The Journal of Laryngology & Otology (2010), ***124***, *1129–1131*.

doi:10.1017/S0022215109992623

Clinical Record

Case report of Zenker's diverticulum in identical twins: further evidence for genetic predisposition

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Abstract

Objective: We report identical twins with Zenker's diverticulum.

Methods: Case report and literature review.

Conclusions: Geographical and racial variation in occurrence, and rare familial cases, suggest that inherited factors play a role in the pathogenesis of Zenker's diverticulum. The identical twins reported here provide further evidence supporting a genetic predisposition.

Key words: Zenker's Diverticulum; Pharyngeal Pouch; Aetiology; Identical Twin

Introduction

Zenker's diverticulum is the most common type of diverticulum of the upper gastrointestinal tract. Typical symptoms include dysphagia, regurgitation of undigested foods, halitosis and weight loss. Zenker's diverticulum has an estimated annual incidence of two per 100 000 in the UK;[1] however, there seems to be geographical variation in its global occurrence, as the condition is more prevalent in Northern Europe.

Several theories on the pathogenesis of Zenker's diverticulum have been proposed, including abnormal function of the cricopharyngeal muscle, and anatomical predisposition due to weakness of the posterior hypopharyngeal wall in Killian's triangle. It is generally accepted that age plays a role, probably causing loss of tissue elasticity and decreased muscle tone.[2]

Very rare familial cases of Zenker's diverticulum have been reported. Namely, only two publications exist on the possible inheritance of Zenker's diverticulum, of which one is a case report.[3,4] Approximately 2 per cent of patients have a positive family history.[2,3] The mode of inheritance in the rare familial cases seems to be autosomal dominant.

Here, we report a case of identical twins with Zenker's diverticulum.

Case report

Both individuals were treated at the Helsinki University Central Hospital. The apparently identical twins were born in 1941 and live in Southern Finland. The clinical picture of these twins is strikingly similar.

Twin A was referred to the department of otorhinolaryngology, Helsinki University Central Hospital, in January 2009 due to Zenker's diverticulum diagnosed with barium swallow study (Figure 1). Symptoms had commenced in 2007 (at 66 years of age), including globus sensation, dysphagia and regurgitation of undigested foods, and had worsened over time. A gastroscopy had been performed in November 2008, but the only finding had been a diaphragmatic hernia and lower oesophageal sphincter dysfunction. The patient is obese and smoked heavily. Previous medical history included high blood pressure, episodes of atrial fibrillation, acute myocardial infarction, coronary artery bypass surgery, erysipelas of the lower extremity and cataract extraction. After receiving a diagnosis of Zenker's diverticulum, twin A informed us that his twin brother had undergone surgery for the same condition. Twin A was successfully treated with stapler-assisted diverticulostomy in August 2009.

Twin B had been diagnosed with Zenker's diverticulum in 1988, based on barium swallow study. This patient's symptoms had began in 1986 (at 45 years of age), and had included dysphagia and regurgitation of undigested foods, worsening over time. In 1989, this patient had been treated with diverticulectomy via an open surgical approach at the department of surgery, Helsinki University Central Hospital. Like his brother, twin B is obese and a heavy smoker. He also shared with his twin brother a previous medical history of high blood pressure, episodes of atrial fibrillation, acute myocardial infarction, coronary artery bypass surgery, diaphragmatic hernia and cataract extraction. Unlike his brother, who had erysipelas of the lower limb, twin B had a history of auricular erysipelas. His past medical history also included chronic obstructive pulmonary disease, gastro-esophageal reflux and surgery for appendicitis.

Both twins completed a detailed questionnaire, including questions on: physical health; family history; the number of first, second and third degree relatives; and any relatives with Zenker's diverticulum or swallowing problems. To our knowledge, there were no other relatives affected by Zenker's diverticulum (Figure 2). The twins gave written, informed consent and permission to publish their medical history with radiographs.

Discussion

The clinical pictures of the two described, apparently identical twins are strikingly similar. In addition to Zenker's

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Accepted for publication: 18 October 2009. First published online 11 January 2010.

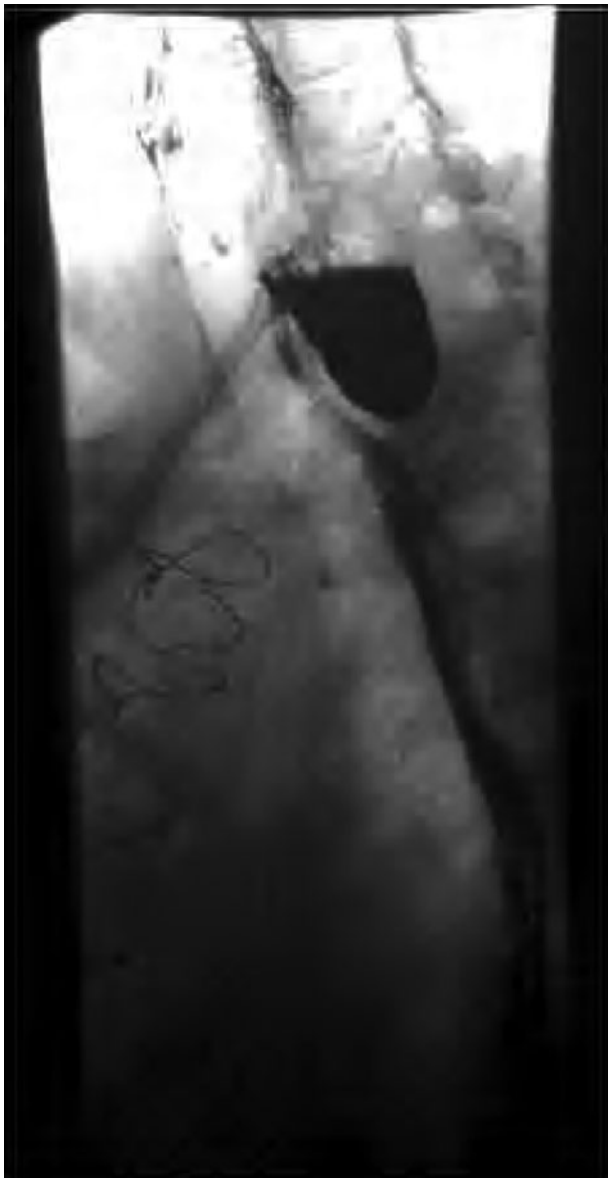

FIG. 1
Barium swallow study for twin A.

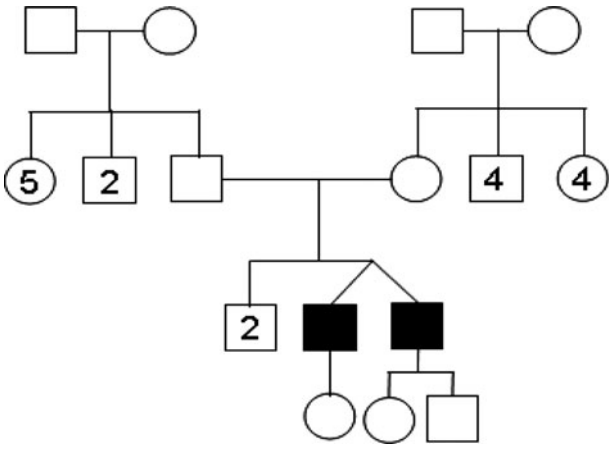

FIG. 2
The twins' family pedigree. Square = male; circle = female; filled symbol = affected; *n* = number of siblings.

diverticulum, they share numerous medical conditions: obesity, heavy smoking, high blood pressure, episodes of atrial fibrillation, acute myocardial infarction, coronary artery bypass surgery, diaphragmatic hernia, erysipelas and cataract extraction. Twin studies have demonstrated that genetic factors play a role in obesity,[5] cigarette addiction[6] and cardiovascular conditions.[7] In addition, cataract[8] and (congenital) diaphragmatic hernia can be genetically determined.[9] Whether the reported twins developed these conditions due mainly to environmental or to genetic factors is purely speculative, but it is likely that at least some kind of genetic predisposition exists for all these conditions.

- **The aetiology of Zenker's diverticulum is probably multifactorial in most patients**
- **Geographical and racial variation in occurrence, and rare familial cases, suggest that inherited factors play a role in pathogenesis**
- **Two identical twins with Zenker's diverticulum are reported, providing further evidence for a genetic predisposition in this condition**

The aetiology of Zenker's diverticulum is likely to be multifactorial in the majority of patients. Geographical and racial variations in incidence, and the rare occurrence of familial cases, suggest that inherited factors play a role in pathogenesis. The identical twins described in this report provide further evidence supporting a genetic predisposition for Zenker's diverticulum. The relative lack of reports describing familial cases indicates that this proposed genetic predisposition is generally not significant; however, in specific patients information on affected relatives may aid early diagnosis. As molecular genetics research expands, genotypes more prone to Zenker's diverticulum may be identified, providing new information on the pathogenesis of this condition.

Acknowledgements

Dr Klockars undertakes ongoing research on the genetics of otorhinolaryngological diseases with the financial support of Finska Läkaresällskapet and Wilhelm och Else Stockmanns Stiftelse.

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https://doi.org/10.1017/S0022215109992623 Published online by Cambridge University Press

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Dr T Klockars takes responsibility for the integrity of the content of the paper.
Competing interests: None declared