FREQUENTLY ASKED QUESTIONS - GENETICS 8.5

Here are some questions that might arise while teaching the genetics unit, and some suggested responses.

1. What is the XY chromosome on the karyotype for? I heard XY were boys and XX were girls. Is this true?"

In humans, we have two distinct characteristics, biological sex and gender. Biological sex is determined by an XX/XY system. However, it is really the SRY gene on the Y chromosome that matters most here. A functional SRY gene, once activated, creates testosterone and anti-mullerian hormone, which typically causes the development of a male reproductive system. Without a Y chromosome and functional SRY, an XX embryos develop into a female. While SRY is central, there are many other genes involved in influencing maleness. When SRY is mutated and produces a non-functional protein, the individual presents externally as female (and is usually raised as a female, that is, has a female gender identity), but has XY and underdeveloped ovaries (therefore the individual cannot bear children). There are several other systems for determining sex, some genetic (like XX/XY) and some environmental, like the effect of temperature on sex of reptiles.

One important point to note is that while *biological sex* is genetically determined, *gender* is not. Gender is a social construct that refers to how individuals position themselves and are positioned by others in relation to roles as female or male. There are more gender identities than just male or female. Gender definitions, expectations, and roles vary in different cultures, with some cultures embracing multiple gender definitions.

2. If Myostatin stops cells from turning into muscle cells, how do we have muscles at all?

Myostatin is one of many, many proteins involved in muscle development, and there are several pathways that interact with the myostatin pathway. Myostatin can also be turned on or off (gene expression) and thus the organism can regulate when myostatin is present. Moreover, there are natural protein inhibitors of myostatin in the blood, and they can also be upregulated (producing more) or downregulated (producing less).

3. If Mendelian genetics is problematic, why are we still teaching it (and why is it in the NGSS)?

Mendel's discoveries of independent assortment and segregation and the idea that we get two variants of each gene (allele) from our parents are relevant to understanding inheritance. However, the discrete phenotypes of simple Mendelian inheritance patterns and their typical ratios are fairly rare. That is, Mendelian inheritance patterns provide a simple model that can explain some rather unique variations in some specific traits. For example, we know that mutations in genes that produce proteins can result in specific phenotypes, and these are largely predictable (e.g. known mutations that cause Cystic Fibrosis). Such knowledge is useful and helpful. However, even in the cases of known genetic disorders, not all genotypes result in the expected (predicted) phenotypes. For example, there are mutations in the CF gene that result in very mild symptoms, while other mutations result in severe symptoms. Moreover, sometimes the same mutation can result in different severity of symptoms in different individuals (Cooper, Krawczak, Polychronakos, Tyler-Smith, & Kehrer-Sawatzki, 2013). Thus, even "simple" Mendelian traits can very rapidly become complex and not so Mendelian.

The larger problem, however, is that most traits are simply not single-gene traits that follow Mendelian inheritance patterns. Most of the traits students (and adults) are familiar with, such as eye color, height, and muscle mass, as well as common diseases like diabetes and heart disease, are multifactorial (influenced by more than one thing, such as one gene). These traits are influenced by many genes (not just one), as well as the internal and external environment of the organism. For most of these traits, scientists cannot accurately predict an individual's phenotype. While teaching all the complexities involved in multifactorial traits is beyond secondary science education, we need to make sure students understand the limitations of the simple Mendelian inheritance that we do teach. Furthermore, we need to go beyond this model to introduce the idea of multifactorial inheritance and the interactional effects of genes (many genes) and the environment in traits.

4. Can a genetic test distinguish paternity between brothers or twins?

The answer depends on the comprehensiveness and sensitivity of the test. Brothers share some, but not all of their genetic material, and there are paternity tests that can distinguish between brothers. Most standard paternity tests cannot, however, distinguish between maternal twins (identical twins). There are more extensive tests that examine more of the

DNA, and that can even distinguish between identical twins, because such twins develop differences in their DNA due to errors in replication and random mutations that can be detected by highly sensitive tests.

5. Do we have 50% of the same genes as a banana? And if so, why don't we look more like bananas?

We share about 1% of the DNA of bananas, but that 1% shared DNA accounts for about 50% of our genes. That is, we do share about 50% of our genes with that yummy fruit. People who share more than 50% are obviously sweeter (just kidding). If we share so many genes with bananas, why don't we look like them? That is because the genes we share, and their protein products, are involved in basic life functions such as cellular respiration. Cellular respiration, protein synthesis, DNA replication, and many other cellular activities are common to many different living organisms including bananas and humans. There are also many genes that we do not share with bananas. We share 96% of our genome with fellow apes (such as chimpanzees, gorillas, and so on), and we look more like apes than bananas.

6. Are race and the color of one's skin the same thing? How is skin color determined?

Race and a person's skin color are not the same thing. The biology of skin color is perhaps related to, but separate from race, which is a social construct. Here is a Scientific American article for more information:
https://www.scientificamerican.com/article/race-is-a-social-construct-scientists-argue/

The issues around race are complex, and should be tackled with specific supports around those issues. This curriculum is not designed to directly address race. The curriculum was designed thoughtfully to avoid certain ways of thinking about genetics that can lead to problematic misconceptions about the nature of human difference. See the genetics unit rationale in the teacher background knowledge section for more information regarding these curriculum design decisions.

There are resources out there to support teachers if you do want to tackle race head-on. Visit the humane genetics website to find out more:
https://bscs.org/our-work/rd-programs/towards-a-more-humane-genetics-education/

Another option for a more in-depth discussion could be to collaborate with the social science department at your school with teachers that have training to address issues around race.

Skin color is largely determined by the types and amount of melanin produced in skin cells called melanocytes. These melanocytes produce melanosomes, which are essentially little membrane-bound packages filled with melanin. The size and distribution of melanosomes also impacts skin color. There are multiple genes that influence skin color, each with multiple alleles. In addition, exposure to UV radiation (tanning) can also alter skin color to some extent (and to different degrees in different people). It is also noteworthy that genes involved in skin color do not all influence hair and eye color; these traits are distinct. Here is a video resource called "How We Get Our Skin Color" from HHMI BioInteractive that explains the biology of skin color if it comes up in your classroom:
https://www.youtube.com/watch?v=VC0TL_lYLm8

7. Are boys more muscular than girls and is this because of genetics?

Males have more testosterone, which increases muscle mass. Females on average have about 40% less muscle than males. Moreover, there are differences in the composition of muscles. Males have more type 2 fast-twitch fibers in their muscles, which makes their muscles stronger. They can produce more force and produce it faster. There are many genes that contribute to muscle mass, and some of these genes are expressed differently in males versus females. However, differences in muscle mass are also attributable to diet, fitness level, and age.

8. If understanding the contribution of genes and the environment to traits is important, why doesn't the unit have anything about twin studies?

Twin studies have been used extensively to try and determine the heritability of traits. Heritability is a statistically estimated measure of how much of the variation in a given trait can be explained by genetic factors. Twin studies compare identical (maternal) and non-identical (paternal) twins to establish an estimate of the heritability measure. A core assumption in twin studies is that maternal and paternal twins share environments to the same extent. That is, identical twins growing up in the same household and paternal twins growing up in the same household share those environments to the same extent. However, this assumption turns out to be rather problematic, and several studies have shown that

identical twins share environments to a greater extent than paternal twins (Moore & Shenk, 2016). This is because identical twins share a placenta (usually) and thus in the womb their environment is more shared than paternal twins who develop in separate placentas. These placental differences do impact gene expression in significant ways. Moreover, similar-looking people (identical twins) tend to evoke similar social responses than different-looking people (paternal twins). Thus identical twins likely have more of a shared environment even outside the womb than do non-identical twins.

9. Can we say what proportion of the variation of a trait is due to genes or environment?
As mentioned above, almost all traits are influenced by *both* our genetic material and the environment of the organism. It may be tempting to think that one can actually separate environmental and genetic influences on traits. But it is too simple to try to say that a certain percentage of a trait is influenced by environment and a certain percentage is influenced by genetics. Rather, all traits are mutually influenced by genes and environment in an integrated way. In other words, it's not like there are two influences that are independent of each other. Instead, it is genes *interacting* with the environment that determines our traits. For example, a particular type of environment might be necessary for a particular genetic variation to make a difference. The notion of heritability as a measure of how much genes *versus* environment influence a trait is therefore misleading.

10. What is the "environment" for a trait?
It is tempting to think of "environment" as the lived-in world that is external to the organism -- things like what predators might be around, variations in food availability, temperature, and so on. It's important to realize that the environment includes some factors that are actually inside the organism. A simple example to think about is the womb in which an embryo is developing. There are many factors that might vary in the womb and affect how genes are expressed. The external environment can even include what is happening in the body of the individual -- that is still external to what is coded in the genes. For example, the concentration of the substances in the cells could impact some kinds of gene expression. A person under a lot of stress may have increased cortisol levels in their blood, which can affect how particular genes are expressed.

References:

Cooper, D. N., Krawczak, M., Polychronakos, C., Tyler-Smith, C., & Kehrer-Sawatzki, H. (2013). Where genotype is not predictive of phenotype: towards an understanding of the molecular basis of reduced penetrance in human inherited disease. *Human genetics*, *132*(10), 1077–1130. doi:10.1007/s00439-013-1331-2

Moore, D.S., & Shenk, D. (2016). The heritability fallacy. *WIREs Cognitive Science, 8*, e1400. doi:10.1002/wcs.1400