Respiratory Medicine CME (2008) 1, 185–187

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respiratory MEDICINE CME

CASE REPORT

Severe mitral regurgitation in a young female with pansinusitis and bronchiectasis

Seong Beom Kim, Jung-Ju Sir*, Bora Lee, Cheol-Hoon Kwak, Sang Min Kim, Wook-Hyun Cho, Suk-Koo Choi

Department of Internal Medicine, Cardiovascular center, Seoul Paik Hospital, Inje University, Seoul, Republic of Korea

Received 12 March 2008; accepted 11 April 2008

KEYWORDS
Primary ciliary dyskinesia;
Bronchiectasis;
Myxoid degeneration;
Mitral regurgitation

Summary
Primary ciliary dyskinesia (PCD) is a disease characterized by symptoms of upper and lower respiratory tract infections due to abnormal structure and function of cilia.
Cardiac involvement is characterized by situs inversus (Kartagener's syndrome in PCD) and other congenital cardiovascular abnormalities. We describe a 34-year-old female with a history of recurrent sinusitis and bronchiectasis but without situs inversus or other congenital cardiac anomalies in whom an association between mitral regurgitation secondary to myxoid degeneration and primary ciliary dyskinesia was suggested.

Introduction

Primary ciliary dyskinesia (PCD) is a disease characterized by symptoms of upper and lower respiratory tract infections due to abnormal structure and function of cilia. Cardiac involvement is characterized by situs inversus (Kartagener's syndrome in PCD) and other congenital cardiovascular abnormalities, including bilateral superior vena cava, inferior vena cava drainage site abnormality, Tetralogy of Fallot, transposition of the great arteries, aortic coarctation, subpulmonic stenosis, atrial isomerism, atrial septal defect, common atrium, atrioventricular septal defect, ventricular septal defect, double outlet right ventricle and left ventricular outflow tract obstruction.[1,2] We report a first case with a history of recurrent sinusitis and bronchiectasis but without situs inversus or congenital cardiac anomalies in whom an association between mitral regurgitation secondary to myxoid degeneration and PCD was suggested.

Clinical presentation

A 34-year-old woman was admitted in our institution with hemoptysis and productive cough. At the age of 17, she had undergone mitral valve replacement (MVR) using a 29-mm Carpentier-Edwards bioprosthesis (Edwards Lifesciences, Irvine, CA) for severe mitral regurgitation secondary to myxoid degeneration. At the age of 27, she required redo-MVR using a

*Corresponding author. Tel.: +82 2 2270 0011; fax: +82 2 2278 0792.
E-mail address: seo0515@medimail.co.kr (J.-J. Sir).

1755-0017/$ - see front matter © 2008 Elsevier Ltd. All rights reserved.
doi:10.1016/j.rmedc.2008.04.007

31-mm Carpentier-Edwards bioprosthesis (Edwards Lifesciences, Irvine, CA) due to prosthetic mitral valve stenosis. She had a history of nasal obstruction, rhinorrhea, and anosmia and had been treated for recurrent sinusitis and otitis media several times since her early childhood. At the age of 22, she had undergone sphenoethomoidectomy with polypectomy for chronic sinusitis with inflammatory polyp. Also, she had suffered from repeated lung infections since childhood and diagnosed as bronchiectasis at the age of 24. Her sister had also suffered from bronchiectasis and had died at 36 years of her age. She was infertile. Upon physical examination, the blood pressure was 120/80 mm Hg, and the pulse rate was estimated at 90 beats/min with irregularly irregular rhythm. She had saddle-nose deformity from collapse of the nasal bridge. Cardiovascular examination showed that the apex beat was prominent in the left fifth intercostal space 1 cm lateral to the midclavicular line. Bilateral coarse crackles with rhonchi were heard on lung auscultation. Chest X-ray demonstrated levocardia, diffuse bronchiectasis, and the gastric air bubble on the left (Figure 1A). Pulmonary function test showed severe airflow obstruction. Routine laboratory tests were normal except mild leukocytosis. Pseudomonas aeruginosa was isolated from sputum culture. A high-resolution computed tomographic scan of the chest revealed tubular bronchiectatic changes of both lobes (Figure 1B). A CT scan of the paranasal sinuses demonstrated chronic pansinusitis with polyps along the nasal septum (Figure 1C). Transthoracic echocardiography showed normally functioning prosthetic mitral valve. The mean transmitral pressure gradient was 4.0 mm Hg, and mitral valve area was 2.45 cm^2. The pulmonary systolic pressure was in the normal range. There were no other combined congenital cardiac abnormalities. As her clinical condition led to the suspicion of PCD, we performed nasal biopsy and ultrastructural analysis of the nasal cilia, and they showed abnormal ciliary structure on electron microscopy, consistent with a clinical picture of PCD (Figure 2). Bronchietasis with acute infectious exacerbation was thought to be the cause of hospital admission. After nine days of intravenous antiobiotics therapy, she gradually improved and was discharged and currently she is on regular follow-up.

Discussion

To our knowledge, this is the first case report of simultaneous occurrence of myxoid degeneration with severe regurgitation of mitral valve and PCD (without situs inversus) in an adolescent patient. In the English medical literature, there was only one case report of simultaneous occurrence of mitral valve prolapse with regurgitation and Kartagener's syndrome (PCD with situs inversus) in a pediatric patient with Down syndrome, described by Kovesi et al.[3] In 50% of the patients, PCD is associated with situs inversus. The large portion of situs abnormalities and congenital cardiovascular anomalies in patients with PCD may be explained by abnormal movement of the nodal cilia during embryogenesis.[2] At this point, it is uncertain if there is a genetic or pathophysiological link between myxoid degeneration of mitral valve and PCD.

PCD is characterized by ultrastructural and functional defects of cilia, which lead to recurrent sinusitis, otitis media and bronchiectasis. The diagnosis relies on a

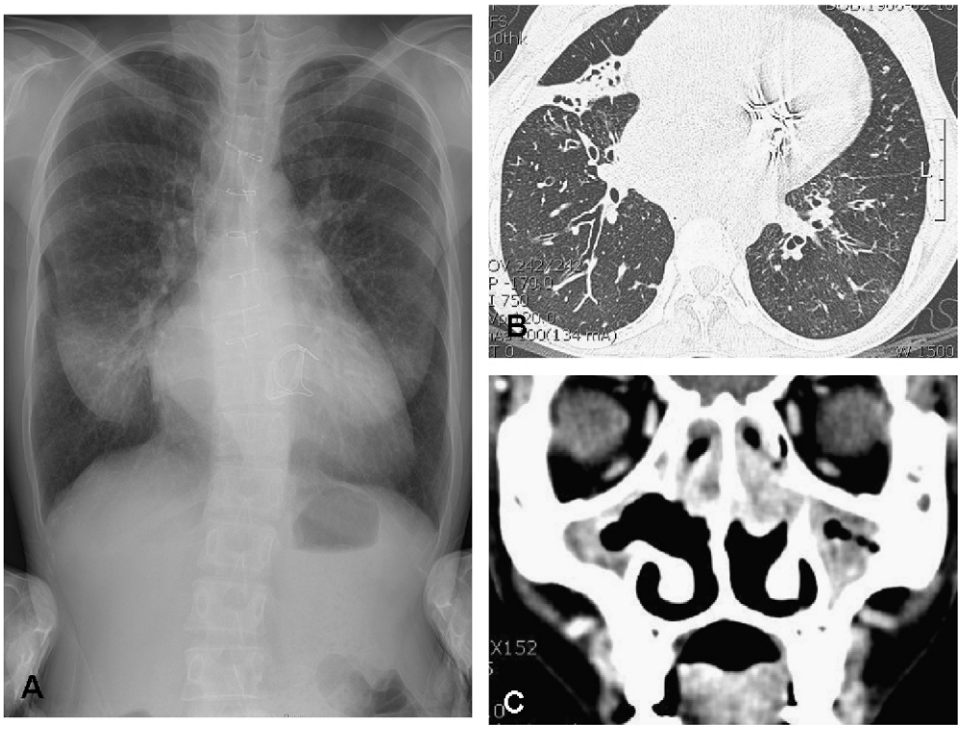

Figure 1 (A) Chest radiograph showing cardiomegaly with levocardia, normal gastric gas bubble on the left side and coarse reticular opacities through both lung fields suggestive of tubular bronchiectasis. (B) High-resolution computed tomography of the chest showing bronchiectatic changes in both lobes. (C) Computed tomographic scan of the paranasal sinuses showing chronic pansinusitis with polyps along the nasal septum.

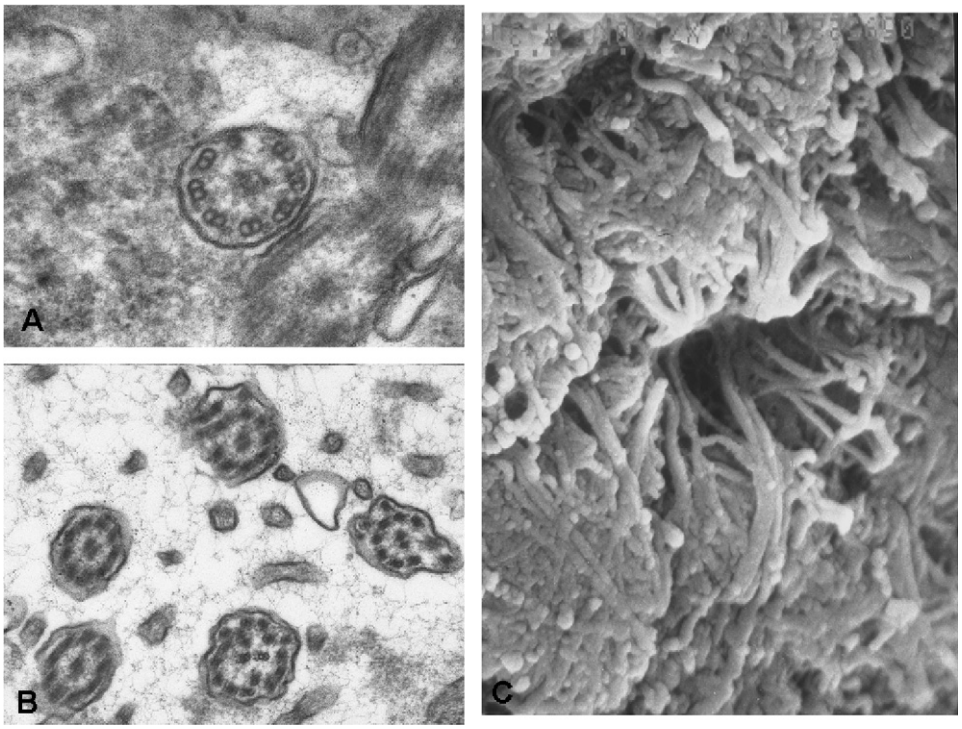

Figure 2 (A) and (B) Transmission electron micrograph of a nasal mucosal biopsy sample which demonstrates that the cilia show partial defect of outer and inner dynein arms and radial spoke. (C) Scanning electron micrograph of the cilia of nasal mucosa shows feeble appearance with variable diameter. Blunt-ended and unstraightened cilia are present.

combination of clinical evaluation and electron microscopic analysis of ciliary ultrastructure. Although most patients with PCD have symptoms from birth or early infancy, the diagnosis is often delayed.[4] Failure to diagnose PCD leads to progressive and permanent lung destruction owing to obstruction of the airways with secretions and subsequent infection, leading to bronchiectasis.[5] Therefore, clinicians should be alert to this possible association between PCD and myxoid degeneration of mitral valve, and a high index of suspicion is necessary for the diagnosis of PCD even in patients without situs inversus as in our case.

Conflict of interest statement

We declare not to have any conflict of interest as we do not have any financial and personal relationships with other people or organizations that could have inappropriately influenced our work.

Disclosure summary

The authors have nothing to declare.

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