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SPECIAL REVIEW

Marathon of eponyms: 5 Ehlers–Danlos syndrome

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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 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 Ehlers–Danlos syndrome.

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Also known as

Chernogubow syndrome
Danlos syndrome
Meekeren–Ehlers–Danlos syndrome
Sack syndrome
Sack–Barabas syndrome
Van Meekeren syndrome I

The condition

Ehlers–Danlos syndrome (EDS) is a group of rare disorders of collagen formation. It is believed to affect one in 5000 people. Gene mutations in EDS affect fibrous proteins (genes COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 and TNXB) or enzymes (ADAM-TS2, PLOD1).

The most common forms of EDS are inherited as autosomal dominant traits, while others are recessive. Currently, according to the Villefranche nosology, the main types are the Hypermobility type (identical with the familial joint hypermobility syndrome); the Classical type (skin manifestations are important) and the Vascular type (may result in fatal bleeding). However, ten subtypes have previously been described; I (gravis) Classical type; II (mitis) Classical type; III (benign hypermobility) Hypermobility type; IV Vascular type; V (sex-linked); VI (ocular or hydroxylysine deficient); VII (multiple congenital dislocation or arthrochalasis multiplex congenital); VIII (periodontal); IX (skeletal and urinary tract dysplasia) and X (fibronectin deficiency).

Different types of EDS show a wide variation in the clinical features, but EDS is characterized by loose-jointedness, hyperextensible skin and propensity to bruising. Hypermobility of the joints is the best known manifestation and may be extreme (''India-rubber'' man). Recurrent, semi-spontaneous joint dislocations may result. Hypermobility of joints is severe in types I, VI and VII and, in all of these, subluxation and dislocation of the temporomandibular joint are common.

The skin is typically soft in EDS, abnormally extensible, feels fine and thin, may appear lax and wounds tend to gape or to split after slight trauma. Healing is slow and may leave fragile stretched scars with a tissue-paper texture. Weakness of the connective tissue component may cause lesions in the gastrointestinal tract, urinary tract and respiratory system.

Easy (spontaneous) bruising, though of vascular origin, may mimic purpura caused by haematological diseases, but unlike in these, there is a combination of subcutaneous, submucosal and deep bleeding. Although platelet defects have occasionally been reported, haemostatic function is usually normal, except in type IV (the vascular type) in which spontaneous rupture of major arteries can cause fatal haemorrhage and internal bleeding, and where mild haemophilia may be associated.

Mitral valve prolapse (floppy valve syndrome) is seen in many types of EDS, particularly type III, and may confer susceptibility to infective endocarditis or lead to rapid development of mitral insufficiency and heart failure. Cardiac conduction defects may be present.

In type VIII EDS and possibly type IV, there is an early onset of periodontal disease, with early tooth loss.

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In type III EDS, local anaesthetics may be less effective, though there is no satisfactory explanation.

The teeth in EDS may be small with short or abnormally shaped roots, and many pulp stones. The deciduous dentition shows abnormal morphology of the molars, obliteration of the tooth pulp and severe enamel attrition. The permanent dentition shows agenesis and microdontia of several teeth. Tooth discolouration, dysplastic roots and tooth pulp obliteration are present in a restricted number of permanent teeth.

Background to eponym

Job Janszoon van Meekeren, a Dutch surgeon, presented the Spanish sailor George Albes – infamous for being able to stretch the skin on his chest out to arm's length – to the Academy of Leiden in 1657. Alexandr Nicolaevich Chernogubow in 1892 presented two patients at the Moscow Dermatological and Venereologic Society and, in Russia, the disorder carries his eponym.

In 1899, Edvard Ehlers presented a patient at the Paris Society of Syphilology and Dermatology. François Henri Hallopeau and Macé de Lépinay presented another patient to that Society. In 1908, Henri-Alexandre Danlos re-presented the same patient drawing attention to extensibility and fragility of the patient's skin and used the term 'cutis laxa'. In 1936, Frederick Parkes-Weber suggested that the disturbance should be named Ehlers–Danlos syndrome. Andras P. Barabas, a vascular surgeon in Suffolk, England, born in Hungary and trained at the medical school in Budapest presented his MD Thesis on the Heterogeneity of the Ehlers–Danlos syndrome, in 1967.

The main persons

Edvard Lauritz Ehlers was born in Denmark in 1863 and grew up in Copenhagen, where his father was Mayor. He studied medicine, qualifying in 1891 and moved to Berlin, Breslau, Vienna and then Paris. In 1906, he was appointed Chief of the dermatology department at the Frederiks Hospital, Copenhagen, and from 1911 was director of the special service at the Copenhagen Municipal Hospital (Kommunehospitalet). He died in 1937.

Henri-Alexandre Danlos, born on 26 March 1844 in Paris, started his education in business, but changed to study Medicine in Paris, and qualified with distinction in 1869 and gained his doctorate in 1874. Danlos initially undertook research in the laboratory of Charles-Adolphe Wurtz (1817–1884) but, in 1881, became Médecin des Hôpitaux and trained with Edmé Félix Alfred Vulpian. In 1885, Danlos became Chef de Service at the Hôpital Tenon for 5 years, followed by 5 years in public service, and then in 1895, was appointed in dermatology at the Hôpital Saint Louis in Paris. He was the first to describe the treatment of lupus erythematosus with radium in 1901. He died on 12 September 1932, in Chatoux.

Associated persons

Andras P. Barabas
Alexandr Nicolaevich Chernogubow
Henri-Alexandre Danlos
Edvard Lauritz Ehlers
Job Janszoon van Meekeren
Georg Sack

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

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