

Chapter 12c

Topic: Multiple alleles, multiple genes

Main concepts:

- In classical Mendelian genetics, each gene has two possible alleles. However, some genes have more than two alleles. In human blood typing (A, B, AB, and O), the gene for the blood type protein has three alleles (A, B, and O). One eye color gene in fruit flies has many alleles.
- However, sometimes for practical purposes, multiple alleles may be classified together as “dominant” or “recessive.” For example, there are over 900 known mutations that cause cystic fibrosis, so technically there are over 900 alleles. However, all mutations that cause a “broken” gene (codes for a non-functional protein) are considered “recessive,” since the presence of one intact (dominant) gene is enough to make a person function normally.
- Human blood types are determined by proteins on the surface of the red blood cells. Alleles A and B, for A-type and B-type glycoproteins, are co-dominant; that is, a person who inherits a A allele from one parent and a B allele from the other parent will have type AB blood. The o allele is recessive. The o allele produces no glycoproteins. Thus a person with the genotype Ao will make some type A glycoproteins, and have type A blood. A person with the genotype oo will make neither the A-type nor the B-type glycoproteins, and will have type O blood.
- Blood donation matching takes these identifying glycoproteins into account when matching blood types. A person with type A blood can receive type A blood, but will make antibodies against type B blood because the B-type glycoproteins will appear foreign. The type A person can also receive type O blood, because there are no foreign glycoproteins on the type O blood cell. But a person with type O blood can only receive type O blood, because both the A-type and B-type glycoproteins would be attacked by their immune system.
- Most human traits are controlled by several genes. Some, such as skin color, eye color, and hair color, are controlled by multiple copies of the same gene. In skin color, for example there are several pairs of genes that code for the pigment melanin. The more copies of the dominant allele a person has, the darker their skin. Some traits, such as human height, are controlled by the activities of many different genes.
- The environment can affect how genes are expressed. Gene-environment interactions affect many aspects of our physical being and our lives. For example, height is controlled by several different genes, but a person who does not get enough food in childhood may not grow to the full height that their genes would otherwise cause them to grow. Another example: people may have a genetic predisposition toward developing Type II diabetes, heart disease, or cancer, but can reduce their chances of developing these diseases by their choices in diet and lifestyle.

Common misconceptions:

- Because Mendelian inheritance is used as the first example in most textbooks, many students get the impression that all traits are controlled by single genes. Most traits, however, are influenced by multiple genes and by the environment, and often the interaction is complex.
- You may have heard that “brown eyes are dominant over blue eyes,” and even textbooks use eye color in this way. However, eye color is controlled by several pairs of melanin-producing genes, and as a result, human eyes come in a range of shades of blue and brown, as well as green, gray, and hazel.

Reading notes:

- Define what is meant by “multiple alleles” as it is used in this reading.
 - List the human blood types (phenotypes) and the genotypes that produce them. State what it is that the blood type gene codes for. (Note: the Rh factor, which makes a blood type positive or negative, is yet another gene).
 - List some examples of polygenic inheritance.
 - Explain what pleiotropy is.
 - List several examples of how the environment can influence how genes are expressed.
-

Problems

1. Mr. and Mrs. Smith have a daughter, Samantha. Mr. Jones, their neighbor, is suing for custody of the child, claiming that he had an affair with Mrs. Smith and that Samantha is his daughter. The judge in the case orders blood tests to determine blood types of all the people involved. The results are: Mr. Smith: Type AB; Mrs. Smith: Type B; Mr. Jones: Type A; Samantha: Type O. Is it possible that Mr. Jones could be Samantha's father?
2. What if Samantha had type AB blood? Who could be her father in that case?
3. Kim's father, Alfred, is rushed to the hospital after a serious car accident, and needs a blood transfusion. Kim offers to be tested to see if she can be a donor. Kim has type A blood. Her father is type O. Kim is surprised, and wonders if she might be adopted and didn't know. Can Alfred be her father?
4. Human eye color inheritance is still not fully understood, but at one point it was believed to be caused by four pairs of melanin-producing genes. There are nine colors of eyes: light blue, medium blue, dark blue (violet), gray, green, hazel, light brown, medium brown, and dark brown. If light blue is caused by having all recessive eye color alleles (aabbccdd), and dark brown by having all dominant alleles (AABBCCDD), give at least one combination of alleles that would produce the other colors.

Useful websites:

- "How are human eye colors inherited?" <http://www.athro.com/evo/gen/inherit1.html> is a discussion of some of the hypotheses around human eye color.
- "Blood types tutorial" http://www.biology.arizona.edu/Human_Bio/problem_sets/blood_types/Intro.html has a guided tutorial and problems involving human blood type, including Rh factor.
- "Human blood types" <http://waynesword.palomar.edu/aniblood.htm> has animations showing a simple test for blood types, and a good explanation of the biology of blood types.

Answers:

1. Consider the phenotypes and possible genotypes of the people in this case:

person	phenotype	genotype
Samantha	Type O	oo
Mr. Smith	Type A	AA or Ao
Mrs. Smith	Type B	BB or Bo
Mr. Jones	Type AB	AB

Samantha had to get one o allele from each of her parents. Mrs. Smith must have a genotype of Ao. Mr. Jones, with his genotype of AB, cannot have given Samantha her other o allele. This does not prove that Mr. Smith is the father, but does show that Mr. Jones is not.

2. If Samantha has type AB, she had to get an A allele from one parent and a B allele from the other. Mrs. Smith can only give her a B or an o, so she must be the source of the B allele. Either Mr. Smith or Mr. Jones could have given her an A allele.
3. light blue: aabbccdd gray: AABbccdd dark brown: AABBCCDD
medium blue: Aabbccdd green: AABBCcdd light brown: AABBCCdd
dark blue: AAbbccdd hazel: AABBCcdd medium brown: AABBCCDd

Note: Eye color inheritance may be more complex than this. Current hypotheses suggest two separate genes, one with brown and blue alleles, the other with green and blue alleles, and possible incomplete dominance involved.
