

TEEN TALK

Genetics 101

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Genetics and Bleeding Disorders

You probably know what bleeding disorders are, and what some of their characteristics are. What you may not know, is how bleeding disorders are carried from one generation to the next. The simple explanation is genetics. Genes are passed from parents to child, and determine the traits of the child, including, among many other things, whether the child has genetically transmitted diseases, such as bleeding disorders. This process is complex, but scientists have been making progress in understanding more about genes in recent years.

To understand how genes work today, it is important to know how we have gained our knowledge of genes. Francis Crick, Maurice Wilkins, Rosalind Franklin, and James Watson discovered the structure of DNA, the double helix. It's structure resembles that of a twisted ladder.



In terms of genetics, this is significant because DNA stores genetic information. Thus, knowing more about the structure of DNA allowed scientists to learn more about what parts of DNA control what characteristics. Matching specific genes to traits is the purpose of the Human Genome Project.

Another important contributor to the study of genetics was Gregor Mendel. His experiments with pea plants were the first scientifically documented observations of heredity, the process of passing traits from parents to offspring. Mendel's work is the foundation of genetics today.

The information for inherited traits is contained in units called *genes*. Genes are made up of the chemical substance deoxyribonucleic acid (DNA), which contains a code that tells cells how to make protein. There are thousands of genes in every human cell. Genes are packaged into threadlike structures called *chromosomes*. Humans have 23 chromosome pairs with the last pair containing the genes that determine our sex. An XX chromosome pair

In This Issue: [Genetics and Bleeding Disorders](#)
[Sports Skinny](#)

[Genetic Research Links](#)

[National News Guest Columnist](#)

TEEN TALK

results in a female and a XY chromosome pair results in a male.

CELLS CONTAIN CHROMOSOMES, WHICH ARE MADE OF DNA, WHICH CONTAINS GENES

Each individual gene contains information that determines a specific trait.

Remember, the DNA of our cells is inherited from our parents. If one of the parents has a gene that is associated with an abnormality, such as a bleeding disorder, that abnormality *may be* inherited by that child if they received that gene. Because a child only receives half of its genetic information from either parent, the child will not necessarily inherit a bleeding disorder from a parent who has the disorder. Even if a gene is inherited, it may not result in the disease occurring.

Why? Some disorders require two genes for the disease to occur. These genes are known as recessive genes. Von Willebrand Type III is an example of a recessive trait, requiring both recessive genes. Other conditions are caused by autosomal dominant genes. These conditions only require one gene in the gene pair to be present for the trait to occur. Von Willebrand Type I and II fall into this category. The other situation involves genes found on the X-chromosomes. These are called sex-linked traits. Hemophilia fits into this category. Remember, there are always exceptions to these scenarios so we encourage you to dig deeper if you have more advanced questions or scenarios.

To reinforce the general “rules” we just discussed relating to gene

inheritance, let’s look at an example. A trait is expressed based on the combination of genes passed on for that trait. This is called our *genotype*.

Remember, since two recessive genes are needed for the trait to be expressed, how we see the trait (*phenotype*) is different than what we see when we look at the gene pair for that trait. The gene pair can only be seen by identifying one’s genes, and not by how you look on the outside. In some cases, the gene is present but is not expressed because it is recessive and hidden by another dominant gene that requires only one gene to be present (an autosomal dominant gene).

An example would be someone who has brown eyes. The gene for brown eyes is an autosomal dominant gene. The gene for blue eyes is a recessive gene. Remember, a child gets one set of genes from their mother and the other from their father. A person may have one gene for blue eyes but this gene is masked by the other gene in the pair which is a gene for brown eyes, an autosomal dominant gene.

The following tables depict this idea.

KEY:

The information in the tables below represent the genotype combinations for blue and brown eyes **each time** a child is conceived depending on the genotype combinations received from the mother and father. The description under each table shows the phenotype seen or how the gene is expressed as blue or brown eyes.

TEEN TALK

- B=brown eyes b=blue eyes
- Remember the gene for brown eyes is an autosomal dominant gene and the gene for blue eyes is a recessive gene

EXAMPLE 1

<i>MOTHER</i>			
B	b		
BB	Bb	B	FATHER
BB	Bb		
		B	

When both parents carry one autosomal dominant gene for brown eyes, 100% of the time a child will have brown eyes

EXAMPLE 2

<i>MOTHER</i>			
B	B		
BB	BB	B	FATHER
BB	BB		
		B	

When both parents carry two autosomal dominant genes for brown eyes, 100% of the time a child will have brown eyes

EXAMPLE 3

<i>MOTHER</i>			
B	b		
BB	Bb	B	FATHER
Bb	bb		
		b	

When both parents carry one autosomal dominant gene for brown eyes and one recessive gene for blue eyes, there is a 75% chance that a child will have brown

eyes and a 25% chance that a child will have blue eyes.

EXAMPLE 4

<i>MOTHER</i>			
b	b		
Bb	Bb	B	FATHER
Bb	Bb		
		B	

When one parent carries two autosomal dominant genes for brown eyes and one parent carries two recessive genes for blue eyes, 100% of the time a child will have brown eyes.

EXAMPLE 5

<i>MOTHER</i>			
B	b		
Bb	bb	b	FATHER
Bb	bb		
		b	

When one parent carries one autosomal dominant gene for brown eyes and one recessive gene for blue eyes and one parent carries two recessive genes for blue eyes, 50% of the time a child will have brown eyes and 50% of the time a child will have blue eyes.

EXAMPLE 6

<i>MOTHER</i>			
b	b		
bb	bb	b	FATHER
bb	bb		
		b	

When each parent carries two recessive genes for blue eyes, 100% of the time a child will have blue eyes.

TEEN TALK

These are simple examples of a very complex process which can include gene mutations and blending of genes.

A short glossary of the terms in this article are provided below for easy reference.

Autosomal Dominant Gene: a gene that, when present, is always expressed. For these types of genes you only need one gene for the trait to be expressed.

Cell: a small unit of living matter that contains DNA

Chromosome: A part of a cell that contains genes

DNA: DNA is found on genes, and the way that the units of the DNA are combined determines characteristics of an organism

Gene: Basic unit of heredity; genes transmit characteristics from one generation to the next.

Genetics: the study of heredity

Genotype: the genetic makeup of an organism

Phenotype: how the genes(s) are expressed and seen in the body

Recessive Gene: A non-dominant gene that is only expressed when two recessive genes are present. It is hidden when paired with an autosomal dominant gene

Sex-linked Trait: A trait inherited through genes located on the X or Y chromosome. Most that are known are located on the X chromosome.

Genetic Research

In order to be able to treat bleeding disorders, and perhaps one day find a cure, scientists need to study the causes of the disease. As established in the previous article on genetics, bleeding

disorders can be passed on from one generation to the next through the genes they inherit. Therefore, scientists want to study genetic data. This will allow scientists to focus on how genetic information is passed on, how diseases are produced, and identify the genes that cause them.

The Human Genetic Cell Repository and the Human Genome Project are two major research projects dealing with the study of genes. The Human Genome Project has a general focus, which is to map the entire human genome in an effort to sequence the 100,000 genes that determine human characteristics. From 1989, when the project began, to 2000, more than 90% of this information was complete. This project has resulted in a rough map of the human gene structure and helped identify which genes have what affect on the body. For instance, we can identify the genes that cause some bleeding disorders.

The Human Genetic Cell Repository is sponsored by the National Institute of General Medical Sciences (NIGMS) and funded by the National Institutes of Health (NIH) and several other foundations. A repository, or cell bank, is a collection of cell lines that contain certain diseases that scientists may wish to study. Based on past scientific information and the Human Genome Project's mapping of the human gene structure, certain diseases can be identified as being caused by certain gene problems. The Coriell Institute for Medical Research, which houses the Coriell Cell Repository, can obtain cell cultures or DNA from just four tablespoons of blood or a skin biopsy and create cell lines for different diseases. These cell lines can be

TEEN TALK

maintained for long periods of time at low temperatures.

The Coriell Institute for Medical Research distributes these cell cultures and DNA for worldwide research when requested by any qualified scientist. By extracting cultures from their Repository and sending cells to researchers, The Coriell Institute allows scientists who cannot grow cells in their own laboratories to receive genetic material from the repository.

In fact, scientists from 55 different countries have used resources provided by the Coriell Institute as the basis of their studies. Over 200,000 cell cultures and DNA samples have been shipped to research facilities. By distributing these cell lines, The Coriell Institute makes possible studies that can result in early diagnosis, prevention, and treatment of diseases.

The Repository has continued to facilitate efforts to understand genetic diseases. The entire collection consists of almost a million vials of cells, representing about 2000 different genetic diseases. They have collected cell lines associated with single gene defects, and have started a new initiative for the cell lines of complex disorders including several types of cancer, Parkinson's disease, multiple sclerosis, asthma, and many more. The Human Genetic Cell Repository also stores cell lines for Factor II coagulation, Factor V deficiency, Factor VII deficiency, and Hemophilia B disorders. The Human Genetic Cell Repository and the Human Genome Project are bringing the world closer to a complete scientific understanding of diseases and the genes that carry them.

More information concerning current genetic research projects can be found at '<http://coriell.umdnj.edu/>' and

www.ornl.gov/sci/techresources/Human_Genome/home.shtml.'

National News **The Genetic Information Nondiscrimination Act-An Update**

In our first issue, we told you about the Genetic Information Nondiscrimination Act, passed unanimously by the U.S. Senate in October of 2003, with a vote of 95 to 0. Now, the House of Representatives must pass it before becoming a law. We described the importance of this act for people with bleeding disorders, as most bleeding disorders are genetically based. This mapping of the human genome creates an urgency associated with this issue, because now we have the technology to do genetic testing before we have dealt with how the information should be used. We would now like to give you an update on new developments concerning this piece of legislation.

The Genetic Information Nondiscrimination Act prevents discrimination based on genetic information. Therefore, under this act employers and insurers could not give preference to someone who does not have a bleeding disorder if that was the only difference between two people applying for insurance or for a job. Also, employers could not purchase genetic information about a current or prospective employee or use genetic information to fire or make decisions about training current employees. For instance, many employers give health care benefits to their employees. If genetic testing reveals a disease, like a bleeding disorder, employers know that

TEEN TALK

health care for that person will be more costly, and therefore may hire somebody else. In addition, insurance companies would not be able to charge those with bleeding disorders more than they would anyone else for insurance coverage.

Since our last discussion of this bill, it has arrived in the House of Representatives for further consideration. Fortunately, the House Employer-Employee Relations Subcommittee heard testimonies in July of 2004 concerning the possible implications of genetic testing. The Chairman of the Subcommittee, Sam Johnson, said, "With this unprecedented potential for discovery comes an equally weighty challenge for public policymakers. That is information that seems to indicate the possibility of illness, disease, or other disorders could be used unjustly against people and their families." Even now, there are some laws that protect the privacy of genetic information, preventing discrimination based on this information. The Health Insurance Portability and Accountability Act, or HIPAA, the 1964 Civil Rights Act, and the Americans with Disabilities Act, or ADA, all govern the privacy of Americans. In addition, 32 states have laws that prevent insurance companies from using genetic information to a client's disadvantage. Furthermore, President Bush has indicated that he supports the Act and will sign it if it passes the House.

Along with the Genetic Information Nondiscrimination Act, these are great steps moving us closer to insuring equal opportunities for those with bleeding disorders or other genetic conditions. Genetic knowledge is valuable for scientific study, but who can have access to this information must be decided, so employers and insurance

companies do not have access to information that creates a bias against those with bleeding disorders or other genetically based condition.

Sports Skinny

With spring, many of us may feel reenergized and ready to tackle something new. If you have that urge, consider tackling a new fitness goal, the MFQ. MFQ stands for Mental Fitness Quotient. Every day, as teens, we are asked to perform and be on top of things. This constant pressure and stress can sometimes seem overwhelming.

Most people know when they are not physically healthy but not many people know when they are not at the peak of mental fitness. The company, Procoach Systems, is all about mental toughness. It is a consulting firm that helps people achieve success professionally and outside of work or school.

Mental fitness is not about how fast you can run the mile, or how long you can swim, it is about enjoying life in a whole new light. When total mental fitness has been reached, many people feel just as energized as they do after a good work out.

If school is stressing you out, and you need to "check-in", consider going to the website for Procoach Systems (www.mentalgamecoach.com) and taking the MFQ quiz or reading some of their free articles. Maybe this will help you shake the winter blues and get ready for spring. The articles and simple quiz found at the Procoach Systems website were written by Bill Cole, MS, MA, the founder and CEO of Procoach Systems.

TEEN TALK

Click on this link, www.MentalGameCoach.com and have fun!

Links

In this issue of Teen Talk, we reviewed the Genetics Home Reference web site at <http://ghr.nlm.nih.gov/>. The National Library of Medicine at the National Institutes of Health, Department of Health and Human Services, introduced this new web site. This site has a lot of great information on all different kinds of genetic disorders.

When reviewing this web site, we were blown away by the detailed information provided. First of all, on the homepage there is tons of different parts. There are browse options, a search engine, resources, a handbook, and some genetics news. For every genetic disorder included on the web site, they had ten other links leading to more information on that particular disorder. This web site tells you everything known about genetic disorders, when they were discovered, who was the discoverer, current related research projects going-on about them, and medically based solutions or preventive measures that should be taken if you have the disorder.

The search engine on this site is one of the most detailed one's we have ever seen. When we searched under bleeding disorders we got 18 hits. Factor V, Ehlers-Danlos syndrome, hemophilia, and thrombophilia all came up. The only thing I noticed that I could not find was von Willebrand Disease. It includes anything even slightly related to what we searched for. This search engine allows you to find things fast and easy.

If you consider yourself a “reader” this site is for you! The reading level on this web site is at a very high level. Unless you already know about a particular genetic disorder, most of the information given to you will be very confusing. This is not a good site for people just starting to learn about a genetic disorder. This site is more for the person who wants to know even more detailed information and specifics about a genetic disorder.

During this review, not one “visual” showed up on the screen. For those teens who are visual learners, this is *not* a good site for you. There are no pictures! The only way you might find this site useful is when you need to write those dreaded research papers or want to check up on research studies related to your condition! Otherwise, this site might not be for you.

Overall, the Genetics Home Reference site is good for detailed information and specific in depth information on the field of genetics. It is very well organized and laid out. The site has new information which can be found using a dynamic search engine. This site is great for someone who wants scientific information written at a high level.

	Excellent	Very Good	Good	Poor
Amount of Info	*			
Info for				*
Easy to		*		
Searchable	*			

Overall rating: Very Good

TEEN TALK

Guest Corner

Teen Talk is still looking for a guest columnist for the next issue! If you write and send us a publishable article concerning Blood 101, you could receive ten dollars! The article can be a story, idea, opinion, or tip! Be creative, it's up to you! The article needs to be well written and polished so we can easily put it in our newsletter. Also, limit your article to 500 words, please. So get those pens out and start writing! You can send those publishable articles to Ali and Derick Stace-Naughton at <pjstacen@wisc.edu> with the subject line "Teen Talk"!

Don't
Forget to
Look for
Our Next
Issue of
Teen Talk
on Gene
Therapy!