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### SPECIAL REVIEW

# Marathon of Eponyms: | Albers-Schönberg disease (osteopetrosis)

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Abstract: The use of eponyms has long been contentious, but many remain in common use, as discussed elsewhere (Editorial: Oral Diseases. 2009 in press). The use of eponyms in diseases of the head and neck is mainly in specialities 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 recognised 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 paper summarises data about Albers-Schönberg disease.

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#### Also known as

Hench-Assman syndrome Osteopetrosis Marble-bone disease

#### The condition

Osteopetrosis is a rare syndrome of excessive calcification of bones causing marble-like appearance with increased radiological density of the skeleton, characterized mainly by multiple fractures. Osteopetrosis is caused by osteoclast defects.

Osteoclasts are derived in the bone marrow from monocyte/macrophage stem cell lines. Osteoclastogenesis requires signals from monocyte-macrophage colony-stimulating factor and also from the receptor activating NF-kappa B ligand (RANKL) acting through its cognate receptor, RANK. Osteoprotegerin, a soluble decoy receptor, can also bind RANKL and restricts its stimulation of osteoclastogenesis. In mouse models at least, disruption of these signaling pathways leads to the osteopetrotic phenotype.

Osteoclasts act to cause bone resorption by first attaching to bone matrix via integrin receptors; then forming a sealing zone within which acids are released. Acidification solubilizes the bone mineral, and proteases, especially cathepsin K, catalyze the degradation of bone matrix proteins.

Onset of clinical features is variable. Adult onset, infantile and intermediate forms are recognized, as well as other rare forms (e.g., lethal, transient, post-infectious and acquired). The autosomal dominant form is clinically innocuous, whereas the autosomal recessive type is lethal in infancy. A distinct form due to carbonic anhydrase isoenzyme II deficiency occurs in association with renal tubular acidosis and cerebral calcification.

Complications may include osteomyelitis of the jaws, especially the mandible (affected in 10%), anaemia, hepatomegaly and cranial nerve compression (visual disorders, hearing disorders, facial paralysis) The osteomyelitis is often precipitated by dental extractions, which are technically challenging in the dense bone.

Investigations include radiography and blood tests (low blood calcium and often elevated serum phosphatase).

Management with vitamin D (calcitriol) may produce modest clinical improvement, not sustained after therapy is discontinued. Gamma interferon has produced long-term benefits by improving leukocyte function, decreasing the incidence of infections, as well as increasing haemoglobin, platelet counts and survival rates. Combination therapy with calcitriol is superior to calcitriol alone.

Erythropoietin can help correct anaemia. Corticosteroids have been used to treat anaemia and stimulate bone resorption.

#### **Background to eponym**

Heinrich Ernst Albers-Schönberg possibly described osteopetrosis in 1898, but his first published report

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was in 1904. The condition was also recognized by Assmann and Hench. Herbert Assmann, born in the year 1882, Danzig, became professor and director of the medical clinic at the University of Königsberg, working on röntgen diagnostics, tuberculosis and joint diseases. Philip Showalter Hench, born in the year 1896, became one of the leaders in American rheumatology. Professor of Medicine at the Mayo Clinic, he received the Nobel Prize for Physiology or Medicine in 1950 'for discoveries relating to the hormones of the adrenal cortex, their structure and biological effects,'with Edwin Calvin Kendall and Tadeus Reichstein.

#### The main person

Heinrich Ernst Albers-Schönberg, originally a surgeon and later Professor of Radiology, was born in Hamburg, Germany on 21 January, 1865 and died on 6 June, 1921, in Hamburg. Albers-Schönberg attended the Gelehrtenschule of the Hamburger Johanneum. He studied medicine, with breaks for military service, in Tübingen and Leipzig where he qualified in 1891. Later that year, under Heinrich Curschmann, he obtained his doctorate on Einige mit Kochschen Tuberkulin behandelte Fälle. He subsequently worked in Professor Max Saenger's women's clinic in Leipzig and in 1892, became assistant physician at the newly established Allgemeines Krankenhaus Hamburg-Eppendorf, where he remained until 1894. In recognition of his self-sacrificing activity during the cholera epidemic, he was given a grant to visit Berlin and Vienna. Following a year as assistant physician at Paul Zweifel's clinic in Leipzig, he settled in Hamburg as a general practitioner in 1895. In 1896, he married the daughter of Senator Dr. Schroeder in Hamburg and his son Ernst was born in 1897.

Albers-Schönberg had immediately recognised the importance of Røntgen's discovery of X-rays and, in 1897, in collaboration with Georg Deycke, he gave up his medical general practice to establish a private radiographic institution, becoming the first specialist and founding the journal Fortschritte auf dem Gebiete der Röntgenstrahlen. In 1903, he was appointed radiologist to the Hamburg Hospital and 2 years later, became the head of the department. The first German Röntgen Congress was held in Berlin from April 30 until

3 May 1905, and the Deutsche Röntgen-Gesellschaft (German Röntgen Society) was founded, with Albers-Schönberg among the founding members. Albers-Schonberg (1904) received the Grand Prize at the world fair in St. Louis and, in 1907, the Prussian Kultursministerium honoured him with the title of Professor. In 1915, he moved to a similar post at Allgemeines Krankenhaus St. Georg, Hamburg. During World War I, Albers-Schönberg was consultant to the Ninth Army Corps and he subsequently received a Red Cross medal. Academic awards followed from the Universities of Würzburg, Heidelberg and Breslau. His career peaked in 1919 when the University of Hamburg bestowed a special honour upon him by electing him as Ordentlicher – full professor, the first full professorship of its kind.

However, his practices in using X-ray equipment were somewhat lackadaisical and he developed radiationinduced neoplasia on his hands in 1908 and his right middle finger and left arm had to be amputated. However, he developed tumours in his thorax and shoulder and died of pneumonia and cardiac failure on 4 June, 1921.

#### **Associated persons**

Heinrich Ernst Albers-Schönberg Herbert Assmann Philip Showalter Hench

## Source Internet sites (accessed 21 February 2009) and further reading

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