CASE REPORT

Primary spontaneous pneumothorax in conjunction with Marfan syndrome

Yannan J Wang,¹ Elsie Negron-Rubio,² Jayanth H Keshavamurthy,³ William B Bates²

¹Department of Radiology, Augusta University Medical College of Georgia, Augusta, Georgia, USA

²Department of Radiology, Augusta University Medical College of Georgia, Augusta, Georgia, USA

³Department of Diagnostic Radiology, Augusta University Medical College of Georgia, Augusta, Georgia, USA

Correspondence to

Dr William B Bates,
wibates@augusta.edu

Accepted 8 February 2018

SUMMARY

A 25-year-old man with a history of Marfan syndrome, asthma and smoking presented with worsening dyspnoea and right-sided chest pain worsened with deep breathing after a fall 2 days prior. Diagnostic imaging revealed a spontaneous right-sided pneumothorax due to ruptured subpleural bullae in the apex of the right lung. Smaller subpleural bullae were also noted in the apex of the left lung. A chest tube was placed to reduce the right pneumothorax successfully.

BACKGROUND

Primary spontaneous pneumothorax (PSP) occurs when air enters the intrapleural space in a patient without a previous trauma or a history of lung disease. A common cause of PSP is the existence of pleural bullae due to subclinical lung disease. Tall, thin, male patients who smoke and are between the ages of 10 and 30 are at higher risk for developing PSP.¹ In this case, since the patient was previously a smoker and falls into the high-risk age group and gender, he had increased likelihood of developing PSP.

Initially, the resident on call did not comment on the bulla as the cause of pneumothorax or link the existence of bulla with the patient's medical history of Marfan syndrome, so it is important to emphasise the need to identify the cause of a PSP.

CASE PRESENTATION

A 25-year-old man presented to the emergency department with dyspnoea and right-sided chest pain worsened with deep breathing. He fell 2 days prior, and has a known history of Marfan syndrome with aortic root dilation and mitral valve prolapse. The patient also has asthma and was previously a smoker.

INVESTIGATIONS

Chest CT was obtained to determine the extent of the pneumothorax (figure 1).

DIFFERENTIAL DIAGNOSIS

Diagnostic imaging revealed a spontaneous right-sided pneumothorax caused by ruptured subpleural bullae in the apex of the right lung. Smaller subpleural bullae were also noted in the apex of the left lung (figure 1). The most likely cause of the pneumothorax was rupture of the bullae caused by the fall 2 days prior.

TREATMENT

A chest tube was placed to reduce the pneumothorax, and the patient underwent bullectomy to address the bullae found on CT.

OUTCOME AND FOLLOW-UP

The patient tolerated the surgery well and was discharged with a stable apical pneumothorax.

DISCUSSION

Studies have also shown that 5%–11% of patients with Marfan syndrome develop spontaneous pneumothorax caused by rib cage abnormalities or apical bullae due to fibrillin abnormalities that cause weakening of the connective tissue in the lungs.^{2,3} In addition, Saita *et al*⁴ observed that patients with PSP have asymmetric and flat thoraces—a clinical feature also present in most patients with Marfan syndrome (pectus excavatum), which predisposes them to the formation of subpleural bullae. Studies have also shown that the risk of PSP is higher in patients with Marfan syndrome who have bullae that are detectable on CT scan.⁵

Figure 1 Axial, coronal and sagittal views of the chest in lung windows. There is a conglomerate of moderate to large right apical bullae. A large pneumothorax is present. Large bore chest tube in place.

Learning points

- ▶ Identifying the cause of primary spontaneous pneumothorax is extremely important for optimal treatment of the patient.
- ▶ Patients with Marfan syndrome are predisposed to the formation of subpleural bullae due to the shape of their thoraces and weakening of the connective tissue in their lungs.
- ▶ Radiologically detectable bullae should be surgically addressed to prevent primary spontaneous pneumothorax.

To cite: Wang YJ, Negron-Rubio E, Keshavamurthy JH, *et al*. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-222354

Contributors JHK and ENR were involved in the care of the patient and thus contributed to the planning of the manuscript and framing of the teaching points. JHK and ENR were also involved in drafting and editing portions of the manuscript involving radiologic interpretation. WBB was involved in editing portions of the manuscript. YJW consented the patient and drafted the case presentation. YJW and JHK are the guarantors.

Funding This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

© BMJ Publishing Group Ltd (unless otherwise stated in the text of the article) 2018. All rights reserved. No commercial use is permitted unless otherwise expressly granted.

REFERENCES

- 1 Sahn SA, Heffner JE. Spontaneous pneumothorax. *N Engl J Med* 2000;342:868–74.
- 2 Neuville M, Jondeau G, Crestani B, *et al.* Manifestations respiratoires du syndrome de Marfan. *Rev Mal Respir* 2015;32:173–81.
- 3 Noppen M. Spontaneous pneumothorax: epidemiology, pathophysiology and cause. *Eur Respir Rev* 2010;19:217–9.
- 4 Saita K, Murakawa T, Kawano H, *et al.* Chest wall deformity found in patients with primary spontaneous pneumothorax. *Asian Cardiovasc Thorac Ann* 2013;21:582–7.
- 5 Karpman C, Aughenbaugh GL, Ryu JH. Pneumothorax and bullae in Marfan syndrome. *Respiration* 2011;82:219–24.

Copyright 2018 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- Submit as many cases as you like
- Enjoy fast sympathetic peer review and rapid publication of accepted articles
- Access all the published articles
- Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow