

THE BLUE PEOPLE OF TROUBLESOME CREEK

They're known as the "**blue people**" in the hills around Troublesome Creek. Most blue people live long lives with blue skin. But, for some blue people there was a pain not seen in lab tests. That was the pain of being blue in a world that is mostly shades of white to black.

There was always guessing about what made the blue people blue: heart disease, a lung disorder, etc. But no one knew for sure, and doctors rarely visited creek-side settlements where most of the "blue Fugates" lived. By the time a young doctor from the University of Kentucky came down to Troublesome Creek in the 1960s to cure the blue people, Martin Fugate's descendants had multiplied their genes all over the area.

"They were bluer than ever," Cawein, the doctor, says. "I started asking them questions: 'Do you have any relatives who are blue?' then I sat down and we began to chart the family" Cawein said. Cawein remembers the pain in their faces. "They were really embarrassed about being blue," he said. "You could tell how much it bothered them to be blue."



After ruling out heart and lung diseases, the doctor suspected methemoglobinemia, a rare hereditary blood disease that results from high levels of methemoglobin in the blood. Methemoglobin, which is blue, is a mutated form of the red molecule that normally carries oxygen in blood. Methemoglobin is the color of oxygen-poor blood seen in blue veins just below the skin. The blue people's blood had accumulated so much of the blue molecule that it showed through their skin, making their whole body as blue as their veins.

Cawein concluded that the condition was *inherited as a simple recessive trait*. In other words, to get the disorder, a person would have to inherit two alleles for it, *one from each parent*. Somebody with only one gene would not have the condition but could pass the gene to a child (a carrier).

Before Cawein ended his study of the blue people, he returned to the mountains to patch together the family history of Martin Fugate's recessive gene. From a history of Perry County and some Fugate family Bibles listing ancestors, Cawein came up with a fairly complete story.

A Story of the Blue Fugates

Martin Fugate was an orphan who moved to Kentucky in 1820 to find a place to live near Troublesome Creek. Family stories say that **Martin himself was blue**. The odds were very much against it, but **Martin Fugate** managed to find and marry a woman who carried the same **recessive gene**. His wife, **Elizabeth**, carried one copy of the blue gene, but her skin was white as snow.

Martin and Elizabeth began a family. Of their seven children (three girls and four boys), they had two blue daughters and two blue sons. **Zach** was one of their favorite sons, and he was blue! He married **Rachel**, *his mother's sister*, and they had three non-blue children—one boy and two girls.

Levy, a non blue, was one of **Zach's sons**. **Levy** married a girl named **Sarah** and bought a large farm. The couple had three children, including **Luna**, the only blue. As it turns out, the girl that Levy married was had Fugate blood in her family four generations back. In fact her great grandfather's brother was Martin Fugate's Grandfather!

A fellow by the name of **John** spotted **Luna** at Sunday services of the Old Regular Baptist Church back before the century turned. **John** dated her and married her. **Luna** was healthy, like most blue people, and had three kids, *none of which were blue*. Her son **Steve** married a local girl named **Susie**, and they had three non-blue kids, including **Alva**. As coal mining and the railroads changed Kentucky, the blue people started moving around and marrying other people. The strain of inherited blue began to disappear as the recessive allele spread to families where it was unlikely to be paired with another copy of the allele.

Not too long back **Alva** found himself a bride in a local woman named **Hilda** and the two had a son named Benjy. You can only imagine how shocked they were when their little son **Benjy** popped out blue!! As it turns out, Hilda has Fugate blood in her from generations back.

YOUR TURN: On the back flap of this paper, create a pedigree for the Blue Fugates.

Directions: Answer the following questions based on the Pedigree of the Fugate Family.

1. How many individuals have blue skin in the seven generations shown in the pedigree?
2. If in generation VI, individual 2, married someone who is not a carrier of the blue people allele, what is the probability that their kids would be blue?

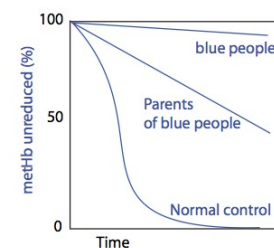
Complete a Punnett Square to support your answer. (Use the letter B)

3. Which of the following is true about people with blue skin?
 - a) the people are homozygous dominant
 - b) the people are homozygous recessive
 - c) the people are heterozygous
 - d) the people are carriers
4. Looking at this pedigree, how can you tell that blue skin is a recessive trait?
5. What are the genotypes of the parents in the first generation?
6. Why are all of Martin and Elizabeth's kids either carriers or blue people? Use a Punnett Square to support your answer.

7. What are the three lines on the graph in Figure 2 telling us?

The graph below in Figure 2 shows enzyme activity over time in people that are blue, people that are not blue but may have blue children, and people that are not blue and never have blue children.

Figure 2. RBC NADH Diaphorase Activity



Red blood cell extracts from three different groups assayed for ability to reduce methHb to Hb.

8. Provide genotype and phenotype designations for each line on the graph in Figure 2.