Chapter 1

Martin Needs Medical Treatment (or does he?)

Martin came home from school the other day with a black eye and broken glasses. Another boy had called him a freak and punched him.

Martin is albino, which means that his skin has no color. He is very pale and his hair is white. His eyes are pink and he doesn't see very well.

Martin's mother loves her son very much just the way he is. But when she sees other children tease him, she wishes he were not so different. If he weren't, then perhaps he wouldn't be picked on so much. It makes her wonder why everyone can't be the same.

Do you ever wonder about this yourself? If it were up to you, would you want everyone to be alike?

The world is filled with nearly 6 billion people, but each and every one of us is different from everybody else.

Only you have your combination of looks, personality, and behavior. As the saying goes, they broke the mold when you were made! There is *no one* in the world exactly like you.

EYES

Genes determine physical traits like eye color.

At the same time, you have traits, or ways of looking, thinking, and being, that you share with some other people on earth. For example, you may look like your father or share your mother's sense of humor.

You also have **traits** that you share with *every* other person on earth. For example, every person has blood, lungs, and a brain. All things considered, you are

more like every other person on earth than you are different from them.

One way that scientists know this to be true is by studying our **genes**. Genes are units of information inside the cells of your body. They contain the instructions for making cells and for doing the work that goes on inside them. It is through the genes that traits are handed down from parents to offspring, in a process called **heredity**. Genes help decide your size, build, coloring, and other features. They make you male or female. Researchers believe that genes also play a part in how you think and behave and in your body's health.

The human body is very complicated, so it makes sense that it needs a lot of instructions. Scientists don't yet know the exact number of genes that humans have, but they think that the number is somewhere around 80,000. Inside each one of those genes are distinct chemical ingredients called **bases**. The bases are linked together in long chains, with thousands or millions of bases per gene. Millions more bases link the genes together. Add up all these small parts, and you have 3 billion separate pieces that make up the human instruction book. Yet of these 3 billion pieces, only about 3 million are different from person to person. These are the parts that make you unique.

Now, 3 million is a huge number, but it is not much compared to 3 billion. What this means is that all human beings are built from nearly the same set of instructions. We are all really quite similar.

Genetics

How does a set of instructions work to create humans and other forms of life? This is the subject of a field of science called **genetics**. Genetics is the study of how traits are passed down, or inherited, from one generation to another. It is the study of how each living thing is similar to others of its kind, but also unique.

MICROSCOPE

For hundreds of years, people have known that traits can be inherited. They observed how looks were passed down from parents to children. They noticed how illnesses run in families. They used their understanding of inheritance to breed plants and animals. But no one really understood how this passing down of traits actually worked.

Then, about 400 years ago, the microscope was invented, and for the first time, scientists could see objects as tiny as a cell. They discovered that living things are created from cells of their parents. They also learned that living things grow when their cells divide to form new cells. As more powerful microscopes were invented, scientists could even look inside cells to watch what happened as they divided and reproduced.

Scientists also learned about heredity by studying plants, fruit flies, and other forms of

life, or species, that produce several generations in a short period of time. They discovered patterns in the way that traits are passed down in a **species** from one generation to the next. And they learned how species change over time, trait by trait, in a process called **evolution**.

Slowly, scientists began to unravel the mystery of genetics. Today, powerful computers and other modern research tools are helping scientists learn a great deal more, at a much faster pace. They are figuring out how genes work to do what they do. And they are uncovering the functions of specific genes.

These discoveries are teaching us a great deal about the genetic instructions that construct and operate the human body. This new information will give us new opportunities to control the destiny of our bodies. But at the same time, it will force us to face new and sometimes difficult choices. Some of these choices will be have to be made by individuals or families. Other choices will be made by all of us together, as a society.

Questions Raised by the "New Genetics"

To get an idea of the many choices that come with the new genetic information, consider Martin, the boy who is albino.

Martin is albino because his genes do not give the right instructions for his body's production of **pigment**, the dye that colors the skin, eyes, and hair. The result is that Martin is very pale. He must avoid the sun because he is at high risk of sunburn and skin cancer. Strong light hurts his eyes, and his vision is poor, so he needs glasses.

Suppose researchers discover a way to treat Martin's genes so that they give the proper instructions for producing pigment. This kind of genetic treatment may be possible someday. It would mean that Martin's skin and eyes would regain color. He no longer would have to stay out of the sun all the time. Plus, he wouldn't stand out from other children. These changes could make a big difference in Martin's life.

Do you think Martin should have the genetic treatment? In other words, do you think being albino is a medical problem that needs fixing? Or would you say the treatment is more along the lines of a nose job or face-lift -- something nice, but not necessary?

Your answers to these questions are important, because genetic treatment could be expensive. Should health insurance pay for it? Maybe you say yes. However, the cost of this treatment for people who are albino may drive up the cost of health insurance for everyone. Would that change your answer?

Think about the choices Martin's mother would have to make. If she loves Martin the way he is, how does she explain a decision to have him treated? But if *he* is unhappy with the way he is, how does she explain a decision *not* to treat him? Also, many medical treatments have side effects. What level of risk is acceptable?

Perhaps when Martin grows up, he will decide that he wants to prevent his children from having the problem he has had. He may decide to have any baby of his tested before it is born, to make sure it is not albino. If it is, he and his wife could choose to have an abortion and try again. What do you think of this choice?

Adoption is another choice Martin and his wife could make, instead of risking bearing children who are albino. With adoption, the children would not be their own, genetically. But Martin and his wife could raise the children as their own, and they would not be albino. What do you think of this choice?

It's possible that when Martin grows up, he will be comfortable with how he looks. He may not care whether his children are born albino. In fact, he may even prefer it because then they would look more like him. What do you think about this? Do you think it is wise to let children into the world with problem skin and poor eyesight if we know how to keep this from happening? Another way to ask this is, Should Martin be prevented from having children who are albino? Who are we to say no to him?

Finally, there is the question of where society should put its time and money. Perhaps along with research into the treatment of genetic conditions, we should put equal effort into teaching children (and adults) to accept those who are different. What do you think?

These questions are just the "tip of the iceberg" when it comes to genetic research. There are many more. One way to explore the topic is to look at it in terms of the **ethical**, **legal**, and **social issues**. Ethical issues concern what is moral or right. Legal issues concern the protections that laws or regulations should provide. And social issues concern how society as a whole (and individuals in society) will be affected by events. To really get into all of these issues, you first need to know some of the basic science of genetics. That comes in our next chapter.

Your Genes, Your Choices is a publication of Science + Literacy for Health, a project of the AAAS

Chapter 2

Priya Should Find Out She Inherited a Fatal Disease (or should she?)

Priya has just lost her mother to an illness called Huntington's disease. It was hard for Priya to watch her mother die. First her mother had strange changes of mood. Then her arms and legs began twitching. Soon she couldn't talk or control her movements. In the end, she was totally bedridden and could barely get food down without choking.

Priya knows that Huntington's disease usually strikes people in middle age. It is always fatal, and there is no treatment. She also knows that since the disease is inherited, she has a strong chance of getting it herself.

Priya just learned about a test she can take. The test will tell if she carries the gene for Huntington's disease.

She is tempted to take the test. She thinks that if she could find out once and for all whether she will get the disease, she could plan for her future. On the other hand, she wonders if it is better not knowing. At least then Priya would still have some hope.

If you were Priya, what would you do?

CHROMOSOMES

Chromosomes

In Chapter 1, we said that genes contain the instructions for making cells and for the work that goes on inside them. The complete set of genes for a human being is called the human **genome**. It may help to think of the human genome as a book of instructions, with each gene a single instruction. You have a copy of this instruction

book inside nearly every one of the trillions of cells in your body. The book is written in an unknown language that only your cells can read.

We will have to learn this language if we want to learn all the secrets of the genes. So this is exactly what scientists from around the world have decided to do. They have set out to learn the language of the genes. This international effort is called the **Human Genome Project**. The United States is spending 3 billion dollars over 15 years on this project. Other countries also are investing large amounts on research. It is a huge undertaking that involves researchers in biology, physics, engineering, computer science, and many other fields.

The task they have taken on is challenging and exciting, but difficult. Genes are made of a threadlike material called **DNA**, which itself contains chemical ingredients called bases. There are only four bases, but they repeat one after the other in an ever-changing order throughout the genes. Think of the four bases as letters of the alphabet, combining together in some strange language to spell out each gene's instruction.

With just four letters, the alphabet of this mysterious language is very short. However, the words written in this language are not short at all. A single gene has thousands of bases. Some genes have millions of bases. So each gene is like a single word with thousands or millions of letters to it. In addition, it is hard to figure out where each gene begins and ends, because the spaces in between also are filled with long strings of letters.

DNA

This close-up shows the four bases that make up the genetic code. The bases are thymine (T), adenine (A), guanine (G), and cytosine (C).

The immediate goal of the Human Genome Project is to put together a copy of the human instruction book, letter by letter. Having this copy to read will make it easier to decipher the language of the genes. But making the copy is very difficult. First, scientists have to get inside a cell. Then they have to get inside the **nucleus** of the cell to the DNA. The DNA is curled into tight coils, so they have to uncurl it. Then they have to look at the DNA to see which of the four bases comes first, which second, which third, and so on. Then they have to write this down.

It may sound simple, but it isn't. This is a job that involves unthinkably small objects and incredibly large numbers. It would seem to be an impossible task. Yet, because of advances in computers, microscopes, chemical analysis, and other tools of science, it is a job that is only a few years from being done.

CELL-NUCLEUS-CHROMOSOME

Your Unique Genome

Once the Human Genome Project has made its copy of the human instruction book, there will still be the task of translating what it means.

And even when this translation has been done, it will only be a model. It will not reveal exactly what is written in your genome or in any other particular person's genome. This is because every human being is different. Each person's genome is unique.

Your unique genome was given to you by your parents. This took place in the process of conception. Conception is when a man's sperm fertilizes a woman's egg.

Sperm and eggs are made from special cells called germ cells. (The word "germ" is from the Latin word for "seed" or "bud".) Germ cells are found in the male's testes and in the female's ovaries. Like most other cells, each germ cell has a copy of the genome inside it. The genome is not one long strand of DNA. Rather, it is divided into separate strands called **chromosomes**, each containing several thousand genes. Human beings have 46 chromosomes.

FERTILIZATION

Conception happens when a man's sperm enters a woman's egg and fertilizes it.

To make sperm (in a man) or eggs (in a woman), a germ cell goes through a complicated process. First, it makes a copy of each chromosome. Then it divides, twice, to form four sperm or four eggs. Its double set of chromosomes is sorted equally among the four sperm or eggs, so that each has a half set of 23.

When a sperm and an egg join together, they combine their half sets to make a unique, new set of 46 chromosomes. There are so many different ways that your parents' chromosomes can be combined, that the chance of the same mix happening twice is close to zero.

The chance becomes truly zero because of something that happens as sperm and egg are formed. That something is called **crossing over**. It happens when a germ cell is making copies of its chromosomes before dividing. In crossing over, a section of one chromosome switches places with the same section from its pair. This makes the copied chromosome a little different from the original. It will carry a slightly different mix of genes.

Along with the random mixing of chromosomes, crossing over contributes to uniqueness. This is what makes you one of a kind.

Dominant and Recessive Genes

The chromosomes you inherit from each parent correspond in size and in the genes they carry. The one exception to this rule involves the two chromosomes that determine sex, nicknamed X and Y. A person who inherits two X chromosomes (XX) is female, while a person who inherits one X and one Y chromosome (XY) is male. The X chromosome is larger and contains genes that are not found on its partner Y chromosome.

DOMINANCE

Dominant Genetic Disorders

RECESSIVENESS

Recessive Genetic Disorders

The fact that you have pairs of chromosomes means that you have pairs of genes for every trait (except for those traits found only on the X chromosome). So which instruction in each pair gets followed? It depends on whether the genes in the pair are **dominant** or **recessive**. In any pair, if one gene is dominant over the other, its instructions are followed. A recessive gene's instructions come into play only if neither gene in its pair is dominant.

For example, the gene for Type A blood is dominant over the gene for Type O blood. Therefore, if you inherit the Type A gene from one parent and the Type O gene from your other parent, you will have Type A blood. The gene for Type B also is dominant over the gene for Type O. So if you inherit one Type B gene and one Type O gene, you will have Type B blood. Because the Type O gene is recessive, you will have Type O blood only if you inherit two Type O genes, one from each parent.

Something very interesting happens, however, if you inherit one Type A gene and one Type B gene. In this case, you will have Type AB blood. The instructions of both genes come into play because neither dominates over the other. In fact, many of your traits are shaped by both genes in a pair. In addition, most traits are affected by more than one gene. For example, the shade of your hair is affected by many pairs of genes working together.

How Genes Instruct Your Body

When a gene becomes active, it leads to the production of a **protein**. Proteins are the basic chemicals that make up the structure of cells and direct their activities. The human body produces thousands of different proteins. Most every protein has a different function, although there is some overlapping of jobs.

For example, one protein carries oxygen in the blood. Another protein regulates the salt in your sweat. For every function of your body, proteins are involved, and the production of these proteins is regulated by genes.

A gene in one person may carry a slightly different instruction than the corresponding gene in another person. Most variations in the same gene don't cause any problems to

health. For example, a person whose genes lead to blue eyes can see just as well as a person whose genes lead to brown eyes.

Mutations

Variations in the instruction carried by a gene come about through a process called **mutation**. A mutation is a change that occurs to the order of the bases appearing in the DNA inside a cell. Mutations can happen to any gene inside any cell of your body at any point in your life. However, most mutations occur as germ cells make copies of their chromosomes before dividing to form sperm or eggs. In this process, millions of bases must be copied in exactly the right order. Mostly, they are. However, with every copy of DNA, there are some errors. A base is put in the wrong place or is left out. Sometimes extra copies are made of a string of bases or of whole chromosomes.

These kind of changes can alter the order of bases in the affected genes. Therefore, the instructions that direct the production of proteins may change. This can affect the traits that show up.

Many mutations happened to the genes of people who lived long ago, and these mutations have been passed down through the generations. Others are new, occurring to a person during his or her lifetime. New mutations may be passed on to the next generation if they appear in sperm or egg cells.

SICKLE CELL

Inherited mutations are carried in the DNA of reproductive cells. When reproductive cells containing mutations combine to produce offspring, the mutation will be present in all body cells of the offspring.

Most mutations are harmless because they don't result in any important changes to traits. Some mutations are important because the new trait is helpful to the survival of a species. However, sometimes mutations cause problems in how your body functions. These problems are called **disorders**.

An interesting example of a mutation is the one that causes red blood cells to take on an unusual "sickle" shape, like the curving blade of a knife called a sickle that is used to cut tall grasses. This gene variation is found mainly in people who live near the equator (or whose ancestors did). Researchers believe that inheriting one gene with this particular variation may be helpful against malaria, which is a disease caused by **parasites** transmitted by mosquitoes into the bloodstream. The parasites feed off red blood cells. However, they have a hard time feeding off sickle-shaped cells. This limits the damage they can do inside the body.

People who inherit two copies of this gene variation, however, are affected by a disorder called **sickle cell anemia**. They have so many sickle-shaped red blood cells that sometimes it is hard for the blood to flow. The symptoms for sickle cell anemia range from mild to severe. Although treatment has improved in the past few decades,

many people still become very sick and die from the disease.

Genetic Testing

In the past few years, researchers have learned how to test for hundreds of genetic disorders. More new tests are coming out all the time. These tests can be very helpful for diagnosing disorders in children and adults. The tests also can be used to predict the chances that a person will come down with a particular disease later in life.

Genetic tests are sometimes used by couples who want to learn their risk of passing on genetic disorders to any children they might have. Other genetic tests are **prenatal**. That is, the tests are done before birth to an **embryo** or **fetus**, to see whether it has any genetic problems.

There are many different kinds of tests. The test used depends on what disorder is being looked for and what is known about the gene, such as its location in the genome or the protein it controls. In some cases, the test results are definite. In other cases, the test results only suggest what the person's *risk* is for developing a disorder.

One way of testing is to do a medical exam. Doctors examine the person to see if he or she shows signs of the disorder. They also may study the person's family history for clues as to how the disorder has been passed down from generation to generation.

To find some disorders, doctors may take a blood sample in order to look at the person's chromosomes under a microscope. They may test the blood sample to find proteins that would reveal a gene at work. Very advanced tests can look inside a section of a chromosome to "read" the DNA.

Concerns About Genetic Testing

Great progress has been made in genetic testing in recent years. However, progress in the treatment of genetic disorders has been much slower. That is the problem for Priya, the woman who fears that she may have inherited Huntington's disease (HD).

HD affects 1 in every 10,000 people. A mutation in one gene means that the protein it instructs the body to make gets produced abnormally. For reasons that are not yet clear, this leads to a breakdown in the parts of the brain that control movement.

The HD gene is dominant. This means that only one gene in a corresponding pair needs to carry the HD mutation in order for the disease to take hold. Priya's mother had the gene and, therefore, the disease. If Priya inherited the HD gene from her mother, she too will someday develop the disease. This will be true even if the corresponding gene

she inherited from her father does not have the HD mutation.

GENETIC TEST

A doctor looks at the results of a genetic test.

Genetic tests are available to tell Priya if she carries the gene for the disorder. However, no treatment is available. If Priya finds out through testing that she has the gene, there is little she can do. No one knows how to ward off the disease or keep it from getting worse. No one can even tell when the disease will hit or how quickly it will take over her body.

In other words, the only real use of the test is to tell Priya how she stands, one way or the other. Such news could change her life, but not necessarily in the way she expects.

For example, you might think that Priya would be happy to find out from testing that she has been spared the disease. However, it is possible that she will feel "survivor's guilt" if other members of her family have not been spared. It also is possible that once Priya stops worrying about HD, she may discover other problems in her life that she has been avoiding. Finding out that she doesn't have HD may be happy news, but it will not necessarily make Priya a happy person.

You might also think that Priya would be sad to find out from testing that she will get the disease. But Priya may discover that it is a big relief to know for sure what she has always feared. It may give her a new appreciation for each day that she has. She may feel that she can now make better decisions for the life she has left.

The fact is, Priya cannot know ahead of time how she will react to the test results. And she cannot predict other problems that may come as a result of testing. For example, in order to find out how HD is passed down in her family, doctors may want to test her close relatives, such as her brothers, sisters, aunts, and uncles. This is called a **genetic linkage** study. Priya will have to ask them to take part in the study. Asking such a thing can be very difficult. It may put extra pressure on her when she already has a lot on her mind. The relatives may feel pressured themselves. Or they may feel guilty if they refuse to take part.

Here's another problem Priya must consider: Suppose the test result says that she has the HD gene. Who else should know? Does her boyfriend have a right to know before they make plans to marry and have children? When does she tell her employer, now or when she becomes ill? What about her health insurance company, which at some point may have to provide expensive medical care for Priya?

Priya must decide how to handle the information in a way that is fair to herself and to others. However, it may not be entirely in her control. She may want to keep it secret, but have a hard time doing so. Or a relative who took part in the testing may reveal

information to others. If information on the results of the test is put into her medical records, both her employer and her health insurance company may find out without her telling them. This could cause her to lose her job, which would be illegal, but still could happen. Or it could cause her to lose her health insurance, which has indeed happened to others in this situation.

Another thing that Priya must think about is that testing could take a lot of time and be expensive. She could have a long, distressing wait for results. The answer may not be clear cut. It is even possible that the answer will be wrong because of a lab error or because the test itself is not perfect.

Priya's decision whether to get tested may be affected by recent news. In 1993, researchers located the gene in which the HD mutation occurs. This discovery means that scientists can develop a simpler and more reliable way to test for HD. The information also is useful for scientists working on a cure. Treatment may come in time to help someone like Priya. Then again, it may not.

COUNSELING

Genetic counselors are trained to help people make decisions about their health.

The Need for Genetic Counseling

HD is unusual because if you carry the gene for the disorder, you get the disease for sure and it kills for sure. Most genetic disorders are not so direct. But the issues surrounding testing remain.

That's why many people feel that **genetic counseling** is extremely important for anyone wanting to get information about their genes. With counseling from professional advisors who are experts on genetics, people like Priya can understand the facts of their situation. Counselors can help them clearly understand the limitations of tests and think through ahead of time how different test results might affect them. Finally, counselors can explain to people what their choices are once they know the results.

However, genetic counseling is a new field, and there aren't very many trained genetic counselors. Genetic testing is fast becoming a billion-dollar industry. New tests are coming out faster than new counselors are being trained. There simply aren't enough genetic counselors for everyone who should have counseling.

This problem is probably going to get worse before it gets better. Companies that sell tests want to make a profit, so they will be marketing them not just to genetics specialists, but to all doctors and directly to the public. As people hear about new tests, demand for them will surely increase. This is especially likely to happen as the tests become cheaper, more accurate, and easier to perform. People getting tested may not realize that they need to have the results explained to them. They may not know how to

ask for this. Also, doctors who perform the genetic tests, but who do not have special training in ,may not be able to answer their patients' questions.

Some people who have genetic tests may not get counseling even if they want it, because their insurance company will pay for testing, but not for the counseling to go with it. All of this means that a lot of people may be getting very serious information from tests without getting the support they need to understand the results and to make good decisions.

There's one more reason why genetic counseling is so important. With many genetic disorders, genes are only one of the factors involved. Other factors, such as lifestyle, play a part. Counselors can help people understand what they can do to avoid triggering a genetic disorder. These kinds of choices are covered in our next chapter.

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Chapter 3

Howard's Health Is Up to Him (or is it?)

Howard will turn 50 soon, and it worries him. His grandfather died of a heart attack in his fifties, and so did his father and uncle.

Several years ago, a doctor told Howard that he was at high risk for heart disease because of his family history. But the doctor said that Howard could improve his chances if he lost some weight, stopped smoking, and exercised. The doctor also told Howard to come back every year for a checkup.

Howard hasn't gone on a diet, and he hasn't given up his cigarettes or taken up exercise. He also hasn't been back to the doctor. He's afraid of what the doctor might find.

Howard can't make up his mind. Sometimes he thinks he should try to take better care of his health. Other times, he thinks that he should just accept the fact that he won't live much longer and should get as much fun out of life while he can.

If you were Howard, what would you do?

Researchers now believe that some of the roots of cancer, high blood pressure, and perhaps even alcoholism are found in the genes. They also believe that genes may play a role in the development of certain cases of obesity, some types of depression, and diabetes. The more they search, the more they are finding a link between genes and disease.

However, the truth is always complicated. It isn't so simple as "if you have the gene for a disease, you will get the disease." Here are some of the reasons why:

- *While genes play a role in many disorders, so do the conditions and circumstances of your life and the decisions you make.* In other words, heredity may influence your health, but so does your **environment**. For example, some people have genes that put them at risk for cancer. However, their chance of actually getting cancer may be much less if they do not smoke. Some people have genes that put them at risk for diabetes. But, they may never become diabetic if they watch their weight.

  Some people have genes that put them at risk for asthma. Still, they may only begin to wheeze and cough when cats are nearby. Diet, exercise, levels of stress, and access to health care are just a few of the many environmental factors that can influence the course of a gene-related disorder. Some factors in your environment are under your control, and some are not. Either way, they can affect the progress of gene-related disorders.

- *Only a few human diseases are triggered by a single gene working by itself. In most cases, a disease results from the actions of many genes.* An example of a single-gene disorder is Huntington's disease, which was discussed in the last chapter. One error in one gene leads to the fatal health problems of HD. Multiple-gene disorders, where several mutated genes come into play to trigger the problem, are much more common. Examples of multiple-gene disorders include breast cancer, asthma, and diabetes.

- *The "strength" of the genes involved in a disorder can affect its progress.* Scientists call this **genetic expression**. Take two people who have the same disease-causing gene and have pretty much the same lifestyle. In one of those persons, the gene "expresses itself" mildly. The failures in its instruction for producing a protein are rather minor. Enough of the protein is produced so that the body can stay healthy for a long time. The disease moves slowly if and when it appears. In the other person, the gene "expresses itself" strongly. The failures in its instruction for making a protein are major. A necessary protein does not get produced or is produced in the wrong amounts. The body cannot stay healthy. That person becomes ill at an early age or comes down with a severe case of the illness.

- *In many cases, different mutations in different genes can lead to the same basic problem.* An example of this is the albino condition that affected Martin, discussed in Chapter 1. The lack of skin, eye, and hair color can be caused by mutations in several different genes that are involved in the making of pigment. In some cases, the mutations result in patches of colorless skin or hair. With other mutations, the entire body has no pigment. Often, a person who has genes that cause albino coloring also has mutations in other genes that lead to mental retardation and growth problems. In each case, the disorder is called "albinism."

However, the genes involved are different, and the result is slightly different, too.

- *While there are many diseases that involve mutated genes, the reverse is not true. Many mutated genes do not lead to disease.* Sometimes people have what look like "problem" genes because they are different from those of many other people. However, these unusual gene variations don't necessarily lead to disease.

- *Some people carry a gene mutation that causes a disorder, but are not at risk for the disorder themselves.* This happens when the gene mutation is recessive. You need to inherit two such mutated genes (one from each parent) for the disease to be triggered. If you inherit only one, you won't get sick. You will, however, be a **carrier** to the next generation. This means that you may pass on the mutated gene to your children without ever showing any symptoms yourself. Your children will be at risk for getting the disease only if they inherit the disease-causing gene mutation from both you and their other parent. An example of a recessive disorder is sickle cell anemia, which was discussed in the last chapter.

- *Some disorders occur when healthy genes become damaged.* Gene damage can be caused by exposure to radiation or by a viral infection. It also can happen if you come into contact with cancer-causing substances, called **carcinogens**. Aging also introduces errors into the DNA. Depending on which genes have been damaged, and how many, different disorders may be triggered. Some people's genes appear to be more easily damaged than others. Such people are at higher risk for disorders caused by damage to healthy genes.

The truth is that everyone has at least a handful of "problem" genes. Your genes may never cause you trouble...

- ...*if* factors in your environment do not "trigger" the genes for a disorder...

- ...*if* you only have some, but not all, of the genes that come into play to cause a particular disease...

- ...*if* your genes for disorders don't express themselves strongly...

- ...*if* you have genes that only lead to a mild form of a disease or disorder...

- ...*if* your unusual genes have no effect on health...

- ...*if* your genes for disorders are recessive and you inherit only one copy, and...

- ...*if* your genes are not damaged by substances in the environment or by aging.

Genetic Determinism

Research tells us that there is no simple link between genes and disorders. Genes have *something*, but not *everything*, to do with disorders. Genes do not equal fate.

However, it is easy to fall into that simple way of thinking. The misunderstanding that genes by themselves can determine what happens to you is called **genetic determinism**. Genetic determinism can lead people to make harmful and unfair judgments about themselves and others.

This kind of simple thinking is leading Howard to fear that, no matter what he does, he will die of a heart attack just like his father, uncle and grandfather. The fact that three of Howard's close relatives died of heart attacks strongly suggests that Howard himself is at risk. He may have inherited genes that make his body less able to resist heart disease.

Researchers don't yet know how many genes are involved in heart disease or how they work to bring about the illness. There is no test yet that can tell Howard whether he has any or all of the genes that can lead to a heart attack.

However, researchers do know something about the kinds of behavior that can trigger heart disease. They know that it occurs more often in people who smoke, have high blood pressure, eat high-fat diets, and do not exercise.

So maybe the reason that Howard's relatives died of heart attacks is that they smoked, worried a lot, ate fatty foods all the time, and sat around too much. Maybe Howard can improve his chances by taking better care of himself.

On the other hand, it is possible that Howard's family carries heart disease genes that express themselves very strongly. It is possible that no matter what Howard does to keep fit, he *can't* stop those genes from bringing on a heart attack.

The fact remains that Howard simply does not know what his risks are. The way that genes and other factors work together to produce heart disease is so complicated that doctors may never be able to make any safe predictions.

Of course, Howard also needs to remember that his cause of death could have nothing to do with genes. A car accident or a bolt of lightning could get him tomorrow. No matter how much Howard learns about his genes, he will never be able to read his future.

Dealing with Genetic Knowledge

Like Howard, we all must make decisions about the way we live our lives. More and more, however, our decisions will be influenced by information we have about our genes.

Not too far in the future could be a single test that will examine thousands of your genes. The test will reveal whether any of these genes are unusual or defective.

Knowing your **genetic profile** could be very helpful to you. It could suggest what health-related behaviors you should follow. It could tip you off to have frequent checkups for genetic conditions for which you are at risk. It could help you plan your life so that you avoid behaviors and substances that trigger diseases.

At the same time, knowing your genetic profile could create problems for you. As we said earlier, everyone has a number of "problem" genes. For the most part, you don't know what your problem genes are, and you never will find out unless a health problem surfaces. A piece of paper that lists these "problem" genes could give you a lot of things to worry about that may never come to pass.

It is possible that people who learn their genetic profile will limit the choices they make based on such fears. People may choose not to marry or build a career because they believe that they are doomed by their "problem" genes. The expectation of disease may ruin their enjoyment of life.

A big question is whether children should be told information about the genes they carry or, if so, at what age they should be told. It can be difficult for children to understand some of the important facts about genetics, such as the difference between a risk and a sure thing. There is the possibility that they will misunderstand what they are told.

There also is the risk that they will not be mature enough to cope with the information. For example, if a girl has the gene that puts her at high risk for breast cancer when she is in her forties, should she be told? It may seem that the best thing is to avoid getting this information. However, many parents want to know if their children are at risk for genetic disorders. That way, the parents can be prepared and get treatment for the children in time. They also could make lifestyle choices to avoid triggering the disorder in their child. Parents will have to decide whether and how to share genetic information with their children. Society also may have some say in how genetic information is shared with children, through standards, laws, and regulations that are developed.

Discrimination Based on Genes

EXPENSE

New gene therapies will be expensive. Who will be able to afford them?

For some people, information about their "problem" genes can bring extra trouble. For example, it can cost them their health insurance. People with "problem" genes have been refused health insurance or dropped from their health plans. In other cases, they have been told that medical expenses for their genetic condition will not be covered. In still others, they have been told that their children will not be covered because they are at risk for inheriting genetic diseases. The number of such cases may increase as genetic testing becomes more common.

You might think that it makes sense to keep genetic information about you to yourself. But this may not be possible. Part of your genetic profile may be obvious to others from your family's medical history. Also, the results of genetic tests usually go into your medical records. Insurance companies may demand to see these records before they will cover you.

Some people are concerned that employers may try to use genetic information to weed out workers who are sick, or who may someday become sick, because of a genetic disorder. Under the 1990 **Americans with Disabilities Act (ADA)**, it is against the law to discriminate against workers who are disabled. The federal Equal Employment Opportunity Commission has ruled that the ADA also protects people from discrimination based on their genetic profile.

However, the fear of being discriminated against may lead people to refuse genetic testing even when it could help diagnose, prevent or treat a health problem. They may be too afraid that the information will be used against them. If that happens, then all the benefits of genetic research could come to nothing.

Genes and Behavior

In Howard's case, it didn't take any special genetic test to reveal that he is at risk for a heart attack. His risk is clear from his family's medical history and from his own smoking, eating, and exercise habits. Given these facts, Howard has a choice: whether or not to reduce his risk by adopting a healthier lifestyle.

But this raises an interesting question: How much control does Howard have over the choices he makes? If Howard decides to change his ways, is that because his genes have made him a careful person? If Howard rejects his doctor's advice, is that because his genes have made him reckless?

The study of whether and how traits for behavior are inherited is called **behavioral genetics**. Scientists have long tried to figure out whether behavior is shaped by our

genes or by how we are raised. It is called the question of "nature versus nurture." For a long time, scientists took one position or the other. They believed that either nature or nurture was responsible, but not both. Today, most scientists agree that both genes (nature) and environment (nurture) help make us who we are. What no one knows is just how nature and nurture work together.

Some researchers believe that genes shape our inborn frame of mind, or **temperament**. According to this theory, we may inherit our tendency to be shy or bold, risk-taking or cautious. However, any temperament we inherit is shaped and altered by our experiences from the moment we are born..For example, a shy boy who is encouraged to try out new situations may learn to be more outgoing. Another shy child who is pushed too quickly into strange situations may always remain timid. So might a shy child who is allowed to hide behind his parents.

There is some evidence that to some degree, abilities also may be inherited. But any such genetic trait is heavily shaped by experience. A girl who is never allowed to play sports may never develop her inborn athletic talent. The reverse also is true. A girl whose genes do not give her any athletic advantage may still become a star if she is encouraged to play, practices hard, and keeps at it.

Many people are interested in how genes shape other personal characteristics, such as sexual orientation, intelligence, and social behavior. Research into these areas is very controversial and raises many questions. For example, some researchers are trying to find out if there are any genes that contribute to homosexuality. But what if there are, and what if there aren't? If homosexual identity is caused in part by the genes, does this mean that society should be more accepting of it? On the other hand, should society be more accepting of homosexual behavior even if it is purely a lifestyle choice?

Other researchers are trying to determine how genes shape intelligence. The question is, what do we do with this information? If intelligence is controlled in part by the genes, should society spend more money educating those who lack genetic smarts to give them a boost? Or should it spend more money on the genetically gifted, who could make more use of the education? And take the question of a genetic link to criminal behavior. If such a link is found, should the police keep close tabs on people with "criminal" genes? Should such people be excused for crimes they commit, since their genes are at fault?

Some people are critical of theories linking genes and certain kinds of behavior. They say that these theories are often based on fanciful thinking or prejudice, not science. They say that this kind of research is easily twisted to support discrimination against minorities. In any event, research suggests that environment is at least as powerful a shaper of behavior as genes. And there is still the role of personal responsibility. Shy or bold, risk-taking or cautious, it is still within Howard's power to choose whether to quit

smoking, to give up sweets, and to start jogging. Most researchers do not believe that our genes fully explain our behavior.

As you can see, genetic research doesn't give us all the answers. But it surely does open up some interesting questions. This is particularly true for the part of health care that has to do with making babies. That's the subject of our next chapter.

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