

40 Genetic Inheritance in Humans

Human traits rarely follow strict Mendelian inheritance patterns.



Young Prince Louis XIII (1603), son of Marie de Medici, of France.

The young prince has a triangular shaped point of hair at the center of his forehead, known as a widow's peak. His mother Marie has one too. The widow's peak is often described as a "textbook" example of Mendelian inheritance. However, like many other traits in humans and other species, the inheritance of this trait is much more complex. Charles Martin, Portrait of Maria de' Medici and her son Louis XIII, 1603.

Topics Covered in this Module

- Inheritance Patterns in Humans
- Methods for Analyzing the Inheritance of Genetic Disorders

Major Objectives of this Module

- List traits, including disorders, that follow Mendelian inheritance patterns.
- Determine how complex inheritance patterns can generate a variation of phenotypes.
- Explain methods used to predict and diagnose genetic disorders.
- Describe inheritance patterns in humans.

There are more than 7 billion people in the world, and almost everyone looks at least slightly different from everyone else. How is this amount of variation possible? With the exception of identical twins, the gene combinations we receive from our parents vary from sibling to sibling. These individual genotypes that make up the genetic profile of the individual are part of the reason why no two humans look exactly the same. Only identical multiples (e.g., identical twins, triplets, etc.) share a genotype, but their phenotypes, or physical appearances, still differ due to other factors, including complex genetic interactions and interactions with the environment. However, look at a multi-generational family photo and it is clear that relatives share characteristics such as freckles, a cleft chin or attached earlobes. Thousands of human traits are at least somewhat inheritable and run in families. Is there a reliable way to predict whether these traits will be inherited?

Inheritance Patterns in Humans

For more than 100 years, scientists have applied the basic principles of inheritance that Gregor Mendel described in pea plants to a large number of human traits, ranging from seemingly unimportant traits such as whether one's hair forms a widow's peak to genetic disorders that can cause debilitating disease and death. Many of these traits are readily observable and have served as teaching tools and textbook examples for decades. For example, eye color, the ability to bend back the thumb nearly 90 degrees (known as hitchhiker's thumb), the ability to roll one's tongue into a U-shape, or whether one's earlobes are attached or free were all thought to have simple Mendelian inheritance patterns (Figure 1). Even though many people (including textbook authors and biology teachers) still portray these traits as having simple Mendelian inheritance characteristics such as dominant and recessive alleles of a single gene, the inheritance patterns of most of these traits are in fact much more complex. Even some traits thought to have somewhat more complicated but still predictable inheritance, such as male pattern baldness (originally thought to be a sex-linked trait passed from the mother's father to her sons), turn out to be determined by much more complex interactions of multiple genes and the environment.



Figure 1: Examples of human traits that were once thought to show Mendelian inheritance.

Common traits that are often described in textbooks and used to teach Mendelian inheritance, but in fact are much more complex, include hitchhiker's thumb (a), tongue rolling (b), and attached earlobes (c).

a) Eder/Shutterstock b) Cordelia Molloy/Photo Researchers, Inc./Science Source c) Will & Deni McIntyre/Science Source

Although the inheritance of these physical traits is not as simple as we used to think, the basic principles of Mendelian genetics can still provide a guide for understanding the inheritance patterns of many traits. Mendelian genetics

is particularly useful when understanding the role of inheritance in genetic diseases, some of which largely conform to simple Mendelian inheritance of dominant and recessive alleles, such as sickle-cell disease and Huntington's disease. Other genetic diseases are somewhat more complex but still follow basic probability rules. For example, hemophilia A is influenced by a gene on the X chromosome. Because males only have one copy of this chromosome (the other being the Y Chromosome inherited from their fathers), they will express the disease phenotype if they inherit the allele on the X chromosome from their mothers. Females, which have two X copies (one from each parent) would need inherit the disease allele from both parents to express the disease phenotype. In 1896, Archibald E. Garrod, a physician in a London hospital, observed a rare congenital disorder called alkaptonuria. He suggested that the incidence of this disorder followed the pattern of recessive inheritance described by Mendel. Because biochemistry was still in its infancy, Garrod's work did not excite his contemporaries much. It was only in the 1950s that his work gained appreciation when the relationship between the genes and the specific proteins responsible for these disorders became known.

What kinds of traits follow Mendelian patterns?

Gregor Johann Mendel, an Augustinian monk, teacher, and avid gardener, played a major role in unlocking the basic principles that govern heredity. His findings ran counter to the more popular idea of the time that the phenotypes of parents blended together and were passed on to the next generation, and it took decades after his death for Mendel's work to be recognized as a basis for understanding patterns of genetic inheritance.

Charles Darwin, the British naturalist who put forward the theory of evolution by natural selection along with fellow Briton Alfred Russell Wallace, believed there were hereditary particles in the blood that made their way to reproductive cells. Although Darwin and Mendel were contemporaries, Darwin was unaware of Mendel's discoveries.

Just how did Mendel discover the principles of heredity? He knew farmers, herders and gardeners had selectively bred crops, livestock, and flowers to improve yield and change appearance. He also knew that their results were sporadic and time consuming, so he turned to the relatively fast-growing garden pea plant, *Pisum sativum*, to perform his own experiments to achieve a deeper understanding of the principles of inheritance.

Mendel undertook breeding experiments with hundreds of garden pea plants and observed the outcomes over many plant generations. He observed the inheritance of characteristics such as stem length, flower color and seed pod shape. Based on the results of his experiments, Mendel determined that each parent passed one allele of a particular gene to their offspring and that alleles could form new combinations during fertilization, leading to phenotypes in offspring that were not observed in the parents. As a result, he deduced that alleles for some phenotypes were **dominant** (if the allele is present, its phenotype is expressed) and others were **recessive** (both recessive alleles must be present for the phenotype to be expressed). Keep in mind that dominance and recessiveness are not intrinsic properties of an allele; they are instead effects that can only be measured in relation to the effects of other alleles of the same gene. Phenotypes that are passed on to the offspring according to the inheritance rules Mendel deduced follow what we call **Mendelian inheritance** patterns.

Mendel observed these inheritance patterns in plants, but he suspected that they also occurred in animals. And in fact, after Mendel, researchers found similar patterns of inheritance in a wide variety of organisms, including humans. With the advance of scientific tools and techniques, including genome sequencing, we now know that many organisms, including Mendel's own pea plants, exhibit complex inheritance patterns. We also know that the

environment plays a strong role in determining phenotype and that some phenotypes can be inherited through mechanisms other than gene sequence variations (known as epigenetics). Asthma, for example, is an inflammatory disease that researchers believe involves both inherited genes and environmental factors. Despite these complexities, the fundamental principles of Mendelian inheritance still provide an important foundation for understanding these more complex patterns.

Mendelian patterns in human genetics.

Human eye color is a trait that has often been described as a simple Mendelian trait, with brown eye color being dominant over green or blue eye color. However, recent studies have shown that two key genes involved in pigmentation of the iris can modify each other, resulting in multiple shades of brown, gray and blue. It's also possible that two blue-eyed parents can have brown-eyed offspring, which is thought to be impossible in a simple Mendelian scenario where blue eyes are recessive to brown eyes.

There are in fact only a few clear cases of classic Mendelian inheritance of human traits. The consistency of earwax is one: it can be dry or wet, and these traits appear to be controlled by two alleles on a single gene. The allele for wet earwax is dominant over the allele for dry earwax. The dry earwax allele appears to have arisen from a mutation in Asia tens of thousands of years ago and is very common among people of Asian descent, relatively rare among those of Western European descent and extremely rare among people of African descent.

Some single-gene genetic disorders or mutations, which can cause debilitating disease and death, also exhibit patterns of inheritance that follow Mendel's rules or nearly so. Although it is much more common for lethal diseases to be caused by alleles that code for recessive phenotypes, Huntington's disease is one example of a rare, fatal neurodegenerative disorder caused by an allele that codes for a dominant phenotype. Because the allele that causes Huntington's disease is dominant, offspring have a 50% chance of inheriting the allele, and therefore the disease, from a parent with the disease. There is, however, variation in the form of the mutation that causes the disease, which influences both the age at which symptoms appear as well as their severity. Because Huntington's disease symptoms do not manifest until later in life, the allele is often passed to the next generation before a diagnosis is made. Legendary United States folksinger Woody Guthrie died from Huntington's disease, which he inherited from his mother and passed to at least two of his children (Figure 2).



Figure 2: Huntington's disease is an autosomal dominant genetic disorder.

Woody Guthrie suffered from Huntington's disease, a rare genetic disease caused by a dominant allele. Huntington's disease is often passed on to offspring because its symptoms do not manifest until later in life, and at least two of Woody Guthrie's children inherited the disease.

New York World-Telegram and the Sun Newspaper Photograph Collection (Library of Congress).

Sickle-cell disease is a blood cell disease that follows Mendelian patterns of inheritance.

What other genetic diseases follow Mendelian patterns of inheritance? Sickle-cell disease is a blood disorder characterized by red blood cells that are distorted into a sickle, or crescent, shape. This shape leads to the blood cells prematurely breaking down or getting stuck in very small blood vessels, leading to anemia. The disorder affects millions of people, mostly those of African descent. Sickle-cell disease differs from Huntington's disease because it is inherited in an autosomal recessive pattern. A person who inherits one copy of the allele for sickle-cell disease is heterozygous. Rather than showing complete dominance, heterozygous carriers express a partial phenotype; some blood cells are normal, while others are distorted, and as a result, heterozygous individuals only sometimes experience moderate symptoms. To have full-blown sickle-cell disease, an individual must have two copies of the allele coding for the recessive sickle-cell phenotype; in other words, he or she must be homozygous recessive. One reason why the sickle cell phenotype appears to have remained present in the human population, particularly those of African descent, is that the intermediate (heterozygous) phenotype confers a survival advantage against malaria, which is particularly prevalent in Africa.

The majority of genetic disorders that resemble Mendel's patterns of inheritance are caused by mutant alleles that code for recessive phenotypes. Albinism (Figure 3) and cystic fibrosis are two other such disorders.



Figure 3: Albinism.

Humans with the albinism phenotype lack pigmentation, which can result in white (or pinkish) skin, white hair, and white eyes. The two albino girls in the center of the picture are natives of Ghana, where this picture was taken.

Photographer's Mate 1st Class (AW) Shane McCoy/U.S. Navy.

Test Yourself

If neither of the parents displays the symptoms of an autosomal recessive Mendelian disorder that runs in their families, what is the likelihood that their child will develop the disease?

Submit

Hemophilia is a disease caused by X chromosome-linked inheritance.

Hemophilia, a bleeding disorder that slows the blood-clotting process, is another genetic disorder that runs in families. It is a recessive disease caused by mutations in the $F8$ gene (which causes hemophilia A) or in the $F9$ gene (which causes hemophilia B); both genes are on the X chromosome. Females who inherit an allele coding for hemophilia from one parent are less likely to express the disease phenotype because they have a second X chromosome from the other parent; because the allele for hemophilia is recessive, the disease will not be expressed in females that are heterozygous. However, these heterozygous females (carriers) can still pass the allele on to their children. Alternatively, males who inherit the hemophilia allele on the X chromosome they inherit from their mothers always show symptoms of the disease. This is because males have a Y chromosome inherited by their fathers that is paired to this X chromosome from their mothers. Because they do not have a second X chromosome that could potentially mask the hemophilia allele, they will always display the hemophilia phenotype even though it is recessive. The British royal family was affected by this genetic disorder, which descended from Queen Victoria. The Queen was a carrier and passed the recessive allele on through her lineage (Figure 4). Does this mean today's British royalty has to worry about hemophilia? No, the current British royal family is descended from Queen Victoria's son, Edward, who did not inherit the allele for hemophilia A.



Figure 4: Hemophilia: The royal disease.

Seated at the bottom row in the center, with a black shawl on her head, Queen Victoria (1819-1901) poses with her family; among them are four of her nine children and six of her 34 grandchildren in 1894.

Science Source

Most human traits result from multifactorial effects.

Geneticists have known for a long time that most human traits result from more than the coded building instructions of single genes that we associate with Mendelian inheritance. Most traits are multifactorial, meaning that several factors contribute to the actual phenotypes that develop. Interactions between genes can play a role in modifying phenotypes, and environmental factors can influence a variety of phenotypes, including altering the likelihood of developing a genetically determined disease. How to pinpoint these complex relationships between genotype and phenotype is a hot topic in genetic research. How do educators go about teaching inheritance and genetic patterns without having a long list of "exceptions to the rule"?

Some educators argue that beginning a genetics discussion with Mendel's simple principles closes students' minds to the variability of traits. Michael Dougherty, Director of Education at the American Society of Human Genetics, has suggested using teaching methods that de-emphasize Mendel and only introducing Mendelian principles later as one of several ways of understanding phenotypes. Other teachers are concerned that Dougherty's suggestion may end up minimizing Mendel's contributions too much.

Is a compromise between these two schools of thought possible? Expanding the teaching of inheritance without minimizing the importance of Mendelian principles is an option. One field that may require more emphasis is **epigenetics**, which examines heritable changes that do not involve changes

in the underlying DNA sequence but instead changes in how those genes are expressed. Likewise, environmental factors can have a variety of complex effects on how and when genes are expressed, which in turn can dramatically alter an organism's phenotype.

Test Yourself

Why can't Mendelian inheritance explain the inheritance patterns of all genes?

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Methods for Analyzing the Inheritance of Genetic Disorders

The human genome contains ~21,000 genes that encode proteins. Each gene sits in a specific location on a chromosome. Humans have 22 pairs of autosomes and one pair of sex chromosomes (XX for females and XY for males). Clinical cytogeneticists are scientists who study chromosomes and detect and analyze hereditary diseases and abnormalities with the help of pedigrees and karyotypes. Molecular tests can also determine the presence of alleles linked to diseases and genetic variations that increase a person's chances of developing a disease. (These molecular tests are commonly called genetic tests.) How do these tools help geneticists study and make predictions about the inheritance of genetic diseases and other traits?

Pedigrees can be used to follow a trait through the generations.

Although it is easy enough for scientists to study the genetics of model organisms such as mice and pea plants, researchers cannot study human genetics by performing controlled crosses on human subjects. Instead, researchers examine patterns of inheritance in an existing population through pedigree analysis. During Queen Victoria's lifetime, it was known that diseases like hemophilia A ran in families. Had Queen Victoria not kept the royal disease a secret, Nicholas II of Russia may not have married her granddaughter Alix for fear of passing the disease on to his heirs. In fact, the couple's only son, the Tsarevich Alex, had hemophilia, and historians have claimed that his disease contributed to the Russian Revolution in 1917. The Bolsheviks murdered the 14-year-old Alex and the rest of his family the following year.

BIOSKILL

How Does One Read a Pedigree?

In cases in which several members of a family have the same disease, a **pedigree** can be used to determine whether the disease has a genetic component and whether it is a recessive or dominant phenotype. In the 1980s, Japanese researchers used a pedigree analysis to pinpoint a sex-linked recessive version of Severe Combined Immunodeficiency Disease (SCID). SCID is also known as "bubble-boy" disease because those who have the condition are highly vulnerable to infectious diseases and in the past had to be isolated from human contact within plastic, bubble-shaped isolation units. Individuals with SCID have defective white blood cells (B- and T-lymphocytes) and thus a compromised immune system. Researchers have now identified several genes associated with SCID. Only 20% to 30% of boys diagnosed with SCID have a family history of immunodeficiency. The researchers started with three sick cousins and found males afflicted with SCID in four generations of their family, outnumbering affected females by more than 4:1. Practice reading a pedigree for SCID in Figure 5.

Figure 5: Family pedigrees for an X-linked version of SCID.

Pedigrees allow scientists to follow the presence of an allele across many generations of a family.

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A karyotype is a way to visualize chromosomes.

What other tools do scientists use to analyze inheritance patterns? A **karyotype** is a tool that profiles a person's chromosomes. It can detect gross genetic and subtle structural changes in the chromosomes of a cell population. The changes are often associated with specific birth defects, genetic disorders, and cancers. A karyogram depicts the karyotype in a snapshot of the 23 human chromosome pairs; the chromosome pairs are usually lined up and numbered by size with the pair of sex chromosomes at the end (Figure 6).

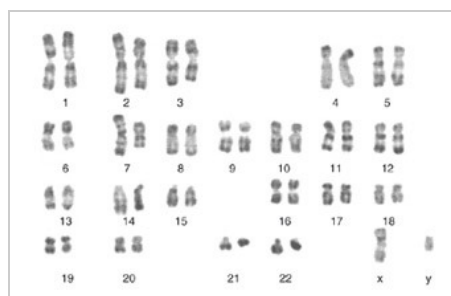


Figure 6: A normal human karyotype.

Displaying the 23 pairs of human chromosomes allows scientists to determine genetic abnormalities. The 23rd pair of chromosomes is an X chromosome and a Y chromosome indicating that this individual is male. This image shows duplicated sister chromatids, which only occurs during

cell division.

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Stamatoullas, A., et al. Conventional cytogenetics of nodular lymphocyte-predominant Hodgkin's lymphoma.

Leukemia 21, 2064–2067 (2007)

doi:10.1038/sj.leu.2404736. Used with permission.

How is a DNA sample prepared for karyotyping? Researchers culture cells from a source such as a tumor, the skin, blood, bone marrow, or the amniotic fluid of a fetus and suspend cell growth and multiplication at a stage when the chromosomes are most condensed. Further chemical treatment allows researchers to determine the number of chromosomes and reveals structural features of the chromosomes as dark- and light-colored bands. The order and width of the bands help determine whether chromosome fragments have moved and attached elsewhere to create chromosomal rearrangements. These rearrangements usually have negative effects on the individual, including infertility or an increased cancer risk.

BIOSKILL

Can We Identify Genetic Diseases Using a Karyotype?

The standard format of a karyotype is suited to detect **aneuploidies**, or abnormal numbers of chromosomes that often occur when chromosomes fail to segregate properly during cell division, resulting in chromosomal errors. The presence of an extra autosome or sex chromosome is often linked to several traits that are inherited together as a syndrome. Large-scale chromosomal rearrangements have happened frequently over the course of billions of years of life on Earth and have played an important role in the diversification of organisms; however, they are very rare and, for the most part, quite deleterious to the individual that acquires them.

Numerical abnormalities relating to autosome pairs 13, 18 and 21 are known (Figure 7). The chromosomal pair abnormality is referred to as trisomy when three chromosomes are present instead of the normal two. When there is only one chromosome instead of the normal two, the condition is described as monosomy. One of the best-known autosomal abnormalities is Down syndrome (often called Down's syndrome) in which affected individuals possess an extra copy of chromosome 21 (trisomy 21). Down syndrome is characterized by multiple physical malformations in some internal organs (such as the heart), and developmental delay caused by abnormalities in the central nervous system. It is a congenital syndrome characterized by certain facial features, including skin folds at the inner part of the eye.

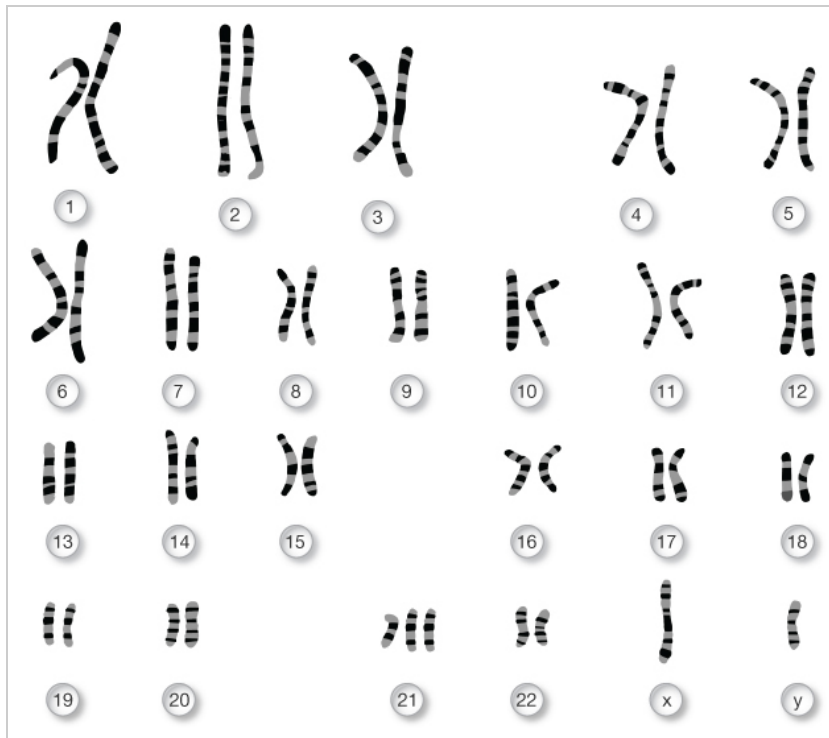


Figure 7: An abnormal human karyotype.

Scientists can determine chromosomal abnormalities using karyotypes.

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Test Yourself

Analyze the karyogram in Figure 7 to determine what chromosomal abnormality the individual has. What genetic disorder is associated with this abnormality?

Submit

BIOSKILL

What is genetic testing?

Genetic tests can also identify small chromosomal changes. The first of these tests appeared in the 1950s to detect suspected genetic diseases such as Duchenne muscular dystrophy and cystic fibrosis. Today, gene, chromosomal and biochemical tests are available to predict thousands of genetic conditions, but their usefulness has limits. A test that came to market in 2007 measured the activity of 70 genes in early-stage breast cancer. The information the test provided allowed oncologists to tailor the course of treatment to each patient's risk of recurrence. Over time, research showed the test was more than 95% accurate in determining those with a low risk of recurrence but only around 20% accurate in determining high-risk cases. Other genetic tests have higher accuracy percentages and are particularly useful for predicting genetic disorders in unborn children.

CAREERS

Genetic Counselors Diagnose and Counsel Those with Genetic Diseases

Parents-to-be can turn to a genetic counselor if genetic testing reveals that their child will have a disease. Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of medical

genetics and counseling. They counsel families affected by or at risk for genetic disorders and serve as patient advocates, educators, and researchers. The human genome project has provided consistent information about gene-related disorders. If a fetus has been found to have a genetic disorder, a genetic counselor can use available information about the human genome to advise the parents-to-be regarding the disorder's likelihood of manifesting, as well as its potential severity and consequences. Most genetic counselors enter the field from other disciplines such as biology, genetics, nursing, psychology, public health, or social work.

The field of genetics has come a long way since Mendel's time, and it continues to grow. Scientists around the world contribute new findings all the time. Gene cards are databases that provide concise information about all known and predicted human genes. They offer information about the human gene products, their functions, and their involvement in diseases using approved gene symbols (unique abbreviation of the full name for each gene) and standard nomenclature. In Europe, clinical utility gene cards (CUGCs) are used to list and assess the inheritance of a wide range of human genetic diseases. The cards, which follow a standard format, help physicians and clinical geneticists understand the validity, clinical usefulness, and interpretation of genetic tests for more than 50 Mendelian inherited diseases. The European Union-funded network that creates the cards is working to expand the service. As more and better technology is developed to study genes and inheritance in humans, doctors come closer to finding ways to prevent and treat potentially lethal genetic disorders.

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IN THIS MODULE

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- ▶ Test Your Knowledge

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Summary

OBJECTIVE Describe inheritance patterns in humans.

In the mid-19th century, Mendel determined the basic principles of heredity using observations and experiments with the common garden pea plant. He found that each parent passes one allele to its offspring and that the combinations of alleles determine the offspring's phenotypes. Most human phenotypes are multifactorial, and although few phenotypes follow true Mendelian inheritance, the principles of dominant and recessive alleles apply to several human characteristics, including several genetic disorders.

OBJECTIVE List traits, including disorders, that follow Mendelian inheritance patterns.

It was previously thought that widow's peak hairline, tongue rolling, eye color and hitchhiker's thumb were inherited according to Mendelian principles, but the inheritance of these phenotypes is more complicated. Human earwax appears to show Mendelian inheritance, along with a few other traits as well as several genetic disorders such as Huntington's disease, sickle-cell disease, albinism and cystic fibrosis.

OBJECTIVE Determine how complex inheritance patterns can generate a variation of phenotypes.

Complex inheritance patterns can generate a variety of phenotypes in offspring due to the independent assortment of alleles.

OBJECTIVE Explain methods used to predict and diagnose genetic disorders.

Family pedigrees, karyotypes and genetic tests help identify and analyze genetic diseases that follow Mendelian inheritance patterns.

Key Terms

aneuploidy

Change in the number of chromosomes in an individual by addition or deletion of chromosome(s).

dominant

Describes an allele for a phenotype that is expressed even when only one copy of the allele encoding for that phenotype is present in the genotype.

epigenetics

Heritable changes in phenotype that do not involve changes in the underlying DNA sequence but instead changes in how those genes are expressed.

karyotype

A diagrammatic representation of the chromosomal composition of an organism.

Mendelian inheritance

Patterns of passing genetic traits as described by Gregor Mendel.

pedigree

Mapping or charting of a family history, showing the occurrence of a trait or characteristic.

recessive

Describes an allele for a phenotype that is masked by the dominant allele and only expressed in the phenotype when the individual is homozygous for the recessive allele.

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Test Your Knowledge

1. Name a human trait that is no longer considered a classic Mendelian trait.
 - eye color
 - cystic fibrosis
 - albinism
 - sickle-cell disease
 - Huntington's disease
2. What did Michael Dougherty, the director of education at the American Society of Human Genetics, suggest regarding the teaching of Mendelian inheritance?
 - Dougherty has criticized the proposal that teachers talk about gene variation before they introduce Mendel.
 - Dougherty suggested that teachers talk about epigenetics instead of Mendel's inheritance patterns.
 - Dougherty suggested Mendel should no longer be taught in genetics.
 - Dougherty suggested that proving cause and effect of gene-environment interactions involves too many challenges to teach students about epigenetics.
 - Dougherty suggested that teachers should talk about gene variation before they introduce Mendel.
3. How does a karyotype differ from a family pedigree?
 - A family pedigree is an older and more accurate tool compared to a karyotype.
 - They are the same.
 - A family pedigree is useful in identifying inherited diseases, but a karyotype is usually not.
 - Family pedigrees require sample preparation in the lab; karyotypes do not.
 - Karyotypes provide details about genes on individual chromosomes to better identify inherited diseases.
4. How did Mendel determine the basic principles of inheritance?
 - He was influenced by Darwin's theory of inheritance.
 - As an avid gardener, he took observational data.
 - He used experimentation, observation, and mathematical analysis.
 - He took basic principles of inheritance from other scientists' published papers on inheritance.
 - He applied Darwin's theory of evolution.
5. Which of the following is the best tool for detecting aneuploidy?
 - pedigree analysis
 - karyotyping
 - genome-wide association studies
 - test crossing
 - None of these answers are correct.
6. Albinism is an autosomal recessive trait. An albino mother has an albino daughter by a healthy man. They plan on having several more children. On average, what proportion of the children are expected to be albino?
 - half of the sons and all of the daughters

- half of the sons and daughters
- all of the daughters and none of the sons
- all of the sons and daughters
- all of the sons and none of the daughters

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