**Human genetics**

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A discipline concerned with genetically determined resemblances and differences among human beings. Technological advances in the visualization of human [chromosomes](http://www.answers.com/topic/chromosome) have shown that abnormalities of chromosome number or structure are surprisingly common and of many different kinds, and that they account for birth defects or mental [impairment](http://www.answers.com/topic/impairment) in many individuals as well as for numerous early spontaneous abortions. Progress in molecular biology has clarified the molecular structure of chromosomes and their constituent genes and the ways in which change in the molecular structure of a gene can lead to a disease. Concern about possible genetic damage through environmental agents and the possible harmful effects of hazardous substances in the environment on [prenatal development](http://www.answers.com/topic/prenatal-development) has also [stimulated](http://www.answers.com/topic/stimulate) research in human genetics. The medical aspects of human genetics have become prominent as nonhereditary causes of ill health or early death, such as infectious disease or nutritional [deficiency](http://www.answers.com/topic/deficiency), have declined, at least in developed countries.

In normal humans, the nucleus of each normal cell contains 46 chromosomes, which comprise 23 different pairs. Of each chromosome pair, one is [paternal](http://www.answers.com/topic/paternal" \t "_top) and the other maternal in origin. In turn, only one member of each pair is handed on through the reproductive cell (egg or [sperm](http://www.answers.com/topic/sperm)) to each child. Thus, each egg or sperm has only 23 chromosomes, the [haploid](http://www.answers.com/topic/haploid) number; fusion of egg and sperm at fertilization will restore the double, or [diploid](http://www.answers.com/topic/diploid), chromosome number of 46. *See also* [Chromosome](http://www.answers.com/topic/chromosome).

The segregation of chromosome pairs during [meiosis](http://www.answers.com/topic/meiosis) allows for a large amount of “shuffling” of genetic material as it is passed down the generation. Two parents can provide 223 × 223 different chromosome combinations. This enormous source of variation is [multiplied](http://www.answers.com/topic/multiply) still further by the mechanism of crossing over, in which [homologous chromosomes](http://www.answers.com/topic/homologous-chromosome) exchange segments during meiosis. *See also* [Crossing-over (genetics)](http://www.answers.com/topic/chromosomal-crossover); [Meiosis](http://www.answers.com/topic/meiosis).

Twenty-two of the 23 chromosome pairs, the autosomes, are alike in both sexes; the other pair comprises the [sex chromosomes](http://www.answers.com/topic/sex-determination-system). A female has a pair of X chromosomes; a male has a single X, paired with a [Y chromosome](http://www.answers.com/topic/y-chromosome) which he has inherited from his father and will transmit to each of his sons. Sex is determined at fertilization, and depends on whether the egg (which has a single [X chromosome](http://www.answers.com/topic/xy-sex-determination-system)) is fertilized by an X-bearing or a Y-bearing sperm. *See also* [Sex determination](http://www.answers.com/topic/sex-determination" \t "_top).

Any gene occupies a specific chromosomal position, or [locus](http://www.answers.com/topic/locus" \t "_top). The alternative genes at a particular locus are said to be alleles. If a pair of alleles are identical, the individual is [homozygous](http://www.answers.com/topic/homozygous); if they are different, the individual is [heterozygous](http://www.answers.com/topic/heterozygous). *See also* [Allele](http://www.answers.com/topic/allele).

Genetic variation has its origin in mutation. The term is usually applied to stable changes in DNA that alter the genetic code and thus lead to synthesis of an altered protein. The genetically significant [mutations](http://www.answers.com/topic/mutation) occur in reproductive cells and can therefore be transmitted to future generations. Natural selection acts upon the genetic diversity generated by mutation to preserve beneficial mutations and eliminate [deleterious](http://www.answers.com/topic/deleterious) ones.

A very large amount of genetic variation exists in the human population. Everyone carries many mutations, some newly acquired but others inherited through [innumerable](http://www.answers.com/topic/innumerable" \t "_top) generations. Though the exact number is unknown, it is likely that everyone is heterozygous at numerous loci, perhaps as many as 20%. *See also* [Mutation](http://www.answers.com/topic/mutation" \t "_top).

The patterns of inheritance of characteristics determined by single genes or gene pairs depend on two conditions: (1) whether the gene concerned is on an [autosome](http://www.answers.com/topic/autosome) (autosomal) or on the X chromosome (X-linked); (2) whether the gene is dominant, that is, expressed in heterozygotes (when it is present on only one member of a chromosomal pair and has a normal [allele](http://www.answers.com/topic/allele)) or is [recessive](http://www.answers.com/topic/recessive) (expressed only in homozygotes, when it is present at both chromosomes). *See also* [Dominance](http://www.answers.com/topic/dominance).

A quantitative trait is one that is under the control of many factors, both genetic and environmental, each of which contributes only a small amount to the total variability of the trait. The phenotype may show continuous variation (for example, height and skin color), quasicontinuous variation (taking only integer values—such as the number of ridges in a [fingerprint](http://www.answers.com/topic/fingerprint)), or it may be discontinuous (a presence/absence trait, such as diabetes or [mental retardation](http://www.answers.com/topic/mental-retardation)). With discontinuous traits, it is assumed that there exists an underlying continuous variable and that individuals having a value of this variable above (or below) a threshold possess the trait.

A trait that “runs in families” is said to be familial. However, not all familial traits are hereditary because relatives tend to share common environments as well as common genes.

The variability of almost any trait is partly genetic and partly environmental. A rough measure of the relative importance of [heredity](http://www.answers.com/topic/heredity) and environment is an index called [heritability](http://www.answers.com/topic/heritability). For example, in humans, the heritability of height is about 0.75. That is, about 75% of the total [variance](http://www.answers.com/topic/variance) in height is due to variability in genes that affect height and 25% is due to exposure to different environments.

**Hereditary diseases**

Medical genetics has become an integral part of preventive medicine (that is, genetic counseling, including [prenatal](http://www.answers.com/topic/prenatal-care) diagnostics). Hereditary diseases may be subdivided into three classes: chromosomal diseases; hereditary diseases with simple, [mendelian](http://www.answers.com/topic/mendelian) modes of inheritance; and [multifactorial](http://www.answers.com/topic/multifactorial) diseases.

One out of 200 newborns suffers from an abnormality that is caused by a microscopically visible [deviation](http://www.answers.com/topic/deviation) in the number or structure of chromosomes. The most important clinical abnormality is Down syndrome—a condition due to [trisomy](http://www.answers.com/topic/trisomy) of chromosome 21, one of the smallest human chromosomes. This chromosome is present not twice but three times; the entire [chromosome complement](http://www.answers.com/topic/chromosome-complement) therefore comprises 47, not 46, chromosomes. Down syndrome occurs one to two times in every 1000 births; its pattern of abnormalities derives from an [imbalance](http://www.answers.com/topic/imbalance) of gene action during [embryonic](http://www.answers.com/topic/embryonic) development. Down syndrome is a good example of a characteristic pattern of abnormalities that is produced by a single genetic defect. *See also* [Down syndrome](http://www.answers.com/topic/down-syndrome).

Other autosomal aberrations observed in living newborns that lead to characteristic syndromes include trisomies 13 and 18 (both very rare), and a variety of structural aberrations such as translocations (exchanges of chromosomal segments between different chromosomes) and deletions (losses of chromosome segments). Translocations normally have no influence on the health status of the individual if there is no gain or loss of chromosomal material (these are called balanced translocations). However, carriers of balanced translocations usually run a high risk of having children in whom the same [translocation](http://www.answers.com/topic/translocation) causes gain or loss of genetic material, and who suffer from a characteristic [malformation](http://www.answers.com/topic/malformation) syndrome.

Clinical syndromes caused by specific aberrations vary, but certain clinical signs are common: low birth weights (small for date); a peculiar face; delayed general, and especially mental, development, often leading to severe mental deficiency; and multiple malformations, including abnormal development of limbs, heart, and kidneys. *See also* [Congenital anomalies](http://www.answers.com/topic/congenital-disorder).

Less severe signs than those caused by autosomal aberrations are found in individuals with abnormalities in number (and, sometimes, structure) of sex chromosomes. This is because in individuals having more than one X chromosome, the additional X chromosomes are [inactivated](http://www.answers.com/topic/inactivate) early in pregnancy. For example, in women, one of the two X chromosomes is always inactivated. Inactivation occurs at random so that every normal woman is a mosaic of cells in which either one or the other X chromosome is active. Additional X chromosomes that an individual may have received will also be inactivated; in trisomies, genetic imbalance is thus avoided to a certain degree. However, inactivation is not complete; therefore, individuals with trisomies—for example, [XXY](http://www.answers.com/topic/xxy-acronym) (Klinefelter syndrome), [XXX](http://www.answers.com/topic/xxx-abbreviation) (triple-X syndrome), or XYY—or monosomies (XO; Turner syndrome) often show abnormal sexual development, intelligence, or behavior.

In contrast to chromosomal aberrations, the genetic defects in hereditary diseases with simple, mendelian modes of inheritance cannot be recognized by [microscopic](http://www.answers.com/topic/microscopic) examination; as a rule, they must be inferred more indirectly from the [phenotype](http://www.answers.com/topic/phenotype) and the pattern of inheritance in pedigrees. The defects are found in the molecular structure of the DNA. Often, one base pair only is altered, although sometimes more complex molecular changes, such as deletions of some bases or abnormal [recombination](http://www.answers.com/topic/recombination), are involved. Approximately 1% of all newborns have, or will develop during their lives, a hereditary disease showing a simple mendelian mode of inheritance.

In medical genetics, a condition is called dominant if the heterozygotes [deviate](http://www.answers.com/topic/deviate) in a clearly recognizable way from the normal homozygotes, in most cases by showing an abnormality. Since such dominant mutations are usually rare, almost no homozygotes are observed.

In some dominant conditions, the harmful phenotype may not be expressed in a gene carrier (this is called incomplete [penetrance](http://www.answers.com/topic/penetrance)), or clinical signs may vary in severeness between carriers (called variable expressivity). Penetrance and expressivity may be influenced by other genetic factors; sometimes, for example, by the sex of the affected person, whereas in other instances, the constitution of the “normal” allele has been implicated. Environmental conditions may occasionally be important. In most cases, however, the reasons are unknown.

X-linked modes of inheritance occur when the mutant allele is located on the X chromosome. The most important X-linked mode of inheritance is the recessive one. Here, the males (referred to as hemizygotes since they have only one allele) are affected, since they have no normal allele. The female heterozygotes, on the other hand, will be [unaffected](http://www.answers.com/topic/unaffected" \t "_top), since the one normal allele is sufficient for maintaining function. A classical example is [hemophilia A](http://www.answers.com/topic/haemophilia-a" \t "_top), in which one of the [serum](http://www.answers.com/topic/serum" \t "_top) factors necessary for normal [blood clotting](http://www.answers.com/topic/blood-clotting" \t "_top) is inactive or lacking. (The disease can now be controlled by repeated substitution of the [deficient](http://www.answers.com/topic/deficient" \t "_top) blood factor—a good example for phenotypic therapy of a hereditary disease by substitution of a deficient gene product.) Male family members are affected whereas their sisters and daughters, while being unaffected themselves, transmit the mutant gene to half their sons. Only in very rare instances, when a [hemophilic](http://www.answers.com/topic/hemophilic" \t "_top) patient marries a heterozygous carrier, are homozygous females observed. *See also* [Sex-linked inheritance](http://www.answers.com/topic/sex-linked-inheritance" \t "_top).

There are thousands of hereditary diseases with simple mendelian modes of inheritance, but most common anomalies and diseases are influenced by genetic variability at more than one gene locus. Most congenital malformations, such as [congenital heart disease](http://www.answers.com/topic/congenital-heart-disease" \t "_top), [cleft lip and palate](http://www.answers.com/topic/cleft-lip-and-palate-1" \t "_top), neural tube defects and many others, fall into this category, as do the constitutional diseases, such as diabetes mellitus, [coronary heart disease](http://www.answers.com/topic/coronary-heart-disease" \t "_top), anomalies of the immune response and many mental diseases, such as [schizophrenia](http://www.answers.com/topic/schizophrenia" \t "_top) or affective disorders. All of these conditions are common and often increase in frequency with advanced age.

**Biochemical genetics**

Biochemical genetics began with the study of [inborn](http://www.answers.com/topic/inborn" \t "_top) errors of [metabolism](http://www.answers.com/topic/metabolism" \t "_top). These are diseases of the body chemistry in which a small molecule such as a sugar or amino acid accumulates in body fluids because an enzyme responsible for its [metabolic](http://www.answers.com/topic/metabolic" \t "_top) breakdown is deficient. This molecular defect is the result of mutation in the gene coding for the enzyme protein. The accumulated molecule, dependent on its nature, is responsible for the [causation](http://www.answers.com/topic/causation" \t "_top) of a highly specific pattern of disease.

The field of biochemical genetics expanded with the recognition that similar [heritable](http://www.answers.com/topic/heritable) defective enzymes interfere with the breakdown of very large molecules, such as mucopolysaccharides and the complex [lipids](http://www.answers.com/topic/lipid) that are such prominent components of brain substance. The resultant storage disorders present with extreme alterations in [morphology](http://www.answers.com/topic/morphology) and [bony](http://www.answers.com/topic/boney) structure and with [neurodegenerative disease](http://www.answers.com/topic/neurodegenerative-disease).

The majority of hereditary disorders of metabolism are inherited in an [autosomal recessive](http://www.answers.com/topic/autosomal-recessive-1) fashion. In these families, each parent carries a single mutant gene on one chromosome and a normal gene on the other. Most of these mutations are rare. In populations with genetic diversity, most affected individuals carry two different mutations in the same gene. Some metabolic diseases are coded for by genes on the X chromosome. Most of these disorders are fully recessive, and so affected individuals are all males, while females carrying the gene are clinically normal. The disorders that result from mutations in the [mitochondrial genome](http://www.answers.com/topic/mitochondrial-deoxyribonucleic-acid) are inherited in nonmendelian fashion because [mitochondrial DNA](http://www.answers.com/topic/mitochondrial-dna) is inherited only from the mother. Those that carry a mutation are heteroplasmic; that is, each carries a mixed population of mitochondria, some with the mutation and some without.

Phenylketonuria (PKU) is a prototypic biochemical genetic disorder. It is an autosomally recessive disorder in which mutations demonstrated in a sizable number of families lead, when present in the genes on both chromosomes, to defective activity of the enzyme that catalyzes the first step in the metabolism of phenylalanine. This results in accumulation of phenylalanine and a recognizable clinical disease whose most prominent feature is severe retardation of mental development. *See also* [Phenylketonuria](http://www.answers.com/topic/phenylketonuria).

The diseases that result from mutation in mitochondrial DNA have been recognized as such only since the 1990s. They result from point mutations, deletions, and other rearrangements. A majority of these disorders express themselves chemically in elevated concentrations of [lactic acid](http://www.answers.com/topic/lactic-acid) in the blood or [cerebrospinal fluid](http://www.answers.com/topic/cerebrospinal-fluid). Many of the disorders are known as [mitochondrial](http://www.answers.com/topic/mitochondrion) myopathies (diseases of muscles) because [skeletal](http://www.answers.com/topic/skeletal) myopathy or cardiomyopathy are characteristic features.

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