Glossary

Definitions useful in understanding pharmacogenetics; Author: D. Farkas
http://www.aacc.org/AACC/members/nacb/LMPG/OnlineGuide/DraftGuidelines/Pharmacogenetics/

allele - version of a gene at a given locus

amplicon - copy of a target DNA created by PCR or other amplification methods

central dogma (of molecular biology) - fundamental tenet of molecular biology stating that DNA is copied via replication, RNA is derived from DNA via transcription, and protein is derived from RNA via translation. The discovery of reverse transcription disrupted the central dogma of molecular biology by showing that genetic information could also flow from RNA to DNA, not just DNA to RNA.

complementary DNA (cDNA) - DNA produced using an RNA template via the enzyme reverse transcriptase

compound heterozygote - individual with two abnormal alleles at a given locus, each with a difference polymorphism or mutation.

deletion - mutation resulting from the removal of base(s)

DNase - enzyme that degrades DNA

DNA ligase - enzyme that joins two pieces of DNA

DNA polymerase - enzyme that uses DNA as a template to produce a complementary strand of DNA; cDNA is made with a type of DNA polymerase called reverse transcriptase (an RNA-dependent, DNA polymerase)

DNA sequencing - base-by-base determination of the exact sequence of target DNA

epigenetic - referring to heritable changes to the genome that do not alter the coding sequence

frame-shift mutation - insertion or deletion of base(s) that alters the reading frame of a coding sequence, thereby changing the amino acids encoded downstream and/or producing a stop codon

gel electrophoresis - separation of DNA by size via migration in an electric field in an agarose or polyacrylamide matrix

gene - segment of DNA transcribed into RNA that (i) is translated into a protein or (ii) forms structures such as ribosomes

genetic variant - alternative forms of a gene which may or may not lead to altered phenotype

genome - all the genetic material of an organism

genotype - the alleles at a given locus in an individual; see also phenotype

haplotype - analogous to genotype, haplotype is the set of alleles (or SNPs) on one chromosome or part of a chromosome that are linked and usually or often inherited together

heterogeneous - regarding PCR, requiring separate amplification and detection steps

homogeneous - regarding PCR, having combined amplification and detection steps

hybridization - process of forming a double-stranded molecule from a single-stranded probe and a single-stranded nucleic acid target

hybridization probe - type of fluorescently-labeled probe used in real-time PCR that produces signal following hybridization to a target DNA

missense mutation -base change resulting in coding of a different amino acid

molecular diagnostics - diagnosis of disease using nucleic acids as analytes, often used synonymously with molecular pathology

mRNA - messenger RNA, translated into protein

mutation - generally harmful DNA sequence change; compare to polymorphism

nucleic acids - deoxyribonucleic acid (DNA) and ribonucleic acid (RNA), the molecules containing the genetic code

nucleoside - nucleotide lacking a phosphate group

nucleotide - building block of nucleic acids composed of phosphate group(s), a five-sided sugar molecule, and a nitrogenous base

oligonucleotide - short sequence of nucleotides, often used as primers for PCR or DNA sequencing

penetrance - percent expression in a population of the phenotype of a given mutant genotype

pharmacogenetics - the hereditary basis for inter-individual differences in drug response

pharmacogenomics - the convergence of pharmacogenetics and genomics used to mean the influence of DNA sequence variation on the effect of a drug on an individual

phenotype - traits resulting from a given genotype

point mutation - mutation that changes a single base

polymerase chain reaction (PCR) - enzymatic in vitro nucleic acid amplification method using temperature cycling to produce repeated cycles of DNA replication

polymorphism - variant DNA sequence change, typically benign, found in 1% or more of individuals; compare to mutation

primer - oligonucleotide used in PCR or DNA sequencing to target an area of interest

primer-dimers - nonspecific products formed during PCR by the interaction of primers

probe - relatively small piece of DNA or RNA used to find or detect a specific piece of target nucleic acid

proteomics - study of the entire complement of proteins in organisms

quantification standard - synthetic nucleic acid standard spiked into samples before processing to serve as a reference in quantitative PCR

real-time PCR - PCR in which detection of product is simultaneous with amplification

replication - process of duplicating DNA with DNA polymerase

restriction endonuclease (RE) - enzyme purified from bacteria that recognizes and cleaves unique sequences

restriction fragment length polymorphism (RFLP) - polymorphism that changes the electrophoretic banding pattern of DNA fragments generated by digestion with a restriction endonuclease

single nucleotide polymorphism (SNP) - polymorphism that is a single base change

Southern blot hybridization - DNA detection method where digested sample is separated by electrophoresis, transferred to a membrane, and probed

transcription - process of producing mRNA from a DNA template

translation - process of converting the information contained in mRNA into protein

uracil-N-glycosylase - enzyme used to prevent amplicon carryover contamination that degrades any DNA containing uracil (uracil-containing DNA is not natural and is produced in vitro during some PCR protocols)

variant allele - specific alternative forms of a gene, generally causing a known alternative phenotype

wild-type - normal allele; compare to mutant or variant