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BIRT-HOGG-DUBE SYNDROME -A CASE REPORT

Respiratory Medicine

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ABSTRACT

Birt-Hogg-Dubé syndrome is an autosomal dominant syndrome characterised by triad of multiple pulmonary cysts with increased risk of pneumothorax, cutaneous manifestations and renal neoplasms. Herein we present a case of young woman who presented with recurrent episodes of pneumothorax.

KEYWORDS

Birt-Hogg-Dubé syndrome, autosomal dominant, pulmonary cysts, pneumothorax

INTRODUCTION

Birt-Hogg-Dubé syndrome, also known as Hornstein-Knickenberg Syndrome, is an autosomal dominant syndrome characterised by triad of multiple pulmonary cysts with increased risk of pneumothorax, cutaneous manifestations and renal neoplasms.

Wide varied manifestations of Birt-Hogg-Dubé syndrome makes this genetic condition to remain often un-diagnosed. Episodes of recurrent pneumothorax, cyst or bullae on HRCT thorax, multiple skin lesions, renal cysts / lesions, and/or family history of pneumothorax are important diagnostic clues for diagnosis of BHDS.

Pulmonary cysts on imaging studies may be the initial manifestation and are often the finding to first suggest the diagnosis, highlighting the role of CT in early diagnosis. This case therefore highlights the need for a high index of clinical suspicion for inherited causes of pneumothorax in young patients.

Case Report

A 26 year old female, non-smoker, presented to emergency department with complaints of left sided chest pain and shortness of breath for 1 day, not triggered by trauma or activity. Patient was admitted and evaluated. Based on the chest x-ray and computed tomography of chest which showed left sided pneumothorax without any evidence of bullae or cysts diagnosis of primary spontaneous pneumothorax was considered. Patient was managed by passing intercostal drain in 5th left intercostal space following which patient showed improvement within 24 hours both clinically and radio-logically and was discharged from the hospital.

After 2 years, patient presented with recurrent episode of shortness of breath and left sided chest pain. During current episode no abnormality was seen on physical examination whereas on systemic examination breath sounds were found to be reduced on left side Initial blood parameters was found to be normal. Chest X-ray showed left sided pneumothorax, however computed tomography of chest this admission showed large bulla in the anterior segment of left upper lobe and few small blebs in the apical region of left upper lobe, lateral and posterior basal segment of right lower lobe.

On further enquiry, it was disclosed that patient's 1st degree relative had similar complaints of shortness of breath and chest pain 6 months back and for same complaints was evaluated and diagnosed as a case of Birt-Hogg-Dube syndrome based on genomic study which showed presence of FLCN gene mutation.

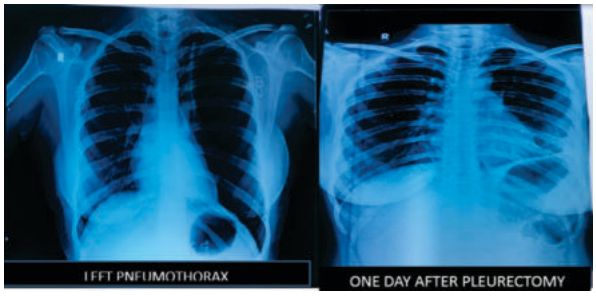
Due to financial constraints, genomic study could not be done in our patient Considering recurrent episodes of pneumothorax, family history and radiological findings a diagnosis of Birt-Hogg-Dube syndrome was made in our patient.

Our patient did not have any skin lesions, and Ultrasonography of abdomen did not show any renal cysts/tumours.

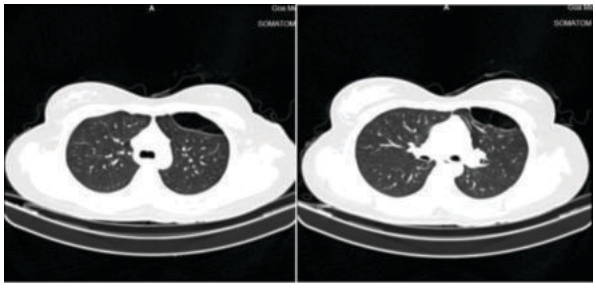
As per the guidelines given by European Birt-Hogg-Dube syndrome consortium, our patient fulfilled 2 minor criteria.

The patient was initially managed conservatively by passing intercostal drain and showed significant improvement. She was then referred to Dept. of Cardiovascular and Thoracic Surgery for further management in view of recurrent episodes of pneumothorax and CT scan showing large bulla along with multiple blebs.

Patient then underwent pleurectomy for the same. There were no post –operative complications and since patient improved she was discharged.



Chest X RAY



CT THORAX

CT scan showing large bulla with internal septations along the anterior segment of left upper lobe

DISCUSSION:

Birt-Hogg-Dubé syndrome (BHDS) was first reported in 1977 and is named after Canadian physicians Arthur R Birt, Georgina R Hogg and W James Dubé.

It is one of the rare cause of spontaneous pneumothorax which is often underdiagnosed and missed and is detected during evaluation of spontaneous pneumothorax or detected incidentally on computed tomography of chest. Positive family history is often seen without any gender predisposition.

BHDS has an autosomal dominant inheritance with deletion mutation in the folliculin (FLCN) gene located on short arm of chromosome 17. Approximately 142 unique DNA mutations of the FLCN gene which have been implicated in causation of BHDS, which results in varied manifestations. Exact function of FLCN is unknown but is thought to be an oncogene suppressor protein.

According to a metananlysis done by Muller et al, the prevalence of BHD in the general population is 1.86 (1.16, 3.00) per million, with values of 1.86 (1.02, 3.39) per million in men, and 1.88 (0.97, 3.63) per million in women

Management depends on the presentation. Cutaneous fibrofolliculomas can be reduced via surgical and laser treatment. To prevent recurrent pneumothorax pleurodesis can be useful and when possible renal tumours to be treated with nephron-sparing surgery. In our patient pleurectomy was done as a method of surgical pleurodesis. Screening of patients is recommended which includes full-body skin examination every 6-12 months for potential risk of melanoma. Annual MRI or ultrasound to screen for renal lesion every 2 yearly if there are no renal lesions and more frequently in presence of renal cysts.

Also patients are advised to avoid use of cigarette smoking, high ambient pressures, and radiation exposure. First-degree relative should undergo molecular genetic testing to look for presence of family-specific pathogenic variant.

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