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A Glossary of Genetic Terms

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A GLOSSARY OF GENETIC TERMS.

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The definitions of the following genetic terms are provided to assist the reader. Citations are given so that the more adventurous can delve more deeply into the mechanics of genetic engineering, the topic of the symposium. A chronology of genetic concepts and discoveries is found in the articles by Rédei and Witkowski.[1]

Active Site[2]: That portion of a protein, usually an enzyme, whose structural integrity is required for function (e.g., the substrate binding site of an enzyme).

Adenine (A)[3]: One of the four nitrogenous bases present in DNA. Adenine is a purine, as is guanine another one of the bases. In the DNA helix, adenine is always connected to, or "paired" with, thymine by two hydrogen bonds.

Alzheimer's Disease[4]: A progressive dementia that involves disturbances of speech, motor activity, and recognition. Early onset Alzheimer's Disease appears to have a genetic origin.

Amniocentesis[5]: A method to detect fetal genetic and developmental disorders. A hollow needle is inserted through the mother's abdominal and uterine wall and into the amniotic sac which surrounds the fetus. Fluid is then withdrawn from the sac. Included in the fluid is fetal urine and cells shed from the embryo. The fluid can be tested for the presence of biochemicals that may reflect a genetic disorder. The chromosomes from the shed cells can be used for karyotypic analysis to detect chromosomal abnormalities.

Autosomal[6]: Of a trait, or character, in which the gene is not carried on the sex chromosomes.

Bacteria[7]: (singular, bacterium): Extremely small, unicellular microorganisms that multiply by cell division. Bacteria are prokaryotes. The genetic material is usually contained in one

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[1]G.P. Rédei, *Steps in the Evolution of Genetic Concepts*, 93 BIOLOGISCHES ZENTRALBLATT 385 (1974); Jan Witkowski, *Fifty Years On: Molecular Biology's Hall of Fame*, 6 Trends in Biotechnology 234 (1988).

[2]GRIFFITHS ET AL., *supra* note 2, at 783.

[3]ANTHONY J.F. GRIFFITHS ET AL., AN INTRODUCTION TO GENETIC ANALYSIS 307-09 (5th ed. 1993).

[4]MICHAEL R. CUMMINGS, HUMAN HEREDITY 446-48 (3d ed. 1994); Richard J. Wurtman, *Alzheimer's Disease*, 252 Scientific American 62 (Jan. 1985).

[5]CUMMINGS, *supra* note 4, at 165-67.

[6]R. REIGER ET AL., GLOSSARY OF GENETICS 44 (5th ed. 1991).

[7]JAMES DARNELL ET AL., MOLECULAR CELL BIOLOGY 111-17 (2d ed. 1990); Academic Press Dictionary of Science and Technology 205 (Christopher Morris ed., 1992).

"chromosome," called a genophore. Although bacteria lack a membrane-bound nucleus, their single "chromosome" usually is found in the central region of the cell, called the nucleoid.

C. elegans (Caenorhabditis elegans)[8]: A nearly microscopic nematode with a highly predictable pattern of cell division and lineage during embryogenesis. The lineage of every one of the several thousand cells is known from the egg, the single-cell stage, to the adult. The organism is a favorite of geneticists for studying development.

Cancer[9]: A malignant tumor. A tumor is an aberrant growth of cells. If a tumor is self-contained and does not spread to other organs and start to grow, then the tumor is benign. Malignant tumors are not self-contained and metastasize, i.e., cells detach from the primary tumor and establish secondary areas of growth. Cancer cells have abnormal and unstable numbers of chromosomes. Several mutations are required for a cell to become cancerous. Cancers are so destructive to the body because their cells will divide endlessly.

Several cancers are known to be associated with genetic translocations. An example is chronic myeloid leukemia that is associated with a translocation of DNA involving chromosomes 9 and 22.

Cell Division[10]: The mechanism by which a single cell divides into two daughter cells. Cell division is the final stage of a cell's life. The normal cell cycle, or life, is divided into four phases. During the first phase, named Gap 1, the cell grows in preparation for the Synthesis phase. During the Synthesis phase the chromosomal material is duplicated (DNA is synthesized). The cell then prepares for nuclear division during the Gap 2 phase. During Mitosis, the final phase, the doubled chromosomal material divides followed by division of the whole cell. The consequence of the doubling and subsequent division is that each daughter cell receives the same amount of chromosomal material as the mother cell.

Chromosomal Abnormality[11]: A defect in the genetic constitution of an individual at the level of the chromosome. Examples are polyploidy, aneuploidy, deletion, and translocation.

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[8]GRIFFITHS ET AL., *supra* note 2, at 640-43.

[9]CUMMINGS, *supra* note 4, at 343-48, 363; DARNELL, *supra* note 7, at 955-60; GRIFFITHS ET AL., *supra* note 2, at 690-91; Academic Press Dictionary of Science and Technology, *supra* note 7, at 349; John Cairns, *The Treatment of Diseases and the War Against Cancer*, 253 SCIENTIFIC AMERICAN 51 (Nov. 1985); Carlo M. Croce and George Klein, *Chromosome Translocations and Human Cancer*, 252 SCIENTIFIC AMERICAN 54 (Mar. 1985); Michael Feldman and Lea Eisenbach, *What Makes a Tumor Cell Metastatic?*, 259 SCIENTIFIC AMERICAN 60 (Nov. 1988); Lance A. Liotta, *Cancer Cell Invasion and Metastasis*, 266 SCIENTIFIC AMERICAN 54 (Feb. 1992); Garth L. Nicolson, *Cancer Metastasis*, 240 SCIENTIFIC AMERICAN 66 (Mar. 1979); Bert Vogelstein and Kenneth W. Kinzler, *The Multistep Nature of Cancer*, 9 TRENDS IN GENETICS 138 (1993).

[10]WILLIAM S. KLUG AND MICHAEL R. CUMMINGS, CONCEPTS OF GENETICS 26 (4th ed. 1994); GEORGE P. RÉDEI, GENETICS 28-43 (1982); John F.X. Diffley and Bruce Stillman, *The Initiation of Chromosomal DNA Replication in Eukaryotes*, 6 TRENDS IN GENETICS 427 (1990); Andrew W. Murray and Marc W. Kischner, *What Controls the Cell Cycle*, 264 Scientific American 56 (Mar. 1991).

[11]CUMMINGS, *supra* note 4, at 140-74; RÉDEI, *supra* note 10, at 209-91 (explaining the effect of chromosomal abnormalities for both humans and other species).

Polyploidy occurs when an individual possesses an extra complete set of chromosomes. (The normal chromosomal constitution in humans is two sets of 23 chromosomes for a total of 46 chromosomes. One set is donated from the mother, and the other set is donated by the father.) Triploidy, having three sets of chromosomes instead of the normal two, occurs in 15-18% of all spontaneous abortions. The few triploid individuals who survive full term die within a month of birth. Tetraploidy, having two extra sets of chromosomes, occurs in 5% of all spontaneous abortions.

Aneuploidy occurs when a single chromosome is either added or deleted. Monosomy, having one less chromosome, is rarely observed in humans. Turner syndrome is an example involving the sex chromosomes. The reason other monosomic conditions are not observed may be that the embryo is lost before pregnancy is recognized. Individuals that are trisomic, having the addition of one extra chromosome, show varying degrees of development. Patau, Edwards, Klinefelter, and Down syndrome are examples of trisomic conditions.

Deletions, loss of part of a chromosome, occur in humans. An example is *cri du chat* syndrome that results from the loss of the short arm of chromosome 5.

Translocations occur when DNA is exchanged from nonhomologous chromosomes. Translocations are implicated in several human cancers.

Chromosome[12]: A single DNA molecule with its associated proteins, some of which are called histones. The structure of a chromosome has several levels. The DNA molecule, itself, is a double helix. In eukaryotic organisms, including humans, DNA is coiled around several histones to form nucleosomes. The nucleosomes are in turn folded among themselves to form a supercoiled molecule called a chromosome.

Cystic Fibrosis (CF)[13]: One of the most common, inherited diseases in Caucasian populations. CF affects approximately one in 2500 children in those populations. Common clinical manifestations are bowel obstruction and chronic obstructive pulmonary disease. Life expectancy for afflicted individuals ranges from the neonatal period to the sixth decade. About half live past age 25.

CF is inherited as an autosomal (i.e., not sex-linked) recessive. The gene has been mapped and cloned, and parents can be screened to determine if they carry the CF gene.

Cytoplasm[14]: The cellular material within the plasma membrane which contains the organelles but excluding the nucleus which contains the chromosomes.

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[12]GRIFFITHS ET AL., *supra* note 2, at 470-74; Michael Grunstein, *Nucleosomes: Regulators of Transcription*, 6 TRENDS IN GENETICS 395 (1990); RÉDEI, *supra* note 10, at 374-78.

[13]Sherman Elias et al., *Carrier Screening for Cystic Fibrosis: A Case Study in Setting Standards of Medical Practice, in* GENE MAPPING 186 (George J. Annas and Sherman Elias eds., 1992); David J. Porteous and Veronica van Heyningen, *Cystic Fibrosis: From Linked Markers to the Gene*, 2 TRENDS IN GENETICS 149 (1986).

[14]DARNELL ET AL., *supra* note 7, at 113-116; Academic Press Dictionary of Science and Technology, *supra* note 7, at 579; David S. Goodsell, *A Look Inside the Living Cell*, 80 AMERICAN SCIENTIST 457 (1992).

Cytosine (C)[15]: One of the four nitrogenous bases present in DNA. Cytosine is a pyrimidine, as is thymine another one of the bases. In the DNA helix cytosine is always connected to, or "paired" with, guanine by three hydrogen bonds.

D. melanogaster (Drosophila melanogaster)[16]: A fruit fly used extensively by geneticists. *Drosophila* was the organism used to demonstrate that linkage of observed genetic traits (in this case eye color) in an individual was associated with the physical crossing over of chromosomes in the parents. The observation led to the conclusion that genes were in the chromosomes.

DNA (deoxyribonucleic acid)[17]: The molecule that stores the "instructions" for development. DNA is comprised of two helical strands which are bound together by hydrogen bonds between pairs of nitrogenous bases. Each strand is a polymer (many copies of a monomer) of four nucleotides. Each nucleotide (the monomers) is comprised of one of four nitrogenous bases, the same deoxyribose sugar, and phosphoric acid. The specific sequence of nitrogenous bases determines which gene is present. Within each strand the nucleotides are bound together by phosphodiester links. That is, the ribose sugars of two adjacent nucleotides are bound together through a phosphate molecule.

Originally, the genes of DNA were envisioned as "beads on a string." With the discovery of controlling elements, so-called jumping genes, DNA is now viewed as a more fluid or dynamic molecule.

DNA fingerprinting[18]: Identifying a particular individual from DNA. Lengths of repeating sequences are compared from approximately 35 different locations throughout an individual's genetic material. No two individuals are thought to possess the same lengths of repeats at all the different locations, called variable number tandem repeat loci. Amounts of DNA sufficient to perform DNA fingerprinting can be obtained from dried blood spots, semen samples, or hair follicles. Typically, samples of DNA found at the crime scene are compared to the suspect's DNA. The exact probability that should attach to a "match," however, is in controversy.

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[15]DARNELL ET AL., *supra* note 7, at 67-69.

[16]GRIFFITHS ET AL., *supra* note 2, at 60-66; RÉDEI, *supra* note 10, at 62, 125-128, 681.

[17] DARNELL ET AL., *supra* note 7, at 66-75; GRIFFITHS ET AL., *supra* note 2, at 309-12; RÉDEI, *supra* note 10, note 15-19; Gary Felsenfeld, *DNA*, 253 SCIENTIFIC AMERICAN 58 (Oct. 1985).

[18]Peter Aldhous, *Geneticists Attack NRC Report as Scientifically* Flawed, 259 SCIENCE 755 (1993); B. Devlin et al., *Statistical Evaluation of DNA Fingerprinting: A Critique of the NCR's Report*, 259 SCIENCE 748 (1993); A.J. Jeffreys and S.D.J. Pena, *Brief Introduction to Human DNA Fingerprinting*, *in* DNA FINGERPRINTING: STATE OF THE SCIENCE 1 (S.D.J. Pena et al. eds., 1993); Samuel Karlin and Volker Brendel, *Chance and Statistical Significance in Protein and DNA Sequence Analysis*, 257SCIENCE 39 (1992); R.C. Lewontin and Daniel L. Hartl, *Population Genetics in Forensic DNA Typing*, 254 SCIENCE 1745 (1991); Kevin C. McElfresh et al., *DNA-based Identity Testing in Forensic Science*, 43 BIOSCIENCE 149 (1993); Mark D. Moody, *DNA Analysis in Forensic Science*, 39 Bioscience 31 (Jan. 1989); Sérgio D.J. Pena and Ranajit Chakraborty, *Paternity Testing in the DNA Era*, 10 TRENDS IN GENETICS 204 (1994).

Dominant[19]: Of a trait (such as a genetic disease) in which an individual with at least one gene will phenotypically display the trait. Since humans have two genes for each trait, the consequence of dominance is that inheritance of the trait will depend upon whether the affected individual has one or two genes. If an affected individual has two genes and mates with a normal or affected individual then all offspring will display the trait. If the affected individual has one gene and mates with a normal individual then each offspring has a 0.5 probability of displaying the trait. If two affected individuals, both carring one gene, mate then each offspring has a 0.75 probability of displaying the trait.

Down Syndrome[20]: One of the most common of all chromosome birth defects. The syndrome occurs in 1 of 900 live births. Afflicted individuals suffer from mental retardation and heart defects. The genetic cause is a trisomy (three copies) of chromosome 21.

Duchenne's Muscular Dystrophy[21]: A genetic disease characterized by progressive wasting of muscle tissue. Death usually occurs by the age of twenty from respiratory infection or cardiac failure. The disease is inherited as a sex-linked recessive.

E. coli (Escherichia coli)[22]: A bacterium found in the intestines of humans and animals. A favorite microorganism of geneticists.

Enzyme[23]: A protein molecule that catalyzes a biochemical reaction without itself being permanently altered or destroyed. Catalysis is the acceleration in the rate of a reaction.

Eukaryote[24]: Organisms that are plants and animals (including man). Some are single celled such as algae and amebae. Eukaryotes differ from prokaryotes in having subcellular structures with membranes such as a nucleus and mitochondria. In addition, the DNA in eukaryotes is associated with histones forming chromosomes.

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[19]GRIFFITHS ET AL., *supra* note 2, at 24; Athel Cornish-Bowden, *Dominance is not Inevitable*, 125 JOURNAL OF THEORETICAL BIOLOGY 333 (1987); Henrik Kacser, *Dominance Not Inevitable but Very Likely*, 126 JOURNAL OF THEORETICAL BIOLOGY 505 (1987); Peter D. Keightley and Henrik Kacser, *Dominance, Pleiotropy, and Metabolic Structure*, 117 GENETICS 319 (1987).

[20]CUMMINGS, *supra* note 4, at 148-49.

[21]CUMMINGS, *supra* note 4, at 119.

[22]Academic Press Dictionary of Science and Technology, *supra* note 7, at 771; RÉDEI, *supra* note 10, at 44.

[23]DARNELL ET AL., *supra* note 7, at 55-65; Academic Press Dictionary of Science and Technology, *supra* note 7, at 755.

[24]DARNELL, *supra* note 7, at 111-17; RIEGER ET AL., *supra* note 6, at 172.

Fragile X Syndrome[25]: Most common cause of familial mental retardation. The syndrome occurs when a region of the X chromosome (bearing a three-nucleotide sequence of nitrogenous bases of cytosine-cytosine-guanine) is repeated between 230 and 2,000 times. In a normal individual the sequence is repeated 6 to 50 times. The number of repeats of the sequence increases when transmitted from mother to child. The exact method of amplification of the sequence is not known.

Gene[26]: The simplest definition is the basic unit of inheritance, or heredity. Alternately, a gene is a particular sequence of nucleotides along a molecule of DNA which represents a functional unit of inheritance. Genes determine the heritable characters observed in the phenotype of an individual.

Gene Amplification[27]: The production of additional copies of a gene. Some cancerous tumors have genes that have been amplified. The result of amplification is an increase in the level of the protein product of the gene.

Gene Cloning[28]: The production of a population of multiple copies of a DNA fragment. Cloning involves the isolation of a fragment of DNA from a chromosome and the copying of the fragment. The fragment of interest will usually code for a gene, such as the gene for cystic fibrosis, or will be closely linked to a gene of interest.

The cloned fragment has several uses. If the cloned fragment codes for the gene, itself, the clone can be used to identify the gene's product. Identifying the gene's product assists in the study of disease. Additionally, the cloned gene can be placed in another individual (this activity is called genetic transformation). The individual receiving the cloned DNA is called transgenic. Finally, the fragment can be used as a probe to determine if a given individual in a population carries the gene.

Gene Therapy[29]: The treatment of a genetic disease, or disorder, by the transfer of a functional gene into the cells of the affected individual.

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[25]Grant R. Sutherland, *The Enigma of the Fragile X Chromosome*, 1 TRENDS IN GENETICS 108 (1985); Grant R. Sutherland and Robert I. Richards, *Dynamic Mutations*, 82 AMERICAN SCIENTIST 157 (1994).

[26]GEORGE J. ANNAS and SHERMAN ELIAS, GENE MAPPING 277 (1992); JURG OTT, ANALYSIS OF HUMAN GENETIC LINKAGE 1 (1985) RIEGER ET AL., *supra* note 6, at 189-90.

[27]GRIFFITHS ET AL., *supra* note 2, at 523-24, 696; RIEGER ET AL., *supra* note 6, at 27.

[28]DARNELL ET AL., *supra* note 7, at 214-19; GRIFFITHS ET AL., *supra* note 2, at 423-34; RIEGER ET AL., *supra* note 6, at 192, 333-34.

[29]Kenneth W. Culver and R. Michael Blaese, *Gene Therapy for Cancer*, 10 TRENDS IN GENETICS 174 (1994); Theodore Fiedmann, *Gene Therapy for Neurological Disorders*, 10 TRENDS IN GENETICS 210 (1994); Mark A. Kay and Savio L.C. Woo, *Gene Therapy for Metabolic Disorders*, 10 TRENDS IN GENETICS 253 (1994); A. Dusty Miller, *Human Gene Therapy Comes of Age*, 357 NATURE 455 (1992); Rachel Nowak, *Genetic Testing Set for Takeoff*, 265 SCIENCE 464 (1994); John Rennie, *Grading the Gene Tests*, 270 SCIENTIFIC AMERICAN 88 (June 1994); Savio L.C. Woo, *Introductory Remarks to the Review Series on Gene Therapy*, 10 TRENDS IN GENETICS 111 (1994); Inder M. Verma, *Gene Therapy* 263 SCIENTIFIC AMERICAN 68 (Nov. 1990).

Genetic Engineering[30]: The deliberate manipulation or transfer of genes among individuals within a species or among species. Some goals of genetic engineering are to cure disease in humans and to increase yield in agricultural crops.

Genetic Predisposition[31]: A greater probability than normal that the genetic disease will develop. Genes for genetic predisposition increase the penentrance, defined as a manifestation in the phenotype, of other genes that code for the genetic disease.

Genetic Transformation[32]: The transfer and incorporation of foreign DNA into the genome of an individual. When the DNA becomes incorporated the individual is transgenic. That is, the individual is the result of genetic transformation.

Genome[33]: The normal chromosome compliment of the either the egg or sperm. That is, a complete single set of genes. The genetically normal individual possess two genomes.

Genotype[34]: The particular assemblage of genes possessed by an individual. The effects of genotype and environment determine an individuals phenotype.

Germline Cell[35]: An ancestral cell to any cell that develops into the gametes, that is, the egg and sperm cells.

Guanine (G)[36]: One of the four nitrogenous bases present in DNA. Guanine is a purine, as is adenine another one of the bases. In the DNA helix guanine is always connected to, or "paired" with, cytosine by three hydrogen bonds.

Heritability (H)[37]: The proportion of variation observed in a trait that is heritable when compared to the total variation for the trait observed in a given population. Highly heritable traits are those in which offspring closely resemble their parents. Conversely, a trait with low heritability are those in which offspring do not closely resemble their parents.

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[30]Griffiths et al., *supra* note 2, at 2-5; Rieger et al., *supra* note 6, at 210.

[31]Ray White and C. Thomas Caskey, *Genetic Predisposition and the Human Genome Project: Case Illustrations of Clinical Problems, in* GENE MAPPING 173 (George J. Annas and Sherman Elias eds., 1992).

[32]RIEGER ET AL., *supra* note 6, at 222-23. *See* Daniel E. Koshland, Jr., *The Engineering of Species,* 244 SCIENCE 1233 (1989).

[33]RÉDEI, *supra* note 10, at 690; RIEGER ET AL., *supra* note 6, at 227.

[34]D.S. FALCONER, INTRODUCTION TO QUANTITATIVE GENETICS 100 (2d ed. 1981).

[35]RIEGER ET AL., *supra* note 6, at 232.

[36]DARNELL ET AL., *supra* note 7, at 68-75.

[37]FALCONER, *supra* note 34, at 148-69; KLUG AND CUMMINGS, *supra* note 10, at 221-23.

Homosexuality[38]: Sexual attraction to the same sex. This trait is a classical example of the controversy surrounding the study of inheritance of complex behavior traits. The genetic research centers on determining the relative effects of nature and nurture. That is, estimating the relative influences of genotype and environment on expression of the trait in an individual.

Huntington Disease (or Huntington's Chorea)[39]: A disease first expressed in mid-adult life having symptoms of involuntary muscular movements. Other symptoms are personality change and dementia. The gene that causes the disease has been mapped to chromosome 4 and is inherited as an autosomal dominant.

Immortalized cell[40]: Cells in culture that are capable of indefinite growth, that is, division. The cells may be obtained from either tumor cells or normal cells that are transformed.

Inheritance[41]: The transmission of genetic information from parent to offspring. Gegor Mendel discovered the laws of inheritance in 1865 working with the garden pea.

Karyotypic Analysis[42]: Examining a picture of an individual's chromosomes for chromosomal abnormalities. The chromosomes used for the analysis are typically obtained from blood. The chromosomes are "spread" on a microscopic slide and stained. The chromosomes are then viewed through a microscope and a photograph is taken.

Linkage[43]: The phenomenon of two genes being on the same chromosome. The consequence of linkage is that the two genes are transmitted together to the offspring more often than random chance would predict.

Liposome[44]: A phospholipid bilayer in the shape of a sphere. The center of the sphere is aqueous and can be used to transport molecules into cells.

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[38]William Byne, *The Biological Evidence Challenged,* 270 SCIENTIFIC AMERICAN 50 (May 1994); Simon LeVay and Dean H. Hamer, *Evidence for a Biological Influence in Male Homosexuality,* 270 SCIENTIFIC AMERICAN 44 (May 1994). *See* Thomas J. Bouchard Jr., *Genes, Environment, and Personality* 264 SCIENCE 1700 (1994) John Hogan, *Eugenics Revisited,* 268 SCIENTIFIC AMERICAN 123 (June 1993); Robert Plomin et al., *The Genetic Basics of Complex Human Behaviors* 264 SCIENCE 1733 (1994).

[39]CUMMINGS, *supra* note 4, at 429, 438.

[40]DARNELL ET AL., *supra* note 7, at 167-68.

[41]GRIFFITHS ET AL., *supra* note 2, at 19-42; RÉDEI, *supra* note 10, at 67-82; RIEGER ET AL., *supra* note 6, at 266-67.

[42]CUMMINGS, *supra* note 4, at 32-33.

[43]OTT, *supra* note 26, at 3-6.

[44]DARNELL ET AL., *supra* note 7, at 77; Academic Press Dictionary of Science and Technology, *supra* note 7, at 1250; Marc J. Ostro, *Liposomes* 256 SCIENTIFIC AMERICAN 103 (Jan. 1987).

Microinjection: Using a very finely tipped needle to physically inject a DNA sequence into a cell.

Monoclonal antibody[45]: A single antibody (a protein produced by the body to "attack" a foreign molecule) produced by a hybridoma. A hybridoma is the result of the fusion of a cancer cell and a normal antibody-producing spleen cell. A hybridoma is immortal and produces only one antibody. Monoclonal antibodies are useful in medicine as diagnostic tools.

Mutation[46]: A change in the DNA sequence of a gene that is inherited in the offspring. The change can be as small as the change in one nitrogenous base to the deletion or translocation of a substantial portion of the DNA sequence. Genetic diseases arise from mutations in normal genes.

Nucleus[47]: The organelle in eukaryotic cells that contains the chromosomes. The nucleus is bound by a membrane.

p53[48]: One of several tumor suppressor genes. About half of all human tumors are found to have a mutation in this gene. The normal product of the gene appears to delay cell division when the DNA has been damaged. During the delay the damaged DNA is repaired. If not repaired, the damaged DNA can engender cancer.

Phenotype[49]: The observed expression of a trait, or character, in an individual. Usually, the phenotype is determined, or influenced, by both an individual's genes and the environment of the individual. Symbolically, this relationship can be expressed:

Phenotype = Genotype + Environment

An example of the interaction of genotype and environment to produce a phenotype is alcoholism. Assuming a genetic component exists for alcoholism, an individual may possess the genes for alcoholism but not express the disease if raised in an environment without access to alcohol. In such a situation an individual would not have the phenotype of alcoholism but would possess the genotype.

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[45]DARNELL ET AL., *supra* note 7, at 172-73.

[46]D.C. Reanney, *Genetic Error and Genome Design*, 52 COLD SPRING HARBOR SYMPOSIA ON QUANTITATIVE BIOLOGY 751 (1987); RIEGER ET AL., *supra* note 6, at 343.

[47]DARNELL ET AL., *supra* note 7, at 113, 134.

[48]Curtis C. Harris, *p53: At the Crossroads of Molecular Carcinogenesis and Risk Assessment*, 262 SCIENCE 1980 (1993); Daniel E. Koshland, Jr., *Molecule of the Year, Id.* at 1953; Jean Marx, *Learning How to Suppress Cancer*, 261 SCIENCE 1385 (1993); Julie Ann Miller, *Genes That Protect Against Cancer*, 40 BIOSCIENCE 563 (1990).

[49]FALCONER, *supra* note 34, at 100; Otto E. Landman, *Inheritance of Acquired Characteristics Revisted*, 43 BIOSCIENCE 696 (1993); Tabitha M. Powledge, *The Genetic Fabric of Human Behavior*, 43 BIOSCIENCE 362 (1993); Tabitha M. Powledge, *The Inheritance of Behavior in Twins*, 43 Bioscience 420 (1993).

Phenylketonuria (PKU)[50]: A disease in which afflicted individuals lack the enzyme that catalyzes the conversion of phenylalanine to tyrosine. The disease is inherited as an autosomal recessive.

Plasmid[51]: A circular strand of DNA not part of the main genetic constitution of an individual. Plasmids are used as vectors in genetic transformation research. In bacteria, plasmids carry genes for resistance to antibiotics.

Polymerase Chain Reaction (PCR)[52]: A method of producing a large number of copies of a specific sequence of DNA from an original template of DNA. In PCR, a strand of DNA with the gene, or sequence of interest, is heated. In the presence of heat, DNA, a double helix, separates into two strands (a phenomenon called melting). When cooled, short lengths of single stranded DNA called primers hybridize, or bind, with the template DNA at specific sites. A polymerase (an enzyme that catalyzes the assembly of DNA) then replicates the primer DNA adding nucleotides that correspond to the sequence of the template DNA. The template/primed duplex is then heated and the duplex separates. Upon cooling the process begins again. After twenty cycles, one million copies of the original sequence can be produced.

Polymorphism[53]: Visible differences in individuals in a population caused by variation in the gene that controls the trait. The gene varies in its specific sequence of nitrogenous bases.

Prokaryote[54]: Any species of bacteria. Prokaryotes lack organelles including a nucleus. Additionally, prokaryotes do not have true chromosomes.

Proteins[55]: A group of organic compounds that consist of amino acids. Amino acids contain carbon, hydrogen, nitrogen, oxygen, and sometimes sulfur. Enzymes and antibodies are examples of proteins.

RNA (ribonucleic acid)[56]: A single stranded polymer of nucleotides. The nucleotides are the same as in DNA except that the nitrogenous base thymine is replaced by another

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[50]CUMMINGS, *supra* note 4, at 261.

[51]GRIFFITHS ET AL., *supra* note 2, at 625.

[52]Kary B. Mullis, *The Unusual Origin of the Polymerase Chain Reaction*, 262 SCIENTIFIC AMERICAN 56 (April 1990); Thomas J. White et al., *The Polymerase Chain Reactions*, 5 TRENDS IN GENETICS 185 (1989).

[53]FALCONER, *supra* note 34, at 42-43.

[54]DARNELL ET AL., *supra* note 7, at 111-17; RIEGER ET AL., *supra* note 6, at 398.

[55]DARNELL ET AL, *supra* note 7, at 44-68; Academic Press Dictionary of Science and Technology, *supra* note 7, at 1742.

[56]DARNELL ET AL., *supra* note 7, at 73, 75; James E. Darnell, Jr., *RNA*, 253 SCIENTIFIC AMERICAN 68 (Oct. 1985).

nitrogenous base, uracil. RNA is an "intermediate" molecule formed during the process of creating protein from genes (encoded in DNA). In fact, in organism the flow of "information" is DNA to RNA to protein.

Recessive[57]: Of a trait (such as a genetic disease) in which an individual must have two genes to phenotypically display the trait. Since humans have two genes for each trait, the consequence of recessiveness is that in a mating of an affected individual and a normal individual the offspring will have zero probability of displaying the trait. In a mating of two individuals both of whom carry a copy of the gene (but do not express the trait) each offspring has a 0.25 probability of displaying the trait. In a mating of two affected individuals (that is both individuals exhibiting the the trait) all offspring will display the trait.

Restriction enzymes[58]: A group of enzymes isolated from bacteria that "cut," or produce double strand breaks in DNA at specific sites in a sequence. Some restriction enzymes' recognition sites in DNA are palindromic sequences.

Restriction Fragment Length Polymorphism (RFLP)[59]: A polymorphism in the length of fragments of DNA created when a restriction enzyme is applied to DNA from several individuals in a population. Individuals differ in the length of specific fragments due to differences in the nucleotide sequences of their genes. RFLPs are heritable. RFLPs can be used, for example, to determine the inheritance of disease genes or to determine paternity. Sex-linked: Of a trait, or character, in which the gene is carried on the sex chromosomes, almost always the X chromosome. The consequece of a trait being sex-linked is that males display the trait at higher frequencies than females.

Somatic Cell[60]: All cells that are not germline, egg, or sperm cells.

Southern Blot[61]: The transfer of DNA fragments onto nitrocellulose or nylon. The DNA fragments are RFLPs that have been separated according to length by electrophoresis. After blotting, a radioactive probe is hybridized to the fragment of interest.

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[57] GRIFFITHS ET AL., *supra* note 2, at 23-25; RIEGER ET AL., *supra* note 6, at 411.

[58] KLUG AND CUMMINGS, *supra* note 10, at 384-86.

[59] CUMMINGS, *supra* note 4, at 212-15; H. DONIS-KELLER ET AL., *Highly Polymorphic RFLP Probes as Diagnostic Tools,* 51 COLD SPRINGS HARBOR SYMPOSIA ON QUANTITATIVE BIOLOGY 317 (1986); Peter J. Neufeld and Neville Colman, *When Science Takes the Witness Stand,* 262 SCIENTIFIC AMERICAN 46 (May 1990); Ray White and Jean-Marc Lalouel, *Chromosome Mapping with DNA Markers,* 258 SCIENTIFIC AMERICAN 40 (Feb. 1988).

[60] RIEGER ET AL., *supra* note 6, at 456.

[61] DARNELL ET AL., *supra* note 7, at 209-10; KLUG AND CUMMINGS, *supra* note 10, at 399-400.

Thymine[62]: One of the four nitrogenous bases present in DNA. Thymine is a pyrimidine, as is cytosine another one of the bases. In the DNA helix thymine is always connected to, or "paired" with, adenine by two hydrogen bonds.

Transgenic[63]: The term used to describe individuals containing a foreign gene introduced into the genome by gene transfer techniques.

Turner Syndrome[64]: A genetic disease in which afflicted females lack one of the two X chromosomes. That is, the female is monosomic for the sex chromosome. Afflicted individuals are short and wide chested.

VNTR (Variable Number Tandem Repeats)[65]: The sequences of DNA of an individual's genome used in DNA fingerprinting.

Vector[66]: The strand of DNA to which a gene that is to be cloned is fused. The vector contains genes for replication.

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[62]GRIFFITHS ET AL., *supra* note 2, at 307-12.

[63]RIEGER ET AL., *supra* note 6, at 487. The inventor of a transgenic mammal, a mouse, received a patent from the United States Patent and Trademark Office in 1988. Michael B. Landau, *Multicellular Vertebrate Mammals as "Patentable Subject Matter" Under 35 U.S.C. § 101: Promotion of Science and the Useful Arts or an Open Invitation for Abuse*, 97 DICK. L. REV. 203 (1993).

[64]CUMMINGS, *supra* note 4, at 152-54.

[65]R. C. Lewontin and Daniel L. Hartl, *Population Genetics in Forensic DNA Typing*, 254 SCIENCE 1745 (1991).

[66]GRIFFITHS ET AL., *supra* note 2, at 420-23.