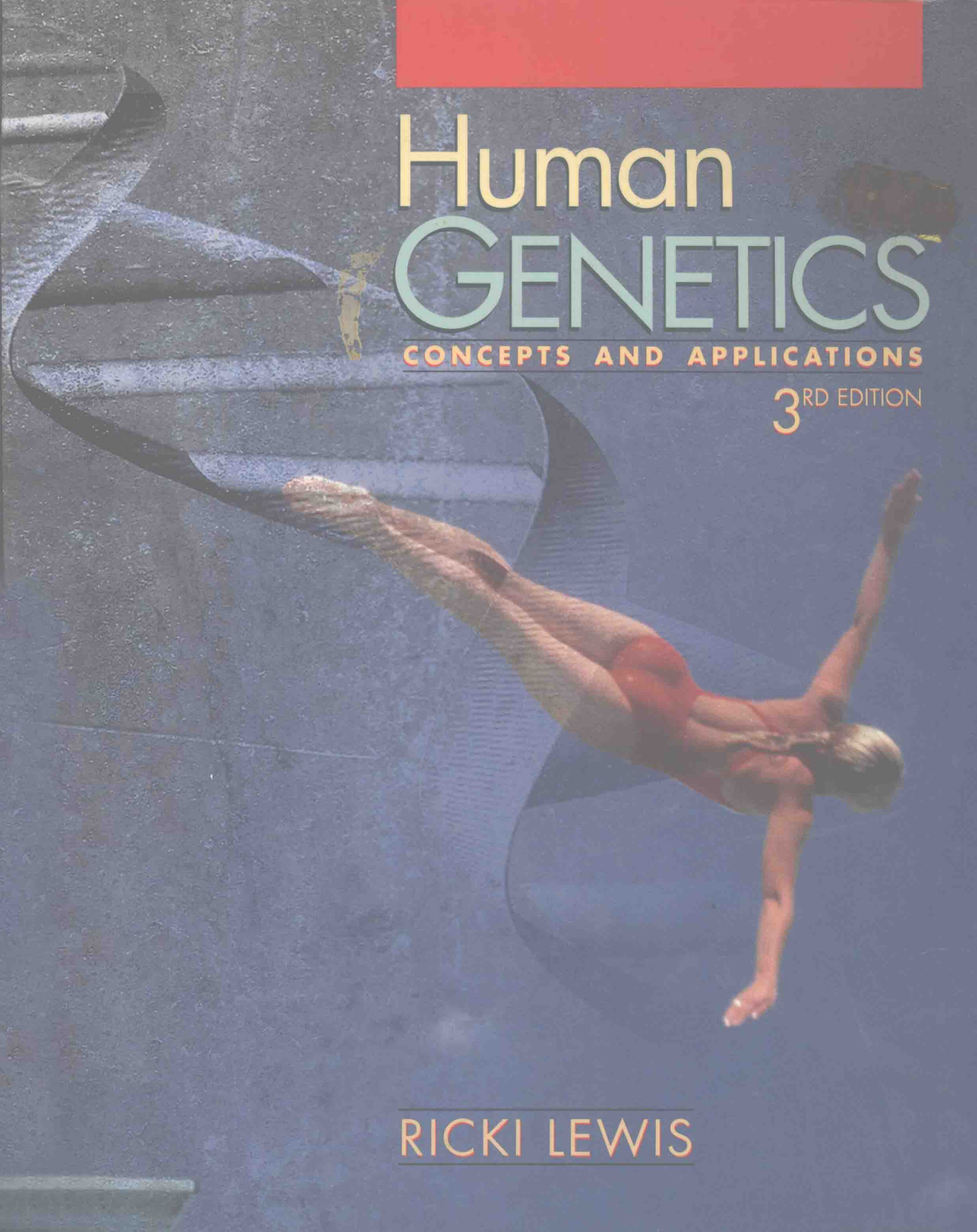

Human GENETICS

CONCEPTS AND APPLICATIONS

3RD EDITION

RICKI LEWIS

HUMAN GENETICS

Concepts and Applications

third edition

Ricki Lewis

The University at Albany
CareNet Medical Group, Schenectady, New York

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HUMAN GENETICS: CONCEPTS AND APPLICATIONS, THIRD EDITION

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preface

I had expected that by the time I wrote the preface to *Human Genetics: Concepts and Applications,* third edition, I would enthusiastically trumpet all of the spectacular new discoveries and describe how they are impacting on our lives. Yet here I sit, befuddled. If we've learned anything about genetics over the past two years, it is that we don't know, or understand, nearly as much as we thought we did.

Consider:

- Testing for inherited breast cancer, and some other disorders, has proven to be much more complex than we expected. Studies on different population groups for inheritance of cancer-causing gene variants yield strikingly different results, making widespread genetic testing a long way off.
- Unraveling the complete genetic blueprints of the simplest species has revealed many genes whose functions we know nothing about. Imagine how little we know of our own genes!
- Many gene therapies, which made perfect sense theoretically, are disappointing in clinical reality.
- "Knocking out" genes in mice to create models of human disease often shows that genes once thought to be vital may not be so.

In the words of Alice, genetics is getting curiouser and curiouser. Added to the recognition of how much we still don't know is society's sometimes negative perception of the field. Information on cloning a mammal jumped to the media and political arena without much explanation and analysis from geneticists, who have been working on cloning for decades. People refuse genetic tests for fear of discrimination by employers or insurers. The possibility of using genetic engineering to create powerful bioweapons lurks. And genetic determinism attempts to anchor all manner of behavioral traits strictly to DNA sequences, downplaying the role of the environment and what happens to us after those initial genetic instructions are set down.

Teaching and learning human genetics amidst these uncertainties and controversies is a daunting challenge. *Human Genetics: Concepts and Applications* confronts that challenge.

What's New in This Edition

Beyond the Single Gene In the early days of human genetic research, and to an extent today, matching gene to protein to disease was paramount. Identifying the abnormal salt channel behind cystic fibrosis, or the protein whose absence causes muscular dystrophy, were important discoveries, but we are learning that gene function often goes beyond a one gene-one protein explanation. And so the most important change in this edition is a subtle shift in emphasis, from considering the gene in isolation, to viewing the gene as an entity that interacts with other genes, and with environmental factors, to mold who and what we are.

In Real Life As in previous editions, true life stories bring the concepts of human genetics alive. New tales include

- Joan/John, who had his penis removed as an infant and fought for years the well-meaning efforts to raise him as a her.

- "A Personal Look at Klinefelter Syndrome" by a young man who discovered that he had an extra chromosome when he tried to become a father.
- "Ashley's Message of Hope" comes from the parent of a child who died because she was missing a small part of a chromosome.

Technology Technology Timelines continue to show, at a glance, how methods to analyze genes and chromosomes, or treat diseases, have matured. A new timeline chronicles the ongoing effort to understand how Huntington disease develops.

Figures highlight technology, too. New figure 11.6 shows the evolution of the karyotype, from crude chromosome spreads done half a century ago, to today's spectacular chromosome paints. Ironically, figure 7.1 shows how some things *don't* change much, such as the continuously varying nature of complex traits. Other technologies updated in this edition include DNA-based forensics, human artificial chromosomes, cloning, umbilical cord stem cell technology, tissue engineering, and intracytoplasmic sperm injection. But the limits of technology are discussed as well. The first chapter addresses the ambiguity of genetic testing, and the final chapter probes genetic discrimination.

Summaries This third edition excels in encapsulating major concepts into summary figures and many new tables. Figure 13.7, for example, summarizes and compares, in one place, how nonrandom

Ashley's Message of Hope

reading 11.3

What is it like to have a child born with cri-du-chat syndrome? How does this affect the family and its future? What kinds of assistance can the medical community offer the family?

The birth of any child raises many questions. Will she have my eyes, her dad's smile? What will she want to be when she grows up? But the biggest question for every parent is "Will she be healthy?" If complications occur during birth or if the child is born with a genetic disorder, the questions become more profound and immediate. "How did this happen?" "Where do we go from here?" "Will this happen again?"

Our daughter, Ashley Elizabeth Naylor, was born August 12, 1988. We had a lot of mixed emotions the day of her birth, but mainly we felt fear and despair. The doctors suspected complications, which led to a cesarean section, but the exact problem was not known. Two weeks after her birth, chromosome analysis revealed cri-du-chat (cat cry) syndrome, also known as $5p^-$ syndrome because part of the short arm of one copy of chromosome 5 is missing. The prognosis was uncertain. This is a rare disorder, we were told, and little could be offered to help our daughter. The doctors used the words "profoundly retarded," which cut like a knife through our hearts and our hopes. It wasn't until a few years later that we realized how little the medical community actually knew about cri-du-chat syndrome and especially about our little girl!

Ashley defied all the standard medical labels, as well as her doctors' expectations. Her spirit and determination enabled her to walk with the aid of a walker and express herself using sign language and a communication device. With early intervention and education at United Services for the Handicapped, Ashley found the resources and additional encouragement she needed to succeed. In return, Ashley freely offered one of her best loved and sought after gifts—her hugs. Her bright eyes and glowing smile captured the hearts of everyone she met.

In May of 1992, Ashley's small body could no longer support the spirit that inspired so many. She passed away after a long battle with pneumonia. Her physical presence is gone, but her message remains: hope.

If you are a parent faced with similar profound questions after the birth of your child, do not assume one doctor has all the answers. Search for doctors who respect your child enough to talk to her, not just about her. Above all, find an agency or a school that can help you give your child a chance to succeed. Early education for your child and support for yourself are crucial.

If you are a student in a health field, become as knowledgeable as possible and stay current with the latest research, but most importantly, be sensitive to those who seek your help. Each word you speak is taken to heart. Information is important, but hope can make all the difference in a family's future.

—Kathy Naylor

figure 1

Ashley Naylor brought great joy to her family and community during her short life.
Courtesy of Kathy Naylor.

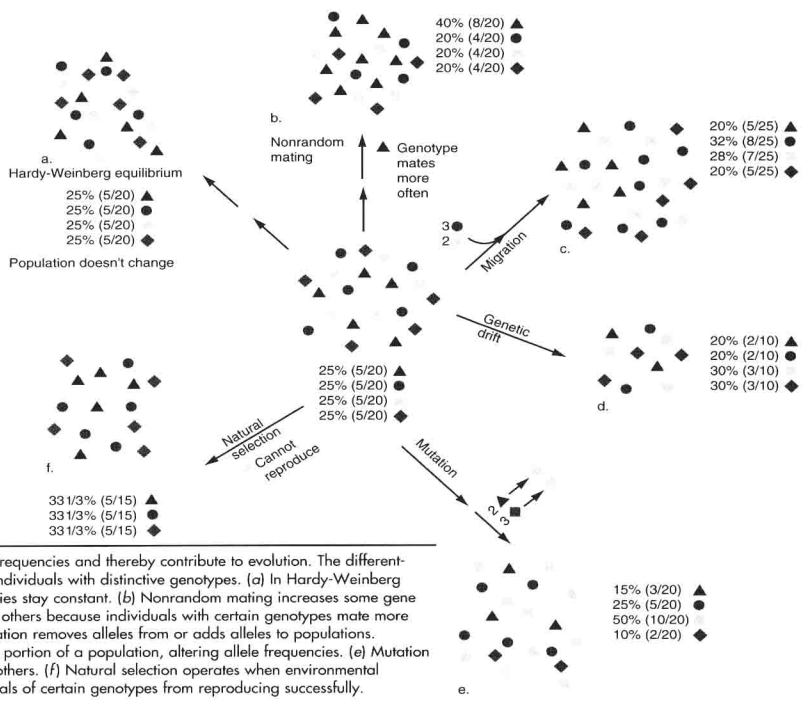

figure 13.7

Several factors alter gene frequencies and thereby contribute to evolution. The different-colored shapes represent individuals with distinctive genotypes. (*a*) In Hardy-Weinberg equilibrium, gene frequencies stay constant. (*b*) Nonrandom mating increases some gene frequencies and decreases others because individuals with certain genotypes mate more often than others. (*c*) Migration removes alleles from or adds alleles to populations. (*d*) Genetic drift samples a portion of a population, altering allele frequencies. (*e*) Mutation changes some alleles into others. (*f*) Natural selection operates when environmental conditions prevent individuals of certain genotypes from reproducing successfully.

Technology

Transplantation

1899	First allograft—a kidney from dog to dog.
1902	Pig kidney is attached to blood vessels of woman dying of kidney failure.
1905	First successful corneal transplant, from a boy who lost an eye in an accident to a man whose cornea is chemically damaged. Works because cornea cells lack antigens.
1940s	First kidney transplants on young people with end-stage kidney failure.
1950s	Blood typing predicts success of potential donor-recipient matches for organ transplants.
1960s	First effective immunosuppressant drugs revive interest in human allografts. Kidney xenografts between baboons and chimpanzees.
1967	First human heart transplant. Patient lives eighteen days.
1968	Uniform Anatomical Gift Act passes. Requires informed consent from next of kin before organs or tissues can be used for organ donation.
1970s	Transplants fall out of favor because they extend life only briefly and do not correct underlying disease, and because surgical complications and rejection reactions are common.
1980s	Improved immunosuppressant drugs, surgical techniques, and tissue matching, plus ability to strip antigens from donor tissue, reawaken interest in transplants.
1984	Doctors at Loma Linda University Medical Center transplant a baboon's heart into "Baby Fae," who was born with half a heart. She lives twenty days before rejecting the xenograft.
1992	Surgeons at the University of Pittsburgh Medical Center transplant a baboon's liver into a thirty-five-year-old man with hepatitis. The man lives for seventy-one days, dying of an unrelated cause.
1995	An AIDS patient receives bone marrow from an HIV-resistant baboon.
1997	Pig cell implants used to treat pancreatic failure and Parkinson disease.

New Summary Tables

5.3	Mechanisms of Non-Mendelian Inheritance
8.1	Experiments that Led to Discovery of DNA's Structure
11.1	Indications for Amniocentesis
11.3	Signs and Symptoms of Down Syndrome
11.4	How Nondisjunction Leads to Sex Chromosome Aneuploids
14.2	Cultural Ages
16.2	Cancer Genes
17.4	Transgenic Mouse Models of Human Disease
17.5	Knockout Mouse Models of Human Disease
18.4	Routes to Gene Therapy
18.5	Ex Vivo, In Situ, and In Vivo Gene Therapy
20.4	Assisted Reproductive Disasters
21.1	Nonhuman Genome Projects

Concepts/Applications Paradigm

Concept Chapter	Application Chapter
2 Cells	16 The Genetics of Cancer
	18 Gene and Protein Therapy
3 Human Development	20 Reproductive Technologies
6 Matters of Sex (linkage)	21 The Human Genome Project
8 DNA Structure & Replication	19 Agricultural & Environmental Biotechnology
9 Gene Function	12 When Gene Frequencies Stay Constant
10 Gene Mutation	13 Changing Gene Frequencies
11 Cytogenetics	14 Human Origins and Evolution
15 The Genetics of Immunity	17 Genetic Engineering

mating, mutation, migration, genetic drift, and natural selection change gene frequencies and alter Hardy-Weinberg equilibrium. In addition, all figures with chromosomes have been redone to emphasize clarity and consistency.

If It Isn't Broken—Don't Fix It!

The organization of concept chapters matched to application chapters continues in the third edition. Review and Applied Questions, Key Concepts, Chapter Outlines, and Suggested Readings remain as well, with appropriate updates.

Acknowledgments

This edition is dedicated to genetic counselors, who link families to researchers and physicians. Genetic counselors explain the science and technology, while comforting, educating, and providing perspective to patients and their loved ones. The scientific field of human genetics could not evolve into a medical specialty without them.

Many thanks to my wonderful family and pets, and to the terrific team at McGraw-Hill—Terry Stanton, Cathy Smith, Jim Smith, and Marty Lange. A special thanks to Toni Michaels, photo editor.

Supplementary Material

Instructor's Manual and Test Item File. A new Instructor's Manual/Test Item File, prepared by the author, is available to instructors and features detailed chapter outlines, answers to chapter questions, additional questions, and their answers. The Test Item File contains 20 to 30 objective multiple choice questions per chapter that can be used to generate exams. Many of the questions have been rewritten to increase rigor, and questions on new material have been added.

Microtest III. Microtest is a computerized classroom management service that includes a database of objective questions suitable for preparing exams and a grade-recording program. The software requires no programming experience and is available in DOS, Windows, and Macintosh formats.

Transparencies. A set of 50 transparencies is available free to adopters and consists of 50 illustrations from the text.

Case Study Workbook. This workbook was written by Ricki Lewis and is available to students and instructors for additional reading and problem solving (ISBN 22287).

Answer Key to the Case Study Workbook. This answer key contains the answers and solutions to the case studies covered in the Case Study Workbook.

Gene Game Software. This software program, written by William Sofer of the State University of New Jersey-Rutgers, is an easy to use, interactive Macintosh software game. It requires students to use critical thinking skills and apply the scientific method in cloning a fictitious fountain of youth gene. The *Gene Game* can be packaged with this text (ISBN 24893).

Explorations in Cell Biology and Genetics CD-ROM. This CD-ROM is an interactive multimedia program developed by George Johnson, of Washington University, and WCB. It calls on students to manipulate variables and examine how they impact the results as they explore genetics-related topics such as: Constructing a Genetic Map, Reading DNA, Exploring Meiosis: Down Syndrome, and more. The CD-ROM is compatible with both Windows and Macintosh systems (ISBN 29214).

Genetic Inheritance: Peas and *Drosophila* Software. This software program, developed by Mark Browning, Purdue University, allows the student to simulate hundreds of genetic crosses right at his/her computer to gain valuable practice in the quantitative aspects of genetics. Both the pea and *Drosophila* experiments investigate Mendel's laws of segregation and independent assortment, and how numbers of offspring affect test results. Once the student has mastered these concepts, he/she can be further challenged with the *Drosophila* experiments that explore the concepts of monohybrid, dihybrid, and trihybrid crosses, as well as the determination of linkage, map distances, and gene order on chromosomes. The software can also be packaged with the text (Macintosh ISBN 28861, Windows ISBN 35225).

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I thank the reviewers of the first and second editions for their valuable observations and suggestions.

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brief contents

chapter one

Overview of Human Genetics

A Look Ahead

The year is 2003. A nurse draws blood from a pregnant woman for a routine prenatal exam. In the blood are a few immature red blood cells from the fetus. The nurse sends the blood sample to a laboratory, where a technologist, using an instrument that recognizes and separates out unusual cells, collects a few fetal cells.

An initial peek within the cells, at the **chromosomes** that carry **genes,** reveals the first piece of information—the individual will be a boy, shown when a fluorescent dye highlights a Y chromosome in each cell. A closer look at specific genes reveals much more (figure 1.1).

Happily, the future child will not have any of the more common inherited disorders. Despite the apparently healthy genetic background, some blood cells from the umbilical cord will be set aside at birth and deep-frozen. Should he one day require a bone marrow transplant to treat life-threatening anemia or cancer, his own cord blood cells will be infused into his body where they will grow new bone marrow, tailor-made for his body.

Other results from the prenatal test indicate that the boy will be able to minimize effects of certain potentially unhealthy inherited characteristics. Tests to type the genes that predispose him to develop heart disease make it clear that a lifelong low-fat diet and regular exercise can extend his life. The same measures can help prevent or delay colon cancer, since he has inherited a pair of susceptibility genes. A computer evaluates how certain genes affect expression of other genes. Many inherited traits are not checked because they will not affect health. Hair and eye color and freckles will remain surprises.

This scenario takes place in the near future, but every one of the tests described is performed today. Other genetic screening tests, for fetuses as well as newborns, have been available for many years. Many new tests to detect genetic disease susceptibilities are being developed. They are raising questions for the health insurance industry on how to handle future illnesses.

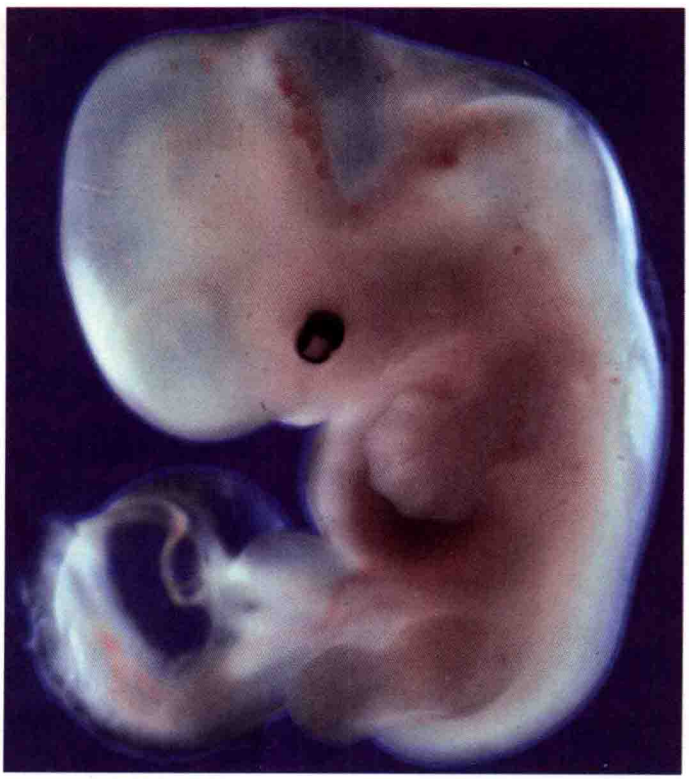
Until recently, we didn't know the nature of most genes that cause disease. However, a global scientific effort called the **human genome project** is rapidly adding more genes to the collection of those we can test for as it deciphers the complete genetic makeup, or **genome,** of humans.

Once a basic life science, human genetics is rapidly becoming a medical discipline. The human genome project is altering the way we view illness and is also revealing the many ways that people differ from each other.

Genetics in the News

Genetics is the study of inherited variation and traits. Genes are biochemical instructions that determine those inherited traits; they consist of sequences of building blocks of **deoxyribonucleic acid** (DNA). Genes strung together make up the larger chromosomes. Human cells have 23 pairs of chromosomes, which include two copies of each of about 70,000 genes.

A gene's sequence of DNA building blocks is like a language that instructs a cell to manufacture a particular protein.



Fetus

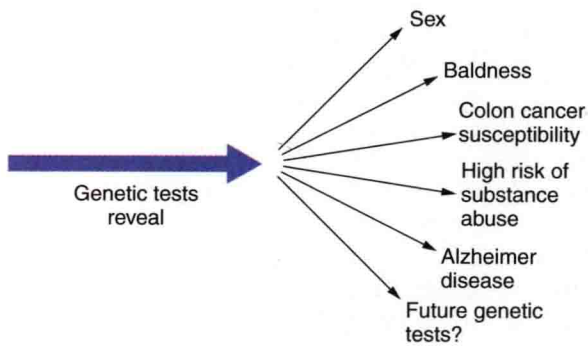


figure 1.1

Genetic tests of a fetus can reveal sex and many aspects of future health. Some traits, however, may be influenced by environmental factors or activities of other genes. Personality traits, for example, are very difficult to analyze.

An intermediate language, encoded in the building block sequence of **ribonucleic acid** (RNA), translates a gene's message into a protein's amino acid sequence. It is the protein that determines the trait.

A gene's DNA sequence can vary in many ways, just as the letters in a sentence can be rearranged to communicate a different message. Variants of a particular gene, different because they include changes in the DNA sequence, are called **alleles.** A change in a gene is a **mutation.** Most mutations we know of are associated with illness, but many are harmless, and some may even be helpful. A mutation, for example, makes a small percentage of the population resistant to HIV infection.

A generation ago, studying genetics meant examining patterns of trait transmission in fruit flies, bacteria, bread mold, corn, and other species whose physical characteristics or chromosomes are easy to study. A generation before that, biologists did not even know what type of chemical comprised the genetic material. While today's genetic researchers still use experimental organisms to unravel the details of how genes control traits, and even to serve as "models" of human disorders, the study of human genetics has grown explosively, touching our lives in a variety of ways. Following are some familiar applications of human genetics.

Establishing Identity

Comparing DNA sequences among individuals can establish, or rule out, that the DNA came from the same person, from blood relatives, or from unrelated people. Such DNA typing, or fingerprinting, has many applications.

In forensics, a DNA match for rare sequences between a tissue sample at a crime scene (blood, semen, skin, bone, or hair) and a blood sample from a suspect is strong evidence that the accused person was at the crime scene. In paternity cases, DNA-sequence matches can establish that a particular man fathered a particular child.

DNA evidence can solve historical mysteries. When anthropologists discovered remains of human infants at the site of a fourth century Roman bathhouse, they expected them to be female, evidence of infanticide in a society that valued males. But analysis of DNA sequences unique to the Y chromosome identified some males. This information, with other evidence, revealed that the bathhouse was the workplace of prostitutes, who disposed of unwanted babies of either sex. DNA typing solves modern mysteries too. Forensic scientists use the technique to reassemble body parts of plane crash victims.

In 1995, the U.S. military began collecting and storing DNA samples from personnel, to identify future casualties. Some people objected, fearing use of their DNA information for other purposes. Two marines who refused to give blood were court-martialed, but the publicity from their objection led to passage of a law to limit the military's use of DNA samples to identifying casualties and criminals. Use of genetic information has emerged as a major issue in many areas.

Agriculture

Agriculture reflects a rich history of controlled breeding to select new combinations of traits in livestock, fruits, and vegetables. Manipulations of individual genes have added a precision not possible with traditional breeding plans, in which trait combinations passed to offspring can be unpredictable.

Genetic alterations can enhance characteristics or add new ones to agriculturally valuable organisms, even from different species. Cotton plants given certain bacterial genes, for example, produce a plastic-like chemical that causes them to produce ultra-warm cotton fibers. Cereal crops are made more nutritious by genetically boosting their output of certain amino acids. Genetically altered trees can resist environmental stress and pests and produce more lignin, a chemical important in the pulp and paper industry. Some consumers, though, are wary of genetically altered crops, as figure 1.2 illustrates.

Crops and animals can also be genetically altered to produce biochemicals of use to humans as drugs, a technology called "pharming." A flock of such genetically-altered sheep, for example, produces the human protein clotting factor that people with hemophilia use.

Defining Race

The *American Heritage and Dictionary of the English Language* defines *race* as a "local geographic or global human population distinguished as a distinct group by genetically transmitted physical characteristics." Shared inherited characteristics, rather than acquired traits like dyed hair color, are used to define race because they indicate that a group of people descended from common ancestors and are therefore closely related by blood. When people tend to mate within their population, certain traits remain in that population, and the combination of traits comes to define a particular race.

Traditional race definitions, however, emphasize skin color, which is only one of thousands of inherited human traits. (Reading 1.1 describes a few of the odder ones.) Why not define race by hair texture,

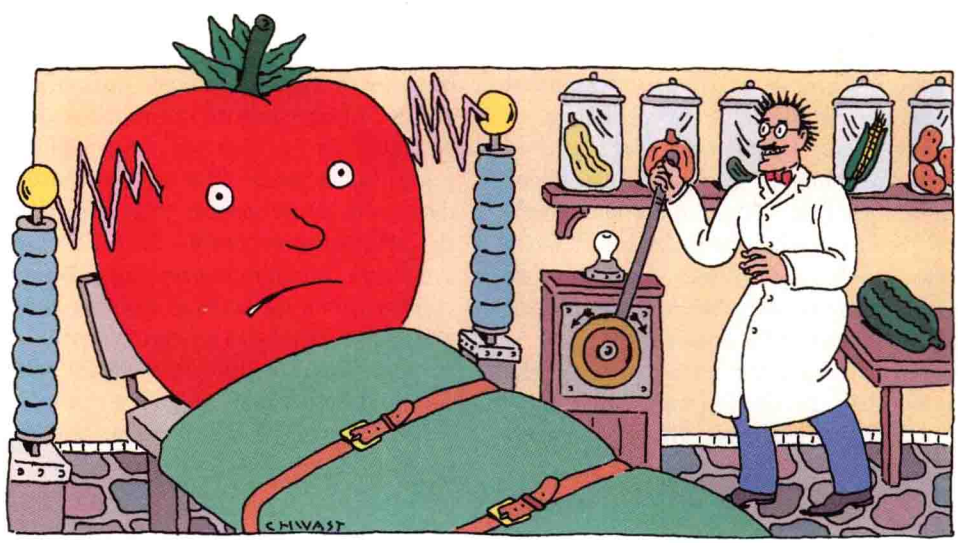


figure 1.2

This cartoon, from *Time* magazine, illustrates the public's fear of genetic manipulation.

© Seymour Chwast, The Pushpin Group.

It's All in the Genes

reading 1.1

Do you have uncombable hair, misshapen toes or teeth, or a pigmented tongue tip? Are you unable to smell a squashed skunk, or do you sneeze repeatedly in bright sunlight? Do you lack teeth, eyebrows, eyelashes, nasal bones, thumbnails, or fingerprints? If so, your unusual trait may appear in a compendium called *Mendelian Inheritance in Man,* or "MIM" for short. A team at Johns Hopkins University led by reknowned geneticist Victor McKusick updates MIM daily. An online version is available at http://www3.ncbi.nlm.nih.gov/omim/. Entering a disease name retrieves a long list of information.

Most of the more than five thousand entries in MIM include family histories, clinical descriptions, the pattern of inheritance, and molecular information on the causative gene. Woven in amidst the medical terminology are the stories behind some fascinating inherited traits (figure 1).

Genes control whether hair is blond, brown, or black, has red highlights, and is straight, curly, or kinky. Widow's peaks, cowlicks, a whorl in the eyebrow, and white forelocks run in families; so do hairs with triangular cross sections. Some people have multicolored hairs, like cats; others have hair in odd places, such as on the elbows, nose tip, knuckles, palms, or soles. Teeth can be missing or extra, protuberant or fused, present at birth, shovel shaped, or "snowcapped." A person can have a grooved tongue, duckbill lips, flared ears, egg-shaped pupils, three rows of eyelashes, spotted nails, or "broad thumbs and great toes." Extra breasts are known in humans and guinea pigs, and one family's claim to genetic fame is a double nail on the littlest toe.

Unusual genetic variants can affect metabolism, producing either disease or harmless, yet noticeable, effects. Members of some families experience "urinary excretion of odoriferous component of asparagus" or "urinary excretion of beet pigment," producing a strange odor or dark pink urine stream after consuming the offending vegetable. In blue diaper syndrome, an infant's urine turns blue on contact with air, thanks to an inherited inability to break down an amino acid.

One bizarre inherited illness is the jumping Frenchmen of Maine syndrome. This exaggerated startle reflex was first noted among French-Canadian lumberjacks from the Moosehead Lake area of Maine, whose ancestors were from the Beauce region of Quebec. Physicians first reported the condition at a medical conference in 1878. Geneticists videotaped the startle response in 1980, and the condition continues to appear in genetics journals. MIM gives a most vivid description:

If given a short, sudden, quick command, the affected person would respond with the

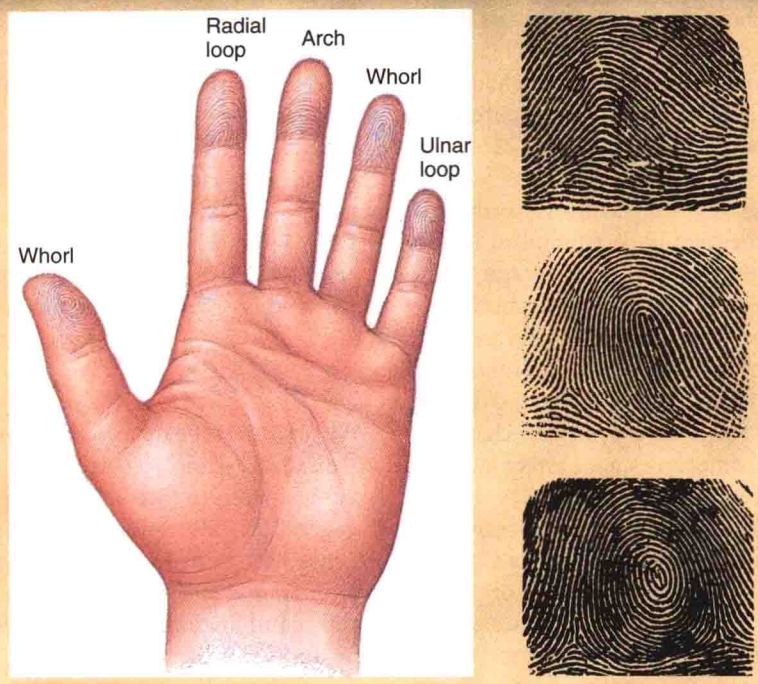


figure 1

Fingerprint patterns form characteristic arches, loops, and whorls. The actions of several genes determine the patterns, along with the fetus's fingertip movements at about 6 weeks of gestation, when these skin ridges are vulnerable to damage. This environmental effect is why identical twins, who have all the same genes, nevertheless have different fingerprints.

Source (for fingerprints only): Kent M. VanDeGraaff, *Human Anatomy,* 4th ed. Copyright © 1995 Times Mirror Higher Education Group, Dubuque, Iowa.

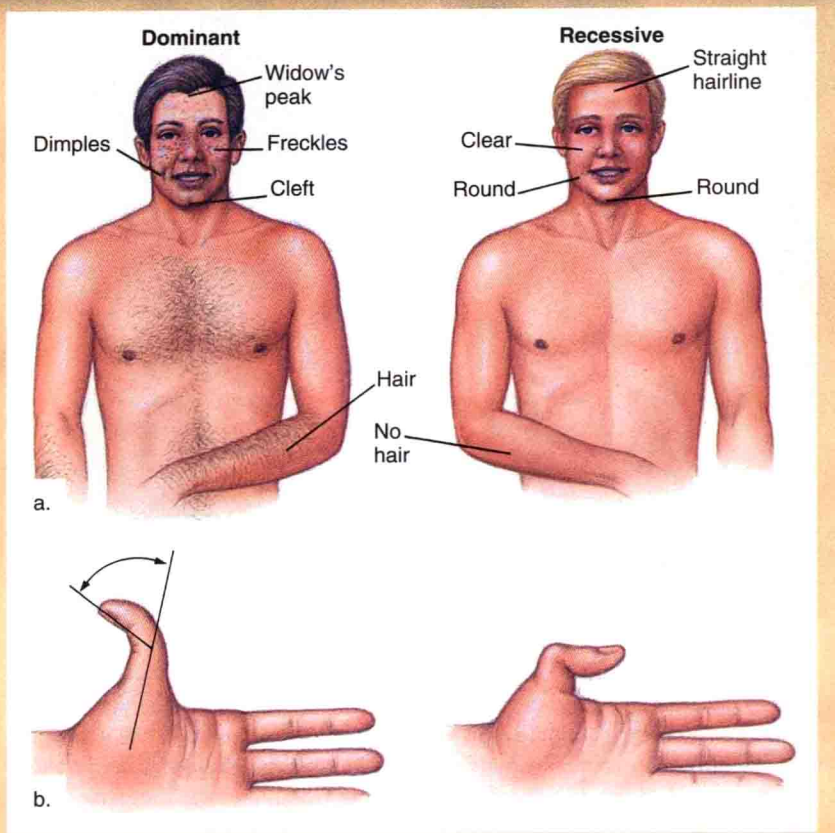


figure 2

Inheritance of some common traits: (*a*) freckles, dimples, hairy elbows, widow's peak, and a cleft chin; (*b*) the ability to bend the thumb backward or forward.

> **appropriate action, often echoing the words of command . . . For example, if one of them was abruptly asked to strike another, he would do so without hesitation, even if it was his mother and he had an ax in his hand."**

The jumping Frenchmen of Maine syndrome may be an extreme variant of the more common Tourette syndrome. Figure 2 illustrates some other genetic variants.

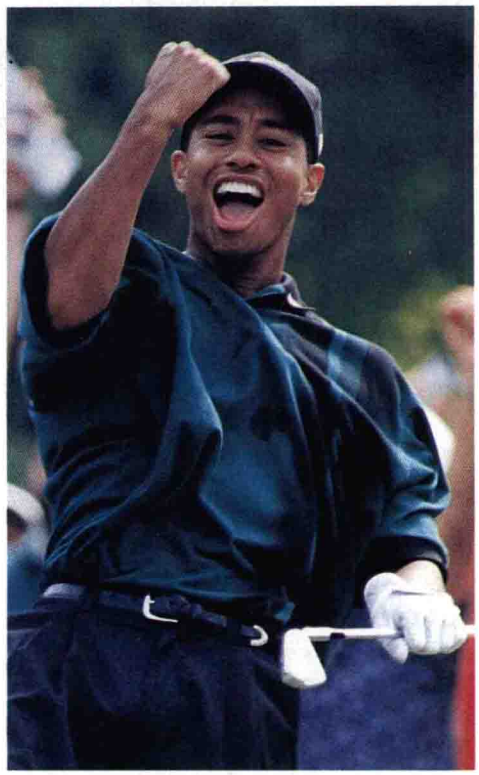


figure 1.3

Skin color is only one way that humans differ from each other. Race based on color is literally only skin deep. Golfer Tiger Woods, for example, objected to being called black, when he is actually part African American, Caucasian, Asian, and Native American.

ear-wiggling ability, or elevated blood pressure? The answer is simply that skin color differences are easy to see; an inherited inability to digest milk sugar is not nearly as obvious (figure 1.3).

Health Care

Inherited illness differs from other illnesses in two important ways. First, because of the laws of inheritance, a genetic condition recurs with a predictable probability—the subject of chapter 4. For example, if parents are each a carrier of a genetic disorder, then each of their children has a 1 in 4 chance of inheriting the disease. In contrast, the risk of passing an infectious disease to a family member depends upon physical contact.

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