VIA MEDICA

CASE REPORT

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Massive nasal polyposis in a patient with newly diagnosed cystic fibrosis

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Abstract

Introduction: Cystic fibrosis (CF) — is the most common fatal autosomal recessive disease in Caucasians. A number of reports have described patients who do not meet diagnostic criteria for cystic fibrosis. Atypical or nonclassic CF is characterised by normal or borderline sweat test, pancreatic sufficiency and a monosymptomatic phenotype. For these reasons clinicians should remain alert to the possibility of the occurrence of CF.

Case report: We described a case presentation of massive nasal polyposis and recurrent sinusitis leading to the diagnosis of cystic fibrosis in a 11-year-old male.

Conclusion: Our study indicates that chronic sinusitis and/or polyposis should raise the clinicians suspicion of a potential presentation of undiagnosed CF and require further investigations.

Key words: cystic fibrosis, nasal polyposis, children, rhinosinusitis

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Introduction

Nasal polyposis recalcitrant to standard treatment has become an increasingly difficult problem in the practice of otolaryngology. In recent years, computerized tomography (CT) scans and endoscopic imaging has greatly aided in diagnosis of the condition [1]. Nasal polyposis is a recognized feature of few genetic diseases including cystic fibrosis (CF). CF is the most common fatal autosomal recessive disease in Caucasians (1: 2,000–4,500 live births). It is caused by mutations in the CF transmembrane conductance regulator (CFTR) gene, which encodes a protein expressed in the apical membrane of exocrine epithelial cells, resulting in changes to the fluid and electrolytes in the cell [2, 3]. CF is characterized by dysfunction of many different organs and systems, which in most cases progress to chronic respiratory infection, pancreatic insufficiency and malnutrition. Coexisting chronic sinusitis or nasal polyps are a frequent complain in CF patients with a reported occurrence of about 40% in patients over 5 years of age. Surgery is required for symptomatic and refractory to standard treatment polyps being the second most common class of operations performed on CF patients [4]. In recent years, the detection of CF has improved with the widespread introduction of infant CF screening. Despite improvements in our understanding of CF genetics, diagnosis of CF in patients presenting with atypical or only the symptom, remains a clinical challenge.

We described a case presentation of massive nasal polyposis and recurrent sinusitis leading to the diagnosis of cystic fibrosis in an adolescent patient.

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Case report

The 11-year-old male was admitted to the Department of Pediatrics and Allergy (Lodz, Poland) due to nasal obstruction and chronic rhinorrhea. Symptoms appeared two years ago which occurred 6 months prior to admission. The boy was diagnosed with allergic rhinitis due to birch allergy at the age of 7. He started regular treatment with nasal steroids and has a two-year history of oral immunotherapy for birch; respiratory symptoms improved temporarily. At the age of 9 years he suffered from nasal obstruction, unresponsive to treatment (despite receiving chronic nasal corticosteroid twice daily and oral antyhistamines symptoms of nasal swelling gradually worsened). The patient had no other associated diseases nor family history of respiratory disorders. Pulmonary function testing FEV_1 was 81% of predicted. The mother reported periodic mild cough coexisting with runny nose in her child.

On physical examination, the patient was in good health status, well-nourished, presented a runny nose, oral breathing and nasal obstruction. An otolaryngological exam showed polyps obstructing the whole bilateral nasal cavities. Paranasal sinus CT scan with axial and coronal views without contrast detected pansinusitis with massive polypoid hyperplasia of mucosa in the maxillary, ethmoidal and sphenoidal sinuses, and a polyp in the nasal cavity obstructing the ostiomeatal complex (Fig.1). Due to bronchial wall thickening on chest X-ray, CT of the chest was performed. The results showed only severe bilateral cylindrical bronchiectasis and mild atelectasis. The child was directed to the Department of Otolaryngology for functional endoscopic sinus surgery (FESS). In the nasopharynx the massive polyp was highlighted, which came from the maxillary sinus and filled completely the nasal cavity (Fig. 2). Histopathologic examination demonstrated mucosal inflammatory infiltrate with polymorphonuclear leukocytes, typical of CF polyps. Methicillin-resistant *Staphylococcus aureus* (MRSA) was isolated from the sample. We observed elevated levels of total IgE and allergen-specific IgE for birch allergen in the serum. There was no hypersensitivity to aspirin nor other nonsteroidal anti-inflammatory drugs. Immunodeficiency, alpha-1 antitrypsin deficiency, celiac disease, infection of Mycobacterium, atypical bacterial infection were excluded. Stool examination for a 72h fat collection were negative, faecal elastase within the normal range. The pilocarpine sweat test was carried out — 85 mmol/l (positive when above 60), which lead to CF diagnosis. In order to confirm the diagnosis, molecular testing for a panel of CFTR mutations was preformed and G542X/3849+10 kb C>T genotype was identified. Subsequently, the child was referred to our CF multidisciplinary team for further treatment, management and follow-up.

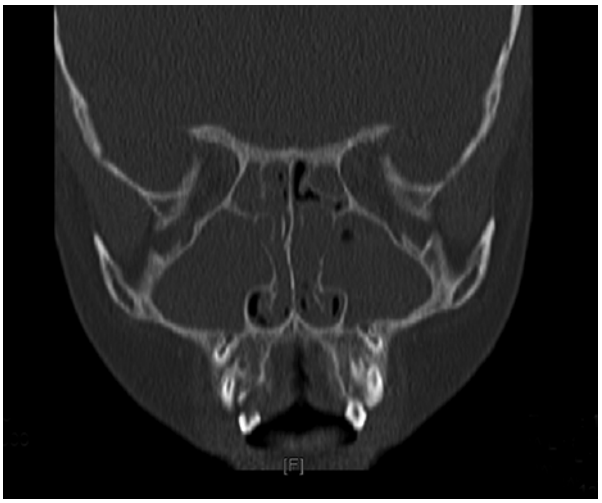

Figure 1. Paranasal sinus computed tomography (CT, coronal image) shows massive polyposis nearly completely obstructing bilateral nasal cavities, maxillary and ethmoid sinuses

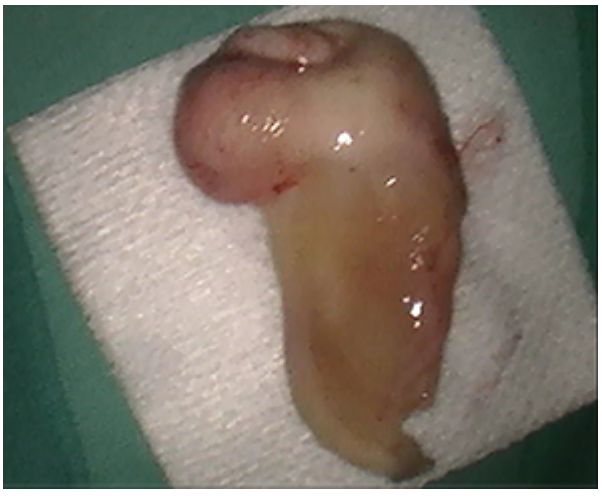

Figure 2. The nasal polyp (5 cm × 3 cm) removed during endoscopic surgery from the nasopharynx

Discussion

In paediatric patients, nasal polyposis massive and resistant to standard treatment are in most cases associated with atopy or cystic fibrosis. [5] While our understanding of CF genetics has expanded in recent decades, the clinical

manifestations of the disease remain highly heterogeneous and physicians with different specialisations are potentially involved in the treatment of CF.

This report described the 11-year-old patient whose the first manifestation of CF was nasal obstruction and signs of chronic rhinosinusitis and, finally, massive polyposis in the nasal cavity and paranasal sinuses that required surgery. The presence of allergic rhinitis and good overall condition delayed the diagnosis of CF. In recent years, improvements in diagnostic testing have allowed identification of milder phenotypes. Going to molecular analysis, our patient bears a G542X/3849+10Kb C>T genotype. In general, 3849+10Kb C>T mutation generally has a mild clinical phenotype [6, 7]. The boy did not present classical symptoms of CF. However, he had mild non-productive chronic cough with significant sinus disease, and ultimately, CT of the chest showed areas of bronchiectasis and fibrous lesions. Even though our patient had airway damage at the time of evaluation, his spirometry was normal.

In the presented case, paranasal sinus CT showed massive polyposis in the nasal cavity and paranasal sinuses. CT scan is the imaging modality of choice to identify location of the polyps and its relationship with surrounding structures [1, 8]. In patients with such hyperplasia of mucosa, it is important to rule out other entities such as meningocele, rhabdomyosarcoma, hemangioma and neuroblastoma [5]. The process of polyp formation due to chronic inflammation is reversible, so the treatment of rhinosinusitis should start very early with nasal washings with saline solution, antibiotics and local steroids. Sinonasal inhalation with dornase alfa is also permissible [9]. However, nasal polyposis in CF is often resistant to standard therapy and requires surgical intervention [4]. FESS is less invasive technique which permits to restore drainage of the paranasal sinuses and ventilation between the nose and sinus cavities and allows shorter hospital stay [1, 8]. In the post-operative period the patient has been recommended to keep on doing frequent nasal washings with saline solution. The boy's nasal obstruction improved significantly, he had no symptoms of rhinosinusitis during the three month follow-up.

Conclusion

We described a case report of a child with cystic fibrosis presenting with nasal polyposis, the manifestation was confounded by a history of seasonal allergy, which delayed the diagnosis. Our study indicates that chronic sinusitis and/or polyposis should raise the clinicians' suspicion of a potential occurrence of undiagnosed CF and require further investigation. When the diagnosis of CF is established, a regular otolaryngology examination is recommended.

We obtained written permission from the parents for the publication of this case history.

Conflict of interest

The authors declare no conflict of interest.

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