Appendix C

Acronyms and Glossary

Acronyms

-/- —negative CF carrier/negative CF carrier (couple)
+/- —positive CF carrier/negative CF carrier (couple)
+/+ —positive CF carrier/positive CF carrier (couple)
ABMG —American Board of Medical Genetics
ASHG —American Society of Human Genetics
BC/BS —Blue Cross and Blue Shield
CF -cystic fibrosis
CFTR -cystic fibrosis transmembrane conductance regulator
DF508 -delta F508 (most prevalent CF mutation)
DF508#i12-delta F508 plus 6 to 12 additional CF mutations
DNA -deoxyribonucleic acid
G542X —a CF mutation
G551D —a CF mutation
Em40 —health maintenance organization
ISONG —International Society of Nurses in Genetics
MSAFP —maternal serum alpha-fetoprotein
N1303K —a CF mutation
NIH —National Institutes of Health (NIH)
NSGC —National Society of Genetic Counselors
OTA -Office of Technology Assessment
R553X —a CF mutation
W1282X —a CF mutation

Glossary of Terms

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.

beta~-thalassemia: An autosomal recessive disorder affecting the red blood cells, resulting in anemia, infections, growth retardation, and other complications. β-thalassemia predominantly occurs among individuals of Mediterranean, Middle Eastern, Asian Indian, Chinese, Southeast Asian, and African descent.

Buccal: Relating to the inside of the cheek. A buccal swab collects cells that can be analyzed for CF mutations.

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

Chest physical therapy (chest PT): A cornerstone of CF therapy that moves the mucus blocking major air passages out of the lungs. A specific form of chest PT is bronchial drainage during which an individual claps on the chest or back of the patient who is usually lying on a table.

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.

Confidentiality: A fundamental component of the health care provider-patient relationship in which the professional has the duty to keep private all that is disclosed by the patient.

Consanguineous: Related by blood or origin, rather than by marriage.

Cystic fibrosis CF A life-shortening, autosomal recessive disorder affecting the respiratory, gastrointestinal, reproductive, and skeletal systems, as well as the sweat glands. CF is caused by mutations in the CF gene that affect the CF gene product, cystic fibrosis transmembrane conductance regulator (CFTR). Individuals with CF possess two mutant CF genes.

Cystic fibrosis carrier: An individual who possesses one CF mutation and one normal CF gene. CF carriers manifest no symptoms of the disorder. See ***carrier.***

Cystic fibrosis carrier screening: The performance of tests on persons for whom no family history of CF exists to determine whether they have one aberrant CF gene and one normal CF gene. See ***cystic fibrosis screening.***

Cystic fibrosis screening: The performance of tests to diagnose the presence or absence of the actual disorder, in the absence of medical indications of the disease or a family history of CF This type of diagnostic screening usually involves newborns, but rarely for CF except in Colorado and Wisconsin. See ***cystic fibrosis carrier screening.***

Cystic fibrosis transmembrane conductance regulator (CFTR): The CF gene product, which regulates chloride (Cl-) conductance and might be a Cl^- ion channel, the structure that governs Cl^- entry and exit in the cell. CFTR produced by a mutant CF gene is frequently impaired, resulting in the medical manifestations of CF in affected individuals.

DF508: A three base pair deletion in the CF gene that results in a faulty CF gene product (i.e., a flawed CFTR). This mutation results in the deletion of one amino acid, phenylalamine, at position number 508 in CFTR. DF508 is the most common mutant allele among the greater than 170 identified in the CF gene.

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.

Discrimination: Differential treatment or favor with a prejudiced outlook or action.

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

DNA: See *deoxyribonucleic acid.*

DNA analysis: A direct examination of the genetic material, DNA, to reveal whether a individual has mutation(s) for CF or other disorders.

DNA probe: Short segment of DNA labeled with a radioactive or other chemical tag and then used to detect the presence of a particular DNA sequence through hybridization to its complementary sequence.

Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotide base pairs to which a specific product or function can be assigned.

Gene therapy: The deliberate administration of genetic material into the cells of a patient with the intent of correcting a specific genetic defect.

Genetic counseling: A clinical service involving educational, informational, and psychosocial elements to provide an individual (and sometimes his or her family) with information about heritable conditions. Genetic counseling is performed by genetics specialists, including physicians, Ph.D. clinical geneticists, genetic counselors, nurses, and social workers.

Genetic screening: The analysis of samples from asymptomatic individuals with no family history of a disorder, groups of such individuals, or populations.

Genetic testing: The use of specific assays to determine the genetic status of individuals already suspected to be at high risk (e.g., family history or symptoms) for a particular inherited condition.

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

Genome: AU the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs. The human genome is 3.3 billion base pairs.

Heterozygote: A heterozygous individual, such as a CF carrier.

Heterozygous: Having two different alleles at a particular locus.

Homozygote: A homozygous individual.

Homozygous: Having the same alleles at a particular locus.

Immunoreactive trypsin (IRT) test: An assay that measures levels of pancreatic trypsin, a digestive enzyme. As a protocol for newborn CF screening, a drop of blood is isolated on a card, dried, and chemically analyzed to detect elevated levels of the enzyme. It is not intended to be a diagnostic test.

Mutation: Changes in the composition of DNA.

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.

Probe: A short segment of DNA tagged with a reporter molecule, such as radioactive phosphorus (^{32}P), used to detect the presence of that particular complementary DNA sequence.

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 phenotypic effect only when present in homozygous form, otherwise being masked by the dominant allele.

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

Sickle cell anemia: An autosoma1 recessive disorder affecting red blood cell flow through the circulatory system, causing complications in numerous organ systems. Sickle cell anemia predominantly occurs in individuals of African descent.

Sickle cell trait: The heterozygous state of sickle cell anemia; sickle cell carrier status.

Single-gene disorder: Hereditary disorder caused by a single gene (e.g., cystic fibrosis, Tay-Sachs disease, sickle cell anemia).

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

Stigmatization: Branding, marking, or discrediting because of a particular characteristic.

Sweat test: An assay used to confirm CF that measures levels of sodium Na^+) and chloride (Cl-) ions. These ions appear in high concentrations in patients with CF Sweating is induced by running a low electric current through a pilocarpine-soaked gauze pad on the individual's arm or back. The amounts of Na^+ and Cl- in the sweat can then be determined to confirm or question a diagnosis of CF

Tay-Sachs disease: A lethal autosomal recessive disorder affecting the central nervous system which results in mental retardation and early death. Tay-Sachs disease predominantly occurs among Jews of Eastern and Central European descent and populations in the United States and Canada descended from French Canadian ancestors.