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Osteochodromyxoma presenting as case of congenital nasolacrimal duct obstruction

Anas Yasin [a,b], Wesley Chow [c], Paul J. Rychwalski [a,b], Samiksha Fouzdar Jain [a,b,*]

[a] Department of Ophthalmology, University of Nebraska Medical Center, Omaha, NE, 68198, USA
[b] Children's Hospital & Medical Center, 8200 Dodge St., Omaha, NE, 68144, USA
[c] Creighton University School of Medicine, Omaha, NE, USA

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ABSTRACT

Purpose: We report the case of a 10-month-old with nasolacrimal duct obstruction (NLDO) associated with osteochondromyxoma (OMX), a very rare bone tumor.

Observations: A 10-month-old boy presented with a 6-month history of right eye epiphora not responding to digital massage and topical steroid-antibiotics eye drops. The ophthalmic exam showed right medial canthal swelling. During the ophthalmic exam an abnormal snoring sound was noted. The mother also reported that patient experienced frequent upper respiratory tract infections. Inspection of nostrils showed a right nasal lesion that upon comprehensive evaluation by ENT and pathology teams turned out to be an OMX with loss of PRKAR1A expression. Further genetic testing confirmed the Carney complex (CNC) diagnosis and the patient was referred to multidisciplinary care. To the best of our knowledge, this is one of the first cases of OMX-induced NLDO, where a typical looking congenital NLDO ended up with a diagnosis of a rare genetic disease.

Conclusion and Importance: We have described a case of OMX of the nasal cavity masquerading as congenital NLDO. This case emphasizes that NLDO is not always congenital if presenting within the first few months of life. It is important to obtain a thorough history and exam to evaluate potential differential diagnoses to guide subsequent decision-making steps.

1. Introduction

Nasolacrimal duct obstruction (NLDO) is the most common disorder of the lacrimal system with approximately 6–20% of newborns affected.[1,2] Congenital NLDO is typically due to a mechanical obstruction at the distal end of the nasolacrimal system at the valve of Hasner, in which lacrimal pooling and epiphora are present within the first weeks or months of life.[3] Acquired causes of NLDO include inflammatory or infectious sources, facial trauma, or malignancy.[4–6] Osteochondromyxoma (OMX) is an extremely rare benign bone tumorthat is always associated with lentigines and other unusual disorders, and it has in fact been called "Carney bone tumor" since it typically presents in cases of Carney complex (CNC),[7] though it is one the least common features of its 11 diagnostic criteria.[8] Here we report a unique case of a patient with a right nasolacrimal duct obstruction due to OMX, a rare manifestation of the CNC.

2. Case presentation

A previously healthy 10-month-old boy presented with a 6-month history of right eye epiphora that persisted despite nasolacrimal duct digital massage and frequent rounds of steroid-antibiotic combination. The ophthalmic exam was unremarkable except for the right eye's increased tear meniscus and cystic swelling at the medial canthus resembling dacryocele (Fig. 1A). During the exam, excessive snoring and nasal grunting sounds were noted, the mother did admit a history of multiple upper respiratory tract infections (URTI). Examination of nostrils reveals a right-sided fleshy nasal polyp (Fig. 1B). The patient was referred to ENT with a working diagnosis of nasal polyp for further evaluation. Orbital CT scan (Fig. 2A) & MRI (Fig. 2B) demonstrated a 3.5 cm well-defined mass occupying the entire nasal cavity and maxillary sinus. Nasal endoscopy (Fig. 3) and endoscopic sinus surgery with excisional biopsy were performed without additional nasolacrimal duct probing or stenting. Histologic tissue examination revealed OMX with loss of PRKAR1A expression. After surgery, breathing normalized, and

* Corresponding author. Children's Hospital & Medical Center, 8435 Cass St, Omaha, NE, 68114, USA.
E-mail address: sjain@childrensomaha.org (S. Fouzdar Jain).

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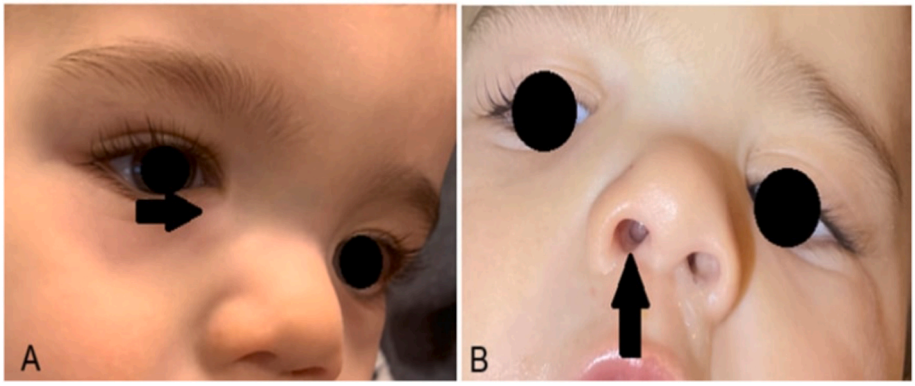

Fig. 1. A. External photograph demonstrating right medial canthal swelling (Arrow).
Fig. 1B. Appearance of an obstructive pink, fleshy mass seen on right nasal passage (Arrow). (For interpretation of the references to colour in this figure legend, the reader is referred to the Web version of this article.)

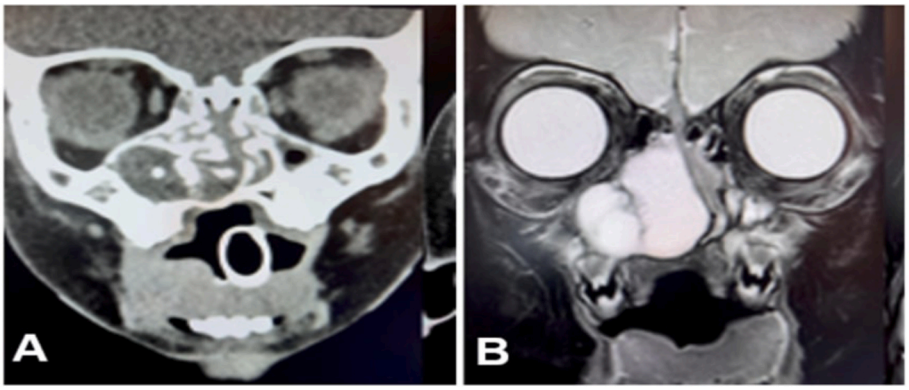

Fig. 2. A,B. Coronal orbital CT (A) and MRI (B) images demonstrating mass in the right nasal cavity and maxillary sinus. The scans demonstrate a significant obstruction with deviation of nasal septum to the left.

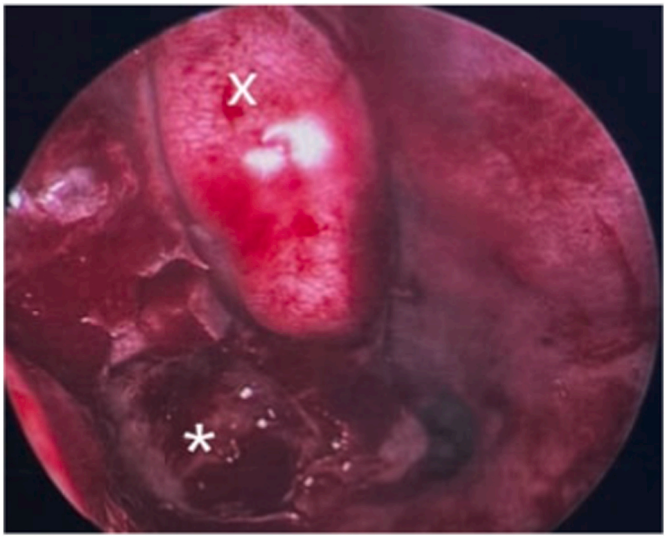

Fig. 3. Nasal endoscopy appearance of osteochondromyxoma (*), purplish mass under the inferior turbinate (X).

epiphora resolved completely. Because of the strong association of OMX with CNC,[7] the patient underwent genetic testing which showed a heterozygous pathogenic variant of the PRKAR1A gene (c.266del; p.P89Qfs*40) providing the molecular confirmation of his CNC diagnosis. Because CNC is an autosomal dominant inherited disease additional testing was performed on his parents, and his father tested positive for the same PRKAR1A variant; this further confirms that his diagnosis is paternally inherited. Given his CNC diagnosis, subsequent evaluation was performed by the cardiology, dermatology, and endocrinology departments. Clinical exams, imaging studies, and blood work all were unremarkable.

3. Discussion

OMX is a rare bone tumor that is classically associated with CNC, OMX has been called carny bone tumor,[9,10] though it presents in only 1 % of all cases of CNC,[7] This makes OMX a rare tumor of a rare condition. Up to 2013, worldwide more than 750 cases of CNC have been reported.[11] CNC is a multiorgan autosomal dominant inherited condition associated with the loss of the PRKAR1A tumor suppressor gene (chromosome 17q22-24). It is characterized by OMX, recurrent cardiac myxomas, cutaneous myxomas, multiple endocrine neoplasms, spotty pigmented mucosal and skin lesions, psammomatous melanotic schwannomas, growth hormone-secreting pituitary adenomas, large-cell calcifying Sertoli cell tumors, breast ductal adenomas and bilateral breast myxomas.[12] Diagnosis is made by the presence of at least two of these criteria, confirmed by histology or one criterion and either a first-degree family history of CNC or a PRKAR1A mutation as is our case.[13,14] Although inherited CNC typically has 100 % penetrance, other reports demonstrate variable penetrance which may explain why the father has not had any clinical manifestations of CNC yet.[15]

OMX typically presents in children younger than the age of 2 years and affects commonly long bones as well as sinus and nasal bones.[7] OMX is typically painless and reported symptoms were due to local mass

effects. Those affecting the nasal bones were presented with facial asymmetry, nasal congestion, or difficulty breastfeeding.[16,17] To the best of our knowledge, NLDO was not reported before as presenting symptom of OMX and subsequent diagnosis of CNC.

Although primary congenital NLDO is a common problem affecting infants, secondary causes should be always kept in mind especially when other symptoms are associated with watery eyes. In our case, abnormal breathing, frequent URTI, and subsequent nasal examination were the keys to identifying such an uncommon presentation to a rare medical condition with more serious systemic manifestations. Both OMX and CNC are very rare conditions, this case with its unique ophthalmic presentation contributes to a better understanding of these entities.

4. Conclusions

We have described a unique case OF OMX of the nasal cavity masquerading as congenital NLDO. OMX can be associated with CNC diagnosis and initiation of the multidisciplinary team approach is crucial in these cases.

Patient consent

Written consent to publish the case report was obtained.

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Authorship

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Intellectual property

We confirm that we have given due consideration to the protection of intellectual property associated with this work and that there are no impediments to publication, including the timing of publication, with respect to intellectual property. In so doing we confirm that we have followed the regulations of our institutions concerning intellectual property.

Research ethics

We further confirm that any aspect of the work covered in this manuscript that has involved human patients has been conducted with the ethical approval of all relevant bodies and that such approvals are acknowledged within the manuscript.

Written consent to publish potentially identifying information, such as details or the case and photographs, was obtained from the patient(s) or their legal guardian(s).

Declaration of competing interest

No conflict of interest exists.

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