

Glossary

AGAROSE: A polysaccharide isolated from seaweed used as a matrix in gel electrophoresis.

ALLELE: One of two alternate forms of a gene occupying a given locus on the chromosome.

ALLOSTERIC CONTROL: The ability of an interaction at one site of a protein to influence (positively or negatively) the activity at another site.

ALU FAMILY: A set of short (*ca.* 300bp) related sequences dispersed throughout the human genome. Refers to the property of these sequences to be cleaved once by the restriction enzyme *AluI*. Genomes of other mammals contain similar families. Their role is unknown.

AMPLIFICATION: The production of extra copies of a chromosomal sequence found either as intra- or extra-chromosomal DNA. With respect to plasmids it refers to the increase in the number of plasmid copies per cell induced by certain treatments of transformed cells.

ANNEAL (RE-ANNEAL): The (re)establishment of base pairing between complementary strands of DNA or a DNA and an RNA strand.

ANOMERIZATION: The interconversion of stereoisomers of a sugar that differ only in the stereochemistry at the carbonyl carbon in their cyclic (furanose or pyranose) form. For D-ribofuranose and D-2-deoxyribofuranose this relates to the α - and β -forms at C-1.

ANTIBODY: A protein that is produced in response to and specifically recognizes and binds to an antigen.

ANTICODON: A triplet of nucleotides in a constant position in the structure of tRNA that is complementary to the triplet codon(s) in mRNA to which the tRNA responds.

ANTIGEN: Any molecule which, upon entry into the organism, causes the production of antibodies (immunoglobulins).

ANTISENSE: A strand of DNA or RNA that has the sequence complementary to mRNA (also non-coding strand).

APOPTOSIS: The programmed death of a cell within a multi-cellular organism, which follows an ordered process.

APTAMER: DNA or RNA molecules that have been selected from random pools based on their ability to bind other molecules.

ARRAY: A spatial arrangement of *e.g.* oligonucleotides or peptides, which can be at high density ($\geq 10,000$ individual sequences).

AUTORADIOGRAPHY: The detection of radioactively labelled molecules present for example in a gel or on a filter by exposing an X-ray film to it.

AUXOTROPHY: The inability of microorganisms to live on minimal medium without supplemented (auxiliary) nutrients.

- BACK MUTATION:** Reverses the effect of a mutation that had inactivated a gene.
- BACTERIOPHAGE:** A virus that infects bacteria; often abbreviated as phage.
- BASE PAIR (BP):** A duplex of A with T or of C with G in a DNA or RNA double helix; other pairs are possible in RNA under some circumstances.
- BLOTTING:** Transfer of DNA, RNA, or protein from a gel to nitrocellulose or other “paper”.
- CAP:** The structure at the 5'-end of eukaryotic mRNA introduced after transcription by linking the 5'-end of a guanine nucleotide to the terminal base of the mRNA and methylating at least the additional G; the structure is $7\text{Me}_G^5' \text{ppp}^5' \text{Np}$.
- CATENANE:** A molecule in which two or more closed rings are interlocked thus holding the structure together without any covalent bond between the separate rings. A DNA catenane is a topoisomer of its components, *i.e.* it is a distinct topological structure that can be acted on by topoisomerase.
- cDNA:** A single-stranded DNA complementary to the RNA synthesized from it by *in vitro* reverse transcription.
- CENTROMERE:** The most condensed and constricted region of a chromosome; point of attachment of the spindle fiber during mitosis.
- CHAIN TERMINATION SEQUENCING:** See Sanger–Coulson sequencing.
- CHROMATIN:** Basic organizational unit of eukaryotic chromosomes; consists of DNA and associated proteins assembled into fibers of average diameter 30 nm that are produced by the compaction of 10-nm nucleosome fibers.
- CHROMOSOME:** A discrete unit of the genome carrying many genes, consisting of a very long molecule of DNA, complexed with a large number of different proteins (mostly histones). Chromosomes are visible as a morphological entity only during the act of cell division.
- cis-ACTING:** The ability of a DNA (or RNA) sequence to effect its influence only on the molecule from which it forms a part. Usually implies that the sequence does not code for a protein. When applied to a protein it means that the protein acts only on the DNA (or RNA) molecule from which it was expressed.
- CISTRON:** The genetic unit defined by the *cis/trans* test; equivalent to gene in comprising a unit of DNA representing a protein.
- CLONE:** A large number of cells or molecules genetically identical with a single ancestral cell or molecule.
- CODON:** A triplet of nucleotides that corresponds to an amino acid or a termination signal.
- COMPETENT:** A culture of bacteria or yeast cells treated in such a way that their ability to take up DNA molecules without transduction or conjugation has been enhanced.
- COMPLEMENTARY SEQUENCE:** Nucleic acid sequence of bases that can form a double-stranded structure by virtue of Watson–Crick base pairing e.g. A-T, C-G.
- COMPLEMENTATION:** The ability of independent (non-allelic) genes to provide diffusible products that produce wild phenotype when two mutants are tested in *trans*-configuration in a heterozygote.
- CONJUGATION:** Directional transfer of DNA between two bacteria.
- CONSENSUS SEQUENCE:** An idealized sequence in which each position represents the base most often found when many actual sequences are compared.
- COPY NUMBER:** The average number of copies of a particular (recombinant) plasmid present in a single host cell. Also used for individual genes.

- COSMIDS:** Plasmids into which phage lambda cos sites have been inserted; as a result, the plasmid DNA can be packaged *in vitro* into the phage coat.
- CO-TRANSFORMATION:** Introduction of two or more genes carried on separate DNA molecules into a cell.
- CROSS-LINKING:** Introduction of covalent intra- or intermolecular bonds between groups that are normally not covalently linked. Used to detect proximity of (parts of) (macro) molecules.
- CUT:** A double-strand scission in the duplex polynucleotide in distinction to the single-strand “nick”.
- DELETION:** The removal of a sequence of DNA, the regions on either side being joined together.
- DENATURATION (OF PROTEIN):** Conversion from the native conformation into some other (inactive) conformation.
- DIFFERENTIAL LYSIS:** A method to enrich for sperm DNA in a mixture of sperm and epithelial cells by preferentially lysing the latter using detergent and protease, so that sperm nuclei can be recovered by centrifugation.
- DIRECT REPEATS:** Identical (or closely related) sequences present in two or more copies in the same orientation on the same DNA (or RNA) molecule; they are not necessarily adjacent.
- DNA FINGERPRINTING:** Generation of a pattern of bands, by Southern blotting and hybridization with a multi-locus probe, which is highly individual-specific.
- DNAZYME:** A short catalytic single-stranded DNA molecule.
- DOMAIN (OF A CHROMOSOME):** Either a discrete structural entity defined as a region within which supercoiling is independent of other domains, or an extensive region including an expressed gene that has heightened sensitivity to degradation by the enzyme DNase I.
- DOMAIN (OF A PROTEIN):** A discrete continuous part of the amino acid sequence that can be equated with a particular function or a particular substructure of the tertiary structure.
- DOMINANT (ALLELE):** Determines the phenotype displayed in a heterozygote with another (recessive) allele.
- DOWNSTREAM:** Sequences that proceed further in the direction of expression; for example, the coding region is downstream from the initiation codon.
- ELECTROPHEROGRAM:** The graphical output of electrophoresis devices in STR (see short tandem repeat) and sequencing analysis, showing fluorescence intensity as a function of molecular weight. The peak at a particular wavelength (colour) corresponds to a specifically labelled molecule of a particular size.
- END LABELLING:** The addition of a radioactively labelled group to one end (5' or 3') of a DNA or RNA strand.
- ENDONUCLEASE:** An enzyme that cleaves bonds within a nucleic acid chain. It may be specific for RNA or for single-stranded or double-stranded DNA.
- ENHANCER ELEMENT:** A DNA sequence that increases the utilization of (some) eukaryotic promoters in *cis*-configuration, but can function in any location, upstream or downstream, relative to the promoter.
- EPIPOPE:** Any part of a molecule that acts as an antigenic determinant. A macromolecule can have many different epitopes each stimulating the production of a different specific antibody.
- EUKARYOTIC:** Any organism that contains a nucleus.
- EXCISION-REPAIR:** A repair system that removes a single-stranded sequence of DNA containing damaged or mispaired bases and replaces it in the duplex by synthesis of a sequence complementary to the remaining strand.
- EXON:** Any segment of an interrupted gene that is represented in the mature RNA product.

- EXONUCLEASE:** An enzyme that cleaves nucleotides one at a time from the end of a polynucleotide chain. Such enzymes may be specific for either the 5'- or 3'-end of DNA or RNA.
- EXPRESSION VECTOR:** A cloning vector designed in such a way that a foreign gene inserted into the vector will be expressed in the host organism.
- FINGERPRINT:** The characteristic array of oligopeptides or oligonucleotides obtained upon two-dimensional electrophoresis of a protein digested with a specific endopeptidase or an RNA digested with a specific endonuclease.
- FOOTPRINTING:** A technique for identification of the site of DNA or RNA bound by some protein by virtue of the protection of bonds in this region against attack by nucleases or by chemicals.
- FORENSIC GENETICS:** The application of genetics for the resolution of disputes at law.
- FUSION GENE:** A recombinant gene constructed from parts of two different genes.
- FUSION PROTEIN:** The protein expressed by a fusion gene containing parts of the coding sequence of two different genes.
- GAPMER:** An antisense oligonucleotide where the central section is either unmodified or contains modifications, such as phosphorothioate, that permit recognition by RNase H, and where the 5'- and 3'-flanking regions contain other chemical modifications.
- GEL ELECTROPHORESIS:** Electrophoresis performed in a gel matrix (usually agarose or polyacrylamide) that allows separation of molecules of similar electric charge density on the basis of their difference in molecular weight.
- GENE:** A DNA sequence involved in the production of an RNA or protein molecule as the final product. Includes both the transcribed region and any sequences upstream and/or downstream responsible for its correct and regulated expression (*e.g.* promoter and operator sequences).
- GENETIC CODE:** The complete set of codons specifying the various amino acids, including the nonsense codons. The code is usually written in the form in which it occurs in mRNA. (It can be different in mitochondrial DNA.)
- GENOME:** The entire genetic material of a cell.
- G-TETRAD:** A structure that involves four oligonucleotide strands in which there is participation from one guanine base in each strand.
- HAIRPIN:** The double-stranded region formed by base pairing of adjacent complementary sequences in the same DNA or RNA strand.
- HAPTEN:** A small molecule that acts as an antigen when it is conjugated to a large (carrier) molecule.
- HETERODUPLEX (HYBRID) DNA:** DNA that is generated by base pairing between partly non-complementary single strands derived from the different parental duplex molecules. It occurs during genetic recombination.
- HOLLIDAY JUNCTION:** A structure that occurs during homologous recombination between two chromosomes; with the two chromosomes side-by-side, one strand of DNA on each chromosome is broken and then attached to the broken strand of DNA on the alternate chromosome. The crossover point is called the Holliday junction.
- HOLOENZYME:** The complete enzyme including all its subunits. Often used in reference to RNA and DNA polymerases.
- HOMOLOGY:** The degree of identity existing between the nucleotide sequences of two related but not complementary DNA or RNA molecules. 70% homology means that on average 70 out of every 100 nucleotides are identical. The same term is used in comparing the amino acid sequences of related proteins.

- HYBRIDIZATION:** The pairing of complementary RNA and DNA strands to give an RNA–DNA hybrid. It is also used to describe the pairing of two single-stranded DNA molecules.
- HYBRIDOMA:** The cell line produced by fusion of a myeloma cell with a lymphocyte. It continues indefinitely to express the immunoglobulins of both parents.
- HYPERCHROMICITY:** The increase of optical density that occurs when DNA is denatured.
- i-MOTIF:** A structure composed of two parallel-stranded duplexes held together in an antiparallel orientation. The structure is stabilised by hemiprotonated C:C⁺ base pairs.
- INCOMPATIBILITY:** The inability of certain bacterial plasmids to coexist in the same cell.
- INDUCER:** A small molecule that triggers gene transcription by binding to a regulator protein.
- INITIATION CODON:** AUG (sometimes GUG), three bases that code for the first amino acid in a protein sequence (*N*-formylmethionine in prokaryotes). This fMet is often removed post-translationally.
- IN SITU HYBRIDIZATION:** A technique in which the DNA of cells is denatured by squashing on a microscope slide so that reaction is possible with an added single-stranded RNA or DNA. The added preparation is radioactively labelled and its hybridization is followed by autoradiography.
- INTASOME:** A protein–DNA complex between the phage lambda integrase (Int) and the phage lambda attachment site (*attP*).
- INTRON:** A segment of DNA that is transcribed, but is removed from within the transcript by splicing together the sequences (exons) on either side of it. The occurrence of introns is almost exclusively limited to eukaryotic cells.
- IN VITRO:** (lit. “in glass”): Any experimental (biological) process that occurs outside the living cell.
- IN VIVO:** Any biological process that occurs within the living cell or organism.
- IPTG:** Isopropyl β-D-thiogalactoside; an artificial inducer of the *lac* operon (physiological inducer: allolactose).
- kb:** Abbreviation for 1000 base pairs of DNA or 1000 bases of RNA.
- KINASE:** An enzyme that catalyzes the transfer of a phosphate group from ATP or GTP to an acceptor, usually a protein or a nucleotide.
- KLENOW FRAGMENT:** An N-terminal truncation of DNA Polymerase I that retains polymerase activity, but has lost the 5′→3′ exonuclease activity.
- LAC OPERON:** An inducible operon in *Escherichia coli* that codes for three genes involved in the metabolism of lactose.
- LEADER SEQUENCE:** The sequence at the 5′-end of an mRNA that is not translated into protein. It contains the coded information that the ribosome and special proteins read to tell it where to begin the synthesis of the polypeptide.
- LIBRARY:** A set of cloned fragments together representing the entire genome.
- LIGASE:** (DNA LIGASE): An enzyme that catalyzes the formation of a phosphodiester bond at the site of a single-strand break in duplex DNA. Some DNA ligases can also ligate blunt-end DNA molecules. RNA ligase covalently links separate RNA molecules.
- LIGATION:** The formation of a phosphate diester linkage between two adjacent nucleosides separated by a nick in one strand of a double helix of DNA. (The term can also be applied to blunt-end ligation and to joining of RNA.)

- LINKER (FRAGMENT):** A short synthetic duplex oligonucleotide containing the target site for some restriction enzyme. A linker may be added to the end of a DNA fragment prepared by cleavage with some other enzyme during reconstruction of recombinant DNA.
- LTR:** An abbreviation for long-terminal repeat, a sequence directly repeated at both ends of a retroviral DNA.
- LYSIS:** The death of bacteria at the end of a phage infective cycle when they burst open to release the progeny of an infecting phage.
- M13:** An *E. coli* phage containing single-stranded circular DNA that forms the basis for a series of cloning vectors.
- MATCH PROBABILITY:** The chance of two unrelated people sharing a DNA profile.
- MAXAM–GILBERT SEQUENCING:** A DNA sequencing technique based on specific chemical modification of each of the four bases.
- MELTING TEMPERATURE (T_m):** The temperature where hyperchromicity is half-maximal.
- MINIMAL MEDIUM:** A chemically fully defined medium containing only inorganic sources of the essential elements as well as an organic carbon source.
- MINISATELLITES:** Loci made up of a number (~ 10 – 1000) of tandemly repeated sequences, each typically 10–100 bp in length, which are usually GC-rich and often hypervariable.
- MODIFIED BASES:** All those except the usual five from which DNA and RNA (A, C, G, T, and U) are synthesized. They result from post-synthetic changes in the nucleic acid or chemical synthesis.
- MONOCLONAL ANTIBODY:** The unique immunoglobulin molecule (1° protein sequence) produced by a clone of cells derived from the fusion of a B lymphocyte with a myeloma cell. The antibody is directed against a single epitope of the antigen used to raise the antibody.
- MULTICOPY PLASMIDS:** Present in bacteria at amounts greater than one per chromosome.
- MULTIPLE DISPLACEMENT AMPLIFICATION:** A method for whole-genome amplification using a highly processive polymerase from bacteriophage $\phi 29$ and random primers to synthesize long molecules from the template.
- MUTAGENS:** Molecules that increase the rate of mutation by causing changes in DNA.
- MUTATION:** Any change in the sequence of genomic DNA.
- NICK TRANSLATION:** The ability of *E. coli* DNA polymerase I to use a nick as a starting point from which one strand of a duplex DNA can be degraded and replaced by resynthesis of new material; is used to introduce radioactively labelled nucleotides into DNA *in vitro*.
- NONSENSE CODON:** Any one of three triplets (UAG, UAA, UGA) that cause termination of protein synthesis (UAG is known as *amber*, UAA as *ochre*, UGA as *opal*).
- NORTHERN BLOTTING:** A technique for transferring RNA from an agarose gel to a nitrocellulose filter on which it can be hybridized to a complementary DNA.
- NUCLEOLUS:** The region in the nucleus where rRNA synthesis takes place.
- NUCLEOSOME:** The fundamental repeating unit of a eukaryotic cell and which consists of DNA and histones.
- OKAZAKI FRAGMENTS:** Separate, contiguous DNA sequences of 1000–2000 bases produced during discontinuous replication; they are later joined together to give an intact strand.

OLIGOMER: Term often used in place of oligonucleotide.

OLIGONUCLEOTIDE: Polymer comprising of nucleotide units (usually less than 50) joined typically by 5'→3' phosphate diester linkages. Those comprised of DNA and RNA can be distinguished where necessary by using 'oligodeoxyribonucleotide' and 'oligoribonucleotide' respectively.

ONCOGENE: A retroviral gene that causes transformation of the mammalian infected cell. Oncogenes are slightly changed equivalents of normal cellular genes called proto-oncogenes. The viral version is designated by the prefix v, the cellular version by the prefix c.

OPEN READING FRAME (ORF): A series of triplets coding for amino acids terminated by a termination codon; sequence is (potentially) translatable into protein.

OPERATOR: The site on DNA at which a repressor protein binds to prevent transcription from initiating at the adjacent promoter.

OPERON: A complete unit of bacterial gene expression and regulation, including structural genes, regulator gene(s), and control elements in DNA recognized by regulator gene product(s).

ORIGIN (ORI): A sequence of DNA at which replication is initiated.

PALINDROME: A sequence of double-stranded DNA that is the same when one strand is read left to right or its complement is read right to left; consists of adjacent inverted repeats.

PATERNITY TESTING: The determination of whether or not a particular man is the father of a child, using genetic analysis. This generally uses similar autosomal markers to individual identification work.

pBR322: One of the standard plasmid cloning vectors.

PCR: Polymerase chain reaction, an *in vitro* amplification of DNA based on primer, template, and a thermostable DNA polymerase.

PCR STUTTER: A PCR artefact in which, as well as a band of the expected size, an additional band is seen that is typically one repeat unit smaller, resulting from slippage synthesis errors by the PCR polymerase.

PHAGE (BACTERIOPHAGE): A bacterial virus.

PLASMID: An autonomous self-replicating extrachromosomal circular DNA.

PLASTID: A family of membrane-bound organelles unique to plant cells; only one type is found in each cell while all types derive from a common precursor organelle called a proplastid.

POLYADENYLATION: The post-transcriptional attachment of up to 200 AMP residues to the 3'-terminus of most eukaryotic mRNAs.

POLYLINKER: A synthetic double-stranded DNA oligonucleotide containing a number of different restriction sites.

POLYMERASE: An enzyme that catalyzes the assembly of nucleotides into RNA or of deoxynucleotides into DNA; usually the enzyme requires single-stranded DNA (sometimes RNA) as a template.

POLYMORPHISM: The simultaneous occurrence in the population of genomes showing allelic variations (as seen either on alleles producing different phenotypes or, for example, in changes in DNA affecting the restriction pattern).

PHOSPHATASE: A class of enzymes that hydrolyses (terminal) phosphoryl groups from nucleotides as well as from proteins.

- PRIMER:** A short sequence (of DNA or RNA) that is paired with one strand of DNA and provides a free 3'-OH end at which a DNA polymerase starts synthesis of a deoxyribonucleotide chain.
- PROBE (HYBRIDIZATION):** A labelled DNA or RNA molecule used to detect a complementary sequence by molecular hybridization.
- PROKARYOTIC:** Any organism that lacks a membrane-enclosed nucleus.
- PROMOTER: (IN BACTERIA):** The region of the gene involved in binding of the RNA polymerase. (In eukaryotes) usually all regions of the gene required for maximum expression (excluding enhancer sequences).
- PROTEIN A:** A protein from *Staphylococcus aureus* that binds specifically to immunoglobulin G molecules. Used in detection of proteins by immunological techniques.
- PROTEINASE K:** A protease used to remove contaminating protein from preparations of nucleic acids. The enzyme also degrades itself.
- PROTEIN KINASE:** A class of enzymes that phosphorylates a protein with the help of ATP, the phosphorylation takes place preferentially at tyrosines.
- PROTOPLAST:** A cell without cell wall but with intact cell membrane; gram-positive bacterium after removal of the cell wall.
- PSEUDOKNOT:** An RNA secondary structure that is minimally composed of two helical segments connected by single-stranded regions or loops.
- QUADRUPLEX:** A four-stranded box-like structure, with a central cavity, composed of successive stacking of two or more G-tetrads.
- RECOMBINANT DNA:** Any DNA molecule created by ligating pieces of DNA that normally are not contiguous.
- RECOMBINATION:** A genetic rearrangement occurring during sperm and egg cell formation.
- RENATURATION (OF DNA OR RNA):** The re-establishment of the DNA duplex or intrastrand hairpin structures in an RNA molecule after denaturation. (Of a protein); the conversion from an inactive into a biologically active conformation.
- REPLICON:** The regulatory unit of an origin and proteins necessary for initiation of replication (specific for this origin).
- REPRESSION:** The blocking of the synthesis of certain enzymes when their products are present; more generally, refers to inhibition of transcription (or translation) by binding of repressor protein to specific site on DNA (or mRNA).
- RESTRICTION ENZYME:** An enzyme that recognizes specific short sequences of (usually) unmethylated DNA and cleaves the respective DNA molecule (sometimes at target site, sometimes elsewhere (in trans), depending on type).
- RESTRICTION FRAGMENT:** A duplex DNA fragment obtained by cutting a larger fragment with either a single or two different restriction enzymes.
- RETROTRANSPOSON:** The major class of eukaryotic transposable elements, which are able to transpose into other genomic DNA sites *via* an RNA intermediate by use of retrotransposon-encoded reverse transcriptase.
- RETROVIRUS:** A virus containing a single-stranded RNA genome that propagates *via* conversion into double-stranded DNA by reverse transcription.

- REVERSE TRANSCRIPTASE:** RNA-dependent DNA polymerase. Originally detected in retroviruses. It is, however, also present in normal eukaryotic cells and even in *E. coli*.
- REVERSION (OF MUTATION):** A change in DNA that either reverses the original alteration (true reversion) or compensates for it (second site reversion in the same gene).
- RIBOSOMES:** Subcellular particles consisting of several RNA and numerous protein molecules. Involved in translating the genetic code in mRNA into the amino acid sequence of the corresponding protein.
- RIBOSWITCH:** A part of an mRNA molecule that can directly bind a small target molecule, where the binding of the target affects the activity of the RNA.
- RIBOZYME:** A naturally occurring folded RNA structure that cuts cognate RNA through an intramolecular *trans*-esterification reaction. Can also refer to any single-stranded catalytic RNA molecule.
- RNA EDITING:** A series of consecutive “cut and paste” reactions carried out by complex cell machinery; results in a change of sequence of RNA following transcription.
- siRNA:** Short interfering RNA; an intermediate in the RNAi process in which the long double-stranded RNA has been cut up into short (~21 nucleotides) double-stranded RNA. The siRNA stimulates the cellular machinery to cut up other single-stranded RNA having the same sequence as the siRNA.
- SANGER–COULSON SEQUENCING:** DNA sequencing technique based on transcription of single-stranded DNA by a polymerase in the presence of dideoxynucleotides. The same technique can also be used for sequencing of RNA.
- SATELLITE DNA:** The many tandem repeats (identical or related) of a short basic repeating unit.
- SDS (SODIUM DODECYLSULFATE):** A detergent.
- SDS GEL ELECTROPHORESIS:** Gel electrophoresis of proteins in polyacrylamide gels in the presence of SDS. Molecules of SDS associate with the protein molecules giving them all a similar electric charge density and thus allowing separation on the basis of differences in molecular weight.
- SELECTION:** The use of particular conditions to allow survival only of cells with a particular phenotype.
- SELEX:** A technique that allows the simultaneous screening of highly diverse pools of different RNA or DNA molecules in order to obtain a particular feature.
- SEQUENCING GEL:** A very thin (0.1–1 mm) high-resolution polyacrylamide gel.
- SHINE–DALGARNO SEQUENCE:** Part or all of the polypurine sequence AGGAGG located on bacterial mRNA just prior to an AUG initiation codon; is complementary to the sequence at the 3'-end of 16S rRNA; involved in binding of ribosome to mRNA.
- SHORT TANDEM REPEAT (STR):** A DNA sequence containing a variable number (typically =50) of tandemly repeated short (2–6 bp) sequences, such as (GATA)_n. Forensic STRs are usually tetranucleotide repeats, which show little PCR stutter.
- SHUTTLE VECTOR:** A vector which is able to replicate in different host organisms *e.g.* *E. coli*, COS cells.
- SIGMA FACTOR:** A subunit of bacterial RNA polymerase needed for initiation; is the major influence on selection of binding sites (promoters).
- SIGNAL HYPOTHESIS:** The process by which proteins synthesized in the cytoplasm are exported either out of the cell or into one of the cellular organelles. The signal peptide of the protein plays an important role in this process.
- SIGNAL PEPTIDE:** The region (usually N-terminal) of a protein that ensures its export out of the cell or its import into one of the cellular organelles (s. leader).

- SIGNAL TRANSDUCTION:** Molecular mechanism of transferring the information from the outside of a cell, a receptor, to the nucleus. The stimulus may be, *e.g.* a hormone or cytokine, the transferring molecules are second messengers, protein kinases, and phosphatases and finally transcription factors.
- SIMPLE STRS:** Short tandem repeat loci composed of uninterrupted runs of a single repeat type.
- SINGLE NUCLEOTIDE POLYMORPHISM (SNP):** A common DNA sequence variation among individuals of the same species.
- SITE-DIRECTED MUTAGENESIS:** Introduction in the test tube of a specific mutation(s) into a DNA molecule at a predetermined site.
- SOUTHERN BLOTTING:** A procedure for transferring denatured DNA from an agarose gel to a nitrocellulose filter where it can be hybridized with a complementary nucleic acid.
- SPLICEOSOME:** A complex of several RNAs and proteins responsible for removing the non-coding parts of RNA (introns) from unprocessed mRNA.
- SPLICING:** Describes the removal of introns and joining of exons in RNA; thus introns are spliced out, while exons are spliced together.
- STEM:** The base-paired segment of a hairpin.
- STOP CODON:** Same as termination codon.
- STRUCTURAL GENE:** Gene coding for any RNA or protein product other than a regulator.
- STUTTER:** See PCR Stutter.
- SUBCLONING:** The cloning of fragments of an already cloned DNA sequence.
- SUPERCOIL:** A closed circular double-stranded DNA molecule that is twisted on itself. Typically a conformation of a circular double-stranded nucleic acid in which strain derived from an excess or deficit of turns of the double-stranded helix is relieved by a counter-helical winding of the circular nucleic acid (imaged as in a skein of wool).
- TAC-PROMOTOR:** A chimeric bacterial promotor of high strength constructed from parts of the Trp and lac promoters of *E. coli*.
- TATA (HOGENESS) BOX:** A conserved A-T-rich heptamer found about 25 bp before the start-point of each eukaryotic RNA polymerase II transcription unit; involved in positioning the enzyme for correct initiation.
- TELOMERE:** A region of highly repetitive DNA at the end of a chromosome.
- TEMPLATE:** Portion of single-stranded DNA or RNA used to direct the synthesis of a complementary polynucleotide.
- TERMINATION CODON:** One of three triplet sequences, UAG (*amber*), UAA (*ochre*), or UGA (*opal*), that cause termination of protein synthesis; they are also called nonsense codons.
- TOLL-LIKE RECEPTOR:** In vertebrates, receptor molecules that are able to stimulate activation of the adaptive immune system, linking innate and acquired immune responses.
- TOPOISOMERASES:** Enzymes that act on the topology of DNA; needed to unravel DNA strands that are topologically linked or knotted; they catalyze and guide the unknotting of DNA.
- TRANS-ACTING:** Referring to mutations of, for example, a repressor gene, that act through a diffusable protein product and can therefore act at a distance not simply on the DNA molecule in which they occur.
- TRANSCRIPTION:** Usually the synthesis of RNA on a DNA template. Also used to describe the synthesis of DNA on an RNA template by reverse transcriptase, the copying of a (primed) single-stranded DNA by DNA polymerase and the copying of RNA by (viral) RNA polymerase.

- TRANSDUCTION:** The transfer of a bacterial gene from one bacterium to another by a phage; phage carrying host as well as its own genes is called transducing phage.
- TRANSFECTION:** The acquisition of native protein-free DNA of a phage by bacteria.
- TRANSFORMATION:** The acquisition by a cell of new genetic markers by incorporation of added DNA. In eukaryotic cells it also refers to conversion to a state of unrestrained growth in culture resembling or identical to the tumorigenic condition.
- TRANSITION:** A mutation in which a purine is replaced by another purine (*e.g.* G to A) or a pyrimidine by another pyrimidine (*e.g.* T to C).
- TRANSPOSABLE ELEMENT:** A heterogeneous class of genetic element that can insert into a new location within chromosomes.
- TRANSVERSION:** A mutation in which a purine is replaced by a pyrimidine or *vice versa*.
- TRIPLET:** A sequence of three nucleotides in DNA or RNA. Usually means the same as codon.
- TWO-DIMENSIONAL GEL ELECTROPHORESIS:** A technique in which a second electrophoretic separation is carried out perpendicular to the first. The two separations are based on different criteria (*e.g.* electric charge and molecular weight).
- UPSTREAM:** Sequences that proceed in the opposite direction from expression. For example, the bacterial promoter is upstream from the transcription unit, the initiation codon is upstream from the coding region.
- WATSON–CRICK RULES:** The base-pairing rules that underlie gene structure and expression. G pairs with C; A pairs with T (A pairs with U in RNA).
- WESTERN BLOTTING:** Transfer of proteins from a gel to a nitrocellulose filter on which they can subsequently be detected by immunological screening.
- WILD-TYPE:** The genotype or phenotype that is found in nature or in the standard laboratory stock for a given organism; the phenotype of a particular organism when first seen in nature.
- WOBBLE HYPOTHESIS:** The ability of a tRNA to recognize more than one codon by non-Watson–Crick (non-G-C, A-T) pairing with the third base of a codon.

