**MEDICAL GLOSSARY**

**Acanthosis nigricans**: skin disease characterised by grey-black warty patches, usually situated in the groin, elbows or knees.

**Acufène**: an inner buzzing noise in the ear of the patient with severe or medium hearing problems. Causes unknown up to now and therefore no effective treatment has been identified. Italian and foreign research has demonstrated that 20%-30% of the population are plagued by this disturbance for a long period and sometimes for their entire lives. Acufène often gets worse with changes in the weather, physical effort, intellectual stimulation or psychological discomfort or uneasiness.

**Amblyopia**: reduced visual acuity, associated with strabismus, vision impairments such as those caused by cataracts, or refractive disorders, even after they have been corrected with glasses, when there is no apparent disease of the eye. It is usually only present in one eye.

**Aphasia:** disturbance due to lesions in the brain centres in particular in the language centres of the cerebral hemisphere (on the left in the right hemisphere). Aphasic disturbances are different from other more simple forms of disturbance because language comes from a complex of symbol activities.

**Alleles:** variant forms of the same gene. Different alleles produce variations in inherited characteristics such as eye colour or blood type.

**Astigmatism**: occurs when the cornea/lens does not have the same degree of curvature in the horizontal and vertical planes, resulting in distorted images.

**Autosome dominant**: passed from one parent, who has the condition, to the child (1:2 chance at each pregnancy).

**Autosome recessive**: passed from both parents (carriers), who themselves have the condition, to the child (1:4 theoretical chance at each pregnancy).

**Karyotype:** the entire chromosome complement of an individual or a cell as it appears during the metaphase.

**Cataract:** crystalline lens opacity preventing a clear retinal image. The removal of the crystalline lens can become necessary when the decrease in vision becomes significant and optical correction is carried out using corrective lenses, contact lenses or intraocular lenses. It can be congenital or a consequence of trauma, disease or aging.

**Cell:** the smallest unit in an organism which can work in an independent way. All living beings are made up of one or more cells: they can be subdivided into unicellular and multicellular organisms.

**Chromosomes:** structure composed of one associated protein and a long DNA molecule that holds the hereditary information of an organism.

**Cellular cycle**: the events that accompany the division of mitotic cells.

**Cytogenetics**: the cytological approach to genetics, that consists mainly in the microscopic study of the chromosomes.

**Cytoplasm:** the cellular substance outside the nucleus in which the cell's organelles are suspended.

**Cloning:** production of identical copies (clones) of molecules, cells or organisms. 1) Isolation of one cell, and therefore of the cells deriving from it to form one cellular line of identical elements (cellular cloning). 2) Employment of the techniques of DNA recombination in order to insert a specific sequence of DNA in a suitable carrier in order to obtain the propagation of numerous copies through the clone amplification of the cells in the carrier in which it has been inserted (properly termed molecular cloning). The term is also used to indicate: 3) Generation of an

individual with identical genetic patrimony to a single parent in the case of organisms that reproduce themselves sexually. In this case the diploid nucleus is introduced into a somatic cell (in the G0 phase, see: cellular cycle) into a female gamete without its own nucleus.

**Cochlear:** part of the inner ear, in the shape of a scroll, situated in the thickness of the temporal bone.

**Coloboma:** absence or defect in one ocular structure, e.g. absence of the lower portion of the head of the optical nerve, the chorioid, the ciliary body, the iris, the crystalline lens and/or the eyelids. Caused from incomplete fusion of the foetal fissure during gestation. It can be associated to other anomalies, like microphthalmia.

**Congenital:** present at birth.

**Cornea**: the transparent front surface of the eye.

**Conserved sequence:** a base sequence in a DNA molecule (or an amino acid sequence in a protein) that has remained essentially unchanged throughout evolution.

**Decibel:** noise measurement unit. Abbreviated to dB (or to DBA). The BIAP (Bureau International d' Audiophonologie) calculates the average total loss on frequencies 500, 1000, 2000, 4000 Hz, the sum of the loss in dB of these frequencies is divided by four to calculate the average loss exactly. Based on quantitative criteria definitions are:
- Normal hearing (auditory threshold inferior to 20 dB).
- Slight deafness (auditory threshold between 20 and 40 dB).
- Medium deafness (threshold between 40 and 70 dB).
- Severe deafness (auditory threshold between 70 and 90 dB).
- Profound deafness 90 and 120 dB. Auditory loss total more than120 dB.

**Diabetes insipidus:** excessive loss of water through urine and increased thirst and drinking as a result of deficiency in the hormone vasopressin.

**Diabetes mellitus:** excessive drinking and urine output due to excess sugar in the blood and urine as a result of difficulty or inability to produce insulin in the pancreas gland.

**DNA:** the substance of heredity; a large molecule that carries the genetic information that cells need to replicate and to produce proteins. DNA is a double-stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.

**External ear**: comprised of the auricular pavilion, which we commonly call the ear, which helps us to establish from where a sound comes. In other words, the auditory canal which includes the thin membrane of the eardrum. When the sounds arrive at the membrane, they are transformed into vibrations that are transmitted to the ear.

**Middle ear**: the middle ear contains three of the smallest bones in the human body in barely one square centimetre: the hammer, anvil and stirrup. Their movements, which initiate from the eardrum, are amplified twenty times and transmit to the inner ear all the sophisticated wealth of sounds, from individual ones to those of an entire orchestra.

**Inner ear**: one small structure called the cochlea or scroll houses forty thousand cigliated cells (twenty thousand in each ear) which are able to divide tasks: some work with the strong sounds, others with the weak. The cigliated cells are responsible for transforming sounds anew from vibrations to electrical impulses. Then through the thin fibres of the acoustic nerve they arrive in the brain, where they determine nerve sensations.

**Electro-oculogram (EOG):** measures the potential difference between the cornea and the back of the eye. It reflects the activity of the retinal pigment epithelium (RPE).

**Electroretinogram (ERG):** records the electrical response of the retina to light. It reflects the activities of many parts of the retina including rods and cones.
Using special techniques the responses of the different photoreceptors, including the rods and cones, can be separated.

**Enzyme**: a protein which serves as a catalyst for chemical reactions in cells.

**Gene:** a unit of inheritance; a working subunit of DNA. Each of the body's 50,000 to 100,000 genes contains the code for a specific product, typically, a protein such as an enzyme.

**Gene mapping:** determining the relative positions of genes on a chromosome and the distance between them.

**Genome**: the totality of the genetic information that belongs to one cell or an organism; in particular the DNA that stores this information.

**Genotype**: the collection of genes forming the genome, discovered through genetic or molecular studies.

**Hypacusia**: slight decrease of auditory sensitivity.

**Transmissive hypacusia**: damage in the external ear to the parts relating to mechanical transmission of the sound. People affected by this type of loss hear all the lower, fainter sounds independent of the type of frequency. They feel like their ears are plugged and speak in a quiet voice because they hear it as unusually strongly.

**Neurosensorial hypacusia**: affects the inner ear so that it becomes incapable of transforming sound vibrations into the correct nerve impulses. People affected by this type of hypacusia feel unable to understand because they do not recognize the sounds.

**Hypacusia of mixed type**: found when the lesion affects the middle ear and the inner ear at the same time. Obviously with this type of loss the effects of the two losses previously listed are all present.

**Central Hypacusia**: the hearing centre is situated in the brain. At times, the sounds sent through the ear are not correctly interpreted by this centre. This is then known as central deafness.

**Metabolism**: The conversion of a chemical from one form to another.

**Mutation:** A change in the number, arrangement, or molecular sequence of a gene.

**Nucleotide:** A subunit of DNA or RNA, consisting of one chemical base plus a phosphate molecule and a sugar molecule.

**Nucleus:** The cell structure that houses the chromosomes.

**Homozygote:** an individual that has identical alleles at one or more loci and therefore produces gametes which are all identical.

**Otitis media with effusion (glue ear):** an accumulation of fluid in the middle ear cavity, which commonly occurs in children with colds or after ear infections. With glue ear the hearing loss is temporary, usually mild to moderate, can affect one or both ears and fluctuate. It usually resolves itself spontaneously but may need specific treatment.

**Ototoxicity**: toxic effects to the inner structures of the ear induced by foreign substances and/or medicines.

**Phenotype:** Observable characteristics of an organism produced by the organism's genotype interacting with the environment.

**Protein:** A large, complex molecule composed of amino acids. The sequence of the amino acids, and thus the function of the protein, is determined by the sequence of the base pairs in the gene that encodes it. Proteins are essential to the structure, function, and regulation of the body. Examples are hormones, enzymes, and antibodies.

**Recessive:** A gene that is phenotypically manifest in the homozygous state but is masked in the presence of a dominant allele.

**Recessive allele:** A gene that is expressed only when its counterpart allele on the matching chromosome is also recessive (not dominant). Autosome recessive disorders develop in persons who receive two copies of the mutant gene, one from each parent who is a carrier. (see dominant allele)

**RNA:** or ribonucleic acid. Nucleic acid containing ribose; involved in the synthesis of proteins, as part of the DNA, the RNA represents the material depositary of the genetic information in some viruses.

**Sensorineural hearing loss:** occurs in or beyond the inner ear, usually in the cochlea, more rarely in the auditory nerve. This type of deafness is permanent, ranges from mild to profound, and affects either one ear (unilateral) or both ears (bilateral) and may be progressive.

**Deafness:** the term "deafness" indicates the total loss or the serious impairment of hearing ability. "Presbycusis" deserves special mention as it is a condition linked to senescence and consists of perception deafness; it is a condition of progressive aging of the ear.

**Syndrome deafness**: deafness associated with pathologies involving other structures like the endocrine, ocular, renal and cardiac systems, etc.

**Non syndrome deafness**: isolated deafness, not associated to other pathologies.

**Progressive deafness**: the auditory loss increases over time.

**Congenital deafness**: present at birth. Not necessarily hereditary.

**Vestibular:** used to refer to either the organ of balance in the ear or the function of balance.

**Virus**: particle that consists of nucleic acid enclosed in a protein and able spread from one cell host to another. Many viruses provoke diseases.