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SPECIAL REVIEW

Marathon of eponyms: 20 Treacher Collins 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 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 Treacher Collins syndrome.

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Also known as

Berry syndrome
Berry-Treacher Collins syndrome
Collins-Franceschetti Syndrome
Franceschetti syndrome I
Franceschetti-Klein syndrome
Franceschetti-Zwahlen syndrome
Franceschetti-Zwahlen-Klein syndrome
Mandibulofacial dysostosis
Thomson complex
Treacher Collins syndrome
Treacher Collins-Franceschetti syndrome

The condition

Treacher Collins syndrome is a rare disorder of craniofacial development, usually evident at birth. Most prevalent in Caucasians and found in 1 in 10 000 births, it is occasionally seen in other ethnic groups. This condition is a result of a defect of the first arch during development.

Treacher Collins syndrome (TCS) is an autosomal dominant condition which results from the loss-of-function mutations in the gene TCOF1 (which encodes a nucleolar protein involved in ribosomal DNA gene transcription) on chromosome 5q32-q33.1. TCOF1 encodes Treacle, a nucleolar phosphoprotein which plays a key role in preribosomal processing and ribosomal biogenesis. In mice, haploinsufficiency of TCOF1 results in a depletion of neural crest cell precursors through high levels of apoptosis in the neuroepithelium, which results in a reduced number of neural crest cells migrating into the developing craniofacial complex. Neural crest cells are a migratory stem and progenitor cell population that generates most of the tissues of the head, including much of the bone, cartilage and connective tissue. It has been hypothesized that mutations in TCOF1 disrupt ribosome biogenesis to a degree that is insufficient to meet the proliferative needs of the neuroepithelium and neural crest cells. This causes nucleolar stress activation of the p53-dependent apoptotic pathway, which induces neuroepithelial cell death. Haploinsufficiency of TCOF1 may perturb mature ribosome biogenesis, resulting in stabilization of p53 and the cyclin G1-mediated cell-cycle arrest that underpins the specificity of neuroepithelial apoptosis and neural crest cell hypoplasia characteristic of TCS.

Facial characteristics of TCS consist of fish-like facies with antimongoloid obliquity of palpebral fissures, notching (coloboma) of lower eyelids, flattening or absence of malar bones, small mandible with receding chin and considerable overbite, high arched palate, macrostomia, absence or malformations of or low-set small ears and prolongation of the hairline on the cheek. There is severe underlying hypoplasia of the facial skeleton including the orbits. The zygomatic bones may be entirely absent. People with TCS usually have normal intelligence. Difficulty in sucking and swallowing, and excessive mucus in the pharynx leads to cyanotic episodes.

In severe cases, the micrognathia may displace the tongue of an affected neonate (newborn) sufficiently to cause obstruction of the oropharynx. The neonate will

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asphyxiate unless a proper airway is established. Death is likely in the first month of life, but survivors of infancy can have a normal life span. The epiglottis can be surgically removed to help alleviate airway obstruction. People with TCS may also need a feeding tube if they cannot swallow. Multiple reconstructive surgeries on the face can improve appearance: bone-anchored hearing aids, and cleft palate surgery may also be indicated.

Background to eponym

There is confusion as to the correct eponymic term for this condition. Treacher Collins syndrome is the term commonly used in Britain and USA, while Franceschetti-Klein syndrome is used in continental Europe.

George Andreas Berry in 1889 first described an abortive form with colobomata of the lower eyelids. In 1900, Treacher Collins presented two similar patients at a meeting of the ophthalmological society, London, and published an account of their features. Franceschetti and Zwahlen in 1944 and Franceschetti and Klein in 1949 published extensive reviews of the condition in which they expanded the phenotype, employing the designation 'mandibulofacial dysostosis'.

The main persons

Adolphe Franceschetti was born on 11 October 1896, in Zurich, Switzerland. He graduated in Medicine from the University of Zurich, and from 1921, he worked in the ophthalmological clinic, but then moved to Basel to work with Richard Max Brückner. Franceschetti obtained his medical doctorate and was habilitated (achieved the highest academic qualification) as a Privatdozent in Basel in 1931, and from 1933 to 1966 he was Professor and Director of the university eye clinic in Geneva. In 1948, Franceschetti created a department of human genetics at his clinic. This was headed by David Klein and became the origin of the first institute of human genetics in Switzerland. Franceschetti published more than 500 articles, and his name is attached to some 10 syndromes. He died on 3 March 1968, in Geneva.

David Klein was born in 1908, in Falkau, Austria-Hungary, and died in 1993.

He was educated in Freiburg and then Basel, graduating from the University of Basel in 1934. He worked at the Rheinau Psychiatric Clinic, Zurich, and moved to Geneva as scientific assistant to Franceschetti. He was appointed as full professor in 1970 and retired in 1978.

Edward Treacher Collins was born on 28 May 1862, in London, UK. He studied Medicine at the Middlesex Hospital, London, graduating in 1883. In 1884, he moved to the Royal London Ophthalmic Hospital, Moorfields, where he spent the next 48 years. In 1886, he was appointed pathologist and curator of the hospital's museum. He remained in this position until 1895 when he was appointed surgeon to the hospital. He also lectured at the Charing Cross Hospital Medical School as well as at the Royal College of Surgeons in 1894 and 1899.

He was a leading figure in the organization of the British Ophthalmological Society as well as in the international council of ophthalmology and was elected president in 1927. He died on 13 December 1932, in London.

Associated persons

George Andreas Berry
Adolphe Franceschetti
David Klein
Allen Thomson
Edward Treacher Collins
Pierre Zwahlen

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

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