International Journal of Pediatric Otorhinolaryngology 103 (2017) 5–9

Contents lists available at ScienceDirect

International Journal of Pediatric Otorhinolaryngology

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

Case Report

Recalcitrant chronic rhinosinusitis in the setting of fucosidosis, a rare lysosomal storage disorder

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ARTICLE INFO

Article history:
Received 27 June 2017
Received in revised form
11 September 2017
Accepted 18 September 2017
Available online 21 September 2017

This manuscript was presented as a poster presentation at the European Society of Pediatric Otolaryngology, June 18–21, 2016 in Lisbon, Portugal.

Keywords:
Fucosidosis
Lysosomal storage disorder
Chronic rhinosinusitis

ABSTRACT

Fucosidosis is an autosomal recessive lysosomal storage disorder caused by the deficiency of alpha-L-fucosidase. We present the case of an affected female in the second decade of life with chronic rhinosinusitis (CRS) including recalcitrant polypoid inflammation, which has not been previously reported in the literature. With the advancement of life-prolonging measures, children with lysosomal storage disorders may suffer increasingly from CRS due to the lymphohistiocytic and macrophage infiltrate of the paranasal sinus mucosa that resembles severe polypoid inflammation.

1. Introduction

Fucosidosis is a very rare, autosomal recessive lysosomal storage disease defined by a severe deficiency in alpha-L-fucosidase, causing severe global developmental delay, abnormal bone development (dysosotosis multiplex), seizures, abnormal muscle stiffness, angiokeratomas, visceromegaly, and recurrent respiratory infections [1]. Its exact prevalence is not known but has been estimated to be less than 1 in 1,000,000 people. Approximately 100 cases have been reported worldwide [2]. We present the case of an affected female in the second decade of life with chronic rhinosinusitis (CRS) including recalcitrant polypoid inflammation, which has not been previously reported in the literature. Informed consent was obtained from the patient's family prior to all procedures. This case report was reviewed by the Institutional Review Board and determined to require no further review or formal exemption.

2. Case overview

2.1. Onset of disease and initial diagnosis

The patient is a 13 year old female with fucosidosis, who experienced initial normal speech development leading up to the age of 18 months, when she experienced rapid speech deterioration over the course of one summer. Fucosidosis was diagnosed on genetic screening for workup of this condition. Her birth history was notable for extreme prematurity at 25 weeks followed by a postnatal NICU stay lasting three months. She was also noted to have significant motor delay with delayed walk until 2 years of age. Both parents were noted to be carriers of her mutation in the FUC1 gene. Her condition has deteriorated since diagnosis, manifesting as inability to walk, contractures, hip dysplasia, and scoliosis. She has protruding tongue and splenomegaly and has angiokeratomas on her extremities and trunk. She has received supportive therapies

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https://doi.org/10.1016/j.ijporl.2017.09.019
0165-5876/

for her multiple sinonasal infections, and has had multiple evaluations with orthopedic surgery for her osteodystosis, for which surgical therapy was not indicated.

2.2. Initial otolaryngologic presentation and first recurrence (August and October 2011)

Our patient first presented to rhinological attention at eight years of age when she was noted to have rapid recurrence of a left sided nasal polyposis within two months after primary endoscopic resection.

She had previously been evaluated by pediatric otolaryngology for recurrent otitis media and adenotonsillar hypertrophy, and had undergone bilateral myringotomy with tube placement and partial tonsillectomy with adenoidectomy one year prior. The patient developed chronic progressive rhinorrhea and was managed with conservative medical management and serial nasopharyngoscopy, when she was noted to have developed purulent mucus and a unilateral watery inflammatory reaction of the left middle meatus. A CT scan was performed at that time, confirming left-sided inflammatory disease (Fig. 1).

She underwent left endoscopic sinus surgery (ESS) including total ethmoidectomy and widening of the left maxillary ostium. Mucosal pathology showed lymphohistiocytic infiltrate and foamy macrophages consistent with lysosomal storage disorders (Fig. 2). Post-operatively, she was maintained on a regimen of nebulized culture directed antibiotic (tobramycin) and budesonide. Over the course of two months, her nasal symptoms worsened, which negatively affected her pulmonary status resulting in hospital admission. Nasal endoscopy at that time demonstrated bilateral purulent rhinorrhea with polypoid inflammation without frank polyposis, and the patient underwent revision left ESS.

2.3. Second recurrence (April 2012)

Following this initial episode, the patient's symptoms were well-controlled with medical management including antibiotic and budesonide sinus rinses as needed. At six months after her first revision, the patient's polypoid inflammation recurred, and she underwent revision left ethmoidectomy and left maxillary antrostomy with tissue removal. Operative cultures from this surgery were positive for *Pseudomonas aeruginosa*. Her symptoms greatly improved after this surgery, and she was maintained on daily gentamicin/tobramycin rinse in the initial post-operative period before transitioning to daily tobramycin/budesonide rinses.

2.4. Third recurrence (September 2014)

The patient's symptoms were well-controlled on this regimen for approximately 18 months until there was recurrence of polypoid inflammation. She underwent revision left ESS, and now, right nasal endoscopy and exploration, with findings notable for acute right maxillary sinusitis. At this time, it was evident that the polypoid inflammation was now bilateral. Operative cultures were positive for *Captocytophaga* spp. She was given one week of Augmentin and restarted on her chronic sinus regimen of daily tobramycin/budesonide rinses.

2.5. Fourth recurrence (December 2014)

Post-operatively, the patient's symptoms improved but incompletely resolved. She was placed on diflucan after growing fungus from a sinus culture obtained by her primary care physician and the decision was made to return to the operating room. She underwent bilateral revision ESS, maxillary antrostomy and total ethmoidectomy. Operative cultures grew *Staph aureus*. She received budesonide/bactroban rinses post-operatively and symptoms improved.

2.6. Fifth recurrence (September 2015)

Symptoms were improved for nine months, at which time the patient's pulmonary status declined, with worsening cough and congestion, home oxygen requirement, and a negative pulmonary workup including two bronchoscopies at an outside institution. She underwent bilateral revision sinus surgery of the maxillary and ethmoid sinuses, as well as frontal sinus exploration. Operative cultures grew *Pseudomonas* spp. and she was placed on gentamicin/budesonide rinses.

2.7. Sixth recurrence (February 2016)

She initially improved on this regimen, then developed recurrent thick nasal mucus discharge and polypoid inflammation despite budesonide/gentamicin rinses and oral Augmentin. She underwent bilateral revision sinus surgery involving the bilateral maxillary, ethmoid, frontal, and sphenoid sinuses. Post-operatively, she was restarted on budesonide/tobramycin daily rinses.

2.8. Seventh recurrence (September 2016)

Her symptoms improved for four months, at which time the

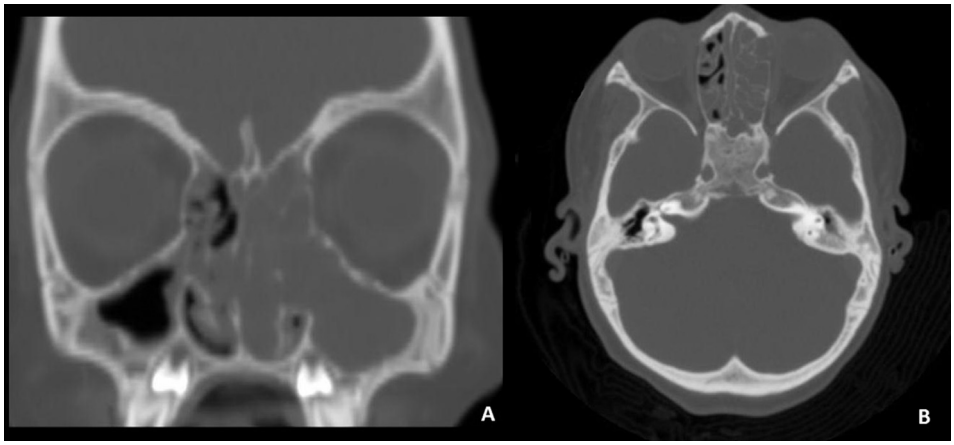

Fig. 1. A CT scan was performed at that time, confirming left-sided inflammatory disease.