



E - Bio Worksheet

Pedigree Analysis In Genetics

Background Reading – pedigree analysis Nelson Biology, Campbell Biology

Purpose - To introduce students to the role of pedigree charts in tracing human traits.

Introduction

Pedigree charts are often constructed to show the inheritance of genetic conditions within a family. Such charts are a great help in determining whether a phenotype is controlled by a dominant, recessive or sex-linked allele. In future activities you will see how gel electrophoresis (DNA fingerprinting) is being used in pedigree analysis. This activity will introduce you to autosomal pedigree charts with sex-linkage being undertaken in a future activity.

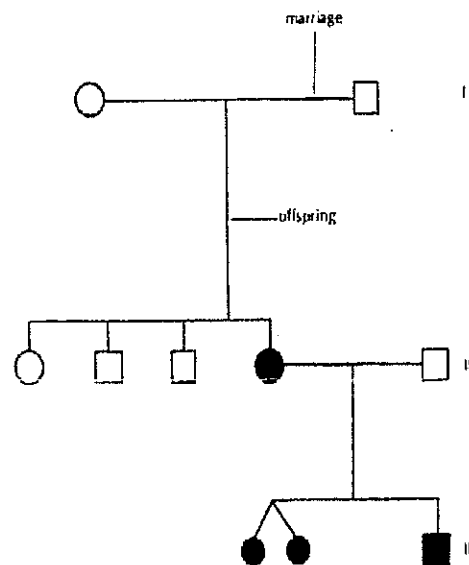
Part I - Understanding The Pedigree

1. **Table I** shows the symbols needed in understanding a pedigree. In **FIGURE I** Generation I is made up of grandparents, Generation II is their children and Generation III is their grandchildren.

Table I - Pedigree Key

Symbol	Meaning
○	Female without trait
□	Male without trait
●	Female with trait
■	Male with trait
●	Female, died in infancy
■	Male, died in infancy
⎓	Identical twins

Figure I - Sample Pedigree



Q1 - How many normal males are represented in Figure I ? Normal females ?

Q2 - How many children did the grandparents have ?

Q3 - How many affected individuals are present ?

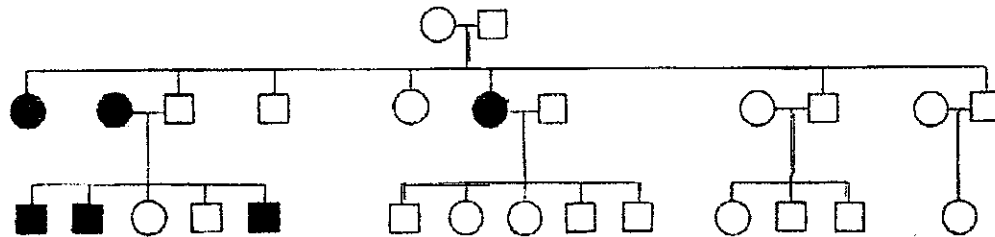
Part III - Trait Analysis

1. The following pedigrees will be used to determine whether the trait is autosomal dominant or autosomal recessive. In tracing autosomal alleles, if both parents have the disorder and the offspring do not, the condition is autosomal dominant. If neither parent shows the disorder but some of their children do, the condition is autosomal recessive. A carrier is an individual who appears to be normal, but who is capable of passing on a gene for the disorder. If the characteristic is dominant, there can be no carriers because only a single gene is needed to show the disorder. **Table II** provides some keys for your answers.

Table II - Pedigree Keys

CHARACTERISTIC	KEY
AUTOSOMAL DOMINANT	AA = AFFECTED Aa = AFFECTED aa = NORMAL
AUTOSOMAL RECESSIVE	AA = NORMAL Aa = CARRIER aa = AFFECTED

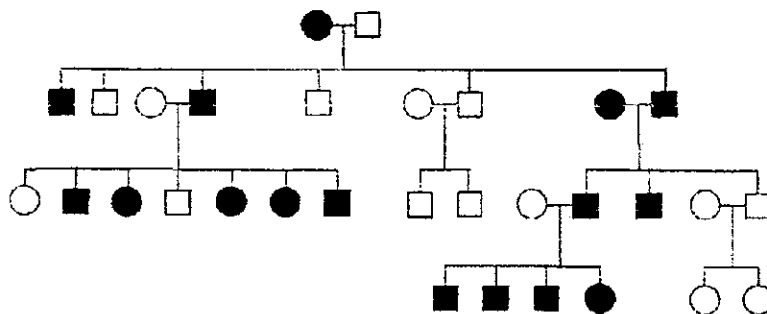
Figure IV - Pedigree 1



Q8 - Is the gene for the condition autosomal dominant or recessive ?

Q9 - Identify the pedigree for each individual using the above table. Use **A_** if undetermined.

Figure V - Pedigree 2



Q10 - Is the gene for the condition autosomal dominant or recessive ?

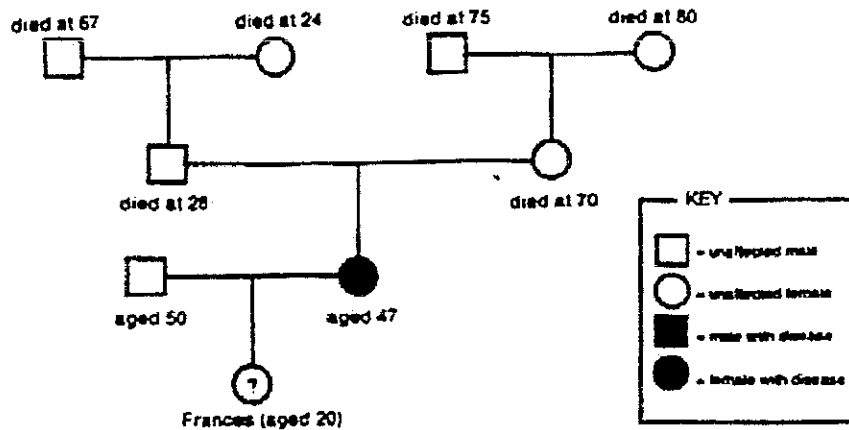
Q11 - Identify the pedigree for each individual using the above table. Use **A_** if undetermined.

Part V - Huntington's Disease

Huntington disease (HD) is an inherited brain disorder affecting the nervous system. It causes progressive deterioration of physical and mental capabilities, leading ultimately to severe incapacitation and eventual death, generally 15-25 years after onset. Primarily, it affects adults, usually appearing between the ages of 30 and 45. Occasionally, HD symptoms appear earlier (before age 20, the juvenile form) or later (after age 50.) Common symptoms of adult-onset HD are involuntary movements, abnormal gait, slurred speech, difficulty with swallowing, cognitive impairment and personality changes. **IT IS A DOMINANT TRAIT.**

Francis knows that her mother has Huntington disease. She deduces that at least one of her maternal grandparents was a sufferer.

- Is her reasoning correct?
- She also deduces that at least one of her great-grandparents was a sufferer. Is she correct?
- Francis draws the family tree found below. Many of the circles and squares are left white because the genotype cannot be determined. Is it possible for Francis not to develop the disease?



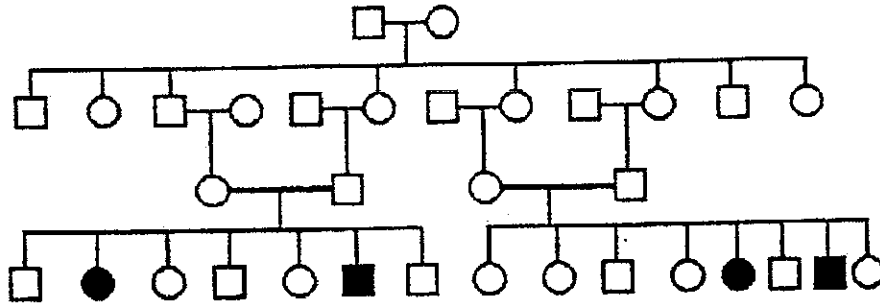
Part IV - The Blue People Of Troublesome Creek

Six generations after a French orphan named Martin Fugate settled on the banks of eastern Kentucky's Troublesome Creek with his redheaded American bride, his great-great-great great grandson was born in a modern hospital not far from where the creek still runs. The boy inherited his father's lankiness and his mother's slightly nasal way of speaking. What he got from Martin Fugate was dark blue skin. "It was almost purple," his father recalls.

Doctors were so astonished by the color of Benjy Stacy's skin that they raced him by ambulance from the maternity ward in the hospital near Hazard to a medical clinic in Lexington. Two days of tests produced no explanation for skin the color of a bruised plum. A transfusion was being prepared when Benjy's grandmother spoke up. "Have you ever heard of the blue Fugates of Troublesome Creek?" she asked the doctors. "My grandmother Luna on my dad's side was a blue Fugate. It was real bad in her," Alva Stacy, the boy's father, explained. "The doctors finally came to the conclusion that Benjy's color was due to blood inherited from generations back."

Benjy lost his blue tint within a few weeks, and now he is about as normal looking a seven-year-old boy as you could hope to find. His lips and fingernails still turn a shade of purple-blue when he gets cold or angry a quirk that so intrigued medical students after Benjy's birth that they would crowd around the baby and try to make him cry. "Benjy was a pretty big item in the hospital," his mother says with a grin. Dark blue lips and fingernails are the only traces of Martin Fugate's legacy left in the boy; that, and the recessive gene that has shaded many of the Fugates and their kin blue for the past 162 years.

Given below is a pedigree of some of the blue people of Troublesome Creek



- The blue trait is inherited as an autosomal recessive trait which means that affected individuals have _____ copies of a mutant gene.
- Indicate which individuals are carriers of the "blue" gene by placing the letters Bb next to their symbol.
- What is the relationship of the parents of the "blue" children?
- What is the probability that the unaffected siblings of the "blue" children are carriers for the "blue" trait?
- What warning does marrying close relatives does this illustrate?



E - Bio Worksheet

Pedigree Analysis In Genetics

Background Reading – pedigree analysis Nelson Biology, Campbell Biology

Purpose - To introduce students to the role of pedigree charts in tracing human traits.

Introduction

Pedigree charts are often constructed to show the inheritance of genetic conditions within a family. Such charts are a great help in determining whether a phenotype is controlled by a dominant, recessive or sex-linked allele. In future activities you will see how gel electrophoresis (DNA fingerprinting) is being used in pedigree analysis. This activity will introduce you to autosomal pedigree charts with sex-linkage being undertaken in a future activity.

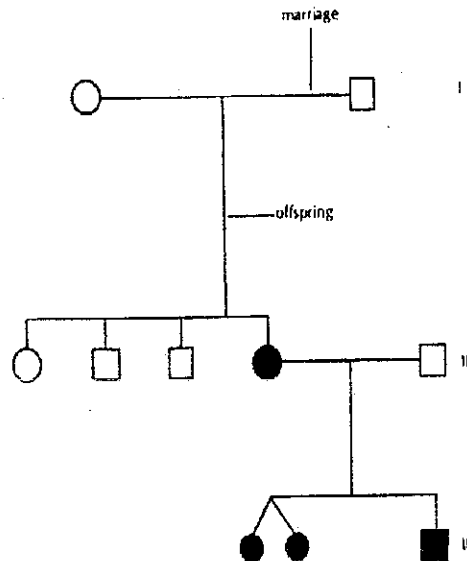
Part I - Understanding The Pedigree

1. **Table I** shows the symbols needed in understanding a pedigree. In **FIGURE I** Generation I is made up of grandparents, Generation II is their children and Generation III is their grandchildren.

Table I - Pedigree Key

Symbol	Meaning
○	Female without trait
□	Male without trait
●	Female with trait
■	Male with trait
●	Female, died in infancy
■	Male, died in infancy
⊙ ⊙	Identical twins

Figure I - Sample Pedigree

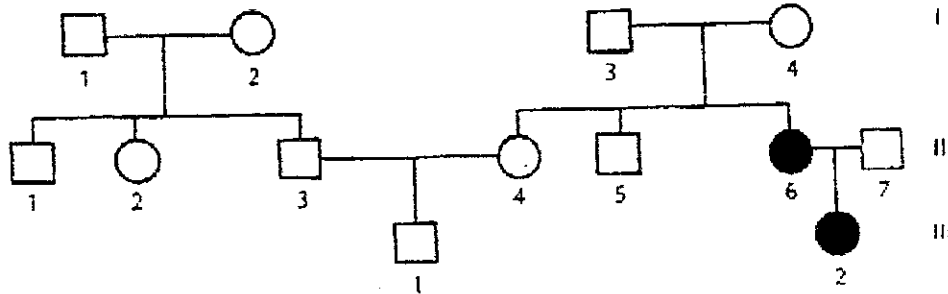


- Q1 - How many normal males are represented in Figure I ? Normal females ?
- Q2 - How many children did the grandparents have ?
- Q3 - How many affected individuals are present ?

Part II - Pedigree Analysis

1. Individuals who lack an enzyme needed to form the skin pigment melanin are called albinos. Normal skin pigmentation is dominant. Use **N** to represent the gene for normal and **nn** to represent the genotype for albinism. If you cannot determine if the dominant trait is heterozygous or homozygous, use **N_**. Refer to **FIGURE II** and identify the genotype of each individual. **Draw a chart listing the individuals and their genotypes.**

FIGURE II - ALBINISM PEDIGREE

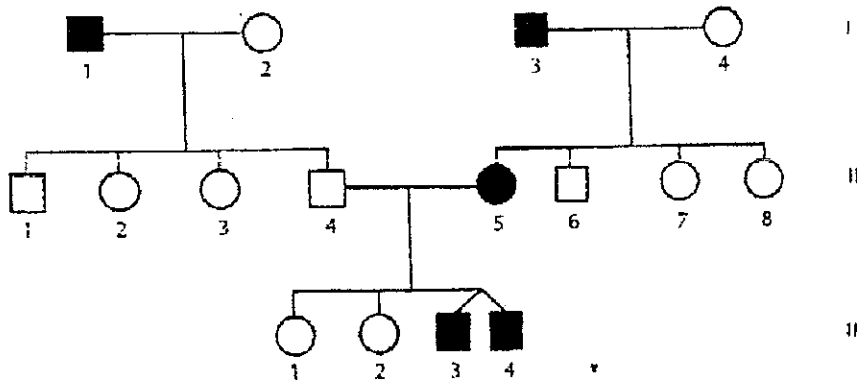


Q4 - How many individuals had the genotype **Nn** ? How many were **N_** ?

Q5 - Using a Punnet square predict the probability of the grandparents having albino children.

2. The following pedigree demonstrates the ability to taste PTC paper. The ability to taste is a dominant trait and is represented by the letter **T**. Nontasters are represented by **tt** and uncertain genotypes as **T_**. Refer to **FIGURE III** and identify the genotypes as you did in the previous pedigree.

Figure III - PTC Tasting Pedigree



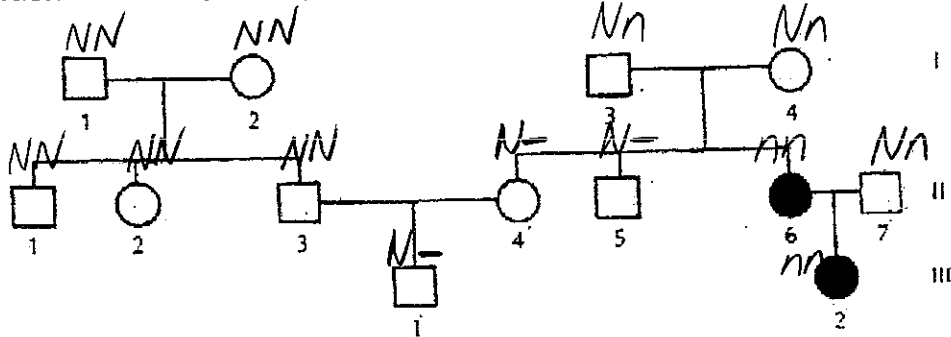
Q6 - How many individuals are heterozygous? How many are homozygous ?

Q7 - What is the probability of grandparents 3 and 4 having nontaster offspring ?

Part II - Pedigree Analysis

1. Individuals who lack an enzyme needed to form the skin pigment melanin are called albinos. Normal skin pigmentation is dominant. Use **N** to represent the gene for normal and **nn** to represent the genotype for albinism. If you cannot determine if the dominant trait is heterozygous or homozygous, use **N_**. Refer to **FIGURE II** and identify the genotype of each individual. **Draw a chart listing the individuals and their genotypes.**

FIGURE II - ALBINISM PEDIGREE



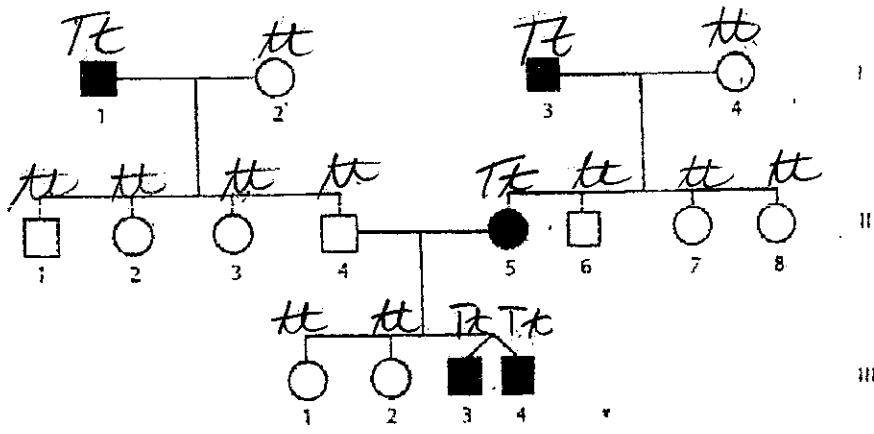
*N - normal
n - albino*

Q4 - How many individuals had the genotype **Nn**? How many were **N_**? **3, 3**
 Q5 - Using a Punnet square predict the probability of the grandparents having albino children.

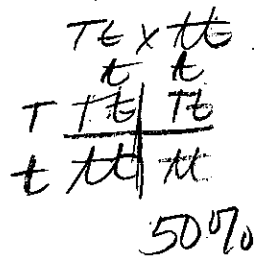
2. The following pedigree demonstrates the ability to taste PTC paper. The ability to taste is a dominant trait and is represented by the letter **T**. Nontasters are represented by **tt** and uncertain genotypes as **T_**. Refer to **FIGURE III** and identify the genotypes as you did in the previous pedigree.

Figure III - PTC Tasting Pedigree

*T - taster
t - non taster*



Q6 - How many individuals are heterozygous? How many are homozygous?
 Q7 - What is the probability of grandparents 3 and 4 having nontaster offspring?



Q5
 G.P. 1+2
 NN
 NN | NN
 NN | NN
 0%
 G.P. 3+4
 Nn
 NN | Nn
 Nn | nn
 25%

Part III - Trait Analysis

1. The following pedigrees will be used to determine whether the trait is autosomal dominant or autosomal recessive. In tracing autosomal alleles, if both parents have the disorder and the offspring do not, the condition is autosomal dominant. If neither parent shows the disorder but some of their children do, the condition is autosomal recessive. A carrier is an individual who appears to be normal, but who is capable of passing on a gene for the disorder. If the characteristic is dominant, there can be no carriers because only a single gene is needed to show the disorder. **Table II** provides some keys for your answers.

Table II - Pedigree Keys

CHARACTERISTIC	KEY
AUTOSOMAL DOMINANT	AA = AFFECTED Aa = AFFECTED aa = NORMAL
AUTOSOMAL RECESSIVE	AA = NORMAL Aa = CARRIER aa = AFFECTED

Figure IV - Pedigree 1

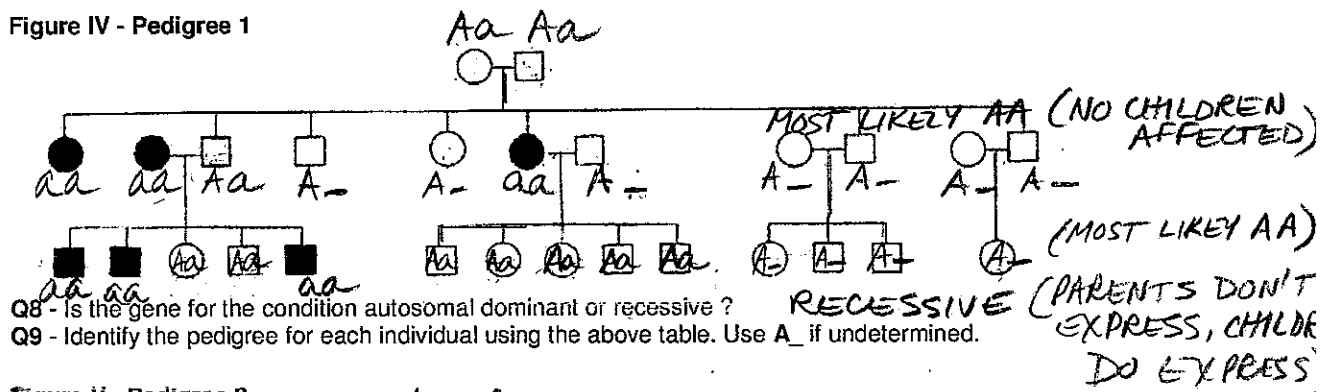
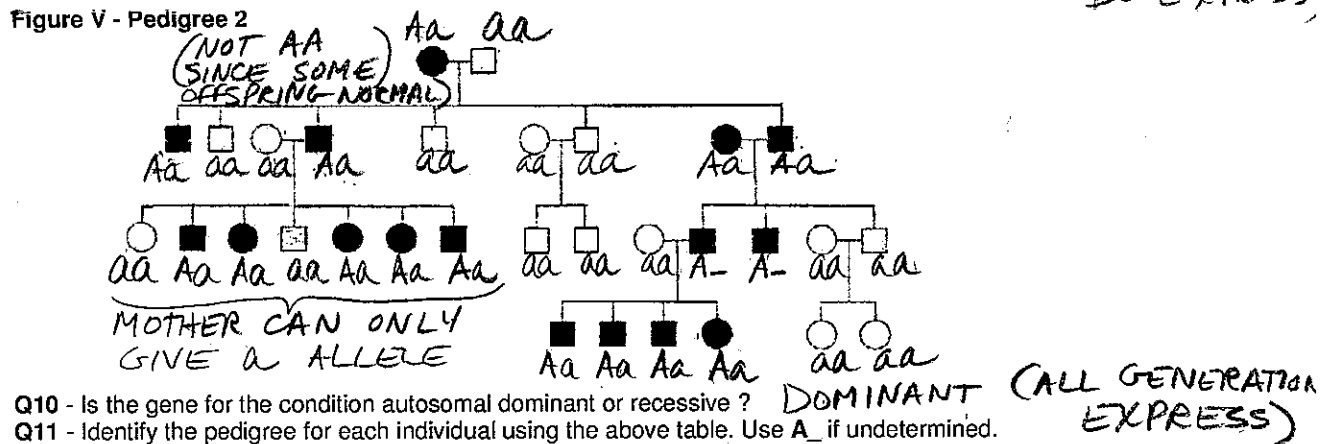


Figure V - Pedigree 2

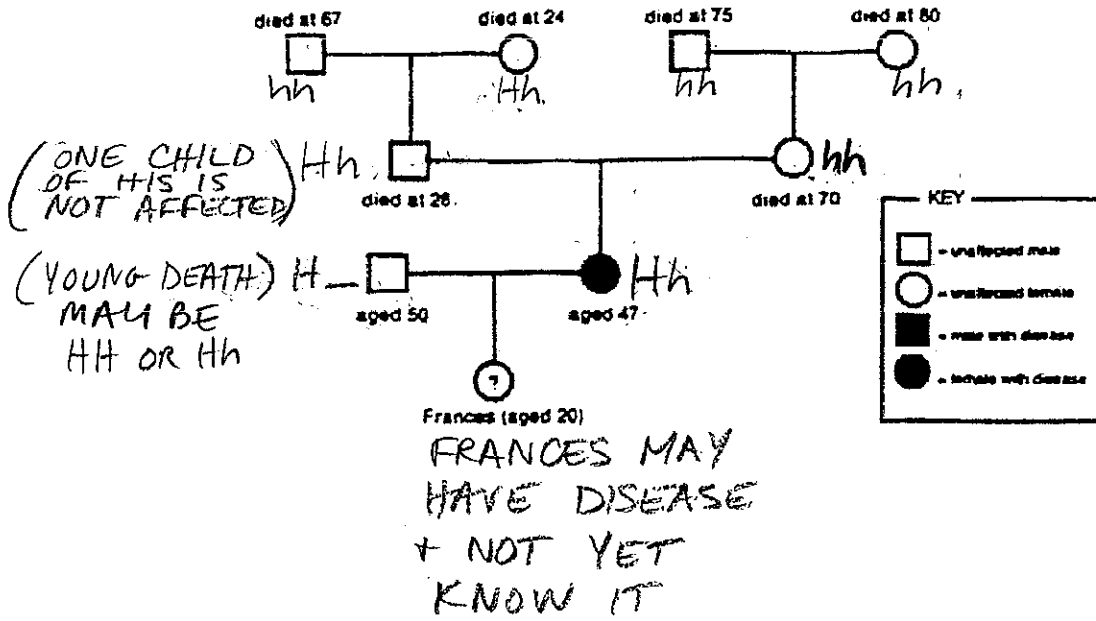


Part V - Huntington's Disease

Huntington disease (HD) is an inherited brain disorder affecting the nervous system. It causes progressive deterioration of physical and mental capabilities, leading ultimately to severe incapacitation and eventual death, generally 15-25 years after onset. Primarily, it affects adults, usually appearing between the ages of 30 and 45. Occasionally, HD symptoms appear earlier (before age 20, the juvenile form) or later (after age 50.) Common symptoms of adult-onset HD are involuntary movements, abnormal gait, slurred speech, difficulty with swallowing, cognitive impairment and personality changes. **IT IS A DOMINANT TRAIT.**

Francis knows that her mother has Huntington disease. She deduces that at least one of her maternal grandparents was a sufferer.

- (A) Is her reasoning correct? **YES, FATHER DIED AT AGE 28**
- (B) She also deduces that at least one of her great-grandparents was a sufferer. Is she correct? **YES PATERNAL GR. GRANDMA DIED AT 24**
- (C) Francis draws the family tree found below. Many of the circles and squares are left white because the genotype cannot be determined. Is it possible for Francis not to develop the disease?



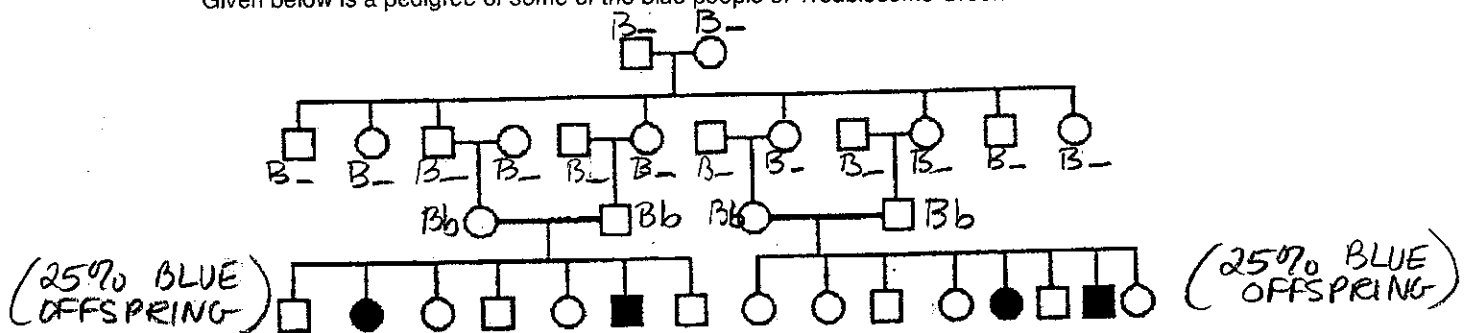
Part IV - The Blue People Of Troublesome Creek

Six generations after a French orphan named Martin Fugate settled on the banks of eastern Kentucky's Troublesome Creek with his redheaded American bride, his great-great-great grandson was born in a modern hospital not far from where the creek still runs. The boy inherited his father's lankiness and his mother's slightly nasal way of speaking. What he got from Martin Fugate was dark blue skin. "It was almost purple," his father recalls.

Doctors were so astonished by the color of Benjy Stacy's skin that they raced him by ambulance from the maternity ward in the hospital near Hazard to a medical clinic in Lexington. Two days of tests produced no explanation for skin the color of a bruised plum. A transfusion was being prepared when Benjy's grandmother spoke up. "Have you ever heard of the blue Fugates of Troublesome Creek?" she asked the doctors. "My grandmother Luna on my dad's side was a blue Fugate. It was real bad in her," Alva Stacy, the boy's father, explained. "The doctors finally came to the conclusion that Benjy's color was due to blood inherited from generations back."

Benjy lost his blue tint within a few weeks, and now he is about as normal looking a seven-year-old boy as you could hope to find. His lips and fingernails still turn a shade of purple-blue when he gets cold or angry a quirk that so intrigued medical students after Benjy's birth that they would crowd around the baby and try to make him cry. "Benjy was a pretty big item in the hospital," his mother says with a grin. Dark blue lips and fingernails are the only traces of Martin Fugate's legacy left in the boy; that, and the recessive gene that has shaded many of the Fugates and their kin blue for the past 162 years.

Given below is a pedigree of some of the blue people of Troublesome Creek



- The blue trait is inherited as an autosomal recessive trait which means that affected individuals have 2 copies of a mutant gene.
- Indicate which individuals are carriers of the "blue" gene by placing the letters Bb next to their symbol.
- What is the relationship of the parents of the "blue" children? **THEY ARE CARRIERS OF GENE**
- What is the probability that the unaffected siblings of the "blue" children are carriers for the "blue" trait? **50% (PARENTS ARE Bb x Bb)**
- What warning does marrying close relatives does this illustrate? **DEFECTIVE GENES ARE MORE LIKELY TO BE EXPRESSED (DON'T DO IT!!)**