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SPECIAL REVIEW

Marathon of eponyms: 18 Robin sequence

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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 Robin sequence.

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Also known as

Pierre Robin syndrome
Robin anomalad
Robin sequence

The condition

'Pierre Robin' syndrome is one of the most readily recognized eponyms, yet there is widespread confusion defining it – which is hardly surprising as it is not a defined syndrome and has no prognostic significance. The original description by Pierre Robin consisted of micrognathia, glossoptosis and compromised airway. Features such as cleft palate or feeding problems are additional to the sequence. This condition is now described as 'Pierre Robin *sequence*' (PRS) as these problems may occur in isolation or as a variable component of many different genetic syndromes, and has inheritance as that of the syndrome with which it is associated.

Neonates with PRS suffer from two main problems – airway obstruction and feeding difficulties – but the severity of these problems varies greatly. Upper airway obstruction in PRS can be treated non-surgically or surgically. Often, respiratory obstruction is relieved by nursing the infant in the prone position or alternatively, a tongue lip adhesion can be performed. If the palatal cleft is very wide, a removable feeding plate assists suckling. In many cases, if the tongue can be kept out of the cleft, the platal shelves spontaneously come together making subsequent surgical closure easier. If the infant can be managed conservatively, mandibular growth rapidly proceeds and alleviates the repiratory and feeding problems.

The syndrome most commonly associated with PRS is Stickler syndrome. Other disorders causing PRS may include velocardiofacial (Shprintzen) syndrome, Treacher Collins syndrome, trisomy 11q syndrome, trisomy 18 syndrome, deletion 4q syndrome, rheumatoid arthropathy, hypochondroplasia, Möbius syndrome, foetal alcohol syndrome and CHARGE association (a chromosome 8 mutation on the CHD7 gene manifesting with Coloboma, Heart defects, Atresia of choanae, Retarded growth, Genital defects and Ear anomalies).

The aetiology of PRS is generally unknown but some factors are suggestive of a genetic basis. Candidate genes for PRS include glutamic acid decarboxylase gene GAD67 on chromosome 2q31, PVRL1 (poliovirus receptor-related 1 gene, which is a member of the immunoglobulin super family that acts in the initiation and maintenance of epithelial adherens junctions) on 11q23-q24 and SOX9 gene (a transcription factor required in chondrogenesis) on 17q24.3-q25.1.

Stickler syndrome is an autosomal dominant, genetic connective tissue disorder characterized by ophthalmological, orofacial, auditory and skeletal anomalies. Ophthalmological features include pathognomonic abnormalities of vitreous gel architecture, usually associated with congenital and non-progressive myopia and a high risk of retinal detachment. Orofacial

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features include a flat midface with depressed nasal bridge, short nose with anteverted nares and micrognathia. Midline clefting, if present, ranges in severity from a soft palate cleft to PRS. Joint hypermobility leads to osteoarthritis, typically in the third or fourth decade. Sensorineural deafness with high tone loss is typically asymptomatic or mild. Stickler syndrome is typically associated with mutations in the COL2A1 (collagen type II) gene. Mutations in COL111A2 can give rise to a syndrome with the systemic features of Stickler syndrome, but no ophthalmological abnormality.

Background to the eponym

The condition was first described by St Hilaire in 1822 and then by Fairbairn in 1846. Lannelongue and Menard in 1891 reported two patients with micrognathia, cleft palate and retroglossoptosis. In 1910 Shukowsky described the condition. In 1923, Pierre Robin published the case of an infant with the complete syndrome. Until 1974, this triad was known as Pierre Robin syndrome.

The main person

Pierre Robin was born in France in 1867. He was a professor at the French School of Stomatology, and from 1914, with Nogue, he was editor-in-chief of the Revue de Stomatologie. His main interest was glossoptosis, on which he published his main paper in 1923 and wrote more than 20 articles and a monograph before he died in 1950.

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

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