Why Is He Different from Both Parents?

The Genetics of ABO Blood Types

by

Jun Liang, Science Department, Borough of Manhattan Community College / City University of New York
William J. Rice, Simons Electron Microscopy Center, New York Structural Biology Center

Part I – Human ABO Blood Type

“But I already know all this stuff! I’ll do well on the test tomorrow, I promise,” begged Kevin. He really wanted to go out that night with his friend, but his mother had other ideas.

“No, absolutely not. You’re going to stay home and study; tomorrow’s test is very important,” his mother replied.

Kevin ended up staying home that night, reluctantly.

The next day Kevin met his best friend Peter. “How’s it going?” asked Peter.

“Not so good. We had a test today. I guess it went all right,” said Kevin.

“How’s your stomach doing? Is it still sore?” Peter asked.

“Oh, it’s better now. You know, there’s something weird going on. Last time I was in the hospital, neither of my parents could donate blood to me. The doc said they aren’t a match for my blood type.”

“They don’t match? What do you mean?” Peter asked.

“Well, the doc said I’m type O positive, my mom is type A positive, and my dad is type B negative. We’re all different, and I don’t get it. They’re my parents—I mean, I should inherit my blood type from at least one of them, if not both, right?”

“That’s a good point,” Peter agreed. “Well, I’ve got a very cool science teacher, Ms. B. She would know better than us. Let’s ask her. She has a class tomorrow. We can meet her afterward. What do you say?”

The next day, Kevin and Peter met Ms. B. and explained Kevin’s puzzle about his family’s blood types.

“Ah, good question,” replied Ms. B. “Let me explain how blood types work so that it will help you to understand why your parents couldn’t donate blood to you and how you can all have different blood types in one and the same family.”

“The ABO blood group system is one of the most important systems that hospitals use to identify individuals for blood transfusion. ABO stands for the type of carbohydrate that sits on the surface of red blood cells; it’s this carbohydrate that allows cells to recognize themselves. They are also called surface markers. Positive (+) refers to the presence of Rhesus (Rh) factor. Some individuals lack Rh factor, and they are negative (−). Rh factor is a predominant protein surface marker on red blood cells.”

“Interesting. What is carbohydrate exactly? The stuff we eat, like bread and sugar?” Peter asked.

“Yes. Sugar is a carbohydrate and bread is made of lots of different carbohydrates. Red blood cells may have carbohydrate A on the cell surface, which results in type A blood; but if they have carbohydrate B, then they become type B blood. Some red blood cells have both carbohydrate A and B, which makes them have type AB blood. When neither carbohydrate A nor B is present on the red blood cell surface, the blood type is considered to be type O.”

That made sense to Peter, who then asked “How do red blood cells get those carbohydrates?”
“That’s a great question. The types of carbohydrate on the red blood cell surface are controlled by genes. In this case, the gene that controls carbohydrate types presented on red blood cell surface encodes enzymes. Enzymes are proteins that catalyze chemical reactions,” Ms. B. continued. “There are two types of such enzymes: enzyme A and enzyme B, which deposit carbohydrate A and carbohydrate B respectively on the red blood cell surface. The three different versions of the gene are called alleles. Allele A codes for enzyme A; Allele B codes for enzyme B; Allele O doesn’t code for a functional protein.”

“Oh, then I must have O alleles since I’m type O!” Kevin was excited to figure out the alleles he had in his body.

Questions

1. What is the human ABO blood group system?
2. What types of biochemicals are surface markers on red blood cells?
3. How many blood phenotypes are there in the ABO blood group system?
Part II – Genetic Inheritance

Ms. B. continued her explanation. “Everyone has two alleles for each gene in their cells. We all inherited one allele for every gene from each of our parents. Since your mom is type A, she could have two copies of allele A or one copy of allele A and one copy of allele O. In either case, she is type A. Your father could …”

“His father could have two copies of allele B or one copy of allele B and one copy of allele O. Either way, he is type B,” Peter said.

“Yes, you’re getting smarter,” smiled Ms. B. “Keep going. Think. If Kevin’s mom has one copy of allele A and one of allele O, and his dad has one copy of allele B and one of allele O, then what possible blood types will their children have?”

“Oh, let me think,” Peter and Kevin said simultaneously.

Questions

1. What is genetic inheritance?
2. What is genetic variation?
3. Among the three alleles of human ABO blood group, which is/are dominant, which is/are recessive?
4. What is homozygous? What is heterozygous? Are Kevin’s parents homozygous or heterozygous for the A or B allele if Ms. B’s assumption is true?
5. Predict the possible blood types of their children from the marriage of Kevin’s parents. Use a Punnett square to solve this problem.
6. What are the probabilities (%) of the children’s possible blood types?
Part III – Blood Transfusion Reaction

“Ok, I get the part that we could have different blood types. So then, how come they couldn’t donate blood to me?” continued Kevin.

“Well, carbohydrates are located on the red blood cell surface; we call them antigens. Meanwhile, corresponding antibodies are created in the blood serum. Antibodies are specific proteins made in the body to bind and react with specific antigens. It’s like a key to a lock. One key can open one lock. One antibody can only react with one antigen. The antibodies won’t react with the body’s own antigen present on red blood cells, but will react with that of incompatible blood from another person. This is part of your normal defense system called the immune response. Have you guys heard of it?”

“Yes,” said Kevin and Peter.

Ms. B continued, “Well, during a blood transfusion, if the donor’s and recipient’s blood are not compatible with each other, the recipient’s immune response will attack the donor’s blood cells and damage them. This is called a transfusion reaction.”

“Oh, that’s serious; I heard of that when I was hospitalized. What types of antibody does everyone have?” asked Kevin.

“Type A blood has antibody B which will react with type B red blood cells; type B blood has antibody A. Type AB blood contains neither antibody; and finally type O blood, which Kevin has, contains both antibodies A and B. Type O blood will react with both type A and type B blood. Since one of your parents is type A and the other is type B, neither of them can donate blood to you.”

“That’s a complicated system,” said Kevin. “But wait a minute, wouldn’t that be a problem while my mom was pregnant with me? I heard that a mother shares everything with her baby before it’s born.”

“Another good question. As a matter of fact, your mother’s blood cells would not cross the placenta so there was no problem. You are fine,” she said.

After their discussion with Ms B, the boys returned home. Mom welcomed Kevin at the door.

“Hi, Kevin. You’re back. How did your test go?”

“Ok. I’m glad that I stayed at home last night. One of questions I studied was on today’s test,” said Kevin.

“Great, I’m glad!” Mom’s eyes were full of happiness.

“What’s for dinner?” Kevin headed to the kitchen.

“Oh, today we are going to have your favorite, pasta with spicy meatballs, and …” said Mom.

Questions

1. What types of antibody are made by each ABO blood group?
2. What is a transfusion reaction?
3. If a patient has type A blood, what would be the donor’s blood type during blood transfusion: type A, type B, type AB, or type O? Why?
4. Some individuals are called universal donors for blood transfusions. What blood type would they be? Explain your answer in terms of antibodies and antigens.
5. Other individuals are called universal recipients for blood transfusions. What would be their blood type? Explain your answer in terms of antibodies and antigens.
Homework

1. What are the enzymes encoded by allele A and allele B in human ABO blood group? What chemical reaction does each enzyme catalyze?

2. Tom with type AB blood marries Anna. They have two children: Eric with type B blood and Jessica with type A blood. Anna's father Paul has type O blood, and her mother Elizabeth has type A blood.
   a. What are the genotypes of Tom and Paul?
   b. What are the possible genotypes and phenotypes of Anna?
   c. What is the genotype of Eric?
   d. Is it possible to define Jessica's genotype and phenotype based on the information provided? Why or why not?

3. (Optional) If possible, identify the ABO blood type genotypes of your family members based on what you have learned. Provide details to support your findings.