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SPECIAL REVIEW

Marathon of eponyms: 21 Urbach-Wiethe disease (Lipoid proteinosis)

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The use of eponyms has long been contentious, but many remain in common use, as discussed elsewhere (Editorial: Oral Diseases. 2009: 15; 185). The use of eponyms in diseases of the head and neck is found mainly in specialties dealing with medically compromised individuals (paediatric dentistry, special care dentistry, oral and maxillofacial medicine, oral and maxillofacial pathology, oral and maxillofacial radiology and oral and maxillofacial surgery) and particularly by hospital-centred practitioners. This series has selected some of the more recognized relevant eponymous conditions and presents them alphabetically. The information is based largely on data available from MEDLINE and a number of internet websites as noted below: the authors would welcome any corrections. This document summarizes data about Urbach-Wiethe disease.

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Also known as

Hyalinosis cutis et mucosae
Lipoid proteinosis
Rössle-Urbach-Wiethe syndrome

The condition

Urbach-Wiethe disease is a rare autosomal recessive genodermatosis, characterized by the deposition of hyaline material in the skin and mucosa, causing generalized thickening and scarring (hyalinosis cutis et mucosae), and in viscera including the brain. The condition is most often seen among people of Dutch or German descent.

The original designation, lipoid proteinosis, refers to the histological features of the deposits, which show similarities to both lipid and protein, although no abnormalities in lipid metabolism have ever been identified. Histological and ultrastructural examinations show deposits of an amorphous eosinophilic, PAS-positive, diastase-resistant, hyaline-like material, and disruption/reduplication of basement membranes at the dermal-epidermal junction and around blood vessels. The exact chemical composition of these deposits remains unclear but the disease is associated with loss of normal function of extracellular matrix protein 1(*ECM1*) caused by loss-of-function mutations in the ECM1 gene on chromosome 1q21. ECM1 proteins are normally expressed in skin, mucosae and other tissues, including heart, liver, small intestine, lungs, ovary, testes, prostate, pancreas, kidneys, skeletal muscle and endothelial cells. ECM1 is a glycoprotein involved in keratinocyte differentiation, in regulation of basement membrane integrity, interstitial collagen fibril macroassembly and growth factor binding. ECM1 has a role in the structural organization of the dermis (binding to perlecan, matrix metalloproteinase-9 and fibulin). ECM1 is also targeted as an autoantigen in lichen sclerosus, and is abnormally expressed in chronologically and light-aged skin and some malignancies.

Early clinical features in Urbach-Wiethe disease include recurrent vesicles, pustules, bullae and haemorrhagic crusts on the skin, mouth and throat. The face and distal extremities are the most common sites of lesions. The infiltrations produce waxiness and thickening of skin and mucosae of the mouth, pharynx, larynx and hypopharynx, causing hoarseness and inability to cry. The classic manifestation is the onset in infancy with a hoarse cry. Skin and mucous membrane changes later become apparent clinically, the most characteristic feature being waxy, yellow, bead-like bumps along the upper and lower edges of the eyelids. Pebbling of the lip mucosa creates a cobblestone appearance, which may also involve the tongue and gingiva. Infiltration of the tongue results in a woody firmness and impaired mobility. Ulceration of the lips and tongue may occur. Teeth, particularly the lateral incisors and premolars, may be hypoplastic. Infiltration of the salivary ducts may cause sialadenitis.

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Respiratory tract involvement may result in airway obstruction. In most cases intracranial calcification occurs, often resulting in damage to the brain amygdala, caudate nucleus and globus pallidus, which leads to difficulties in discriminating facial expressions and in making realistic judgements about the trustworthiness of other people. Over time it is associated with learning disability and grand mal seizures and rage attacks. Virtually any organ may be involved, but visceral involvement rarely leads to other clinically significant consequences, and lifespan is otherwise normal.

In severe cases, diffuse infiltration of the pharynx and larynx can cause respiratory distress, at times requiring tracheostomy. Treatment otherwise is usually limited and variable results have been obtained with dimethylsulphoxide, penicillamine and etretinate.

The main persons

Erich Urbach was born on 29 July 1893, in Prague, Czechoslovakia (now Czech Republic). He studied Medicine at the University of Vienna for 2 years, until the Great War broke out when he served in the Austrian army as a lieutenant and a member of the surgical group of Professor Anton Freiherr von Eiselsberg and was decorated for bravery. Urbach graduated as doctor of medicine in 1919 and worked in departments of the Wiener Allgemeines Krankenhaus under Jakob Pal, Wilhelm Schlesinger and Salomon Ehrmann. He worked at the Breslau skin clinic under Josef Jadassohn, and at the skin department of the Vienna Rothschild hospital under Hans Königstein. In 1929, he was habilitated (achieved the highest academic qualification) for skin and venereal diseases at the University of Vienna, becoming Dozent. He was subsequently an assistant physician at the II skin clinic with Wilhelm Kerl. From 1936 to 1938, he was chief physician at the department of dermatology and allergy at the Merchant's hospital, Vienna, but he migrated to USA in 1938 to become an associate in dermatology at the University of Pennsylvania. From 1939, he was Chief of the allergy department of the Jewish Hospital, Philadelphia. He died in 1946.

Robert Rössle was born on 19 August 1876, in Augsburg, Germany. He studied Medicine at the universities of Kiel and Strassburg, receiving his doctorate at the University of Munich in 1900. In 1904, he received the venia legendi for general pathology and pathological anatomy, and in 1909 became ausserordentlicher Professor at Munich. In 1911, he was appointed at the University of Jena as ordinarius Professor, and in 1922 moved to ordinarius Professor at Basel until 1929, when he moved to the Chair of pathology at Berlin. Rössle died in 1956.

Camillo Wiethe was an Austrian otologist during 1888–1949.

Associated persons

Robert Rössle
Erich Urbach
Camillo Wiethe

Source internet sites (accessed 21 February 2009) and further reading

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