Genetics & Public Policy CENTER

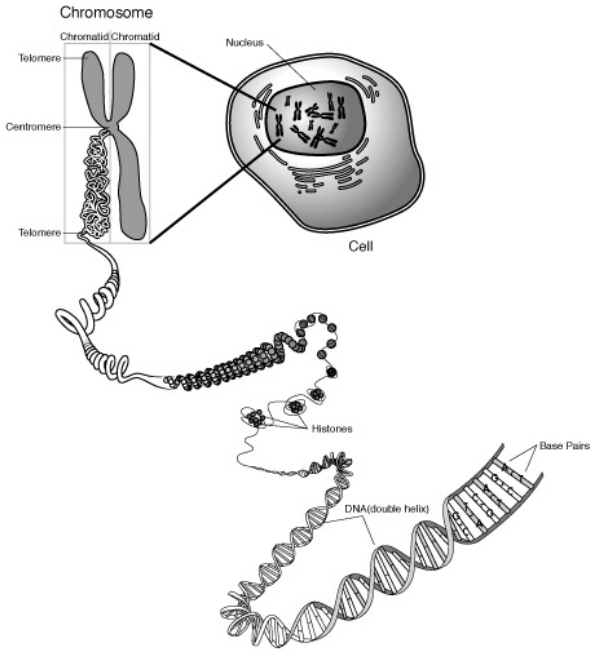
Technical Brief

The Basics of Genetics and Human Reproduction

Genes are the body's unique code that is inherited from an individual's biological parents. Genes are made of a substance called DNA, which stands for deoxyribonucleic acid. Every human cell (with the exception of mature red blood cells, which have no nucleus) contains the same DNA.

DNA is made up of two long strands of four different nucleotides, adenine, thymine, cytosine, and guanine (or A, T, C, and G). An A on one strand always pairs with a T on the other strand, and C always pairs with G. These base pairs form the rungs of the twisting DNA ladder. The twisted chain structure of DNA, known as the "Double Helix," was first identified by James Watson and Francis Crick in 1953. The exact order of the bases in the DNA molecule is what determines the genetic information it carries.

A gene is a segment along the DNA chain that encodes an instruction that allows a cell to produce a specific product, typically, a protein that initiates one specific action or forms a specific structure in the cell. There are about 30,000 genes spread out over the 3 billion base pairs in each cell. The complete set of genetic instructions within a cell is called the genome. All of this genetic information resides in the nucleus, or command center, of the cell.



Legend: In the nucleus of every cell is a set of chromosomes, which are made of densely packed DNA. Illustration courtesy of National Human Genome Research Institute.

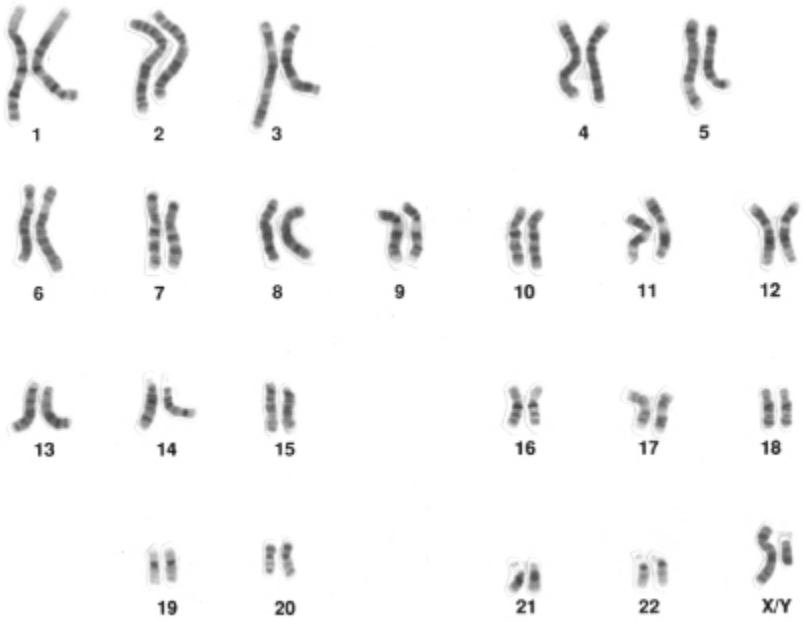
Although each cell contains a full complement of DNA, cells use genes selectively. Some genes enable cells to make proteins needed for basic functions. Called "housekeeping" genes, they are active in many

types of cells. Other genes, however, are inactive most of the time. Some genes play a role in early development of the embryo and are then shut down forever. Many genes encode proteins that are unique to a particular kind of cell, giving the cell its specialized character, for example, a skin cell versus a bone cell. A normal cell activates just the genes it needs at the moment and actively suppresses the rest.

A DNA misspelling, or mutation, in a gene, for example have an "A" where there should be a "G," can cause the protein encoded by that gene to malfunction or not work at all. DNA misspellings in specific genes or combinations of genes cause many diseases. More than 4,000 diseases are associated with misspellings in specific genes inherited from one's mother and/or father. In addition, common disorders such as heart disease and most cancers arise from a complex interplay among multiple genes and between genes and factors in the environment.

Chromosomes

The DNA in human cells is packaged into 23 different chromosomes--22 autosomes and an X or Y sex chromosome. Most human cells contain two sets of chromosomes, one set inherited from the mother and one from the father. Cells that contain 46 chromosomes are called "diploid" for "two sets." (Females inherit an X from each parent, while males get an X from the mother and a Y from the father.) The exception to this are the gametes--sperm and egg cells--which carry a single set of chromosomes. Gametes or germline cells are called "haploid" for "one set."



Legend: Each human cell contains 23 pairs of chromosomes, which can be distinguished by size and by unique banding patterns when stained. When organized by a technician into their pairs as in this figure, they comprise a "karyotype." This karyotype is from a male, since it contains a Y chromosome. Females have two X chromosomes. Photo courtesy of National Human Genome Institute.

Cell Division

Most cells of the body reproduce themselves at some point in the lifespan (some are continuously reproducing). A cell with 46 chromosomes (a diploid cell) reproduces itself by replicating its DNA, then splitting into two cells, each with a complete set of 46 chromosomes. This process is called "mitosis." In contrast, reproductive cells (sperm, ova) contain only 23 chromosomes and are formed through a different

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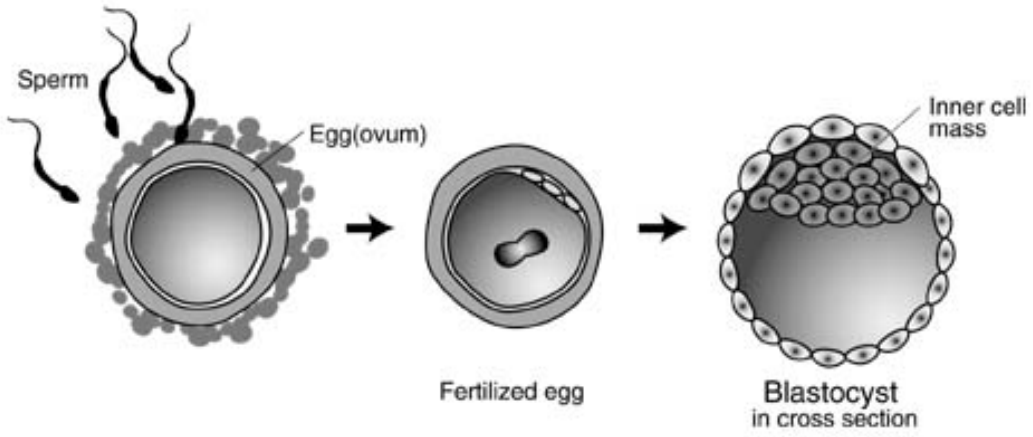
process of DNA replication and cell division called "meiosis." Meiosis involves a reduction in the amount of genetic material by half. It entails two successive nuclear divisions with only one round of DNA replication. When cell division is complete, one diploid parent cell has produced four haploid daughter cells. Daughter cells have half the number of chromosomes found in the original parent cell.

The first cell division (meiosis I) produces one large cell, the secondary oocyte, and a "polar body." Upon fertilization, the secondary oocyte divides (meiosis II) again, unequally, producing a second polar body and the mature haploid oocyte.

Fertilization and Embryogenesis

Sexual reproduction involves the joining of gametes (egg and sperm) to form the first cell of a new individual, called the "zygote." Because each gamete contains 23 chromosomes, the newly joined gametes form a unique diploid cell.

In the first week after fertilization, the zygote undergoes a series of mitotic divisions known as "cleavage." Thus, one diploid cell becomes 2, then 4, then 8, then 16, and so on. Cleavage continues as the zygote moves through the fallopian tube toward the uterus. By the time it reaches the uterus, the zygote is a hollow ball of cells, called a "blastocyst." About six days after fertilization the blastocyst burrows into the lining of the uterus in an event called implantation. There it will begin to grow and develop, capable of forming a child.



Legend: The steps from fertilization to cleavage and the zygote's transformation into a blastocyst. Illustration courtesy of National Human Genome Research Institute.

Human development takes about nine months, a period known as "gestation." The nine months of pregnancy are often divided into three trimesters. For the first eight weeks of pregnancy the developing human is called an embryo; thereafter it is called a fetus.

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