

Glossary of Terms

Biomarkers

A biomarker, or biological marker, is in general a substance used as an indicator of a biological state. It is a characteristic that is objectively measured and evaluated as an indicator of normal biological processes, pathogenic processes, or pharmacologic responses to a therapeutic intervention.

DNA (deoxyribonucleic acid)

A polymeric molecule consisting of deoxyribonucleotide building blocks that in a double-stranded, double helical form is the genetic material of most organisms.

Gene

Segments of DNA that carry separate information packets of the genome. This information is read when the two strands of DNA “unzip” and the series of bases representing a gene are copied into the related nucleic acid RNA.

Gene expression

Process by which information in genes coded in DNA is used to synthesize the functional gene product. It is used to provide information on the roughly 30,000 genes within the human genome. Life science researchers use gene expression profiling to study the differences in expression of genes in a normal versus a disease state. The challenge for scientists is to delineate the associated genes’ expression patterns and their relationship to disease.

Genotyping

The process of determining the genotype of an individual by the use of biological assays.

microRNA

microRNAs are small non-protein-coding single-stranded RNA molecules of 21-23 nucleotides in length that function as negative regulators of gene expression by targeting specific messenger RNAs. This either inhibits translation or promotes messenger RNA degradation. Cancer diagnosis, prognosis, and treatment are important potential clinical applications of microRNA profiling.

Microarray

A technology used to measure gene expression. By attaching a fluorescent dye, scientists determine which genes are active in a given cell. Although microarrays are the dominant technology used today, they have limited sensitivity, accuracy and dynamic range. Thus microarrays capture only 20-40% of the expressed genes. Consequently, scientists obtain only a partial view of the expression profile when utilizing microarrays due to the limited sensitivity. These overlooked genes may be important in a particular disease state. As a consequence of these limitations, the discovery of genes identified by microarray technology requires further validation using real-time PCR.

Personalized Medicine

Use of information about a person's genes, proteins, and environment to prevent, diagnose and treat disease. Sequencing of the human genome has brought about a new healthcare paradigm in which disease is understood at the molecular level, providing the potential for a patient to be diagnosed according to genetic information and treated with therapeutics designed to work on specific molecular targets. With personalized medicine, researchers can target patients' specific diseased genes compared to healthy genes and develop therapies to address specific diseased genes. The optimum result will be therapies less expensive to develop and more targeted and more likely to be effective and therefore become a successfully approved drug. Thus drug companies will not have as many failures. This has led to the need for accurate, highly sensitive, high-throughput gene expression data by researchers, clinicians and pharmaceutical companies.

Pharmacogenomics

The study of how genes affect the way individuals respond to drugs.

Real-Time PCR

A technology used to measure gene expression and genotyping. By using a heating and cooling process to separate copies of the DNA helix, real-time PCR represents a sensitive and accurate method to measure gene expression. PCR is an enzymatic process to increase the number of copies of DNA for easier detection. Real-time PCR chemistries allow for detection of the reaction in the early phase rather than the late phase of the reaction. The polymerase enzyme "reads" an intact DNA strand as a template and uses it to synthesize a new strand, which sets in motion a chain reaction in which the DNA template is exponentially amplified, generating millions or more copies of the DNA piece. Real-time PCR simultaneously amplifies and quantifies (as an absolute number of copies or relative amount) a targeted DNA molecule in real time after each amplification cycle.

RNA (ribonucleic acid)

Any of a group of nucleic acids, present in all living cells, that play an essential role in the synthesis of protein.