Chapter 6-McGraw Hill-Ryerson

The Human Genome Project

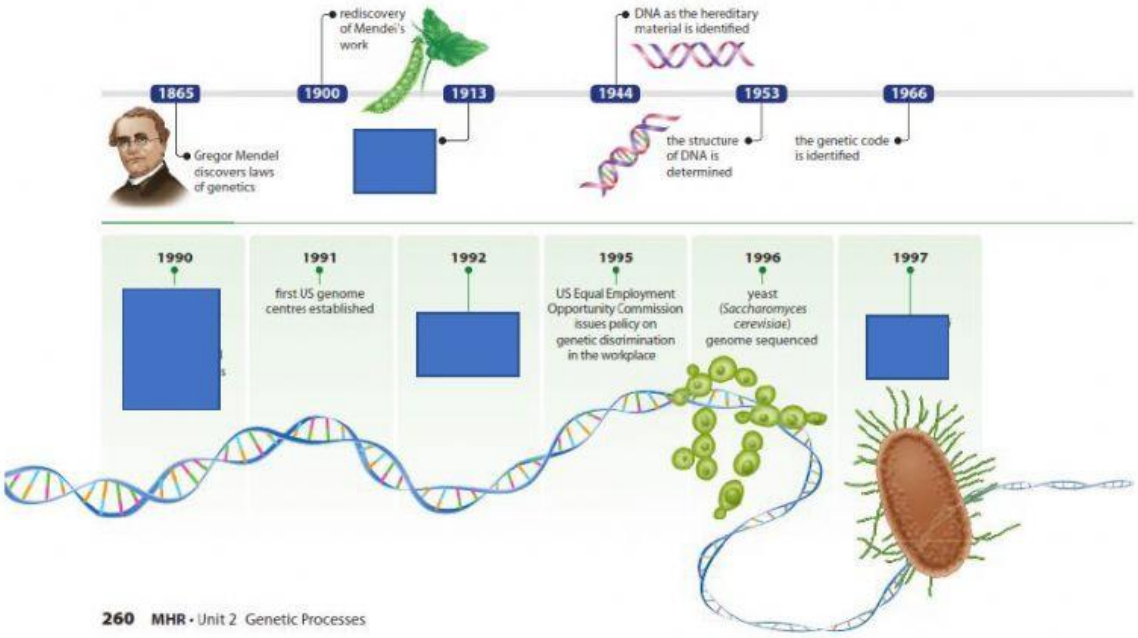
In the opener for this chapter, you were introduced to the Human Genome Project.

Determining the DNA sequence of the human genome is one of the most pivotal contributions to science ever made. Nevertheless, achieving this scientific landmark depended on many discoveries that came before it.

Figure 6.17 highlights only a small number of developments since Mendel's work that formed the foundations of this project.

An important component of the _____Human Genome Project was determining the _____of other organisms. This allows scientists to make comparisons between species and learn even more about important features of genomes. Overall, identifying the genome sequences of humans and many other organisms allows for a much more comprehensive understanding of biological systems. This knowledge will have a wide range of applications in fields such as _____, _____ and _____.

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What's in Our Genome?

In addition to determining the actual sequence of the _____in the human genome, scientists had to make sense of the sequence. Trying to make sense of the sequence can be compared to reading a _____ written in a language nobody knows or understands.

Imagine the _____ as words in a book written without capitalization, punctuation, or breaks between words, _____, or paragraphs. Also, suppose there are strings of additional letters _____ randomly between and within sentences.

Figure 6.18 shows how a page from such a book might look. To understand what is written, you must decode the jumbled text. Similarly, scientists had to decode the sequence of our DNA to learn about the human genome. When the Human Genome Project began, there was a great deal that was not known about our genome. For example, it was not known

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how many _____ humans had and how much of our DNA is part of those genes.

After sequencing the entire human genome, scientists observed many things that surprised them. Some of these discoveries include the following:

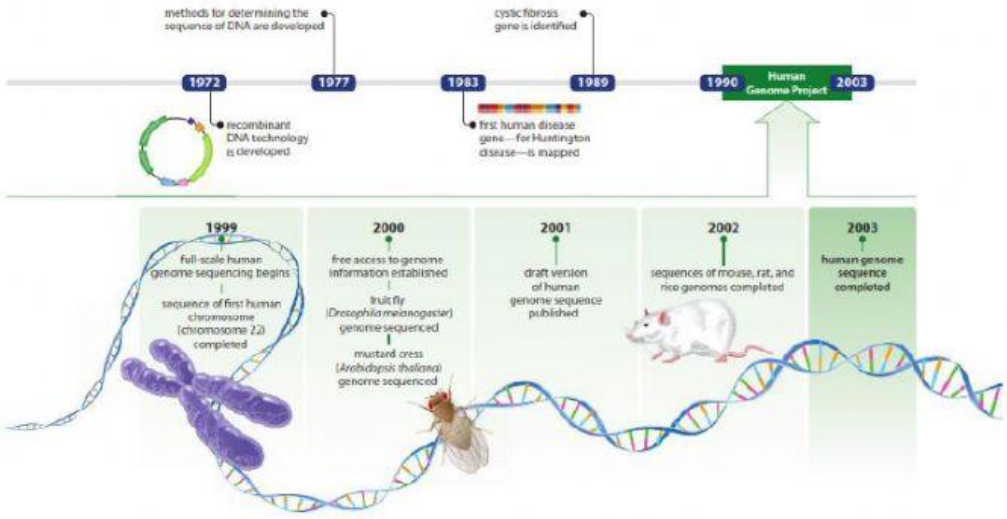
• Only about _____ of the nucleotides in the human genome make up our genes and code for all the _____ in the body.

• The estimated _____ total number of genes is much less than scientists predicted. Previous estimates were between _____ and _____.

• Over _____ percent of our DNA consists of stretches of _____ sequences.

• There is very little genetic _____ within our species. About _____ percent of the DNA sequence is almost the same in all people.

Having the sequence of the human genome only represents a _____ point. It is like being given the pages of an instruction manual for the human body. The next steps involve figuring out how to interpret all the information and use that to understand how everything works together. Scientists agree that this process will take many more years of research.



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The Development of Bioinformatics

Sequencing the human genome and the genomes of other organisms generated exceptionally large amounts of _____ that needed to be _____ and _____ among labs around the world. A new field of study, called _____, arose from this need.

Bioinformatics is a branch of biology that deals with applying _____ technology to create and maintain databases of _____ that can be _____ to better understand _____ processes.

Bioinformatics is a relatively new branch of biology. American chemist Margaret Dayhoff , is the founder of bioinformatics. Her work, which began in the late _____, involved creating a computerized _____ and _____ sequence database—the first bioinformatics project. Today's bioinformatics exists because of simultaneous advances in three areas: _____ to sequence _____ molecules such as DNA and proteins, _____ database _____ to sort and store massive amounts of genetic information, and _____ technology to share information around the _____ efficiently. Today, there are many on-line genetics databases available that allow easy access to vast amounts of genetic information by all members of the public—not just scientific c researchers.

Bioinformatics is just one of several newly developed fields, all of which involve using computers to study biological problems. For example, computational biology involves developing _____ models and computer simulations of biological processes.

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Genomics: The Study of Genomes

Just as genetics is the study of genes, _____ is the study of genomes and how genes work together to control phenotype, as illustrated in **Figure 6.20**. Although some traits are determined by only one _____, most traits involve _____ genes. To understand how an individual gene produces a specific _____, researchers such as Mendel and Morgan chose one gene and studied it and its phenotype across many individuals. A significant advantage that came from the Human Genome Project was the ability to _____ multiple genes and the genome as a whole. This allows scientists to study the _____ among many genes and how they all contribute to a phenotype. Computer technology and fields such as _____ play a vital role in this by allowing scientists to analyze large amounts of information from a variety of sources.

Although there is little variation in the sequence of the human genome, it is important to keep in mind that the _____ percent difference represents potential for variation in about _____ million nucleotides. Some of this variation is associated with many _____. Scientists believe that almost all human diseases have a genetic component, either _____ or _____. Comparing genome sequences has been particularly useful in studying the genetic basis for many human diseases, such as cancer. For example, bioinformatics and computational biology have been used to compare the DNA sequences of certain regions of the genome in individuals affected by a particular type of _____ with the DNA sequences of the same regions in those who are not. Differences in DNA sequence indicate a potential genetic basis for the disease. While this represents a good starting point for the study of the genetics of a disease, scientists are discovering that many

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diseases are the result of a _____ array of factors, and studying them requires more _____ methods.



Linking Genetic Variations to Disease

In previous sections, you learned about diseases that are associated with a mutation or mutations in a single gene, such as _____ cell anemia. Many other diseases, such as cancer, stroke, heart disease, diabetes, and asthma, are influenced by a combination of _____ and _____ factors.

Many scientists consider determining what variations in DNA sequence contribute to different diseases to be one of the best opportunities to understand the complex causes of many human diseases. The most common type of variation between people is differences in _____ nucleotides, as shown in **Figure 6.21**. For example, one person may have a C at a certain location, while another person may have a T. This type of genetic variation is called a

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_____ nucleotide polymorphism, or SNP (pronounced "snip"). A SNP can act as a marker for a gene or be associated with a gene if it is genetically linked to it. Recall that sequences of DNA are genetically linked when they are physically _____ to each other on a chromosome and tend to be _____ together. For example, if a SNP is common among people with high blood pressure, that could provide a marker for the location of a gene that is involved in the disease. However, there are almost _____ million different SNPs that commonly occur in the human genome. Testing all of these is not feasible. Nevertheless, SNPs that are near each other on a chromosome tend to be _____ together. These regions of genetically linked variations are called _____. Certain tag SNPs can uniquely identify these haplotypes. Since there are far fewer of these types of _____, they can be used as a basis for comparing genetic variations and identifying genes that influence the health of an individual.

In 2002, an international group of researchers from Canada, the United States, Japan, China, Nigeria, and the United Kingdom collectively began the International _____ Project. The major aim of this project is to develop a haplotype _____ (HapMap) of the human genome, which represents a map of the _____ in the human genome. This can then be used by other scientists to identify the genetic basis for many human _____.

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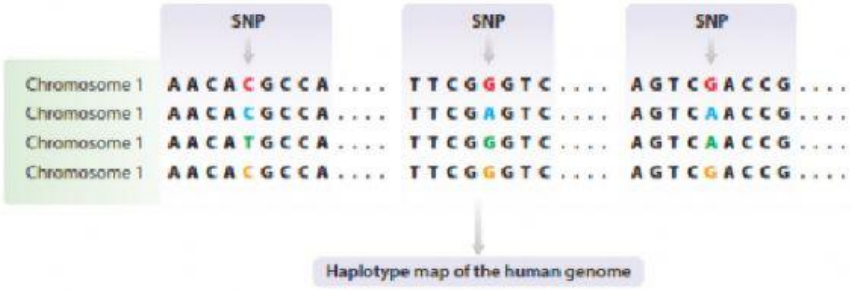


Figure 6.21 A haplotype map is constructed by identifying single nucleotide polymorphisms (SNPs) among a number of individuals.

Genetic Information: Public Benefits and Concerns

Some of the most important benefits of genetic research are in the area of human

_____.

Figure 6.23 illustrates this link between genetics and medical treatment.

Studying the human genome as a whole may make it possible to develop drugs that are tailored to the _____ of the genes associated with particular _____, and to the unique genome of a _____.

In the future, researchers hope to use established links between genetic variation and risk of disease to provide better medical advice to patients. If the cost of DNA sequencing continues to _____, individuals may have access to their **genetic** _____—their complete genotype, including all of the various _____ linked to disease. Currently, doctors are only able to make generalized risk assessments based on medical history. Armed with a genetic profile, however, genetic counsellors and doctors will be able to provide more specific risk assessments, design _____ prevention plans, and design genetically precise treatment programs.

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What Can Happen to Information from a Genetic Profile?

Establishing genetic profiles for individuals and making these profiles available to health-care providers, also creates ethical concerns. For example,

• Could insurance companies _____ coverage to people who have a genetic _____ for a particular disease?

• Could potential _____ have access to an individual's genetic profile and use it in assessing whether to _____ the person?

• Should _____ be allowed to use the genetic profiles of individuals to help them better understand the link between genome and phenotype?

The central issue in all of these ethical questions is _____ should have _____ to the information in a genetic profile.

Ownership of Genetic Information

All the data gathered through the Human Genome Project is _____ available. Having access to the data made it possible for scientists to share what they learned about human genetics. In other areas of genetics research, however, the relationship between public and private information is more complex.

In _____, the National Geographic Society and the IBM company jointly launched the _____ Project. This project uses DNA samples provided by _____ of thousands of volunteers around the world to learn more about the migrations of _____ peoples. Using high-tech genetics tools and computer facilities, DNA sequences of the individuals are analyzed to better understand human genetic roots and how we all "connect" at the level of our DNA.

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