UNIT 2

THE REPRODUCTIVE SYSTEM

wonders of the HUMAN BODY

THE REPRODUCTIVE SYSTEM

Introduction

Throughout the *Wonders of the Human Body* series we are examining the organ systems of the body, both how they have been designed by the Master Creator and how they function. No doubt, you have been awed by the complexity of the body and have been puzzled at how anyone could believe these systems could have come into existence by chance. In many ways, this volume of the *Wonders* books is no different. Here we will be exploring the reproductive system and how it works.

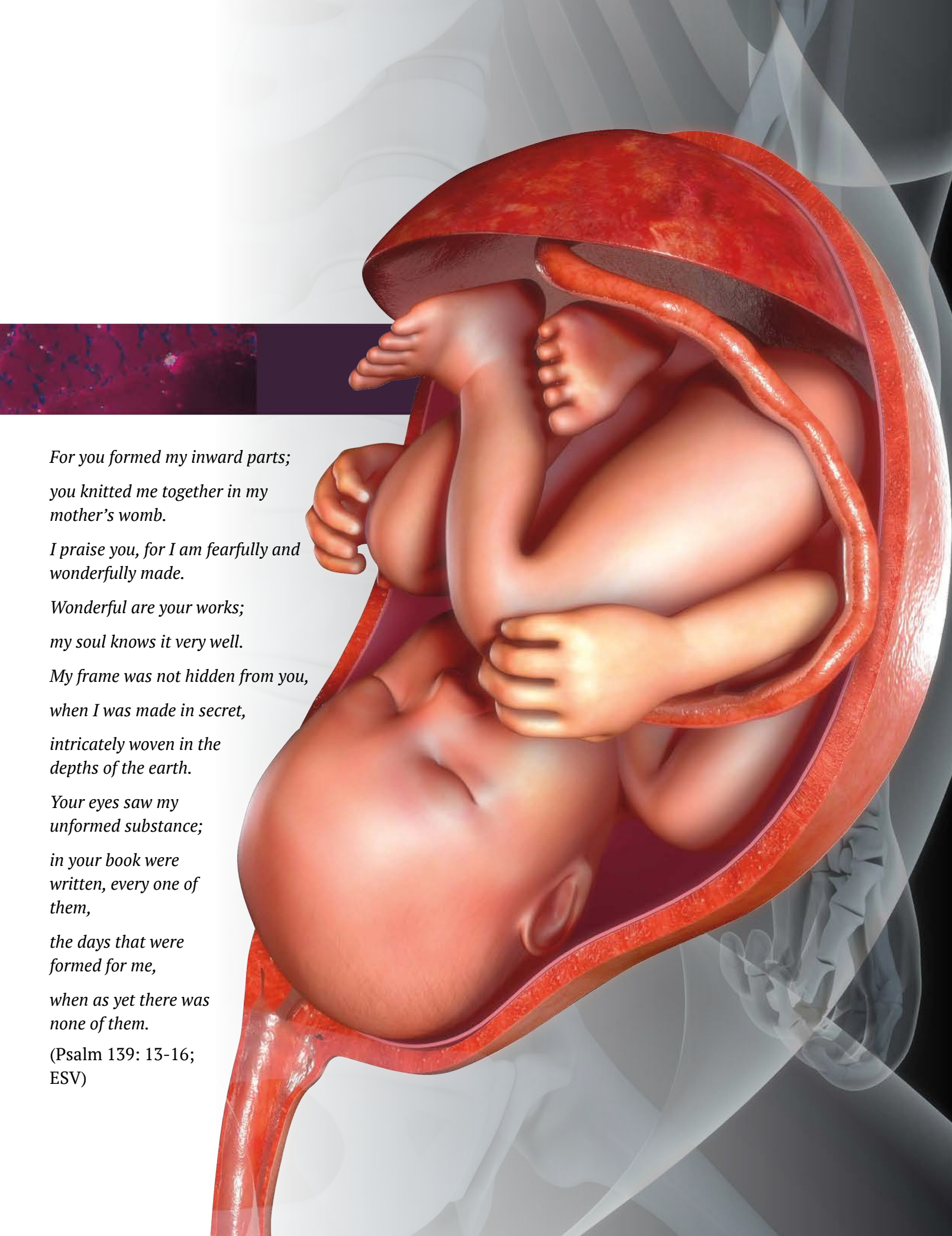
But there IS something different here. In this volume we will take a look back in time, back to about nine months before you were born. It's time to learn how you acquired the body you were born with. It's time to learn how you became you.

In the pages following, we will learn about the complex but orderly processes God designed to make the human body form correctly. Processes that are the same in everyone (assuming things work correctly, that is) but still allow each person to turn out to be a unique individual. This journey into your past will start with a cell, the basic building block of life. We will learn about the genetic code contained in your body's cells and how the collection of genes in your cells came to be. Then we will learn about the marvelous process God designed to form you from components of your mother's and your father's cells. Finally, we will delve into the secrets of the womb, where a baby forms in an unseen place for nine months to prepare him or her for life in this world.

In this volume, we will learn about many ways human rebellion against God and sin's curse have made things go wrong. Once we explore the complexity of God's plan for human reproduction, I think you will be amazed that things usually go right!

Psalm 139

You are the only you, and you were unique from the start. Here you will learn about the science behind this beautiful prayer poem about life before birth — poetry that God inspired in the Book of Psalms.



For you formed my inward parts;

you knitted me together in my mother's womb.

I praise you, for I am fearfully and wonderfully made.

Wonderful are your works;

my soul knows it very well.

My frame was not hidden from you,

when I was made in secret,

intricately woven in the depths of the earth.

Your eyes saw my unformed substance;

in your book were written, every one of them,

the days that were formed for me,

when as yet there was none of them.

(Psalm 139: 13-16; ESV)

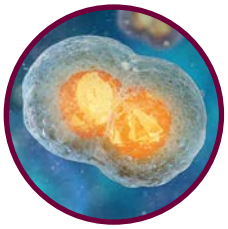
1

THE GENETIC BLUEPRINT FOR THE BODY

The Genome

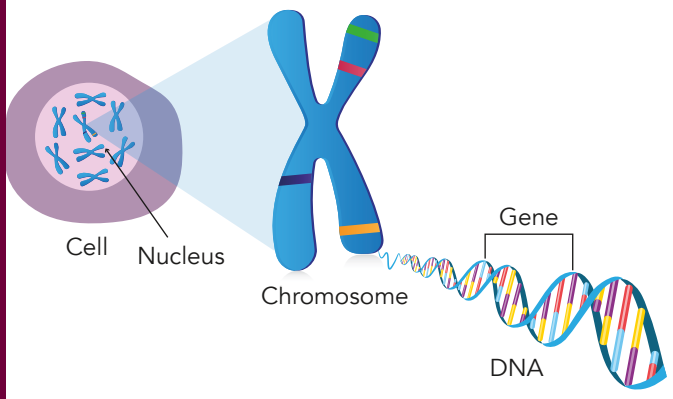
When God designed plants, animals, humans, and even single-celled forms of life like bacteria, He gave each kind of living thing its own genetic blueprint. That blueprint — also called a *genome* — is a complex storehouse of information. It is written in a special code, called the genetic code, on the DNA (deoxyribonucleic acid) molecules inside an organism's cells. The genetic blueprint inside an organism's cells determines how that organism develops and functions. The individual instructions encoded in the genome are called genes.






TAKING A CLOSER LOOK

The Genome



Genes are specific units of instructions that direct how a living organism is made and how its cells function. Each *gene* is coded into the organism's DNA molecules. An organism's DNA is divided up and packaged as *chromosomes*. One complete collection of chromosomes is like a set of encyclopedias for that organism. The full set is housed in a cell, even if a specific cell only needs to use a small fraction of the information in the set.

Each chromosome contains many, many genes. Each gene contains the instructions for making a particular protein important for that organism. The proteins in a cell, working together, do the work that cell must do. Of course, as you may have already learned in the other volume of the *Wonders of Human Body,* many different sorts of cells make up a multicellular organism, such as yourself. Yet whatever particular traits characterize an organism are largely the result of the proteins made by that organism's genes. That's why we can say that the genes are like a blueprint directing how an organism develops and how its cells function.

The DNA in a cell is copied and passed on whenever cells divide. Ultimately, the genetic blueprint — or *genome* — for a particular kind of organism is passed to offspring during reproduction.

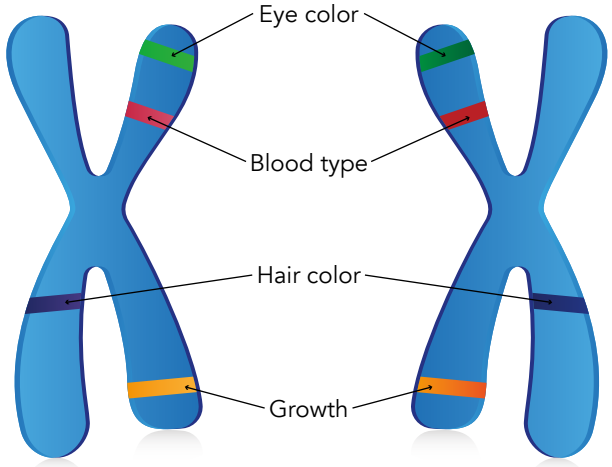
According to Their Kinds

The Bible tells us in Genesis chapter one that God created living things according to their kinds. We observe in biological science that each created kind has its own genome. This genome is the genetic blueprint that produces that kind of organism. Within a created kind's genome, there are many possible variations, such as those that determine our eye color and skin pigmentation. Many genes — the DNA instructions to make particular proteins and the traits associated with them — exist in varying forms, called *alleles*. The shuffling and mixing of these alleles during reproduction produces amazing variety, but never a new and different kind of organism. And never a more complex one.

This is precisely what is seen. No scientific observation has ever been made of one "kind" of creature turning into another "kind." Not once. Ever. Thus, every actual observation ever made shows that molecules-to-man, goo-to-you evolution — the process that some people believe made life evolve from chemicals, and people evolve from ape-like ancestors — has no basis in science and has never happened.

TAKING A CLOSER LOOK

Alleles



Thus, cats give birth to cats, dogs give birth to dogs, and humans give birth to humans. You may have noticed that kittens are not exact copies of either of their parents. And dogs are not exact copies of their parents. Human babies are not exact copies of either of their parents either. Cats' babies are cats, dogs' babies are dogs, and humans' babies are humans, but the offspring always differ from each of their parents. Why is that? The answer has to do with the way the genes from each parent are sorted and combined to make the genetic blueprint for a unique individual, different from either parent. As it happens, even identical twins, once thought to have completely identical genetic make-up, have some subtle differences, but we will go into that later. For now, let's learn more about the DNA in each of your cells. Then we will see how each parent's contribution produces a new, unique human being.

Genesis 1:20–25

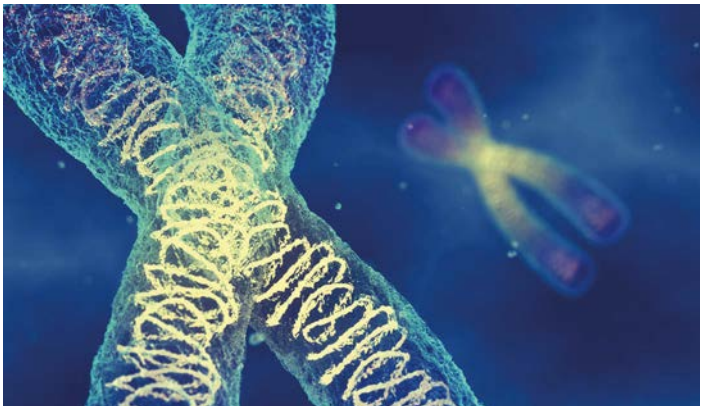
Then God said, "Let the waters abound with an abundance of living creatures, and let birds fly above the earth across the face of the firmament of the heavens." So God created great sea creatures and every living thing that moves, with which the waters abounded, according to their kind, and every winged bird according to its kind. And God saw that it was good. And God blessed them, saying, "Be fruitful and multiply, and fill the waters in the seas, and let birds multiply on the earth." So the evening and the morning were the fifth day.

Then God said, "Let the earth bring forth the living creature according to its kind: cattle and creeping thing and beast of the earth, each according to its kind"; and it was so. And God made the beast of the earth according to its kind, cattle according to its kind, and everything that creeps on the earth according to its kind. And God saw that it was good (Genesis 1:20–25).

God's incredible design allows living things, including humans, to vary a great deal within their created kinds. This variation depends on the particular combination of genetic material an organism possesses. However, there is nothing in the process of passing genetic information to subsequent generations that would allow one "kind" of creature to turn into a different "kind."

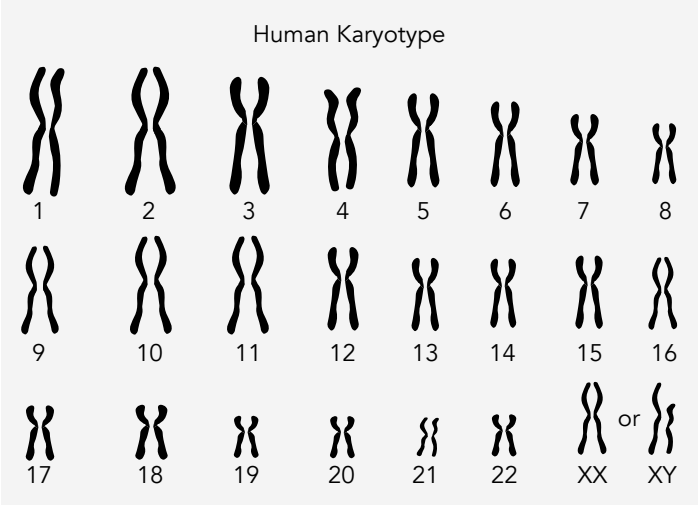
Basic Genetics — The Answer Is Forty-Six

The cells in your body contain your copy of the human genome. This genome, with all its variations that make you unique, is encoded on your DNA. Your DNA is coiled tightly and packaged as 46 chromosomes in the nucleus of each of your body's cells. You might be thinking, "But wait, my red blood cells don't contain any nuclei!" I'm proud of you for remembering that! Red blood cells in your



bloodstream do not contain nuclei, but they do contain nuclei when they are being formed in the bone marrow. Once the red blood cells are made, they eject their nuclei to save space.

Each nucleated human body cell contains 46 chromosomes. (A "body cell" is not the same as a "reproductive cell," as we shall soon see.) Chromosomes come in pairs, so you have 23 pairs of chromosomes. They are known by their number — 1 through 22 — which is so much easier to remember than some special Latin name. The 23rd pair is the chromosome pair that determines whether you are male or female, so the chromosomes in that pair are called the "sex chromosomes," even though they also contain genes that affect things other than your gender. A person with two X chromosomes is female, and a person with one X and one Y chromosome is male.



Dominant and Recessive Alleles

Each pair of chromosomes contains two copies — or *alleles* — of each of the genes on them. In many cases, the alleles vary a bit. They are still the same gene, but with slight variations. Those slight variations may cause the protein produced by the gene to vary slightly. Proteins are molecules made of long chains of many amino acids. Our DNA contains codes for the 20 different amino acids used to build our proteins. Just exactly which amino acids are hooked together to make a particular protein is determined by the gene directing its formation. Sometimes, changing just one amino acid in a protein destroys its ability to function. Changing another amino acid, located elsewhere in the protein, may impair its ability to function efficiently. And changing an amino acid in various other locations in the protein molecule may not hurt its function at all. Such changes are the genetic variations we keep mentioning.

In a gene pair — the two copies (alleles) of a gene located on a chromosome pair — one variant may have more effect than the other. We call the one that "rules" the *dominant* variation, or the *dominant allele.* The variant that is over-ridden by the dominant one is called *recessive,* because its effects "recede" into the background and are not noticed.

Let's consider, for example, the nature of your earwax. Earwax is produced by glands in the outer third of the external auditory canal. Those glands secrete several types of alcohol and lipid molecules, the chemical components of earwax. This oily substance lubricates the lining of your ear. It also helps clean away debris and dead skin cells. Your eardrum constantly makes new cells, and the worn-out dead ones would accumulate in your ear if not removed. These dead skin cells are moved along the ear canal as your jaw moves. They, along with any debris that may have gotten into your ear, stick to the earwax, which also moves gradually toward the outside.

Earwax can be wet, sticky, and brown. It can also be dry, flaky, and gray. Whether your earwax is wet or dry is not determined by your age or how often you shower. It is determined by a gene located on chromosome 16. Depending on which particular "letter" in the genetic "alphabet" is present — and we'll talk more about that alphabet soon — either the amino acid glycine or arginine is inserted at a certain crucial location in the protein encoded by that gene.

The allele that causes glycine to be present is the dominant variation. Therefore, people who have two copies of the glycine-variation — we can call them GG — have wet earwax. And people who have one copy of the glycine variation and one of the arginine variation — the Ga people — also have wet earwax. This is because the arginine allele is recessive and is "overpowered," if you will, by the dominant glycine allele. People who have two copies of the recessive arginine variation — the aa people — have dry earwax. The aa people have dry earwax because they have no dominant G allele.

EARWAX	AMINO ACID	GENE ALLELES
Wet 	Glycine	Glycine-Glycine (GG)
Wet 	Glycine	Glycine-Arginine (Ga)
Dry 	Arginine	Arginine-Arginine (aa)

Genetic Expression

We've said that a complete copy of the human genome is present in every nucleated body cell. But that is an enormous amount of information for one cell to handle! Therefore, in a particular cell, most of that information remains unused. The genes needed by that particular cell are the ones that are *expressed*. God designed a special group of molecules associated with the chromosomes to regulate which genes are expressed in each cell type.

Protein-Building Amino Acids

Some lists now say that 21 amino acids are used in human proteins. This is done by including selenocysteine, which has been found in fewer than 100 of our proteins. However, unlike other protein-building amino acids, selenocysteine is never stored in cells, for it is highly reactive. Its synthesis is directed by the cell's amino acid-building machinery in an unusual way on an as-needed basis. Further, selenocysteine is not represented directly in the genetic code.

For our purposes, we'll just stick with 20 amino acids. That seems sufficient for now.

More About Genetic Expression

In the case of earwax — you didn't think I was through talking about earwax, did you? — the wet/dry gene is expressed by the cells in the glands in your external auditory canal. Depending on whether you are a GG or Ga or aa person, your earwax will contain a slightly different assortment of molecular secretions. And it is those different molecules, and different amounts of those molecules, that make your earwax wet or dry.

It happens that the "wet/dry earwax gene" is expressed by cells in one other location in your body — the cells in the glands in your armpit! People who have wet earwax produce stronger smelling molecules in their armpit perspiration. They tend also to perspire more than dry earwax people. In the rest of your body's cells, the wet/dry earwax gene — which is called the ATP-binding cassette C11 gene, if you must know — is not expressed. It just remains silently in its place in chromosome 16, getting copied and passed on every time a cell divides.

Population Genetics

Scientists can track the historical path of population movements through earwax genetics. It turns out that people with dry earwax are far more prevalent in East Asian, Northeast Asian, and Native American populations than in the rest of the world. Remember, to have dry earwax, you must have two copies of the recessive allele. That means that you must have received the recessive version from both of your parents. You see, during the reproductive process, a special cell from your mother (an egg), containing only 23 chromosomes — not the usual 46 — combines with a cell from your father (a sperm) that also contains only 23 chromosomes. When those cells — called *gametes* — come together, a single 46-chromosome cell called a *zygote* is produced. *This is called fertilization.*

That zygote was the beginning of you. Every bit of genetic information required to produce the body you were born with and to enable you to grow and function was contained in the 46 chromosomes you received from your parents. We'll cover more about that later. For now, let's focus on earwax.

A person who has the aa gene pair on chromosome 16 is *homozygous* for the recessive "a" allele of that gene. He or she received an "a" from mom and an "a" from dad. Since having just one copy of the "G" allele makes you have wet earwax, you can deduce that if everyone in a particular people group has dry earwax, that people group must have gotten isolated from the wet earwax people at some time in their history. Any idea when that might have happened? Yep — after God's judgment on people at the tower of Babel.

Genesis chapters 10–11 describe the centuries soon after the worldwide Flood of Noah's day. God had told people in Genesis 9:7 to spread out and populate the earth. Instead, many people gathered themselves together on a plain in the land of Shinar. They were determined to remain together, building a powerful nation that they thought would be secure and great enough to enable them to do anything they pleased. They all spoke the same language. They started building a great tower to symbolize their united stand against their Creator's command. God foiled their plans by confusing their languages. Suddenly, various groups of people were unable to understand what other groups were saying. No longer able to easily work together, groups of people who spoke the same language would have banded together and left the region to make a home elsewhere in the great, now unfamiliar, world. In this way, people dispersed throughout the post-Flood world.

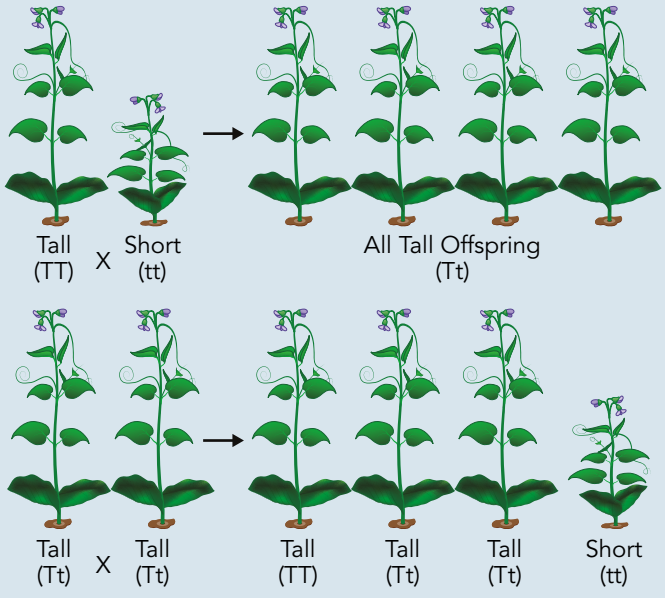
The original group of people at the Tower of Babel would have, in their combined gene pool, all the genetic variety in the human population descended from Noah's family — the people on the Ark. If they had remained forever together, most variations would have been passed around pretty evenly. However, once small groups moved out into the world, *only the variations present in the gene pools of those smaller groups would be available to the children born in those groups*. Thus, if a group of people that moved toward eastern Asia happened to have mostly folks with dry earwax, East Asian people descended from that group would have dry earwax, the "G" allele having been lost to their gene pool. Such genetic isolation is one way that variations characteristic of a particular population group develop.

Origins of Human Genetic Variety

So you must surely be asking, if we are all descended from just two people, Adam and Eve, how did people end up with different variations of the same gene? There are a couple of different ways. It happens that many of our characteristics are actually regulated by more than one pair of genes. We call those characteristics *polygenic*. ("Poly" means "many" — so "polygenic" means "many genes." Get it?)

Gregor Mendel

The existence of the simplest of inheritance patterns — the dominant-recessive pattern of inheritance of traits (phenotypes) controlled by a single gene — was discovered in the 1800s by a monk named Gregor Mendel. He grew pea plants. Those plants had a number of characteristics — such as their height, whether the flowers were purple or white, and whether or not the peas were wrinkled — that were controlled by single genes functioning with dominant-recessive inheritance patterns. Mendel cross-pollinated plants with certain characteristics to see what would happen. He kept careful records. By analyzing those observations, he figured out that the characteristics he was observing were passed on in a simple, logical fashion. Some were dominant, and some were recessive. It is good for the history of genetic science that Mendel just happened to experiment with characteristics that are passed on through simple *Mendelian genetic* patterns. If he had chosen traits that turned out to be polygenic, our appreciation of the power of genetics may have been delayed a good deal longer.



The particular collection of variations you happen to inherit for a polygenic trait determines the polygenic characteristics you have. Eye color is a great example. Scientists used to think that something as simple as blue versus brown eyes was inherited as simply as the nature of earwax. Eventually we learned that there are several genes located on more than one chromosome that work together to determine eye color. Therefore, for many characteristics, the variations present in Adam and Eve's original human genome was sufficient to mix and match as they were shuffled between people over the years to produce the enormous variety of people in the world.

Mutations — Another Source of Genetic Variation

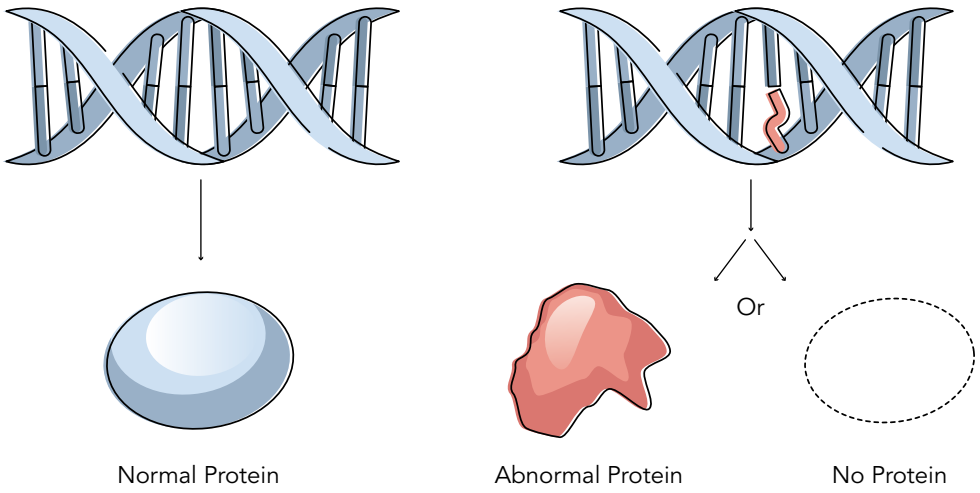
Other variations have come into our human gene pool through mutations. A *mutation* is a genetic mistake. We know that in the beginning God saw that all He had made was very good. After man sinned, God's judgment allowed things to go wrong, and sickness and death entered the world. Genetic mutations are some of the things that go wrong, and they are the cause of many problems.

Many types of mutations occur, and we will cover how they happen in more detail later. Most cells in your body divide frequently, making copies of themselves quite often. (Your nerve cells are one big exception.) When cells divide, their DNA must be duplicated. If something goes wrong when DNA is being copied — if some information is left out, damaged, scrambled, substituted, or even duplicated — that change is a mutation. The cell has proof-reading mechanisms in place — ways that the DNA-duplication process polices itself and self-corrects. However, a mutation that is not caught and corrected by these proof-reading safeguards may be passed on when that cell divides.

If a mutation occurs in one of your skin or bone or blood cells, that mutation cannot be passed on

TAKING A CLOSER LOOK

Normal Genes vs Mutated Genes



to your children. You may develop some problem yourself. You might even develop a cancer, if that mutation is a serious one associated with uncontrolled acceleration of cell division. But you cannot pass on that sort of mutation to anyone else.

If a mutation — a DNA copying error — occurs in reproductive cells, then the mutation may be passed on to offspring. Just as the normal genetic variants for eye color and skin tone are passed on to offspring, so a mutation for a trait like color-blindness or sickle cell hemoglobin may also be passed on. Mutations that occur in a *gamete* — one of those 23 chromosome-containing cells we mentioned earlier — can be passed on to your children. *Sperm* are the gametes produced in males. *Ova* — or eggs — are the gametes produced in females. If a sperm or an *ovum* (singular of ova) contains a mutation, and that gamete is joined to another gamete to produce a zygote, the zygote will contain that mutation.

Some mutations are deadly. When a person with a deadly mutation is conceived, he or she will at some point die as a result of the malfunction caused by the mutation. Some mutations are harmful but not deadly. An example is the sickle cell mutation. People who have the sickle cell mutation in one or both of their copies of their hemoglobin-producing genes are harmed by the presence of an abnormality in their hemoglobin.

Many mutations, however, even though they are genetic mistakes, cause no harm at all. A harmless mutation might result in a noticeable but harmless characteristic. The results of a harmless mutation might not even be noticeable. These insignificant mutations are useful to scientists trying to track population movements through history. People descended from the same isolated people group will often have many of the same mutations in their genes. These can be traced through DNA analysis even if the people have no noticeable manifestation of the mutations.

While on the subject of what sorts of things mutations can do, let's get straight about one thing mutations never do. Mutations can damage or destroy some of the genetic information in a set of chromosomes. However, mutations can never supply the necessary genetic information for one kind of organism to evolve into a new, more complex, different kind of organism. Thus, many variations in people have occurred since God created Adam and Eve about 6,000 years ago. But all people that have ever lived — from Cain to Noah to Neanderthals to your brother — have had a *completely human* genome containing the variations developed from the shuffling of genetic material passed down from Adam and Eve and from the mutations that have occurred over the past 6,000 years.

Genotype and Phenotype

By now you've seen that two people with different genetic variations are sometimes indistinguishable from the outside. We have good words to describe this. *Genotype* is the particular collection of genetic variations present in an organism's DNA. *Phenotype* is the resulting characteristic in the organism. With earwax for instance — you didn't *really* think I was finished with earwax, did you? — a person's genotype might be GG, Ga, or aa. People with the GG and the Ga genotypes have the *same* phenotype — wet earwax and smelly sweaty armpits. People with the aa genotype have the other phenotype — dry earwax and less smelly sweaty armpits. (Yes, I do have a way with words. . . .)

GENOTYPE	PHENOTYPE
Glycine-Glycine (GG)	Wet 
Glycine-Arginine (Ga)	Wet 
Arginine-Arginine (aa)	Dry 

The *genotype* is the *information in the genes*. The *phenotype* is *the way that information ultimately gets expressed.* In dominant-recessive inheritance patterns, any genotype containing the dominant gene manifests the "dominant" phenotype. And only genotypes containing two recessive alleles manifest the recessive phenotype.

There is a simple way to keep track of the genotypes of two parents — whether people or pea plants — and those of their offspring. This is called the Punnett square. For the gene under consideration, the genotype of each parent is written above and to the left of a square divided in fourths. Remember, we said that each parent gives just one of his or her two alleles for this gene to a particular one of their offspring. Write each parent's genotype with one allele above each little box. Then imagine combining each allele from one parent with an allele from the other parent. This is sort of like flipping a coin. Each flip of a coin has two possibilities. But two coins must be flipped at the same time to get the composition of each result. Each "flip of the genetic coins" — the coming together of a randomly selected allele from each parent to form the genotype of the offspring — therefore has *four* possibilities.

Let's take our earwax example. (You knew that was coming.) If both parents have the GG genotype, we write the GG above and to the left of the four-patch square. No matter what allele is selected from each parent, the outcome is always GG. All four "possibilities" are the same — the GG genotype and the wet wax phenotype. The same is true if both parents are aa. The only possibilities for their children are the aa genotype, and the dry wax phenotype.

		FATHER'S GENES (GG-WET) 	
		G	G
MOTHER'S GENES (GG-WET) 	G	GG (Wet)	GG (Wet)
	G	GG (Wet)	GG (Wet)

		FATHER'S GENES (aa-DRY) 	
		a	a
MOTHER'S GENES (aa-DRY) 	a	aa (Dry)	aa (Dry)
	a	aa (Dry)	aa (Dry)

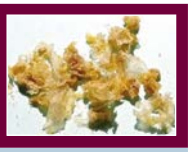
However, let's see what the possibilities are if each parent has wet earwax, smelly armpits, and the Ga genotype. We write the G and the a for each parent above the little squares. Then we match up the possibilities. Do you see that each child of two Ga parents has a 25% (one-in-four) chance of being GG, a 25% chance of being aa, and a 50% (one-in-two) chance of being Ga? Since only the child with aa will have dry earwax and minimal smell to his or her armpits, there is a three-in-four chance that any child the Ga parents have will have wet earwax and smelly armpits like the parents.

		FATHER'S GENES (Ga-WET) 	
		G	a
MOTHER'S GENES (Ga-WET) 	G	GG (Wet)	Ga (Wet)
	a	Ga (Wet)	aa (Dry)

It is important to understand that this does not mean that if the parents have four children, one will be aa, one GG, and two Ga. The odds "reset" for each child. If we were talking about the color of a garden full of flowering peas and hundreds of seeds — like those Gregor Mendel experimented on — then, statistically speaking, about a fourth of the seeds would have the recessive phenotype, and about three-fourths would exhibit the dominant phenotype. But in a family of just four children — instead of hundreds — it is important to remember that the possibilities apply to each individual child.

Of course, there are additional possible combinations. Consider a wet earwax GG person paired with a wet earwax Ga person. This couple's children would all have wet earwax, but each would have a 50% chance of having a GG genotype and a 50% chance of having a Ga genotype. On the other hand, a wet-dry couple with Ga and aa genotypes would produce children with a 50% chance of having dry earwax (and genotype aa, of course). And any wet earwax children they have will have the Ga genotype.

		FATHER'S GENES (GG-WET) 	
		G	G
MOTHER'S GENES (Ga-WET) 	G	GG (Wet)	GG (Wet)
	a	Ga (Wet)	Ga (Wet)

		FATHER'S GENES (aa-DRY) 	
		a	a
MOTHER'S GENES (Ga-WET) 	G	Ga (Wet)	Ga (Wet)
	a	aa (Dry)	aa (Dry)

Diseases Inherited through Dominant-Recessive Patterns

In the 6,000 years since Adam sinned, many mutations have occurred. Some of those mutations have resulted in diseases. Of course, the first time a mutation occurs, the person thus afflicted cannot be said to have inherited the resulting disease from anyone. But once a mutation gets passed on to children and children's children, its inheritance pattern is observable.

Disease-causing mutations may occur in a single gene, or disease may result from a collection of mutations that appear in many genes on different chromosomes. Disease-causing mutations also include mutations that produce extra copies or loss of a particular chromosome.

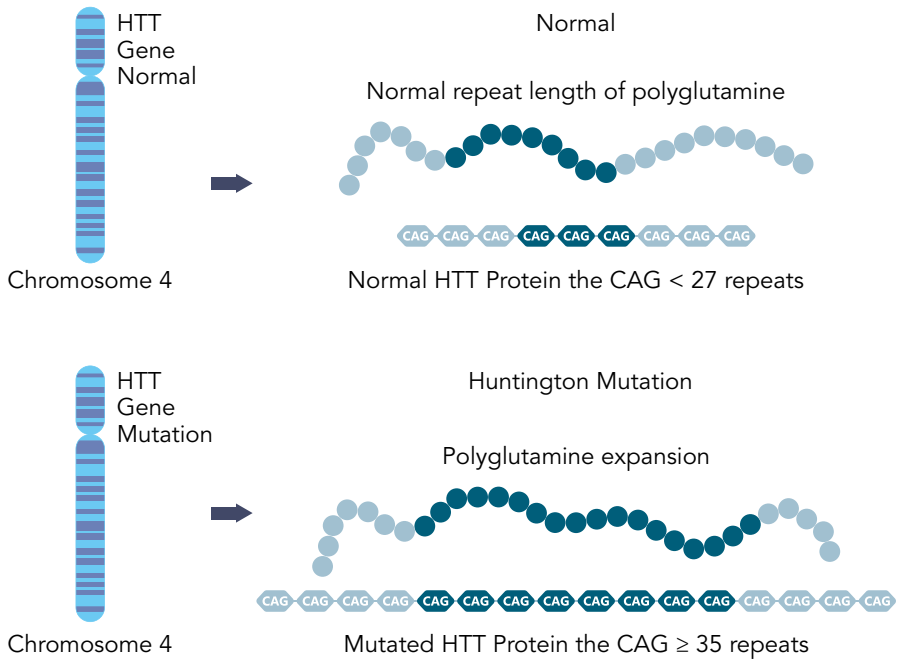
A few of the diseases caused by a mutation on a single gene are cystic fibrosis, polycystic kidney disease, Tay-Sachs disease, Huntington's disease, Marfan syndrome, and sickle cell disease. Like earwax — but with much greater consequences — most of those on this list are inherited with a dominant-recessive pattern. (Sickle cell disease is passed on with a different pattern, which we will discuss farther on.)

Huntington's disease is a classic example. Here the presence of a single allele with the disease variation means the person will ultimately have that disease. Huntington's disease is caused by a mutation in the HD gene located on chromosome 4. The mutant allele causes the protein thus formed to be defective.

The abnormal protein clumps in the brain and causes nearby nerve cells to die. Eventually, a person carrying the dominant mutant gene will develop severe neurological disease involving excessive involuntary movement and a deterioration of mental function. Because this does not usually occur until adulthood, people afflicted by the disease have usually had children of their own by the time the disease affects them. Each of their children has a 50% chance of inheriting the dominant mutant form of the HD gene and of therefore also developing the disease.

TAKING A CLOSER LOOK

Huntington's Disease



Let's look at a Punnett square to see how this works. We usually use an uppercase letter to denote dominant conditions, so H = the Huntington's dominant mutation. Hh would thus be the genotype of a parent carrying one copy of the dominant mutation, and hh would be a parent without the mutation. Here in this Punnett square you see there is a 50% chance that any child of this couple will inherit the H allele, and a 50% chance he/she will not.

		PARENT WITH HUNTINGTON'S DISEASE (Hh)	
		H	h
PARENT WITHOUT HUNTINGTON'S DISEASE (hh)	h	Hh (has disease)	hh (no disease)
	h	Hh (has disease)	hh (no disease)

Because the dominant mutant allele for this disease exists on chromosome 4, not one of the X or Y chromosomes, we say that Huntington's disease is *autosomal dominant*. The word *autosomal* means the genotype in question is on one of the 22 "autosomal" chromosomes, not an X or Y chromosome.

Tay-Sachs disease is another disease passed on through a mutation on a single autosomal gene, located on chromosome 15. A mutation in the Hex-A gene prevents the production of the enzyme Hex-A. Hex-A removes a particular lipid from the nervous system. Because this mutation is *recessive*, so long as a person has one normal allele, he or she makes enough of the enzyme to remove the lipid. However, without the Hex-A enzyme, that lipid builds up in the nervous system. That buildup soon begins causing deterioration in the brain. By the time a baby is just a few months old, many problems develop, including seizures, loss of the ability to move normally, and intellectual disability. Tay-Sachs disease usually kills in the first few years of life.

The mutation causing Tay-Sachs disease is *autosomal recessive*. A parent carrying one copy of the mutation has no symptoms. But if each parent happens to carry one copy of the mutant allele, it is possible for their child to receive the mutant allele from both of them. That child will have Tay-Sachs disease. It is also possible for their child to receive a normal allele from each of them. That child will not develop Tay-Sachs disease and cannot pass it on to his or her children. Finally, it is possible for their child to receive one normal and one mutant allele. That child will never develop Tay-Sachs disease, but his or her children may eventually receive the mutant allele from them.

What are the odds of two Tay-Sachs "carrying" parents giving birth to a child with the disease? About 25%. In other words, there is a one-in-four chance that any child they have will have the disease. There is also a one-in-four chance that any child they have will have neither the disease nor the abnormal gene. And there is a 50% — a one-in-two — chance that any child they have will be a carrier of the disease.

With an autosomal recessive condition, anyone carrying two copies of the recessive allele will have the condition. People who do not have the condition may be free of the allele — in which case they cannot pass it on to their children. Or they may have one copy of the recessive copy of the allele. Anyone with a copy of the recessive allele may pass it on to their children. However, only couples in which both parents carry the recessive allele can produce a child with the condition.

Autosomal Dominant Inheritance

Can you use a Punnett square to determine the likelihood of an autosomal dominant condition — like Huntington's disease — being passed on to a child if *both parents* carry the dominant gene? Use Hh as the genotype for each parent.

		PARENT WITH HUNTINGTON'S DISEASE (Hh)	
		H	h
PARENT WITH HUNTINGTON'S DISEASE (Hh)	H		
	h		

Here you see that if both parents carry an autosomal dominant allele (like H), then each of their children has a 75% chance of inheriting the dominant allele and therefore the condition it causes.