Romanian Journal of Rhinology, Vol. 1, No. 3, July - September 2011

SYNDROMES AND EPONIMES

Syndromes and eponimes in Otolaryngology

Achondroplasia is a genetic autosomal dominant condition that results in abnormally short stature and is the most common cause of short stature with disproportionately short limbs. Although achondroplasia literally means „without cartilage formation," the defect in achondroplasia is not in forming cartilage but in converting it to bone, particularly in the long bones.The baby with achondroplasia has a relatively long, narrow torso (trunk) with short extremities (arms and legs) and a disproportionate shortening of the proximal segments of the limbs, with prominence of the forehead (frontal bossing), hypoplasia of the midface with cheekbones that lack prominence, and a low nasal bridge with narrow nasal passages, lordosis and frequent hypocusis .

Bowen's disease is medically the same as „squamous cell carcinoma in situ." The hallmark of Bowen's disease is a persistent, progressive, slightly raised, red, scaly or crusted plaque. Bowen's disease may occur anywhere on the skin surface (or on mucosal surfaces such as in the mouth). The differential diagnosis should be made with psoryazis.

Caldwell-Luc operation is a surgical procedure designed to treat chronic sinus disease located in the maxillary sinuses.

CHARGE association:

A constellation of congenital malformations. The name of the condition is an acronym of some of the most frequent features:

- **C** = **C**oloboma
- **H** = **H**eart malformation,
- **A** = Choanal **A**tresia
- **R** = **R**etardation of growth after birth, **R**etardation of development
- **G** = **G**enital hypoplasia
- **E** = **E**ar malformations and/or deafness

Fanconi anemia: genetic disease that affects all of the bone marrow elements, is associated with a great diversity of malformations as well as pigmentary changes of the skin, and predisposes to malignancy. Facial skeleton malformation's, skin pigment changes (cafe-au-lait spots on the skin, brownish pigmentation of the body so it has a suntanned look), short stature, vertebral and spinal malformations, mental retardetion, prolonged bleeding time, thrombocytopenia, decreasing number of megakariocytes.

Giraldes ostium – maxillary sinus accessory ostium

Grisel's syndrome: Non-traumatic atlantoaxial subluxation, may occur secondary to head and neck infection or following routine otolaryngological procedures (amigdalectomy, rhinosinusal pathology, nasopharyngeal infections).

Haller cells - are also known **as** infraorbital ethmoidal air cells **or** maxilloethmoidal cells. They are extramural ethmoidal air cells that extend into the inferomedial orbital floor and are present in 2 - 45% of patients.

Kiesselbach's area, also Kiesselbach's plexus, Kiesselbach's triangle, and Little's area, is a region in the anteroinferior part of the nasal septum, where four arteries anastomose to form a vascular plexus called Kiesselbach's plexus. Ninety percent of nose bleeds (epistaxis) occur in this area.

Langer's lines, sometimes called cleavage lines, are topological lines drawn on a map of the human body. They technically are defined by the direction in which the skin of a human cadaver will split when struck with a spike and have relevance to forensic science and the development of surgical techniques.

Mikulicz cell - round or oval macrophage with a small nucleus that is found in the nodules of rhinoscleroma and contains the causative bacterium (*Klebsiella rhinoscleromatis*).

Mott cells are abnormal plasma cells characterized by the presence of Mott bodies or Russell bodies, which are rough endoplasmic reticulum-derived vesicles containing immunoglobulins of the IgM class. It is found in rhinoscleroma.

Ogston – Luc surgery technique - surgical procedure for frontal sinus disease; a skin incision is made from the inner third of the edge of the orbit toward the root of the nose.

Ohngren's line is the oblique plane joining medial canthus of the eye with the angle of the mandible. It divides the maxilla into the infrastructure and superstructure.

Onodi cell - is defined as an ethmoidal air cell that lies posteriorly, and sometimes, superiorly to the sphenoidal sinus. As a result of its location the optic nerve, and less commonly, the internal carotid artery, are very closely related with.

Osler – Webber – Rendu Syndrome (**Hereditary hemorrhagic telangiectasia):** a genetic disease characterized by the presence of arteriovenous malforma-

tions (AVMs) which involve direct connections between arteries and veins without the usual intervening capillaries. Recurrent nosebleeds are common, particularly at night, beginning at about the age of 12, due to the rupture of telangiectases within the nose. Large AVMs can bleed in the gastrointestinal (GI) tract, brain, spine, lung, liver and other sites and create major, sometimes life-threatening, problems.

Paget's disease is a chronic condition of bone characterized by disorder of the normal bone remodeling process. Normal bone has a balance of forces that act to lay down new bone and take up old bone. The symptoms of Paget's disease depend on the bones affected and the severity of the disease. Enlarged bones can pinch adjacent nerves, causing tingling and numbness. Bowing of the legs can occur. Hip or knee involvement can lead to arthritis, limping, as well as pain and stiffness of the hip or knee. Headache, loss of vision, and hearing loss can occur when bones of the skull are affected.

Russell bodies are eosinophilic, large, homogenous immunoglobulin-containing inclusions usually found in a plasma cell undergoing excessive synthesis of immunoglobulin; the Russell body is characteristic of the distended endoplasmic reticulum. This is one cell variation found in rhinoscleroma.

Sluder's neuralgia, also known as Sluder's syndrome, sphenopalatine ganglion neuralgia. Pain is said to begin in the root of the nose and to spread to the orbit, causing extreme soreness of the eyeball, nose, upper teeth, zygoma, palate, and pharynx, and even the shoulder and arm. Rhinorrhea, lacrimation, sneezing, photophobia, and salivation often occur, but no trigger point is found. A unilateral neuralgia can be caused by a septoturbinate contact.

Tay-Sachs disease: A genetic metabolic disorder caused by deficiency of the enzyme hexosaminidase A. The child usually develops normally for the first few months, but head control is lost by 6 to 8 months of age; the infant cannot roll over or sit up, spasticity and rigidity develop, and excessive drooling and convulsions become evident. Blindness and head enlargement occur by the second year. The disease worsens as the central nervous system progressively deteriorates. After age 2, constant nursing care is needed. Till age 3 deafness is present. Death generally occurs by age 5, due usually to cachexia (wasting away) or aspiration pneumonia.

Treacher–Collins syndrome (TCS), also known as Treacher-Collins–Franceschetti syndrome, or mandibulofacial dysostosis is a rare autosomal dominant congenital disorder characterized by craniofacial deformities, such as absent cheekbones. The typical physical features include downward slanting eyes, micrognathia (a small lower jaw), conductive hearing loss, underdeveloped zygoma, drooping part of the lateral lower eyelids, and malformed or absent ears.

Waldeyer's tonsillar ring (or **pharyngeal lymphoid ring**) is an anatomical term describing the lymphoid tissue ring located in the pharynx and to the back of the oral cavity.