

# It's in Your Genes

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## Abstract

*There's a vast misunderstanding of how cells function amongst the general public. Many people understand that our genes are what make us who we are. However, many people don't understand how. Proteins are the functional units of the cell that allow the genome to express itself. The Central Dogma of biology is that DNA is transcribed to messenger RNA and then translated to protein, and everything about our physical being is a result of how proteins function—from the color of our eyes to life-threatening diseases that we endure, such as cancer. It is this idea (that the function of proteins is what determines our biological fate) that I believe is important for the general public to understand in regards to how they view their health and well-being.*

At some point in our lives, we have probably all asked the question, why do I look the way I do? Why are my eyes blue? Why is my hair brown? Why am I so short or tall? Typically, the answer is “because of your genes.” And as a matter of fact, everything about our bodies is the result of our genes—we are the result of the DNA inside every cell in our bodies. However, that's not the full story. What must be discussed is how cells function as a result of the DNA stored inside their nucleus. What often goes unnoticed is that in order for cells to function they must make proteins, and the proteins that cells make is determined by their DNA. Thus, it is safe to say that DNA stores genetic information (i.e., it is the genetic code) while proteins display said information. In this article, I am going to discuss how this process (going from DNA to protein) works, what can go wrong during this process, and some possible outcomes.

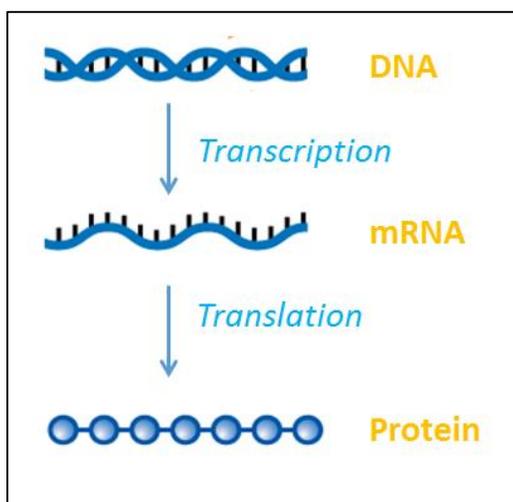


Figure 1.

How does DNA make us who we are? It is known that every single cell in our bodies has its own copy of DNA, and each cell has a way of expressing the genes that the DNA encodes via proteins. This is called the “Central Dogma” of Biology. In the nucleus, a gene on a segment of DNA is transcribed into messenger RNA (mRNA) and

then that mRNA leaves the nucleus and flows into the cytoplasm (the space outside the nucleus) where it is translated into protein via ribosomes (figure 1). Proteins function in many different ways, such as catalyzing metabolic reactions, responding to stimuli, and transporting molecules from one location of the cell to another.

Let us use eye color as an example. One's eye color is the result of how much melanin the melanocytes in the eye produce. The amount of melanin produced is largely dependent upon one thing—the proper functioning of the proteins that are responsible for melanin production. Here's how it works: melanin is made up of long chains of the amino acid tyrosine, and this stringing together of tyrosines takes place in special compartments of melanocytes called “melanosomes.” As long as tyrosine makes it into the melanosomes, the correct amount of melanin will be produced and the result will be brown eyes (yes, brown eyes are normal). But what if the correct amount of tyrosine doesn't make it to the melanosomes? What if the protein that's responsible for tyrosine shipment is defective? Well, it turns out that the protein OCA2 is responsible for distributing tyrosine to the melanosomes, and when it is defective, the result is less melanin production and thus blue eyes [1]. That's right; you do not have a “blue eye” or “brown eye” gene. You have an OCA2 gene that produces a functional or dysfunctional protein which effects melanin production. If you have blue eyes you are the result of a genetic mutation. How cool is that?

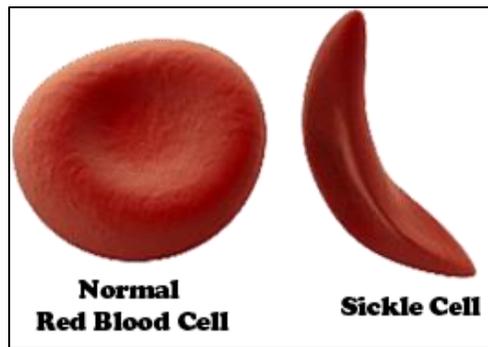


Figure 2. Normal red blood cell versus sickle cell anemia

Not all genetic mutations are harmless, however. Many diseases are the result of simple genetic mutations that produce dysfunctional proteins. A good example of this is sickle cell anemia. Sickle cell anemia is a disease that causes one's red blood cells to be misshaped. Instead of the normal round shape of red blood cells found in individuals without sickle cell anemia, people with sickle cell have sickle-shaped red blood cells (figure 2). The sickle shaped red blood cells are prone to clogging in the capillaries of the circulatory system, which can result in the blocking of circulation and even organ damage [2]. Sickle cell anemia is the result of a dysfunctional protein called hemoglobin. Hemoglobin is the protein found in red blood cells responsible for oxygen transport. In people with sickle cell anemia, their hemoglobin is misshaped when it is not bound to oxygen, and this is what causes the red blood cells to be sickle-shaped [2].

Another example of a disease that is a result of a defective gene is cancer. Now, before I continue I want to clarify that cancer is a very complicated disease, and by no means am I trying to make it sound simple, or to oversimplify my explanation of it. There is, however, a gene that is defective in more than 50% of cancers called “p53.” Cancer is a disease that results in the uncontrolled proliferation of cells, which are more commonly referred to as “tumors.” Normally, cells proliferate in a highly controlled manner, and therefore do not cause tumors. This process is controlled by “tumor suppressor” proteins. That is exactly what p53 is.

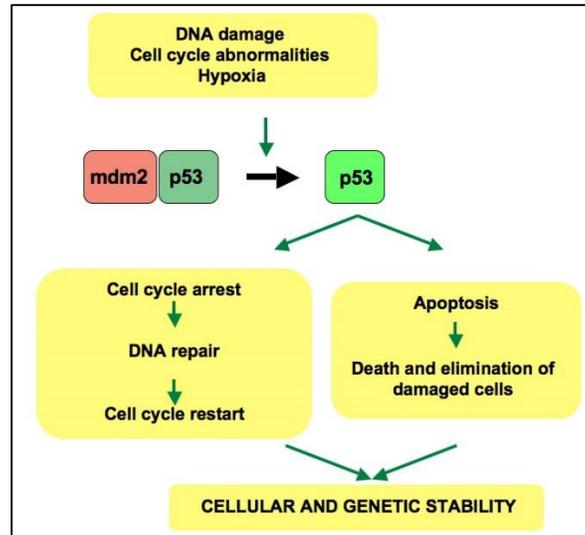


Figure 3.

In order for cells to proliferate, they have to go through what is called the cell cycle, which is a series of events that take place in a cell leading to its division and duplication of its DNA to produce two cells. P53 regulates the cell cycle in order to avoid the uncontrolled growth of the cell. This is how it works: in a normal cell, p53 is inactivated. However, in response to cellular stress, such as DNA damage, p53 becomes activated and does one of two things: 1) halts the cell cycle in order to repair the damaged DNA or 2) causes the cell to undergo apoptosis (cell death) in order to eliminate the damaged cell (figure 3) [3]. When p53 is dysfunctional, however, the cell cycle becomes uncontrolled and ultimately results in cancer. Once again, this is not always the case in cancer and there are many other factors that are involved in cancer progression. This is just a simple example of a dysfunctional protein that can result in a life-threatening disease, such as cancer.

There is one thing that I hope I have made clear: every protein has a segment of DNA (a gene) that transcribes it, and ultimately, it is how proteins function that determines our physical state, from our eye color to the diseases we are susceptible to. So hopefully next time you wonder about your physical being, you will be able to appreciate the beautiful complexity behind what makes you, you. Or even better, the next time a child asks you why they are the way that they are, you can give them a more detailed and accurate answer, not a superficial one like, “because of your genes.”

## References

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