Anterior laryngeal membrane and 22q11 deletion syndrome

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INTRODUCTION

Anterior laryngeal webs (ALWs) are uncommon abnormalities consisting of membranous tissues on the supraglottic larynx, and can be found at birth.¹⁻² These webs answer for about 5% of laryngeal malformations,³ depending on how extensively they are, airway obstruction may ensue, resulting in symptoms such as cough, stridor, dysphonia, and respiratory dysfunction.³ 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.³⁻⁴

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 188400/192430).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 3.130 gr (P50), measuring 50 cm (P25-50), with a 35 mm 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 neurophysiologist, behavioral, and speech development was compromised as the patient grew, requiring drug therapy - haloperidol and biperiden. The physical examination at age 12 years and 2 months was as follows: weight - 47 kg (P75-90), height - 147 cm (P50), cephalic perimeter - 55 cm (P2-50), elongated face, narrow palpebral fissures, bilateral epicanthic folds, hypoplastic nasal alae, dental malocclusion, prognathism, overfolded helices (horizontal and vertical rami), punctis carenati, protruding navel, right cryptorchidism, and protruding navel. The child did not present dyspnea. At this point the decision was in favor of watchful waiting. Abdominal ultrasound revealed only an accessory spleen. The blood calcium level was within normal limits. The initial cytogenetic evaluation - high resolution GTG band karyotyping (≥ 550 bands) - was normal (46,XY). Investigation of the 22q11.2 microdeletion with the fluorescent in situ hybridization technique (FISH), using a DNA DiGeorge/VCFS Goliath, Probe (TUPLE 1) DNA probe, confirmed the diagnosis of the SD22q11.

Figure 1. Craniofacial features of the patient at different ages: 16 days (A), 1 year and 11 months (B), 4 years (C), and 13 years (D).

DISCUSSION

ALWs are a mild form of laryngeal atresia (type III).⁶ Its association with the SD22q11 microdeletion has been described in a few published papers within the past two decades; most of these reports had a small number of patients.⁷⁸ Miyamoto et al. (2004)⁷ reported a 65% frequency of the SD22q11 in 17 patients with ALWs, one of few case series. Studies of patients with the SD22q11 have noted that this malformation has been described in 1 to 2% of cases.

The SD22q11 is a relatively common genetic disease; it is caused by a deficiency in region 11 in the long arm of the chromosome 22. Although most cases of the SD22q11 are sporadic (because of new mutations), patients with this deletion have a 50% chance of passing it onto their offspring. The clinical features of this syndrome are varied; the phenotype includes several types of orofacilaryngeal alterations. These changes have been given several names given before the syndrome was described in the beginning of the 1990s, such as the DiGeorge syndrome, the velocardiofacial syndrome, and Shprintzen’s syndrome. These names reflect the perception of different specialists about the same disease.⁷⁻⁸ It should be noted that in some cases these features may not be so evident, especially younger children, making it more difficult to raise the possibility of this syndrome⁷⁻⁸ (see Fig. 1). Hypocalcemia may be latent, as in this case, and may become apparent only after surgery.

REFERENCES


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⁷ Neurophysiologist, behavioral, and speech development was compromised as the patient grew, requiring drug therapy - haloperidol and biperiden. The physical examination at age 12 years and 2 months was as follows: weight - 47 kg (P75-90), height - 147 cm (P50), cephalic perimeter - 55 cm (P2-50), elongated face, narrow palpebral fissures, bilateral epicanthic folds, hypoplastic nasal alae, dental malocclusion, prognathism, overfolded helices (horizontal and vertical rami), punctis carenati, protruding navel, right cryptorchidism, and thin fingers of the hand. The palate was cleft, but there were no associated abnormalities such as velopharyngeal insufficiency. Figure 1 shows