International Journal of Pediatric Otorhinolaryngology 95 (2017) 114–116

Contents lists available at ScienceDirect

International Journal of Pediatric Otorhinolaryngology

journal homepage: http://www.ijporlonline.com/

Case Report

Refractory sleep-disordered breathing due to unilateral lingual tonsillar hypertrophy in a child with Proteus Syndrome☆

Courtni R. Salinas [a], Brian A. Nuyen [a], Aria Jafari [b], Javan Nation [a, b, c, *, 1]

[a] School of Medicine, University of California San Diego, La Jolla, CA, USA
[b] Department of Surgery, Division of Otolaryngology-Head & Neck Surgery, University of California San Diego, San Diego, CA, USA
[c] Department of Otolaryngology, Rady Children's Hospital San Diego, University of California San Diego, San Diego, CA, USA

ARTICLE INFO

Article history:
Received 5 December 2016
Received in revised form
10 February 2017
Accepted 11 February 2017
Available online 14 February 2017

Keywords:
Sleep disordered breathing
Apnea
Hypertrophy
Lingual tonsillectomy
Proteus
Syndrome

ABSTRACT

Proteus Syndrome (PS) is a rare congenital overgrowth disease affecting bones, skin, adipose and the central nervous system. The result is asymmetric, disfiguring hypertrophy which can manifest as craniofacial dysmorphia and aerodigestive tract abnormalities. We report the case of obstructive lingual tonsillar hypertrophy resulting in residual sleep disordered breathing after adenotonsillectomy in a child with PS, a previously unrecognized manifestation of the disease. Endoscopic treatment with coblation effectively and safely treated the obstructive symptoms.

1. Introduction

Proteus Syndrome (PS) is a sporadic congenital condition affecting less than one in a million individuals. The disease is characterized by disfiguring, asymmetric overgrowth of all germ layer derived tissues and typically involves the unilateral bones, skin, adipose and central nervous system [1].

The general diagnostic criteria of PS requires mosaic distribution of lesions, progressive course, and sporadic occurrence. In addition, specific diagnostic criteria must be met, such as epidermal nevus, asymmetric disproportionate overgrowth, tumor susceptibility, dysregulated adipose tissue, vascular malformations, lung cysts, and a facial phenotype [2]. Ninety-percent of cases meeting these criteria carry a somatic mosaic mutation of *AKT1,* which causes cells to abnormally proliferate and divide [1,3]. Physical manifestations occur at 6–18 months of age, predominantly as skeletal overgrowth of unilateral hands or feet, but rapidly progressing to involve other tissues [1]. Up to 15% of children with PS are estimated to have developmental delay [4].

Head and neck manifestations of PS are variable in site and morbidity. Otolaryngologists play an integral role in management for airway safety, excision of masses, facial reconstruction and bone plating, hearing loss and dental abnormalities, as well as in the evaluation of dysfunctional speech, swallowing, and sleep.

Sleep disordered breathing (SDB) due to obstructive adenotonsillar hypertrophy has been described in a small number of PS patients. However, SDB due to unilateral lingual tonsillar hypertrophy (LTH) has not been reported. Pediatric SDB is associated with significant cardiovascular and metabolic morbidity, nocturnal enuresis, and impaired neurocognitive and behavioral functioning [5]. Evaluation and treatment of SDB is particularly important in syndromic children, as they are at a potentially higher risk of neurobehavioral dysfunction due to developmental delay.

Here we report the presentation and management of a nine-year-old child with PS and a history of palatine tonsillectomy and adenoidectomy seven years prior, found to have progressive SDB and obstructive left unilateral lingual tonsillar hypertrophy (LTH).

2. Case report

A nine-year-old male with known PS was referred to the otolaryngology clinic after a left base of tongue mass was noted

☆ Poster presented at American Society of Pediatric Otolaryngology Combined Otolaryngology Spring Meeting on May 18, 2016, Chicago, IL (#ASPO20160307).

* Corresponding author. Rady Children's Hospital San Diego/University of California San Diego, USA.
E-mail address: javan.nation@gmail.com (J. Nation).
[1] Present/permanent address: 3030 Children's Way Suite 402, San Diego, CA 92123.

http://dx.doi.org/10.1016/j.ijporl.2017.02.015
0165-5876/

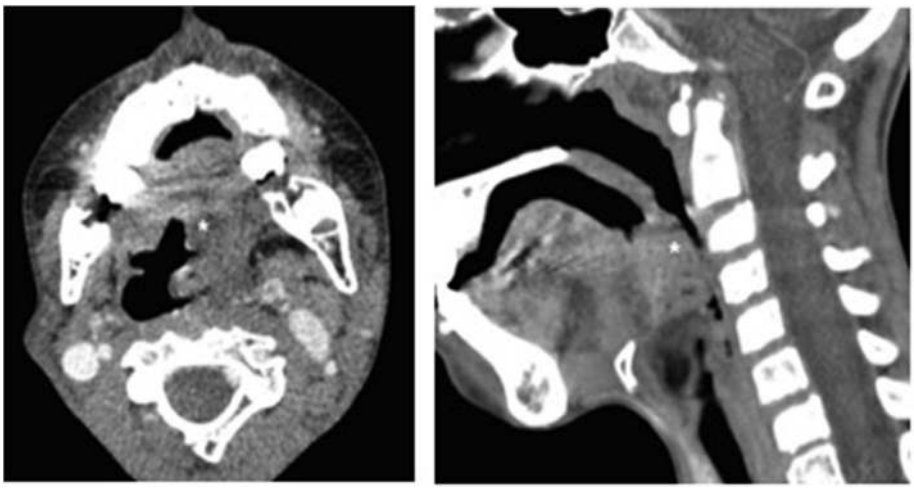

Fig. 1. CT scan of the neck with contrast demonstrated large obstructive left lingual tonsil*.

during a difficult intubation for a dental procedure. The child's parents reported nighttime symptoms of apnea and gasping, and daytime symptoms of excessive sleepiness and muffled voice. At two years of age, he underwent formal complete tonsillectomy and adenoidectomy for treatment of SDB with initial success, but had since developed recurrent symptoms.

On examination of the head and neck, the child was small for age, with left-predominant hemihypertrophic features and asymmetric facial skeletal framework. Fiberoptic nasolaryngoscopy revealed a large left-sided posterior tongue mass. Otologic exam showed left external auditory canal stenosis via bony overgrowth causing moderate to severe conductive hearing loss. The remainder of the exam was notable for a left orbital lipodermoid cyst, and left lateral neck verrucous skin plaques.

The tongue mass resembled lingual tonsillar tissue, and we decided to obtain pre-operative imaging to rule out carcinoma, sarcoma, fibroma, lingual thyroid, and other causes of tongue mass because of the unilateral nature. Computed tomography (CT) imaging of the neck with contrast demonstrated ill-defined soft tissue mass along the left oropharynx and hypopharynx with significant airway narrowing (Fig. 1). Enlarged left tonsillar tissue was in continuity with the lymphoid tissue along the left Waldeyer's ring. The CT noted other asymmetric findings including left parotid and submandibular gland enlargement, asymmetric vessels in left parapharyngeal space, asymmetric left carotid artery, dysplastic mandible with prominent irregularity of left parasymphyseal aspect, and expanded left temporomandibular joint. History and clinical findings indicated SDB secondary to unilateral LTH. The patient underwent endoscopic lingual tonsillectomy.

Intraoperatively, a 30-degree nasal endoscope revealed an obstructive left lingual tonsil originating from the left tongue base extending superiorly into the tonsillar fossae and inferiorly into the hypopharynx, abutting and deforming the epiglottis (Fig. 2A). A biopsy of the mass was obtained and histopathology was obtained identifying follicular lymphoid hyperplasia, confirming the unilateral mass as lingual tonsil. A coblator device was used to perform a complete left lingual tonsillectomy down to the level of the underlying tongue base, ensuring complete lingual tonsil tissue removal and an unobstructed view of the epiglottis. Laryngoscopy at the end of the procedure demonstrated grade II Cormack-Lehane view (Fig. 2B). After one day of uncomplicated hospital observation, the patient was discharged.

During his one-month post-operative visit, the patient reported complete resolution of snoring, a resolution of his muffled voice and more restful sleep. Flexible laryngoscopy demonstrated patent airway and no regrowth of lingual tonsil (Fig. 3).

3. Discussion

In the pediatric population, the leading cause of SDB is adenotonsillar hypertrophy. First line treatment is typically tonsillectomy and adenoidectomy, with 60–85% of patients reporting resolution

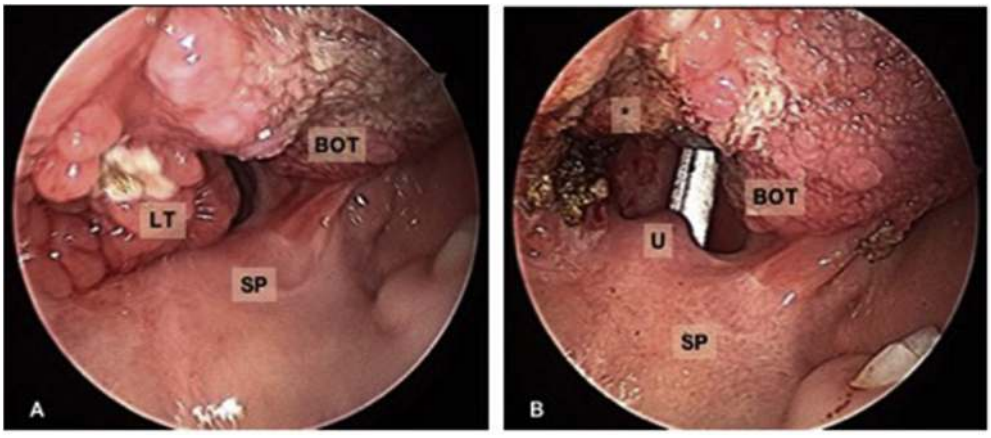

Fig. 2. Intraoperative endoscopic view of obstructive left lingual tonsil (A). Grade II Cormack-Lehane view was obtained following the procedure* (B). (LT – left lingual tonsil, BOT – base of tongue, SP – soft palate, U – uvula).

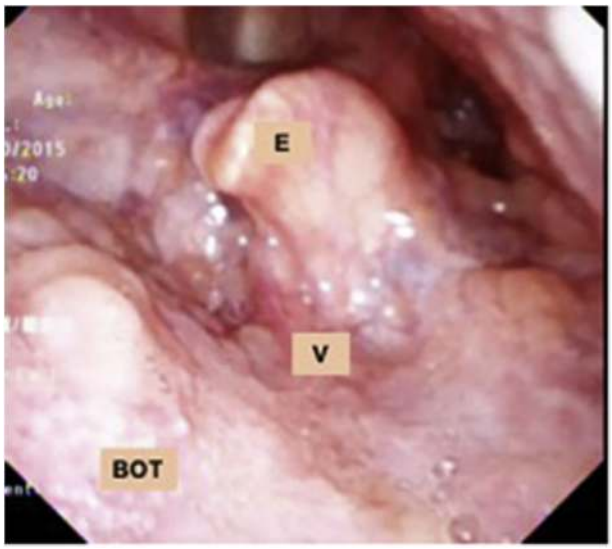

Fig. 3. Laryngoscopic view one month after bilateral endoscopic coblation lingual tonsillectomy (E – epiglottis, BOT – base of tongue, V – vallecula).

of symptoms [6].

Here we present the case of a nine-year-old male with PS who was found to have left LTH resulting in recurrent SDB seven years after treatment with adenoidectomy and tonsillectomy.

In children with craniofacial dysmorphologies, such as those seen in PS, SDB presents a therapeutic challenge because the symptoms recur more frequently after adenotonsillectomy and have more variable etiology [7]. For example, obstructive macroglossia, palatal narrowing, enlarged tongue base, shallow nasopharynx and supraglottic collapse may contribute to upper airway symptoms [6,8]. Moreover, 10–15% of children with PS will have developmental delay or intellectual disability [4]. The impaired neurocognitive and behavioral functioning associated with untreated SDB may exacerbate cognitive difficulties. Thus, monitoring for recurrent airway obstruction is of heightened importance in this population.

When pediatric SDB persists or recurs after adenotonsillectomy, LTH should be investigated as the etiology [9]. LTH is an increasingly recognized source of airway obstruction in SDB. Up to 50% of patients with Down Syndrome and 2% of non-syndromic children with persistent obstructive sleep apnea after adenotonsillectomy have LTH (>10mm) [10,11]. Several case series examining LTH treatment in children with recalcitrant obstructive sleep apnea reported lingual tonsillectomy significantly or completely resolved symptoms in 26/26 and 10/16 cases [8,9]. Partial improvements of apnea and snoring were achieved in patients with syndromic craniofacial abnormalities [8]. Our patient experienced resolution of apnea, gasping, daytime sleepiness, and muffled voice following treatment.

Three case reports have described unilateral palatine tonsillar hypertrophy presenting as dysphagia or snoring in PS, however this is the first documented case of unilateral LTH causing SDB in a patient with PS [12–14].

4. Conclusion

LTH can be a cause of residual SDB in children after adenotonsillectomy. For children with symptoms of SDB after adenotonsillectomy, a full airway evaluation should be performed. Unilateral lingual tonsillar hypertrophy is previously unreported, but logical in a child with PS because of the unilateral nature of the syndrome. Endoscopic treatment with coblation can effectively and safely treat obstructive symptoms.

Funding

None.

Conflict of interest

None.

Acknowledgements

The authors thank Matthew Brigger, MD, MPH, for editorial assistance.

References

[1] L.G. Biesecker, J.C. Sapp, Proteus syndrome, GeneReviews® [Internet], University of Washington, Seattle; 1993-2017, Seattle (WA), 2012, Aug 9. Available from: https://www.ncbi.nlm.nih.gov/books/NBK99495/ [PMID: 22876373].
[2] J. Turner, M.M. Cohen, L.G. Biesecker, Reassessment of the Proteus syndrome literature: application of diagnostic criteria to published cases, Am. J. Med. Gen. 103A (2) (2004) 111–122 [PMID: 15372514].
[3] M.J. Lindhurst, J.C. Sapp, J.K. Teer, et al., A mosaic activating mutation in AKT1 associated with the Proteus syndrome, N. Engl. J. Med. 365 (2011) 611–619.
[4] L.G. Beisecker, Proteus syndrome, in: S.B. Cassidy, J.E. Allanson (Eds.), Management of Genetic Syndromes, second ed., Wiley & Sons, Hoboken (NJ), 2005, pp. 449–456.
[5] H.-L. Tan, D. Gozal, L. Kheirandish-Gozal, Obstructive sleep apnea in children: a critical update, Nat. Sci. Sleep 5 (2013) 109–123 [PMID: 24109201].
[6] C.G. Sulman, Pediatric sleep surgery, Front. Pediatr. 2 (2014) 51 [PMID: 24926473].
[7] D. Rosen, Management of obstructive sleep apnea associated with Down syndrome and other craniofacial dysmorphologies, Curr. Opin. Pulm. Med. 459 (2011) 1 [PMID: 21918449].
[8] M. Abdel-Aziz, N. Ibrahim, A. Ahmed, M. El-Hamamsy, M.I. Abdel-Khalik, H. El-Hoshy, Lingual tonsils hypertrophy; a cause of obstructive sleep apnea in children after adenotonsillectomy: operative problems and management, Int. J. Pediatr. Otorhinolaryngol. 75 (9) (2011) 1127–1131 [PMID: 21737150].
[9] A.C. Lin, P.J. Kotai, Persistent pediatric obstructive sleep apnea and lingual tonsillectomy, Otolaryngol. Head. Neck Surg. 141 (1) (2009) 81–85 [PMID: 19559963].
[10] L.F. Donnelly, S.R. Shott, C.R. LaRose, et al., Causes of persistent obstructive sleep apnea despite previous tonsillectomy and adenoidectomy in children with down syndrome as depicted on static and dynamic cine MRI, Am. J. Roentgenol. 183 (2004) 175–181 [PMID: 15208134].
[11] B.L. Fricke, L.F. Donnelly, S.R. Shott, et al., Comparison of lingual tonsil size as depicted on MR imaging between children with obstructive sleep apnea despite previous tonsillectomy and adenoidectomy and normal controls, Pediatr. Radiol. 36 (2006) 518–523 [PMID: 16596369].
[12] J.K. Sahni, S. Kumar, V. Wadhwa, G. Kathuria, Proteus syndrome with huge tonsillar mass causing dysphagia: a rare case, J. Laryngol. Otol. 120 (5) (2006) 408–410 [PMID: 16556346].
[13] N.D. Zingade, N.N. Zingade, A rare case of Proteus syndrome, J. Laryngol. Otol. 122 (3) (2008) e7 [PMID: 18279570].
[14] E. Cantone, M. Cavaliere, G. Castagna, A. Marino, L. Del Vecchio, M. Iengo, Operative management of OSAS in a complex case of Proteus syndrome, Case Rep. Otolaryngol. (2015) 137589 [PMID: 26199778].