Appendix J

Acronyms and Glossary of Terms

Acronyms

A — adenine
ADA — Americans with Disabilities Act
AHH — aryl hydrocarbon hydroxylase
AIDS — acquired immunodeficiency syndrome
ALA — aminolevulinic acid
AMA — American Medical Association
ANFI — absolute nuclear flourescence intensity
AOMA — American Occupational Medical Association
APKD — adult polycystic kidney disease
AS — ankylosing spondylitis
ASO — allele-specific oglionucleotide
AT — ataxia telangiectasia
ATSDR — Agency for Toxic Substances and Disease Registry (PHS, DHHS)
BC/BS — Blue Cross/Blue Shield
BLS — Bureau of Labor Statistics (DOL)
C — cytosine
CA — chromosomal aberration
CAD — coronary artery disease
CDC — Centers for Disease Control (PHS, DHHS)
CEHIC — Center for Environmental Health and Injury Control (CDC, DHHS)
CEO — Chief Executive Officer
CHD — coronary heart disease
CML — chronic myelogenous leukemia
COPD — chronic obstructive pulmonary disease
DHHS — U.S. Department of Health and Human Services
DNA — deoxyribonucleic acid
DOD — U.S. Department of Defense
DOE — U.S. Department of Energy
DOL —U.S. Department of Labor
EPA — U.S. Environmental Protection Agency
FA — Fanconi's anemia/syndrome
FPP — fetal protection policy
G — guanine
G-6-PD — glucose-6-phosphate dehydrogenase
GM — General Motors
GSH — reduced glutathione
HCS — hereditary cancer syndrome
HD — Huntington's disease
HDL — high-density lipoprotein
HERL —Health Effects Research Laboratory (EPA)
HHMI — Howard Hughes Medical Institute
HIV — human immunodeficiency virus
HLA — human leukocyte antigen
HPRT — hypoxanthine-guanine phosphoribosyltransferase
IDDM — insulin-dependent diabetes mellitus
IgA — immunoglobulin A
IOH — Institute of Occupational Health (Finland)
LDL — low-density lipoprotein
MRP — medical removal protection
NADH — nicotinamide adenine dinucleotide, reduced form
NAS — National Academy of Sciences
NCI —National Cancer Institute (NIH, DHHS)
NCTR — National Center for Toxicological Research (FDA, DHHS)
NIEHS — National Institute of Environmental Health Sciences (NIH, DHHS)
NIH —National Institutes of Health (DHHS)
NIOSH — National Institute for Occupational Safety and Health (CDC, DHHS)
NLRA — National Labor Relations Act
NLRB — National Labor Relations Board
NOES — National Occupational Exposure Survey (NIOSH)
NRC —National Research Council (NAS)
NSF — National Science Foundation
NTP — National Toxicology Program (PHS, DHHS)
OCT — ornithine carbamoyl transferase
OSH Act — Occupational Safety and Health Act
OSHA — Occupational Safety and Health Administration (DOL)
OSHRC — Occupational Safety and Health Review Commission (DOL)
OTA — Office of Technology Assessment
PCBs — polychlorinated biphenyls
PCR — polymerase chain reaction
PHS —U.S. Public Health Service (DHHS)
PKU — phenylketonuria
PTL — phenylthiourea
Px — peroxidase
Rb — retinoblastoma
RFLP — restriction fragment length polymorphism
RNA — ribonucleic acid
SARA — Superfund Amendments and Reauthorization Act of 1986
SAT — serum alpha-l-antitrypsin
SCE — sister chromatid exchange
SIC — standard industrial code
SOD — superoxide dismutase
SRBI — Schulman, Ronca, & Bucuvalas, Inc.
T — thymine
TSCA —Toxic Substances Control Act
TSD — Tay-Sachs disease
VA — U.S. Department of Veterans Affairs
XP — xeroderma pigmentosum

Glossary of Terms

Acetylation: The introduction of one or more acetyl groups into an organic compound.

Acquired immunodeficiency syndrome (AIDS): The most severe clinical manifestation of immune dysfunction caused by the human immunodeficiency virus (HIV).

Allele: Alternative form of a genetic locus (e.g., at a locus for eye color there might be alleles resulting in blue or brown eyes); alleles are inherited separately from each parent.

Amino acid: Any of a group of 20 molecules that combine to form proteins in living things. The sequence of amino acids in a protein is determined by the genetic code.

Autosome: Chromosome not involved in sex determination. In a complete set of human chromosomes, there are 44 autosomes (22 pairs).

Base pair: Two complementary nucleotides (adenosine and thymidine or guanosine and cytidine) held together by weak bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Biochemical genetics: The analysis of mutant genes on the basis of altered proteins or metabolizes.

Carcinogen/carcinogenesis: A chemical or physical agent that causes cancer.

Carrier: An individual apparently normal, but possessing a single copy of a recessive gene obscured by a dominant allele; a heterozygote.

Cell: The smallest component of life capable of independent reproduction.

Cell culture: Growth in the laboratory of cells isolated from multicellular organisms. Each culture is usually of one cell type (e.g., lymphocytes, fibroblasts, etc.).

Chromosomal aberrations: An abnormal chromosomal complement resulting from the loss, duplication, or rearrangement of genetic material.

Chromosome: A threadlike structure that carries genetic information arranged in a linear sequence. In humans, it consists of a complex of nucleic acids and proteins.

Clastogens: Chromosome-damaging agent.

cloning: The process of asexually producing a group of cells (clones), all genetically identical to the original ancestor. In recombinant DNA technology, the process of using a variety of DNA manipulation procedures to produce multiple copies of a single gene or segment of DNA.

Cytogenetics: The study of the relationship of the microscopic appearance of the chromosomes and their behavior to the genotype and phenotype of the individual.

Deoxyribonucleic acid (DNA): The molecule that encodes genetic information. DNA is a double-stranded helix held together by weak bonds between base pairs of nucleotides.

DNA: See *deoxyribonucleic acid.*

DNA adducts: The binding of exogenous and xenobiotic materials to DNA to form additional products. They can be viewed as markers of exposure to specific toxicants.

DNA probes: Segments of single-strand DNA that are labeled with a radioactive or other chemical marker and used to identify complementary sequences of DNA by hybridizing with them.

DNA sequence: Order of nucleotide bases in DNA.

Dominant: An allele that exerts it phenotypic effect when present either in homozygous or heterozygous form.

Dosimeter: Device or methodology for measuring the dose of a chemical or ionizing radiation to a biological system.

Double helix: The shape in which two linear strands of DNA are bonded together.

Electrophoresis: Technique used to separate molecules such as DNA fragments or proteins. An electric current is passed through a medium containing the mixture, and each kind of molecule travels through the medium at a different rate, depending on its electrical charge and size. Separation is based on these differences.

Enzyme: A protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds, without being permanently altered or consumed by the reaction so that it can be used repeatedly.

Epidemiologic studies: Studies concerned with the relationships of various factors determiningg the frequency and distribution of diseases in a human population.

Gamete: Mature male or female reproductive cell with a haploid set of chromosomes (23); that is, a sperm or ovum.

Gel: The semi-solid matrix used in electrophoresis to separate molecules.

Gene: The fundamental unit of heredity; an ordered sequence of nucleotide base pairs to which a specific product or function can be assigned.

Genetic monitoring: Involves periodically examining employees to evaluate modifications of their genetic material+. g., chromosomal damage or evidence of increased occurrence of molecular mutations-that may have evolved in the course of employment. It ascertains whether the genetic material of the group of individuals has altered over time.

Genetic screening: A process to examine the genetic makeup of individuals for certain inherited characteristics. It can be used to detect occupationally and nonoccupationally related traits.

Genetic testing: Technologies that determine a person's genetic makeup or that identify changes (damage) in the genetic material of certain cells. As used in the

workplace, it encompasses both genetic monitoring and screening.

Genetics: The study of the patterns of inheritance of specific traits.

Genome: All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

Genotype: The genetic constitution of an organism, as distinguished from physical appearance, or phenotype.

Germ cell: The male and female reproductive cells; egg and sperm.

Hemoglobin: The oxygen-carrying molecule found in red blood cells.

Hemoglobinopathies: A collection of hereditary disorders of hemoglobin structure and/or function. Examples are sickle cell anemia and thalassemia.

Hemolysis: Condition involving the destruction of red blood cells.

Heterozygous: Having two different alleles at a particular locus.

HLA: see *human leukocyte antigen.*

Homozygous: Having the same allele at a particular locus.

Human Genome Project: Research and technology development efforts aimed at mapping and sequencing some or all of the genome of human beings and other organisms.

Human immunodeficiency **virus (HIV): The** retrovirus that is the etiologic agent of AIDS.

Human leukocyte antigen (HLA): Located on the surface of most cells, except blood cells, these protein-sugar structures differ among individuals and are important for acceptance or rejection of tissue or organ grafts and transplants.

Hybridization: The process of joining two complementary strands of DNA, or of DNA and RNA, together to form a double-stranded molecule.

In vitro: Literally, "in glass," pertaining to a biological process or reaction taking place in an artificial environment, usually a laboratory.

In vivo: Literally, "in the living," pertaining to a biological process or reaction taking place in a living cell or organism.

Linkage: The proximity of two or more markers (e.g., genes, RFLP markers) on a chromosome; the closer together the markers are, the lower the probability that they will be separated during meiosis and hence the greater the probability that they will be inherited together.

Locus: A specific, physical position on a chromosome.

Lymphocyte: One of the major groups of white blood cells.

Marker: An identifiable physical location on a chromosome (e.g., restriction enzyme cutting site, gene, RFLP marker) whose inheritance can be monitored. Markers can be expressed regions of DNA (genes) or some segment of DNA with no coding function but whose pattern of inheritance can be determined.

Metaphase: see *mitosis.*

Micronuclei: Result from the exclusion of fragments or whole chromosomes from nuclei formed at mitosis. Their presence can be taken as an indication of the previous existence of chromosomal aberrations.

Mitosis: The process of division involving DNA replication that results in two daughter cells with the same number of chromosomes and cytoplasmic material as the parent cell.

Mutagen/mutagenicity: A substance capable of inducing a heritable change in the genetic material of cells.

Mutation: Changes in the composition of DNA.

Neoplasm: A localized population of proliferating cells in an animal which are not governed by the usual limitations of normal growth. The neoplasm is said to be benign if it does not undergo metastasis and malignant if it undergoes metastasis.

Nucleotide: The unit of DNA consisting of one of four bases—adenine, guanine, cytosine, or thymine—attached to a phosphate-sugar group. The sugar group is deoxyribose in DNA. (In RNA, the sugar group is ribose and the base uracil substitutes for thymine.)

Oncogene: A gene, one or more forms of which is associated with cancer. Many oncogenes are involved, directly or indirectly, in controlling the rate of cell growth.

Phenotype: The appearance of an individual or the observable properties of an organism that result from the interaction of genes and the environment.

Polymerase chain reaction (PCR): An in vitro process, through which repeated cycling of the reaction reproduces a specific region of DNA, yielding millions of copies from the original.

Polymorphism: The existence of more than one form of a genetic trait.

Protein: A biological molecule whose structure is determined by the sequence of nucleotides in DNA. Proteins are required for the structure, function, and regulation of cells, tissues, and organs in the body.

Recessive: An allele that exerts its phenotype effect only when present in homozygous form, otherwise being masked by the dominant allele.

Recombinant DNA technology: Processes used to form a DNA molecule through the union of different DNA molecules, but now commonly used to refer to any techniques that directly examine DNA.

Reliability: The ability of a test to accurately detect that which it was designed to detect and to do so in a consistent fashion.

Replication: The synthesis of new DNA from existing DNA. PCR is an in vitro technology based on the principles of replication.

Restriction endonuclease: An enzyme that has the ability to recognize a specific DNA sequence and cut

it at that sequence.

Restriction enzyme: See *restriction endonuclease.*

Restriction fragment length polymorphism (RFLP): Variations in the size of DNA fragments produced by a restriction endonuclease at a polymorphic locus.

RFLP analysis: DNA techniques using single-locus or multilocus probes to detect variation in the DNA sequence by revealing size differences in DNA fragments produced by the action of a restriction enzyme. See *restriction fragment length polymorphism.*

Sensitivity: The ability of a test to identify correctly those who have a disease.

Single gene disorders: Hereditary disorders caused by a single gene (e.g., Duchenne muscular dystrophy, retinoblastoma, sickle cell disease).

Sister chromatid exchange: Crossing over between the sister chromatics (two daughter strands of a duplicated chromosome) during cell division (mitosis).

Somatic cells: Any cells in the body except reproductive cells and their precursors.

Southern blot: The nylon membrane to which DNA has adhered after the process of Southern blotting.

Specificity: The ability of a test to identify correctly those who do not have the trait or disease which is being tested.

Teratogen/teratogenesis: A physical or chemical agent (e.g., radiation, alcohol) that can cause congenital abnormalities as a result of exposure in utero.

Trait: A distinguishing feature; a characteristic or property of an individual.

Validity: The extent to which a test will correctly classify true susceptible and true nonsusceptible individuals; sensitivity and specificity are components of validity.