Name_____ Class_____ Date _____

Summary

14–1 Human Heredity

Biologists can analyze human chromosomes by looking at a karyotype. A karyotype is a picture of the chromosomes from a cell arranged in homologous pairs.

Humans have 46 chromosomes. Two of these chromosomes, X and Y, are the sex chromosomes. Females have two X chromosomes (XX). Males have one X and one Y chromosome (XY). The other 44 chromosomes are called autosomes.

Human genes are inherited according to the same principles of genetics described by Mendel. To study the inheritance of human traits, biologists use a pedigree chart. A pedigree shows the relationships within a family. The inheritance of a certain trait in a family can be traced using a pedigree. From this, biologists can infer the genotypes of family members.

It is difficult to associate an observed human trait with a specific gene. Many human traits are polygenic, meaning that they are controlled by many genes. The environment also influences many traits.

Some of the first human genes to be identified were those that control blood type. Red blood cells can carry two different antigens, called A and B. Antigens are molecules that can be recognized by the immune system. The presence or absence of the A and B antigens produces four possible blood types: A, B, AB, and O. The ABO blood types are determined by a single gene with three alleles.

In addition to the ABO antigens, there is another antigen on red blood cells called the Rh antigen. People who have the Rh antigen are Rh positive. People without it are Rh negative. A single gene with two alleles determines the Rh blood group.

There are several human genetic disorders, including phenylketonuria (PKU), Huntington's disease, and sickle cell disease. PKU is caused by a recessive allele. It is expressed only in individuals who have inherited a recessive allele from each parent. Huntington's disease is caused by a dominant allele. It is expressed in any person who has that allele. Sickle cell disease is caused by a codominant allele.

Scientists are beginning to understand which changes in the DNA sequence cause certain genetic disorders. Cystic fibrosis is caused by the deletion of three bases in the middle of the sequence for a protein. This deletion inactivates the protein, which causes the symptoms of this disorder. Only one DNA base is changed in the allele that causes sickle cell disease. This base change produces a blood protein that is less soluble than normal.

14–2 Human Chromosomes

The two smallest human chromosomes, chromosomes 21 and 22, were the first chromosomes to have their DNA sequences identified. Both have many genes important for health. Both have regions of DNA that do not code for proteins.

Genes located on the X and Y chromosomes, the sex chromosomes, are said to be sex-linked. They are inherited in a different pattern than genes located on autosomes. For example, all alleles linked to the X chromosome, including those responsible for color blindness, hemophilia, and Duchenne muscular dystrophy, are expressed in males even if they are recessive alleles. However, in order for these recessive alleles to be expressed in females, there must be two copies of them.

Females have two X chromosomes. Males have only one. To account for this difference, one X chromosome in females is randomly turned off. The turned-off chromosome forms a dense region in the nucleus known as a Barr body. Barr bodies are not found in males because their single X chromosome must be active.

The most common error during meiosis is nondisjunction. Nondisjunction is the failure of chromosomes to separate properly during meiosis. It causes abnormal numbers of chromosomes to find their way into gametes. This may result in a disorder of chromosome number. An example of autosomal nondisjunction is Down syndrome, in which there is an extra copy of chromosome 21. Nondisjunction can also occur in sex chromosomes. In Turner's syndrome, a female has only one X chromosome. In Klinefelter's syndrome, there are extra X chromosomes.

14–3 Human Molecular Genetics

Biologists can use techniques in molecular biology to read, analyze, and even change the DNA code of human genes. Genetic tests are available to test parents for the presence of recessive alleles for genetic disorders.

In a process called DNA fingerprinting, individuals can be identified by analyzing sections of DNA that have little or no known function. These sections of DNA vary widely from one person to the next.

In 1990, scientists around the world began the Human Genome Project. The goal was to identify the DNA sequence for the entire DNA in a human cell. In 2000, the human genome was sequenced. Now the project goal is to analyze these sequences. One way scientists are analyzing the DNA is by looking for genes. To do this, they look for promoter sequences. These are sequences that bind RNA polymerase.

Information about the human genome can be used to cure genetic disorders by gene therapy. In one method of gene therapy, a virus is used to deliver the normal gene into cells to correct the genetic defects. The virus is changed so that it cannot cause disease. The normal gene is attached to the DNA of the virus. The inserted gene can make proteins that correct the genetic defect.

There are risks and problems with gene therapy. Having the power to manipulate human DNA doesn't necessarily make it right. People in a society are responsible for making sure that the tools made available by science are used wisely.