bb) for the trait.

B = _____

b = _____



Dominant Genetic Disorders in Humans

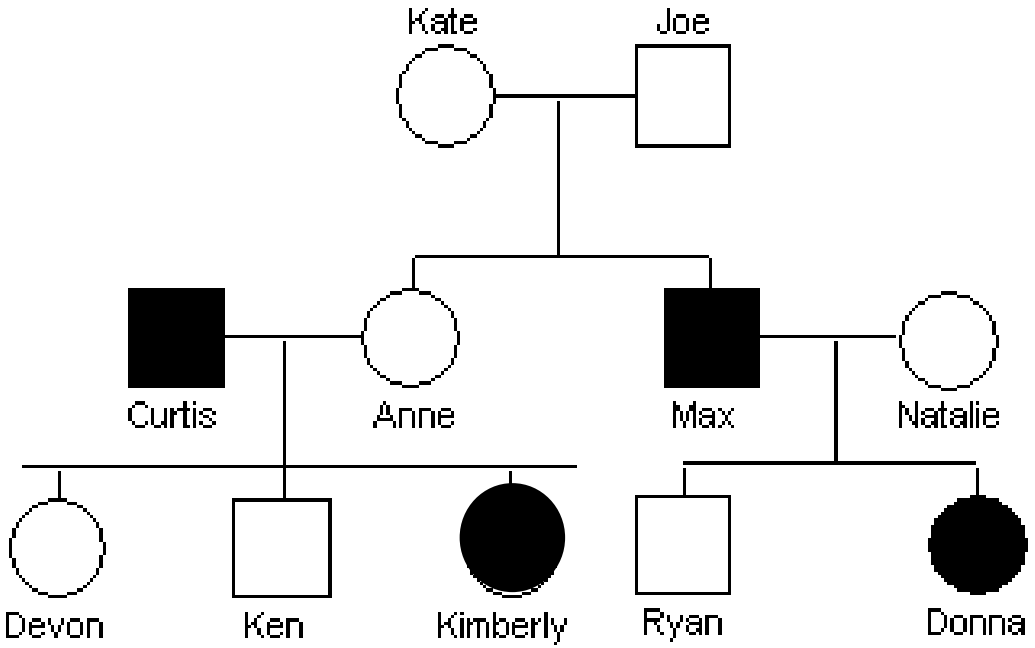
Disorder	Occurence in the U.S.	Cause	Affect	Cure/Treatment
Huntington's disease	1 in 10,000	A gene affecting neurological function is defective.	• Decline of mental and neurological functions • Ability to move deteriorates	• No cure or treatment
Achondroplasia	1 in 25,000	A gene that affects bone growth is abnormal.	• Short arms and legs • Large head	• No cure or treatment

INFERRING GENOTYPES & PREDICTING DISORDERS

- Pedigrees are used to infer genotypes.
- Pedigrees help genetic counselors determine whether inheritance patterns are dominant or recessive.
- If good records have been kept, disorders in future offspring can be predicted.

Filling Out Pedigrees Example:
This pedigree shows the pattern of albinism, a recessive disease. Write the genotypes for each person.

Pick the letter of the disease

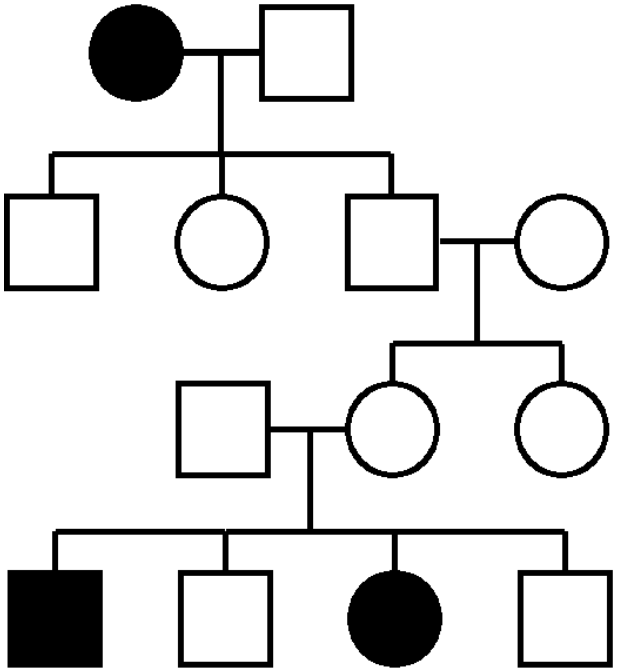


RECESSIVE VS. DOMINANT PEDIGREES

RECESSIVE

If both parents do not have the disorder, but some of the children do

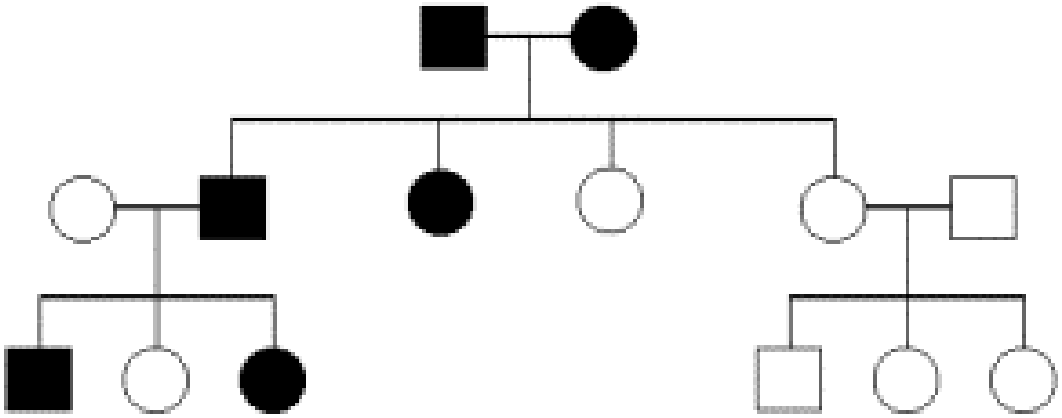
The disease can skip generations



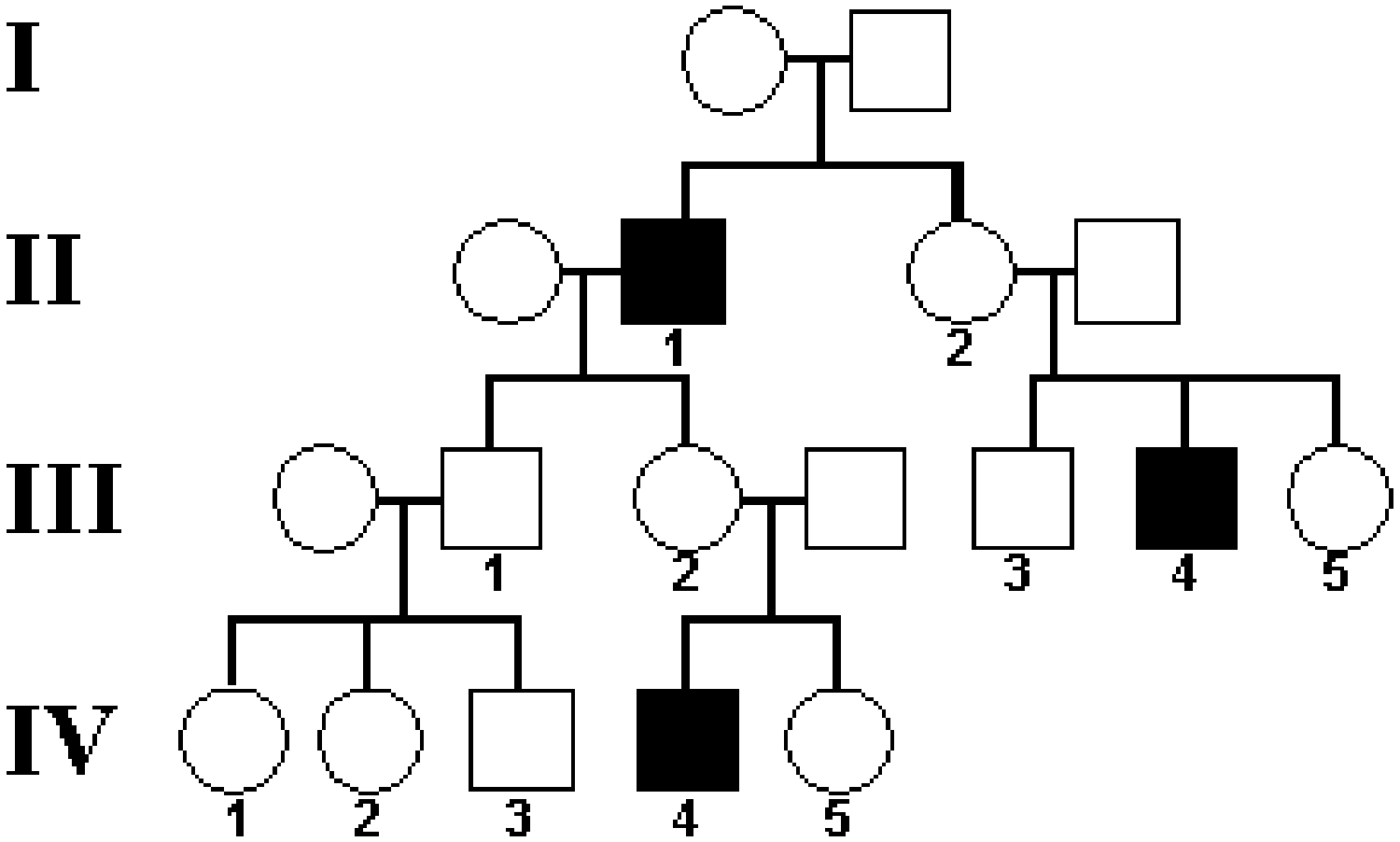
DOMINANT

If both parents have the disorder, and one of the offspring does not have it

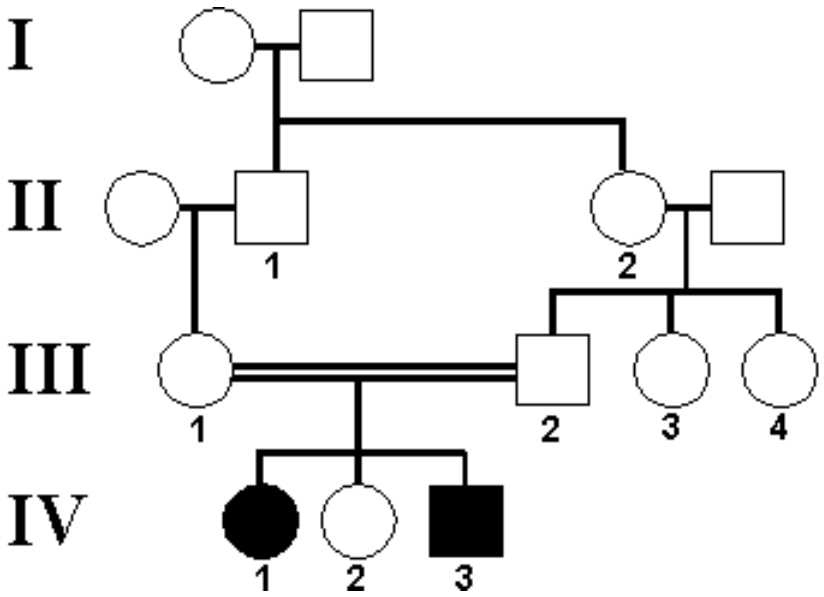
The disease is present in every generation, because if a child has the disease, one of the parents must have it also



What is the inheritance pattern?
What is the genotype of III-1, III-2, and II-3?
What are the odds that IV-5 would have an affected son?



What is the pattern of inheritance?
What are IV-2's odds of being a carrier?



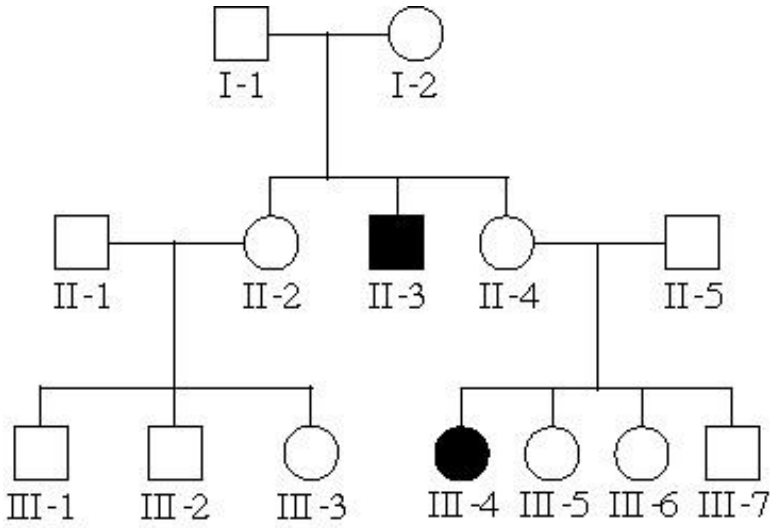
Sample pedigree - cystic fibrosis

What can we say about I-1 and I-2?

What can we say about II-4 and II-5?

What are the odds that III-5 is a carrier?

What can we say about gene frequency?

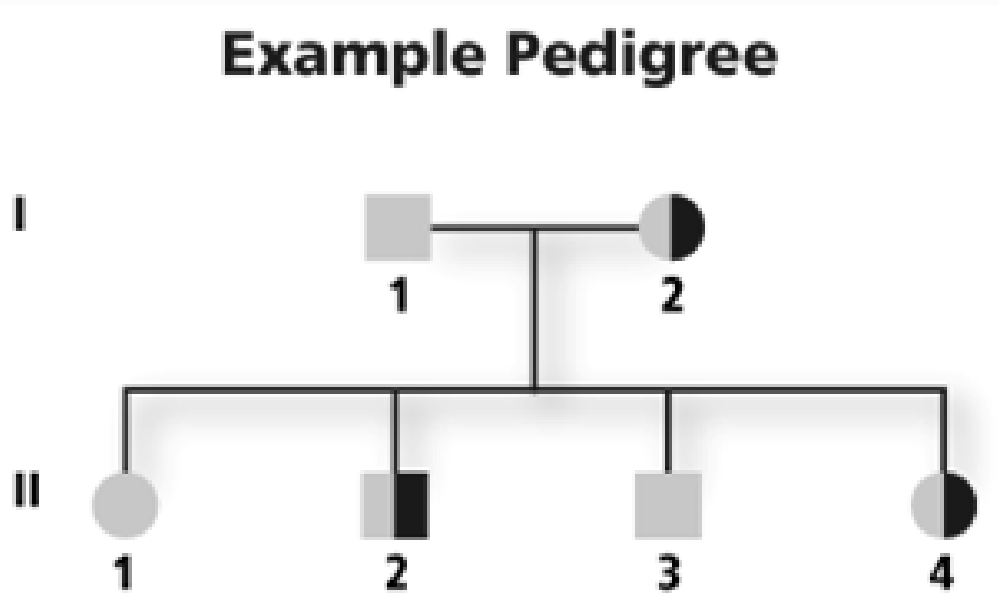


STUDENT NOTES

PEDIGREES

Pedigree: _____

_____.



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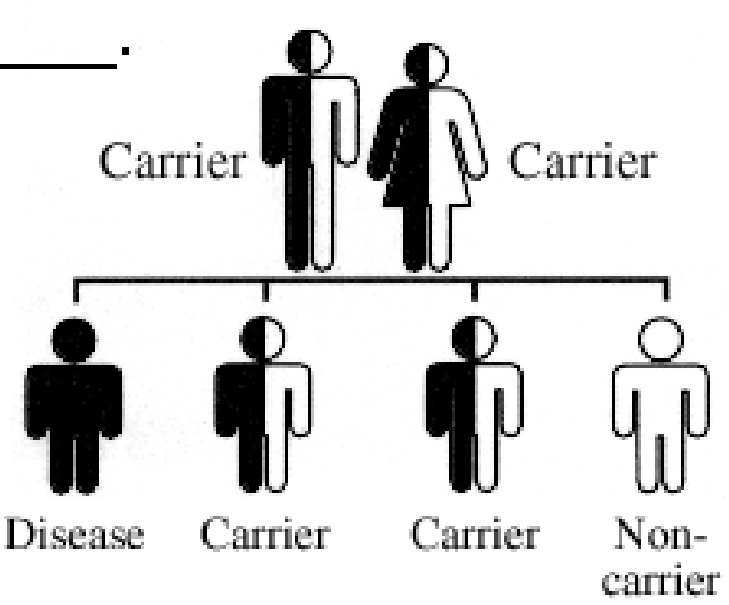

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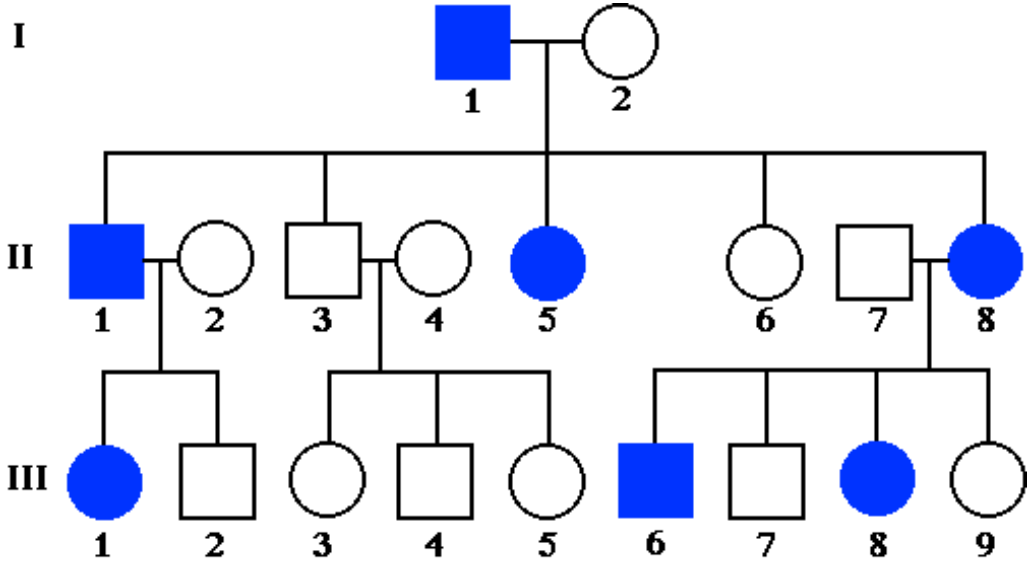

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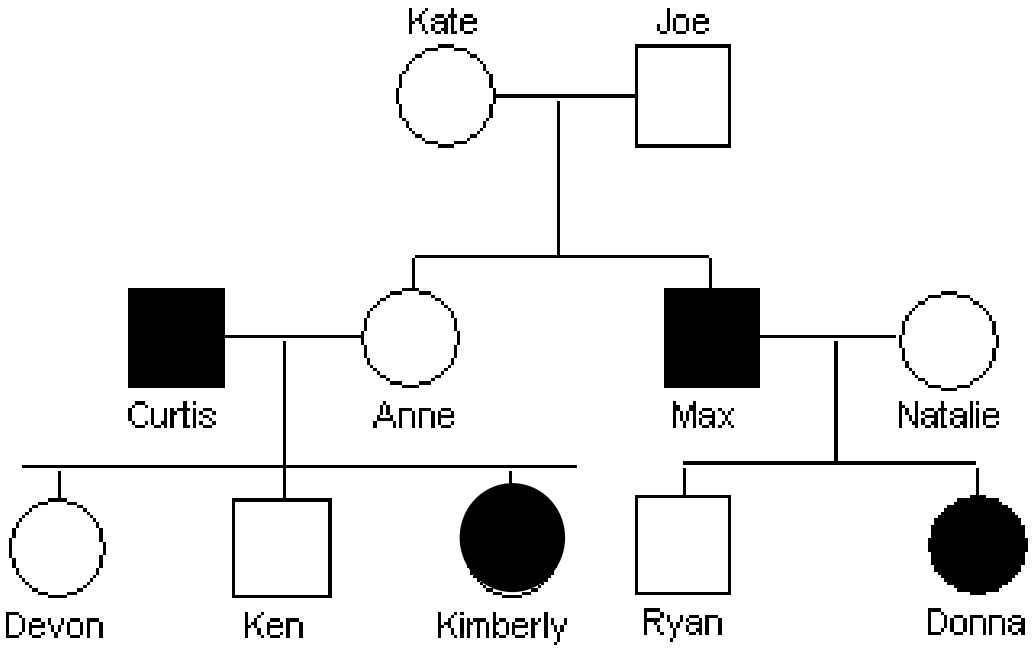


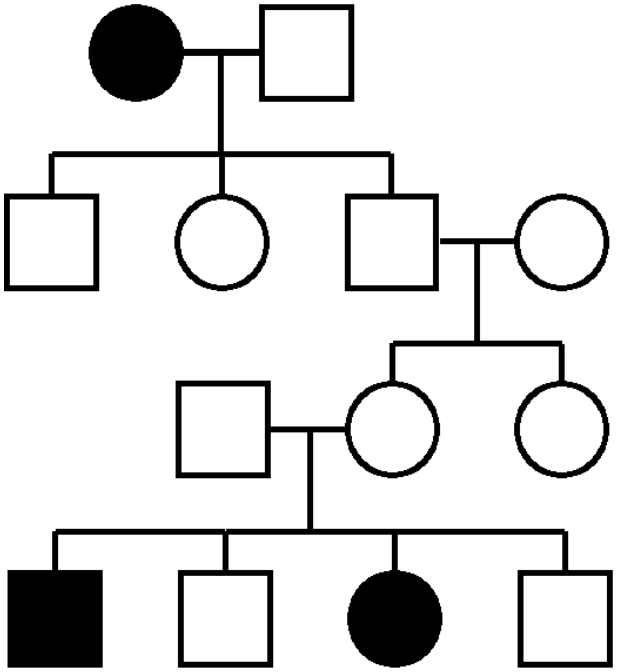
RECESSIVE VS. DOMINANT PEDIGREES

RECESSIVE

If both parents **do not** have the disorder, <u>but some of the children do</u>.

_____

_____.



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The disease is present in every generation, because if a child has the disease, _____

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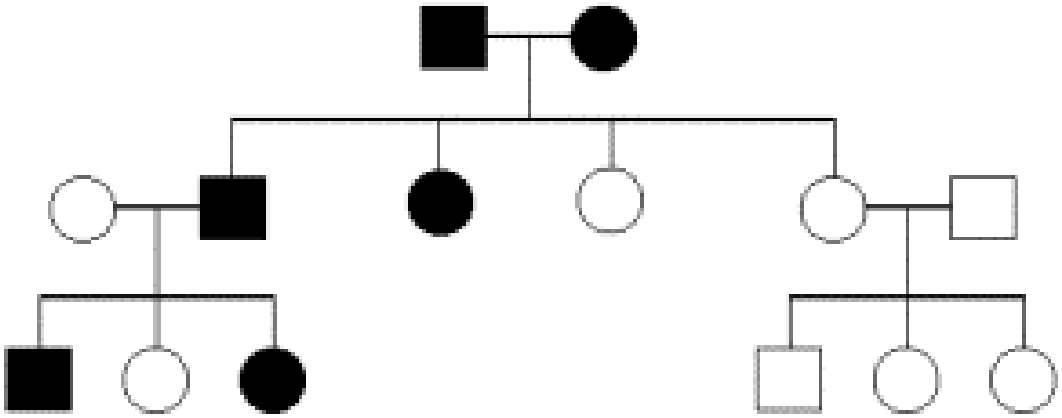

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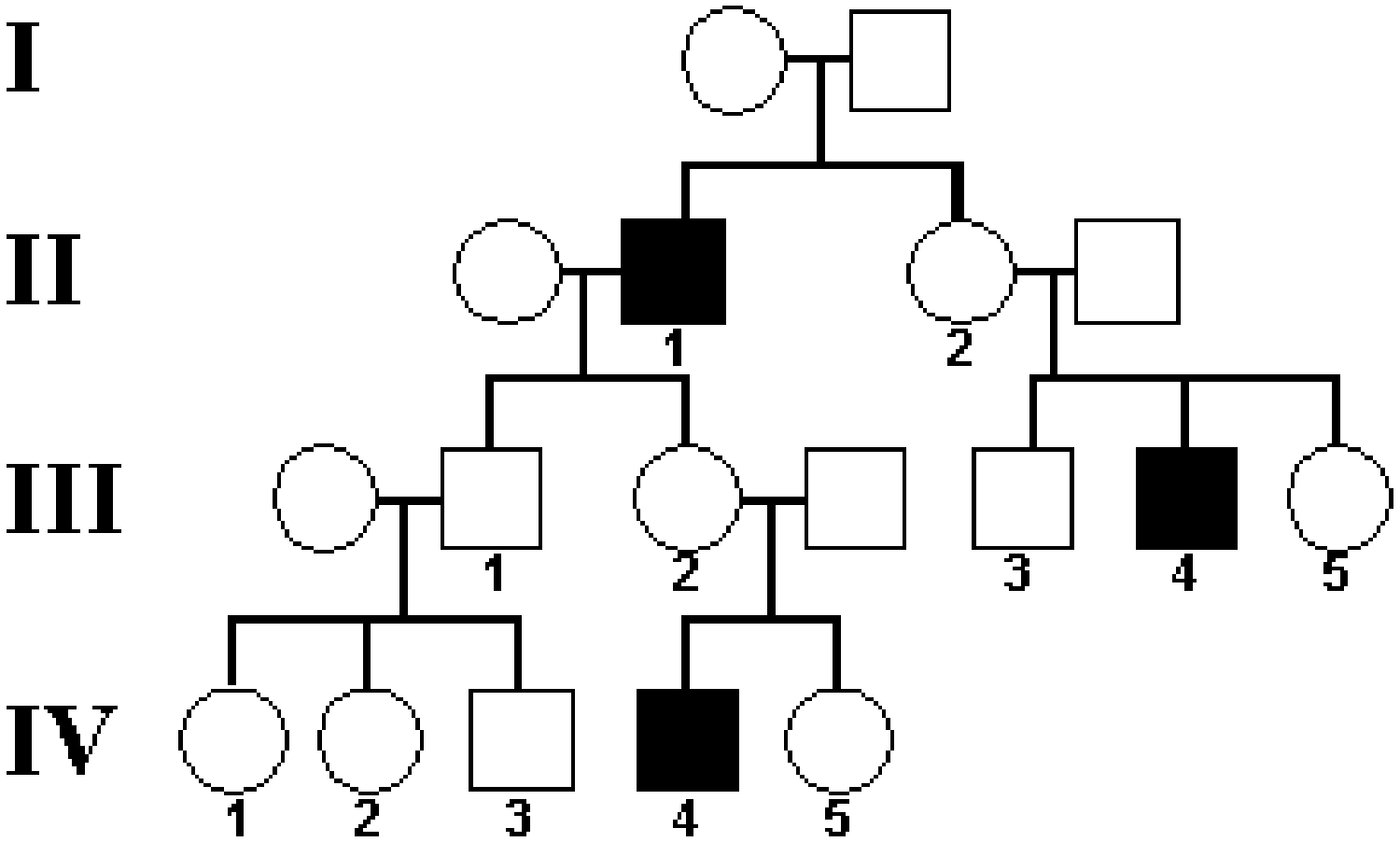

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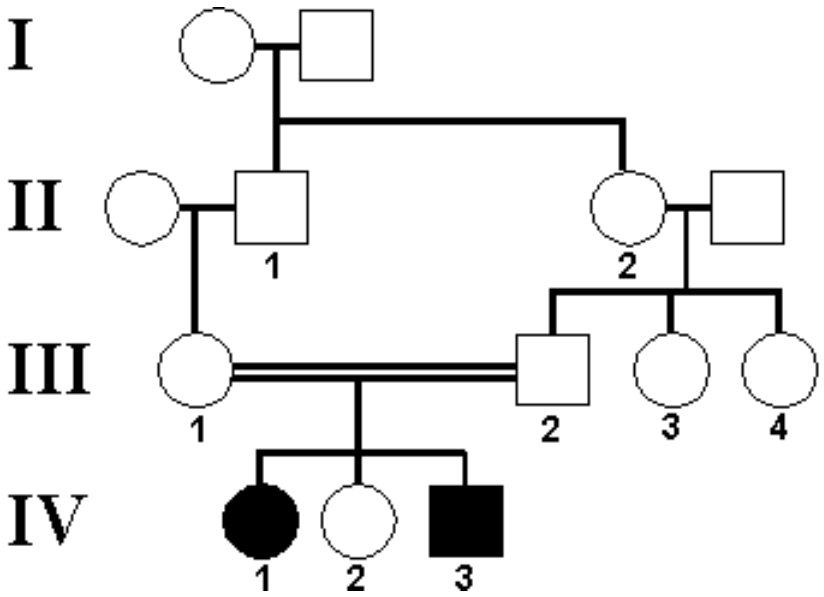

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