Case Report

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Sinus mucocele leads to unilateral proptosis in an infant patient with cystic fibrosis: a literature review and a case report study

Narges Lashkarbolouk, MD[a,b], Mahdi Mazandarani, MD[a,b], Parastou Biazar, MD[c], Lobat Shahkar, MD[d,*]

Introduction and importance: Cystic fibrosis (CF) is a widespread life-shortening recessive genetic disease and can present with sinus mucocele. Sinus mucocele is a rare condition, with limited prevalence data on unilateral proptosis.

Case presentation: The authors present a case of a 19-month-old boy with CF who experienced worsening proptosis and exotropia in his right eye. A brain and orbit MRI revealed diffuse polypoid mucosal thickening, possible dense fungal deposit, deformity of the mid face, especially on the right side, with more prominent bulging of medical and inferior walls of the right lobe, a right ethmoidal mucocele causing ocular globe displacement, medial rectus compression, and optic nerve. An examination of the eye fundus showed disc edema and vascular congestion. Endoscopic sinus surgery successfully drained the mucocele, and treatment with antibiotics and corticosteroids led to symptom improvement and resolution of proptosis within 3 weeks.

Clinical discussion: Mucoceles represent an uncommon complication associated with CF in pediatric patients. Consequently, any child presenting with this issue should undergo evaluation for CF. Investigating this infrequent condition's underlying mechanisms and consequences may improve treatment approaches and outcomes for those impacted.

Conclusion: Sinus mucocele with unilateral proptosis in CF patients is uncommon, and endoscopic sinus surgery appears to be an effective cure for this complication, even in the pediatric population at high risk, like CF patients.

Keywords: children, cystic fibrosis, ophthalmology, proptosis, sinus mucocele

Introduction

Cystic fibrosis (CF) is one of the autosomal recessive diseases that primarily affects exocrine glands, resulting in a reduced lifespan. CF is a result of mutations in the CF transmembrane conductance regulator (CFTR) gene, leading to mucus buildup in different organs, particularly the respiratory and digestive systems. CF epithelial cells cannot release chloride or bicarbonate properly when stimulated by cyclic adenosine monophosphate signals. In respiratory cells, excessive sodium absorption occurs. This genetic anomaly leads to highly concentrated mucus, causing airway damage like bronchiolar obliteration, bronchiolectasis, cysts, abscesses, fibrosis, and bronchiectasis[1,2].

The progression rate of CF disease is the main factor determining morbidity and mortality. As CF advances gradually, symptoms such as chronic cough, sputum production, exercise intolerance, and shortness of breath emerge. In later stages, physical signs include an increased anteroposterior chest diameter, clubbing scattered or localized coarse crackles, and hyper resonance. Expiratory wheezes may be present, indicating airway inflammation and edema with or without bronchodilator response[3,4].

HIGHLIGHTS

- As far as current knowledge extends, mucoceles affecting the paranasal sinuses in pediatric patients with cystic fibrosis (CF) have been documented in a limited number of case reports and small case series within the past decade.
- Sinus mucocele with unilateral proptosis in CF patients is uncommon, and endoscopic sinus surgery appears to be an effective cure for this complication, even in the pediatric population at high risk, like CF patients.
- Mucoceles is a rare complication of CF in children; therefore, every pediatric patient with this ENT complaint should be assessed for CF. Exploring the mechanisms and implications of this rare condition could enhance treatment strategies and outcomes for affected individuals. Healthcare providers must be suspected of CF when encountering mucoceles in pediatric patients. Early detection and proper management can greatly impact the quality of life of these individuals.

[a]Endocrinology and Metabolism Research Center, Endocrinology and Metabolism Clinical Sciences Institute, Tehran University of Medical Sciences, Tehran, [b]Golestan University of Medical Sciences, Gorgan, [c]Azar Hospital, Golestan University of Medical Sciences and [d]Taleghani Pediatric Hospital, Golestan University of Medical Sciences, Gorgan, Iran

N.L. and M.M. have contributed equally to this work and share first authorship.

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*Corresponding author. Address: Taleghani Pediatric Hospital, Golestan University of Medical Sciences, Gorgan, Iran. Tel.: +98 911 175 9597.
E-mail: Lobatshahkar@yahoo.com (L. Shahkar).

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The paranasal sinuses are typically opacified radiographically, and acute sinusitis or recurrent rhinosinusitis is generally observed. Nasal obstruction and rhinorrhea are frequent due to inflamed, swollen mucous membranes or nasal polyposis. A possible sinonasal manifestation of CF is mucocele, a cyst filled with secretions lined with upper respiratory epithelium that can develop in the paranasal sinuses through a slow concentric growth. Usually benign, they can increase in size due to secretion buildup and potentially displace and damage nearby bone, leading to local, orbital, or even intracranial complications. Mucoceles can present with symptoms such as nasal obstruction, facial pain, headaches, and recurrent sinus infections. Surgical intervention may be necessary to remove the cyst and prevent further complications[5,6].

As far as current knowledge extends, mucoceles affecting the paranasal sinuses in pediatric patients with CF have been documented in a limited number of case reports and small case series within the past decade. We presented the case of an ethmoidal mucocele leading to ocular manifestations in a 19-month-old boy with CF with no previous serious complications.

Case report

A 19-month-old boy who was diagnosed with CF and was being closely monitored at a pediatric hospital was referred to the ophthalmology department for evaluation due to a worsening condition of proptosis and exotropia in his right eye. The patient was born at full term without any complications during pregnancy and delivery. His physical growth measurements were above the fifth percentile, including height, weight, and head circumference. There was no reported history of congenital anomalies or respiratory issues in siblings.

In physical examination, the patient exhibited nonaxial proptosis, limited adduction, and exotropia in the right eye. His pupils were reactive and round, with no signs of chemosis or congestion. His heart sounds (S1 and S2) were also regular, and his vital signs were within normal range. A genetic diagnosis of CF was confirmed at 14 months old in the setting of chronic steatorrhea.

Following the patient's admission, a chest radiograph yielded normal results. The pediatric cardiology consultant's ECG indicated a normal sinus rhythm and axis. Additionally, the ophthalmologist requested brain and orbit MRI with and without contrast. It showed diffuse expansive polypoid mucosal thickening occupying all paranasal sinuses and both nasal cavities. Central low T2 signal intensity with associated enhancing wall thickening was observed, which might be a result of dense fungal deposits. As a result of abnormal findings, deformity of the midface has resulted, especially on the right side, with concomitant, more prominent bulging of medial and inferior walls of the right orbit and resultant more obvious right-sided proptosis. In addition, an extension of soft tissue changes is also seen in to supero-medial portion of the right orbit. It identified a right ethmoidal mucocele causing displacement of the ocular globe and compression of the medial rectus and the optic nerve (Fig. 1). Disc edema and diffuse vascular congestion were observed in the eye fundus examination.

Laboratory tests demonstrated leukocytosis, with lymphocytes accounting for 71% and thrombocytosis. The erythrocyte sedimentation rate was measured at 42, and the C-reactive protein test returned a positive result.

Subsequently, endoscopic sinus surgery was scheduled, and dilated ducts filled with thick secretions, strongly indicating CF, were identified upon biopsy. In the details of the endoscopic procedure, a small incision was made in the mucosa to access the mucocele (mucosal incision). Then, the mucocele is carefully drained. This involves removing the cyst wall to prevent recurrence (drainage). The obstructed sinus ostium (opening) is reestablished to ensure proper drainage and aeration of the sinus cavities. This step is crucial in reducing the likelihood of recurrence (duct re-establishment). The sinus was irrigated to remove any residual mucus and debris (irrigation and debridement). In endoscopic procedures, the incisions were closed with absorbable sutures (closure).

The mucocele drainage was assessed microbiologically, revealing the presence of *Escherichia coli* (E. coli) and *Staphylococcus saprophyticus* and fungal infection in the culture. Treatment was with ciprofloxacin, metronidazole, vancomycin, caspofangin, voriconazole, amikacin, and dexamethasone. After 2 weeks of antibiotic treatment and surgery, his symptoms and the proptosis improved, and the patient was discharged in good general condition. Following discharge, the administration of oral antibiotics, including ciprofloxacin and linezolid, along with the use of an amikacin nebulizer, was maintained for a duration of 1 week.

In summary, we presented a 19-month-old boy diagnosed with CF, complaining of worsening proptosis and exotropia in his right eye. Examination revealed nonaxial proptosis, limited adduction, and exotropia in the right eye. A brain and orbit MRI showed diffuse expansive polypoid mucosal thickening occupying all paranasal sinuses and nasal cavities. Central low T2 signal intensity with associated enhancing wall thickening was observed, which might be a result of dense fungal deposits. Deformity of the mid face, especially on the right side, with concomitant, more prominent bulging of medial and inferior walls of right orbit and resultant more obvious right-sided proptosis also observed. In addition, an extension of soft tissue changes is also seen in to supero-medial portion of the right orbit. A right ethmoidal mucocele causing displacement of the ocular globe and compression of the medial rectus and the optic nerve is notable. An eye fundus examination revealed disc edema and vascular congestion. Endoscopic sinus surgery identified dilated ducts filled with thick secretions, drained the mucocele, and treatment with antibiotics, antifungal treatment, and cortisone resolved proptosis.

Discussion

A typical presentation of chronic/recurrent rhinosinusitis in CF patients is commonly seen and may be linked to nasal polyps. Additionally, imaging often reveals hypoplasia/aplasia of certain paranasal sinuses. The studies reported that children are seldom impacted by mucocele; if seen at a young age, there is typically an underlying cause, and the prevalence of it in CF patients is in up to 4% of patients[1,3].

Paranasal sinus mucocele is a cystic lesion lined with respiratory epithelium filled with mucoid secretions commonly found in the ethmoid or frontal sinuses. Factors such as inflammation, trauma, fibrosis, neoplasm, and prior surgery contribute to its development. Mucoceles can present with symptoms such as headaches, nasal congestion, facial pain, and in some cases, mucoceles can become infected, leading to more severe symptoms

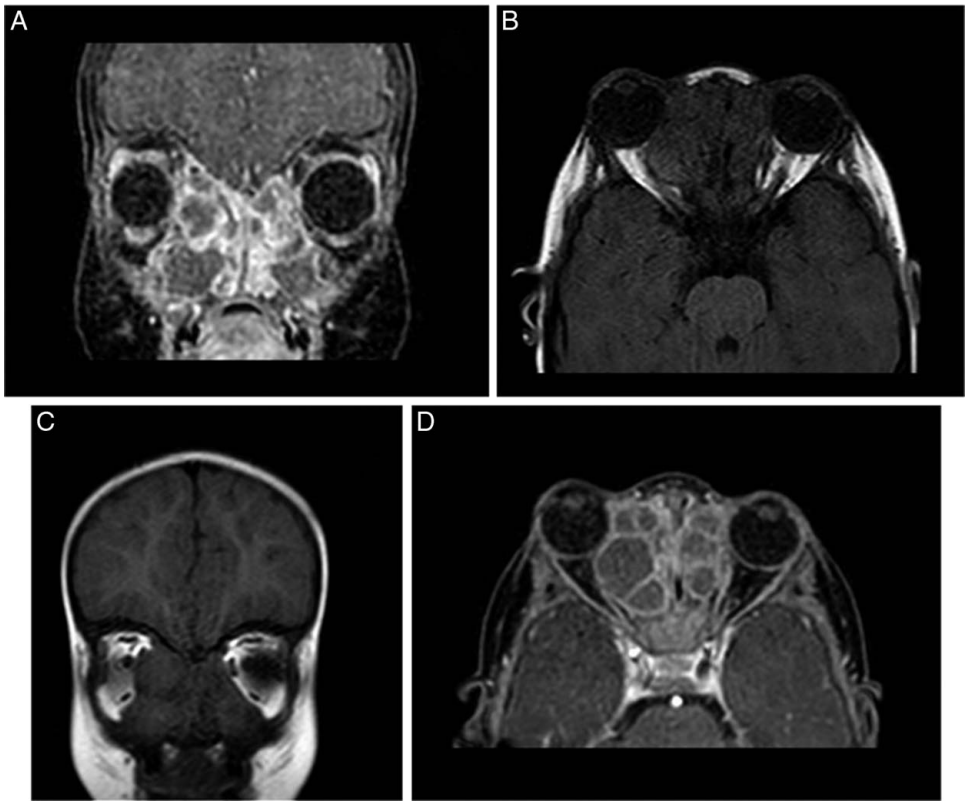

Figure 1. They illustrated widespread, polyploid mucosal thickening affecting all paranasal sinuses and nasal cavities. This has led to deformity in the midface, particularly on the right side, resulting in a noticeable bulging of the right orbit's medial and inferior walls and subsequent increased proptosis on the right side. A right ethmoidal mucocele has been identified, causing displacement of the eye globe and compression of the medial rectus and optic nerve.

such as fever, increased facial swelling, pus drainage from the nose, and proptosis[4,5].

Paranasal sinus mucoceles typically act as benign masses but can grow, potentially displacing and damaging nearby bone with local, orbital, or even intracranial complications. In severe cases, they can lead to visual disturbances and even cranial nerve palsies. Treatment for mucoceles in pediatric patients with CF may include antibiotics to treat any infection, nasal decongestants to help reduce congestion, and surgical intervention to remove the mucocele and improve sinus drainage. Surgical intervention involves marsupialization and drainage, with endoscopic surgery being the preferred method[6,7].

During the surgical procedure, an endoscope, a thin, flexible tube with a camera at its end, is inserted through the nostrils to reach the sinuses. The surgeon will then extract the mucocele and any damaged tissue, as well as widen the sinuses to facilitate proper drainage and ventilation. Endoscopic sinus surgery is commonly conducted under general anesthesia and is deemed a safe and efficient method for treating mucoceles[7,8].

Treating mucocele in CF patients can present several challenges due to the underlying condition and potential complications that may arise. CF patients might have compromised lung function, increasing the risk of surgical complications and prolonging recovery time. The anesthesia and the surgical procedure itself can pose additional risks, and surgery may be challenging because of their small size and fragile anatomy. Nevertheless, like any surgery, there are risks involved, such as bleeding, infection, and harm to nearby structures. CF patients are already susceptible to respiratory infections, and surgery to address proptosis can further increase the risk of infection. Also, mucoceles often recur in infants, as their salivary glands are still developing and may be prone to blockages. In our study, the surgery was performed well, and the child recovered without complications. Therefore, consulting with your healthcare provider to thoroughly discuss the possible complications and benefits of the surgery before making a decision is crucial.

Some findings suggest a potential link between CF and ocular health. Several clinically evident ocular complications of CF, including xerophthalmia, functional optic nerve deficiencies, papilledema, nyctalopia, retinal hemorrhages, edema, proptosis, and glaucoma have been reported. Ocular surface abnormalities like goblet cell loss, epithelial cell enlargement, and keratinization were also observed. A mucocele may follow these symptoms due to pressure effects or glycoprotein and electrolyte changes (e.g. vitamin A) in the CF lens. Ophthalmologic care of CF patients could play a crucial role in overall clinical management. Supplemental therapy with artificial tears may be advisable for eye involvement in the disease[8,9].

The prognosis of mucoceles in CF patients depends on factors like lung function, disease severity, and existing complications. Key prognostic factors: 1. early intervention: timely diagnosis and management improve outcomes. 2. Lung function: patients with better pulmonary function generally have a more favorable prognosis, as lung health directly influences overall outcomes. 3. Chronic disease progression: mucoceles may indicate advanced disease and are associated with increased morbidity. 4.

Complications: infections or issues like pancreatitis related to mucoceles can worsen the prognosis. In conclusion, while mucoceles present challenges for CF patients, outcomes can improve with comprehensive care, timely intervention, and personalized treatments. Regular follow-ups and prompt reporting of new symptoms are essential for effective management.

One strength was the patient's ability to undergo surgery despite their age and the complexity of the procedure given their underlying disease. However, a limitation of our study was that the patient had to be referred to a more advanced center for the surgery. Further long-term studies on larger pediatric CF studies are necessary to determine the true prevalence of this complication, identify the optimal conservative medical treatment, and clarify which patients require surgery.

Conclusion

Mucoceles is a rare complication of CF in children; therefore, every pediatric patient with this ENT complaint should be assessed for CF. Exploring the mechanisms and implications of this rare condition could enhance treatment strategies and outcomes for affected individuals. Healthcare providers must be suspected of CF when encountering mucoceles in pediatric patients. Early detection and proper management can greatly impact the quality of life of these individuals.

Ethical approval

Golestan University of Medical Sciences approved this research with ethics code IR.GOUMS.REC.1403.212.

Consent

Written informed consent was obtained from our patient`s legal guardians to publish this report and any accompanying images. A copy of the written consent is accessible for review by the Editor-in-Chief of this journal. The purpose of this case report was fully explained to the guardians. They were assured that the researchers would maintain the confidentiality of their information. The principles of the Declaration of Helsinki conducted this case report.

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Author contribution

L.S.: advised the case report study; M.M., N.L., and P.B.: gathered patient's medical and health records; P.B.: also did the surgical treatment; M.M. and N.L.: wrote the first draft of the manuscript, and all authors commented on previous versions. All authors read and approved the final manuscript.

Conflicts of interest disclosure

The authors declare no conflict of interest.

Research registration unique identifying number (UIN)

This is a case report study. The datasets used during the current study are available from the corresponding author on reasonable request.

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Availability of data and material

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