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From the Files of BSKI Genetic Counseling Service.

Welcome to the files of the BSKI genetic counseling service. The following is a case study along with background information that you, as a genetic counselor, need to know in order to chart the pedigree of the following family.

This is a story of Tanya and Tim Turkie, and their family. Tanya first made contact with our service a week after she successfully delivered Tom, their son. Tanya was concerned since they received notification that Tom tested positive for phenylketonuria. She wanted to know what it was and if there was a treatment for the disease.

PKU or phenylketonuria is a autosomal recessive disease that affects an enzyme, phenyl hydroxylase. In PKU, not enough of this enzyme is produced. The enzyme is necessary to convert phenylalanine (an amino acid) into tyrosine (another amino acid). In PKU people, phenylalanine starts to build up and prevents the normal brain and central nervous system development.

PKU happens in 1 out of 41,000 people in the U.S. If PKU is undetected, it can lead to severe mental retardation, which is not reversible. However, if PKU is detected within the first 30 days of life, then the child can be put on a phenylalanine free diet and can lead a normal healthy life.

Tim and Tanya were relieved that their other children had not come down with PKU.

Question 1: What are the odds that the other two normal children will be carriers.

Question 2: What are the odds that Tim and Tanya's next child will have the disease?

Six weeks after that incident, BSKI received another call from Tanya. Tony, their 8 year old son, had broken his hand at school. In treating Tony, the hospital discovered that he had hemophilia. As you know, hemophilia is a sex-linked trait that is recessive.

In all sex-linked traits, the gene that is associated with hemophilia is only found on the X chromosome. Since this is the case, then the gene is never passed on from the father to the son and the incidence of the trait is much higher in males than females.

In hemophilia, the female is usually the carrier, and the males are the ones that show the symptoms. Hemophilia is a blood disorder in which the blood fails to clot due to a protein deficiency. A person with hemophilia will need medical attention for external and internal injuries. A simple bruise can prove fatal.

Tom was immediately tested for the disease but was negative. Tisha, their 4 year old daughter, also negative. Since Tim never showed any signs for the disease, BSKI showed concern toward Bitsy Blake, Tanya's sister. Bitsy had 2 children both females (Betty, and Betsy) and was pregnant with a third. The clinic wanted to contact Bitsy, but Tanya was still bitter over a broken dinner engagement and refused the clinic the needed permission.

Question 3: Why was BSKI concerned about the Blake's daughters?

While all this was taking place, Bitsy Blake made an appointment to see a counselor at BSKI. Bitsy had an amniocentesis to confirm the sex of her child. It seemed that her husband, Bob, really wanted a boy and had threatened to walk out on her if she had a girl.

Question 4: Who's fault is it if the Blake's have another girl?

Three months later, Tanya Turkie called again. Tim had died due to the fact that his aorta had exploded. It seems that Tim had Marfan Syndrome, and she was very concerned about the chance that her children would have it. You see, Tim's family had a history of Marfan Syndrome. Marfan Syndrome is an autosomal dominant disorder that strikes 1 out of 20,000 people each year. The defective gene is believed to form a weaker protein that makes connective tissue weaker than usual. Connective tissue is the material that holds the body tissues together. People with Marfan are usually tall, slender, and loose-jointed. They are prone to lung collapses. Their heart and heart valves are affected to the point that they have a heart murmur. The aorta, which is the largest blood vessel in the body, is also affected. A defective aorta can split and has caused instant death.

Tim's mother remarried after Tim's father ran out on her 10 years ago. As of now, there is no way to trace the genes of the father's family. Tim had a brother Dirk who shared Tim's father. Tim's mother, Francine, remarried a man named Mark Fields. Mark and Francine had four more children: Glen, Mary, April and Ted.

BSKI contacted the Fields family. Glen died at the age of 21 in a bizarre gardening accident. Mary entered the nunnery while Ted entered the monastery. Both had taken a vow of isolation and could not be tested. April and Francine both tested negative. Based on this information, it seems that Tim's father supplied the Marfan Syndrome gene to the family.

Question 5: What is the chance that Tony and Tisha may have Marfan Syndrome?

After Tim's death, Tanya had a major break down and made a confession. It seems that she had an affair before Tisha was born. She did not know if Tim was the father of the child. Tim had type A blood while Tanya had type B blood. Tim's father and mother were blood type AB. Tisha was tested and she had type B blood.

Question 6: Was Tisha Tim's child?