Faith & Genetics

Genetics 101

Why do certain characteristics run in families?

What does it mean when someone says, "You have your mother's eyes?"

For all of recorded human history, and no doubt longer than that, humans have always been curious about inheritance. Until the 1800s, the mechanism of how traits were passed from parents to children was debated by philosophers and theologians, but almost no scientific analysis was performed.

In the 1800s Gregor Mendel was born in what is now the Czech Republic. Public schools were not available and private schools were expensive, but the parents of young Gregor recognized how bright he was and sent him to a local monastery to be educated. Gregor excelled, particularly in mathematics. He remained at the monastery as an adult and began performing experiments with sweet peas in an attempt to determine how certain traits such as flower colour, plant height, and pea shape were passed from parent plants to offspring plants.

Although there was as yet no knowledge of genes or DNA (which stands for deoxyribonucleic acid), Mendel was able to develop certain principles of inheritance that, with some refinement and modification, remain true today and, more importantly, are now known to apply equally to humans and sweet peas.

It was nearly 100 years after Mendel's experiments that Francis Crick and James Watson discovered the structure of DNA. The development of molecular genetics has allowed scientists to understand the composition and function of genes in much greater detail. The relationship between genes and characteristics that they express is much better known but we also know that the interaction between genes, the environment, and the development of diseases is often complex.

To understand the importance of genetics its relationship to health and the environment, one needs to understand some anatomy at the organ and cellular levels.

The human body is made up of organs like the brain, heart, and lungs. They fulfill necessary normal functions. These organs are made up of different types of cells that function differently but work together to keep an organ functioning well. An adult human body has several trillion cells. They make up our skin, muscle, liver, brain, eye, bones, stomach, heart, lungs, kidneys; everything is made from cells. Cells in different parts of the body have different structures and compositions, but they also all share some attributes.

The nucleus, which is found in every cell, acts as a control centre. Inside the nucleus is a full set of chromosomes, also known as a genome. The nucleus contains 22 pairs of chromosomes plus another single pair that determines the gender of the person, for a total of 23 pairs. Each chromosome pair has genes, each of which guides the formation of proteins that are building blocks for cell structure and functioning. Each gene has two identical or variant forms, known as alleles. One allele resides on each chromosome of a pair. Thus, each allele of each gene has an identical or largely similar "twin" on the other chromosome in that pair. Chromosomes are composed of chromatin, a complex made up of DNA that is wrapped around histone proteins a small amount of DNA also exists outside of the nucleus. This will be discussed in a later section). This chromatin is packaged into chromosomes which are the paired structures that replicate, divide, and are transmitted to our offspring during conception. DNA is the molecular backbone of genes and contains all the information needed for genetic inheritance. DNA is translated in the cell into proteins that create vital structures and serve many essential functions in our bodies. Each gene produces a distinctly different protein or set of proteins from other genes. Since each gene has wo alleles, each of which might produce the same or a different protein, the structural or functional characteristic attributed to that gene pair is determined by the differences or interactions between the proteins produced by that pair of genes. We will talk more about such proteins later.

An illustration of a generic animal cell

The genome of each person is identical in the nucleus of every cell in each person's body. So your brain cells have exactly the same genes as the cells in your lungs, as the cells in your inner ear, as the cells in the tendons of your big toe. However, your brain is different structurally and functionally from your big toe because not all of the same genes are at work in your brain as in your toe. That is, during development, inherited genetic instructions turn off the functioning of some genes while allowing the functioning of others.

Each organ has a different set of functioning and non-functioning (or silent) genes. We are beginning to understand the mechanisms by which our developing cells know which genes to turn on and off as our cells grow and differentiate into different functions. It has recently been shown that the histone proteins associated with DNA can change and alter the function of certain genes. These chemical changes in histones are known as epigenetic changes (see glossary). So we now know that not all changes that direct gene function involve DNA as previously thought. Such epigenetic changes are involved in the control of cell division and thus tissue growth. They can also be involved in disturbances of cell growth, as in cancers, where epigenetic changes are associated with uncontrolled expression of genes that signal cells to divide or with genes that fail to suppress growth when it is time for a cell to stop dividing. Therapies are currently available and in clinical development that are designed to destroy or alter these epigenetic changes in an effort to control growth of tumours or to destroy them.

The structure of DNA is called a double helix, which looks like a twisted ladder. This is a simple illustration of a very complex macromolecule.

If the DNA in a cell changes, that change (or mutation) will be inherited by the new cells created when that cell divides. Whether that mutation results in a change in the structure or functioning of the new cells containing the inherited mutation will depend on a number of factors including the type of gene that was mutated, whether the gene was a functioning or silent gene, what function it had before it was mutated, and so on.

Genetic information passed on to one's offspring consists mainly of DNA. This substance contains important information for the body. This information is translated in the body into proteins that build structures and fulfill vital functions for the body. These structures and functions are evident to us in characteristics such as eye colour, facial appearance, height, and other features. Because we get our DNA from both of our biological parents, our features often are similar to those of one or both of our parents. This means that you don't really have your mother's eyes or your father's nose but rather you have genetic information from your parents that determines your eye colour and shape of your nose. A difference or change in the molecular structure of a gene is a genotypic difference or change while a difference or change in the expression of a gene such as eye colour is a phenotypic difference or change.

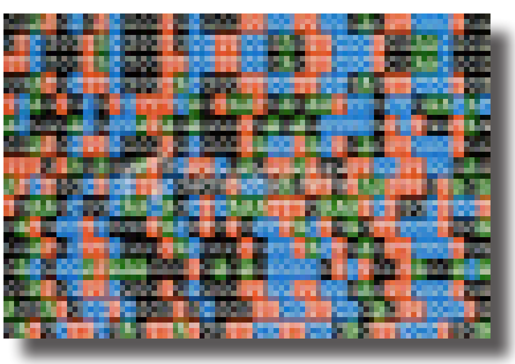
Chromosomes are generally shaped like elongated 'X's' - like the ones in this illustration.

Different Proteins for Different Functions

Proteins made in your muscle cells interact with one another to allow your muscles to contract; proteins in neurons (that is, the main cells that make up the nervous system) produce the neurotransmitters that allow one to know what to say next; proteins in the retina of your eye produce molecules that change shape when light hits them, allowing you to see. Fingernails and hair are composed of the protein keratin. Collagen is a protein that allows your muscles to attach to your bones and keeps your skin from sliding off. Hemoglobin is a protein that transports oxygen from your lungs to the rest of your body. Amylase is a protein made by saliva-secreting cells in your mouth that allows you to digest starch in various foods such as potatoes or crackers.

How is it that DNA contains information for the proteins involved in movement, speech, sight, and all the other workings of our bodies? DNA is a type of molecule called a polymer. Polymers are molecules made up of repeating subunits, like beads on a string. DNA has four subunits: adenine, guanine, cytosine, and thymine (or A, G, C, and T). These subunits are like a recipe for building proteins. Different combinations of subunits code for different proteins. Therefore, arranging the subunits of DNA – A, G, C, and T – in different ways will give rise to the full complement of proteins found in the human body.

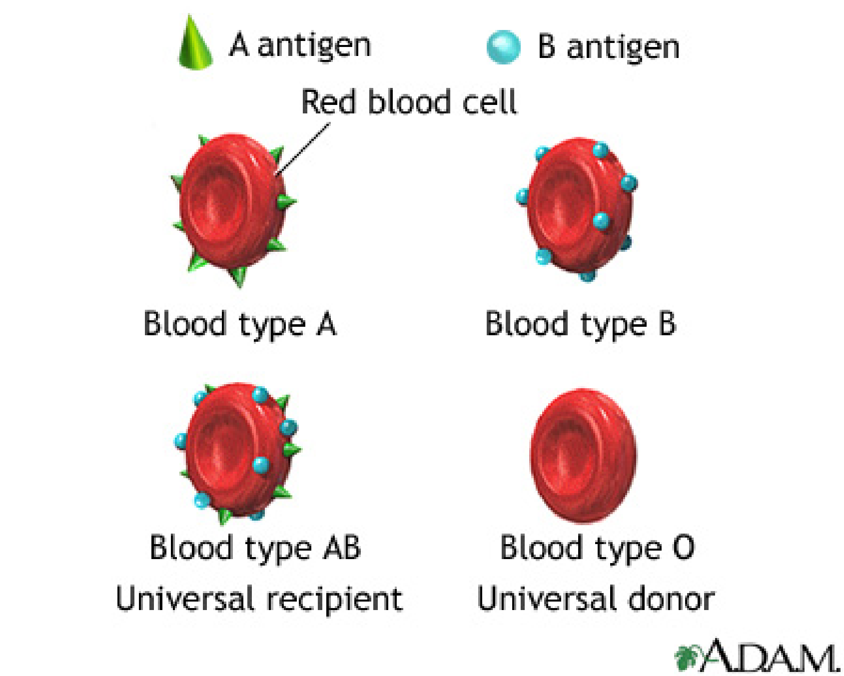
The order of subunits in a protein determines its structure and function. Alterations in the order of subunits in DNA—a mutation—can sometimes result in an altered subunit of its protein with altered structure and function. Not all mutations result in abnormalities or diseases. In fact, occasionally mutations result in new desirable qualities or functions. Some mutations result in a change in a characteristic like hair or eye colour while others result in no noticeable changes at all. This is because the gene in which the mutation occurs, and the protein that the gene produces, continue to function normally despite the mutation.



The four base units of DNA - A,G,C,T - can be arranged in millions of ways to create all the diversity we see in humans and in other species. Switching, inverting, deleting, or repeting any part of the code, however small, can have be effects on the organism.

Implications of Genetic Variation: Why We Have Different Blood Types

For example, there is a gene on one of the chromosomes that contains instructions for producing a protein whose function is to add sugars to the surface of red blood cells. Like other genes, each gene has two alleles and each allele can be one of three variants. The A variant has the recipe for an enzyme that adds A-type sugars to the red blood cells. A mutation that occurred in the far distant past results in a B variant that adds B-type sugars instead of A-type sugars to the surfaces of red blood cells. Yet another variant also arose by mutation in the far distant past. This third variant (or O variant) produces a non-functional enzyme (that is, it does not add either type of sugar nor seems to have any other function). The gene can have identical alleles of any of these three variants (i.e., a pair of A, B, or O alleles) or the gene can be composed of two different alleles (i.e., A and B alleles, B and O alleles, or A and O alleles).



Think of the red blood cell as an oatmeal cookie. The allele for blood type A as a recipe for oatmeal cookies with raisins. The slight variant allele that results in type B blood is analogous to a slight change in the cookie recipe, resulting in oatmeal cookies with chocolate chips instead of raisins. Both cookies are recognizable as oatmeal cookies, but there are clear differences. The variant that results in type O blood is sometimes referred to as a knock-out mutation or a nonsense mutation. Here, the recipe is so changed that cookies can be made but are just plain, with less flavourful distinctiveness! If you have A-type and only A-type sugars on your red blood cells, you'll have Type A blood. If you have B-type and only B-type sugars on your red blood cells, you'll have Type B blood. If you have neither A-type nor B-type sugars on your red blood cells, you'll have Type O blood.

Image from /www.pennmedicine.org/health_info/images/19450.jpg

Because you have two copies or alleles of each gene, one from your mother and one from your father, you could inherit an A allele from each parent. Similarly, you could inherit B alleles from both parents, or O alleles from both parents. If you have identical alleles on any gene, you are homozygous for that gene. If you inherited different alleles from each parent, you are heterozygous for that gene. Remember, alleles are the paired copies of a gene, each of which may vary slightly if a previous mutation or deletion of a small amount of DNA has occurred in one of the alleles. (See the glossary to review these terms and their meanings).

So How Do I Know Which Blood Type I Might Have?

Some alleles are expressed more dominantly than other alleles of the same gene. Such dominant alleles can be expressed (that is, result in a characteristic such as eye colour or a particular disease) in both homozygotes and heterozygotes. Other alleles are less dominant and can be expressed only in homozygotes where a more dominant allele is not present. Alleles that cause a given trait or disease even when only one copy is present are said to be dominant, while alleles that result in a trait or disease only in homozygotes (that is, when a single type of allele is present in both chromosomes) are said to be recessive. In blood typing, the A and B alleles are both dominant and the O allele is recessive.

In the ABO blood typing system, if you are homozygous for any of the three alleles mentioned above, you will express only that allele and it will determine your blood type (that is, AA = blood type A; BB = blood type B; OO = blood type O).

If you are heterozygous and have one dominant and one recessive allele (that is, AO or BO), only the dominant allele will have an effect and will determine your blood type (that is, AO = blood type A; BO = blood type B. If you are heterozygous and have two dominant alleles, they will both be expressed and you will have Type AB blood, or oatmeal cookies with both raisins and chocolate chips.

So, to summarize, you can have blood type A with either the AA or AO combination of alleles and blood type B with either the BB or BO combination. You can only have blood type O with the OO combination of alleles and blood type AB only with the AB combination.

The Punnett Square is a helpful way to explain the relationship between dominant and recessive genes. Named after Reginald Punnett, a biology professor who helped to develop the science of genetics in the early 1900s, it is still in use today. The Punnett Square is used to predict the genetic contribution or genotype of offspring. In other words, the square is a tool that helps us understand the odds of acquiring particular alleles that sometimes result in different physical attributes, such as eye colour or blood type.

Individuals pass on only one copy of each of their genes to their children. If two people with the blood types A and B have a child, the results can be predicted using a Punnett square. The mother's known alleles are placed along the top of the square and the father's known alleles are placed along the side of the square. The four squares represent the possible genotypes of their children. Here we see that this couple could have children with any of the four possible blood types and that each time they have a child, there is a 1 in 4 or 25% chance that the child will have blood type AB, O (represented by OO), B (BO) or A (AO).

Let's try another combination of alleles in the parents. If the parents each have the A and O alleles and therefore each have type A blood (that is, each have AO), plug the letters into the Punnett square. You will see that there is a 75% chance of their child having blood type A (a 25% chance of having two A alleles and 50% chance of having the A and O alleles together) and a 25% chance of having blood type O (or both alleles are O). This type of inheritance, where one gene controls one measurable characteristic is called simple Mendelian inheritance, after the monk, Gregor Mendel, who first deduced the mechanism of single-gene inheritance.