INTRODUCTION

Anterior laryngeal webs (ALWs) are uncommon abnormalities consisting of membranous tissues on the supraglottic, glottis, and/or subglottic at birth. These webs answer for about 5% of laryngeal malformations; depending on how extensive they are, airway obstruction may ensue, resulting in symptoms such as crying, stridor, dysphonia, and respiratory dysfunction. Individuals with this condition often present other concomitant anomalies, such as congenital heart defects, and palatine anomalies, which often are part of known genetic syndromes.

This paper presents a case report of a patient with an ALW and the 22q11 deletion syndrome (SD22q11), also known as the velocardiofacial syndrome or DiGeorge syndrome (OMIM 192430).5

CASE REPORT

A male Caucasian patient aged 12 years and 2 months was first admitted to hospital for surgery to correct a fossa ovalis type interatrial communication. The patient was the first child of young, healthy, and non-consanguineous parents. The family history was negative for congenital defects or genetic diseases. Pregnancy coursed uneventfully. The patient was delivered vaginally, at term, cephalic presentation, weighing 3,430 gr. (P50-98), height 53 cm (P2-50), with a 35 cm cephalic perimeter (P50-98). The baby was cyanotic and did not cry during birth. Oxygen therapy was required, and the patient remained in hospital for 15 days after birth. At the age of 3 months, a laser laryngeal procedure was done to remove a subglottic membrane; at this point the patient had episodes of hypocalcemia that required treatment with calcium gluconate.

The SD22q11 is a relatively common genetic disease; it is caused by a deficiency in region 11 of chromosome 22 (22q11.2 deletion) and occurs in about 1 in 10,000 births. The SD22q11 is typically present at birth,1 but in some cases it may be latent, as in this case, and may become apparent only after birth.2

COMMENTS

As has been reported in the literature,1,4 these findings suggest that patients with ALWs and other congenital malformations such as heart defects - should always be tested for the SD22q11 deletion syndrome. This has important implications for the management of patients and genetic counseling of the family.

REFERENCES

1. Stoler JM, Ladoulis M, Holmes LB. Anterior laryngeal webs and 22q11 deletions. Am J Med Genet. 1998;79:152.
2. McLellinney DB, Jacobs I, McDonald-McGinn DM, Zackai EH, Goldmuntz E. Chromosomal and cardiovascular anomalies associated with congenital laryngeal web. Int J Pediatr Otorhinolaryngol. 2002;66:237-8.
3. Miyamoto RC, Cotton RT, Rope AF, Hopkin RJ, Cohen AP, Shott SR, et al. Association of anterior laryngeal webs with velocardiofacial syndrome (chromosome 22q11.2 deletion). Otolaryngol Head Neck Surg. 2004;130:415-7.
4. Fokstuen S, Bottani A, Medeiros FPV, Antonarakis SE, Stoll C, Schinzel A. Laryngeal atresia type III (glotic web) with 22q11.2 microdeletion: report of three patients. Am J Med Genet. 1997;70:130-3.
5. Ordine Mendelian Inheritance in Man, OMIM (TM) [homepage on the Internet]. Baltimore e Bethesda: BeMcKusick-Nathans Institute for Genetic Medicine, Johns Hopkins University and National Center for Biotechnology Information, National Library of Medicine[cited 2010 Aug 10]. Available from: http://www.ncbi.nlm.nih.gov/omim/.