Case Report

Hereditary Haemorrhagic Telangiectasia with Mitral Valve Prolapse and Straight Back Syndrome: A Rare Association

Suhas HS, Unnati Desai and Jyotsna M. Joshi

Department of Pulmonary Medicine, Topiwala National Medical College and BYL Nair Hospital, Mumbai, India

Abstract

Hereditary haemorrhagic telangiectasia (HHT), synonymously known as Osler-Weber-Rendu syndrome, is a rare autosomal inherited disorder characterised by abnormal blood vessel formation in the skin, mucous membranes, and organs including the lungs, liver and central nervous system. Straight back syndrome is characterised by the loss of normal upper thoracic spinal curvature, *i.e.* thoracic kyphosis, resulting in reduced antero-posterior diameter of thorax, often associated with cardiac murmurs on auscultaion and cardiomegaly on chest radiograph. The association of these abnormalities occurring simultaneously has been rarely reported. We present a case of HHT and mitral valve prolapse (MVP) associated with straight back syndrome. The patient also had respiratory failure. **[Indian J Chest Dis Allied Sci 2018;60:155-157]**

Key words: Hereditary haemorrhagic telangiectasia, Mitral valve prolapse, Straight back syndrome.

Introduction

Hereditary haemorrhagic telangiectasia (HHT), an autosomal dominant multi-organ vascular dysplasia, is characterised by the presence of multiple arteriovenous malformations (AVMs) that lack intervening capillaries and results in direct connections between arteries and veins. On the other hand, straight back syndrome presents with loss of normal upper thoracic spinal curvature associated with radiographic cardiomegaly and is considered a form of 'pseudo heart disease'. The pseudocardiomegaly has been attributed to the compression of the heart in the reduced antero-posterior diameter of the chest. This is also accompanied by a leftward displacement of the heart, resulting in cardiac murmurs, chest pain and tracheal compression. Mitral valve prolapse (MVP) has been reported in 64% of patients. We present a rare case of association of the three abnormalities concurrently occurring in a patient. *To the best of our knowledge, this is the first case report of this trilogy of disorders presenting simultaneously*.

Case Report

A 37-year-old female presented with complaints of dry cough without a history of substance abuse; alongwith dyspnoea on exertion (grade 1) as per modified Medical Research Council (mMRC) scale. She had no history of chest pain or palpitations in the past. However, she had aggravation of symptoms three years back during her pregnancy for which she was managed conservatively. She had recurrent episodes of epistaxis since childhood. She denied any history of haemoptysis or wheeze. However, there was family history of recurrent episodes of epistaxis for her sister.

On examination, pulse rate was 100 beats per minute regular with no radioradial delay or radio-femoral delay, blood pressure was 120/70mmHg, respiratory rate was 24 per minute, with an oxygen saturation of 86% on pulse oximetry; refractory to oxygen supplementation with the presence of orthodeoxia. There was no evidence of telengectasia in the muco-cutaneous junction. Respiratory system examination revealed a decrease in antero-posterior diameter (AP diameter) of the chest with a loss of thoracic kyphosis, resulting in a flat chest wall abnormality. Rest of the systemic examination was within normal limits.

Arterial blood gas analysis revealed Type 1 respiratory failure with partial pressure of oxygen in arterial blood (PaO_2) of 48.5mmHg. Haematological and biochemical blood investigations were within normal limits. Chest radiograph showed a well-defined lobulated homogeneous opacity in the right lower zone. Postero-anterior and lateral views showed a decreased distance measured from T8 thoracic vertebra to the line joining the T4 and T12 vertebra (Figure 1 A and B). The distance was 0.6cm indicating straight back syndrome. There was reduction in the AP diameter of the chest as diagnosed by De Leon's criteria.[2] There was loss of normal dorsal kyphosis suggestive of straight back syndrome.

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Correspondence and reprint requests: Dr Jyotsna M. Joshi, Department of Pulmonary Medicine, 2nd Floor, OPD Building, Topiwala National Medical College and BYL Nair Hospital, Dr AL Nair Road, Mumbai Central, Mumbai – 400 008, India; E-mail: drjoshijm@gmail.com

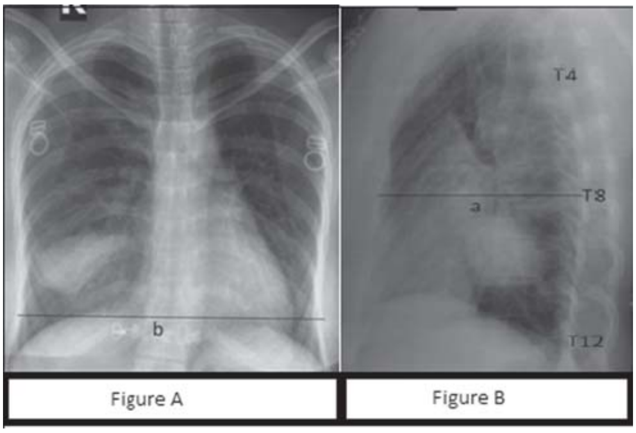

Figure 1 (A&B). Chest radiograph (A) (postero-anterior view) showing a well-defined lobulated homogeneous opacity in the right lower zone and (B) (lateral view) showing decreased distance measured from T8 thoracic vertebra to line joining the T4 and T12 vertebra.

Contrast-enhanced computerised tomography (CECT) of thorax revealed a well-defined lobulated homogeneously enhancing lesion in the right lower lobe connected to the right pulmonary veins. On the basis of radiological findings, a differential diagnosis of arteriovenous malformation (AVM) was considered; and a computerised tomography with pulmonary angiography (CTPA) was done including abdomen to look for other sites of AVM. CTPA showed a well-defined homogeneously lobulated soft tissue density lesion (6.3cm × 4.9cm × 3.9cm) in the right lower lobe with intense enhancement on pulmonary angiography with four feeding arteries. These four feeding arteries were segmental and sub-segmental branches of the right descending pulmonary artery. Early drainage of the lesion into the right middle and inferior pulmonary vein suggested AVM (Figure 2).

In addition, reduced AP diameter of thorax was confirmed and multiple ill-defined subcentimeter sized AVMs were noted in the liver and in the body of pancreas. Magnetic resonance imaging of brain and spine did not show AVMs.

Figure 2. Computed tomography with pulmonary angiography showing the right lower lobe arteriovenous malformation.

Two-dimensional echocardiography showed myxomatous mitral valve with prolapse of anterior and posterior mitral leaflet with grade 2 mitral regurgitation, mild tricuspid regurgitation and pulmonary hypertension with a pressure of 35mmHg with good ventricular function. Spirometry was done in view of dyspnoea which showed a restrictive pattern with FEV_1/FVC of 77%, FVC of 1.7L (60% of predicted value) and FEV_1 of 1.31 L (54% of predicted value).

Due to the large size of AVM; the patient was referred for embolisation. However, embolisation was not performed due to the development of thrombus within the pulmonary AVM. Therefore, the patient was initiated on anticoagulants for the same (Figure 3).

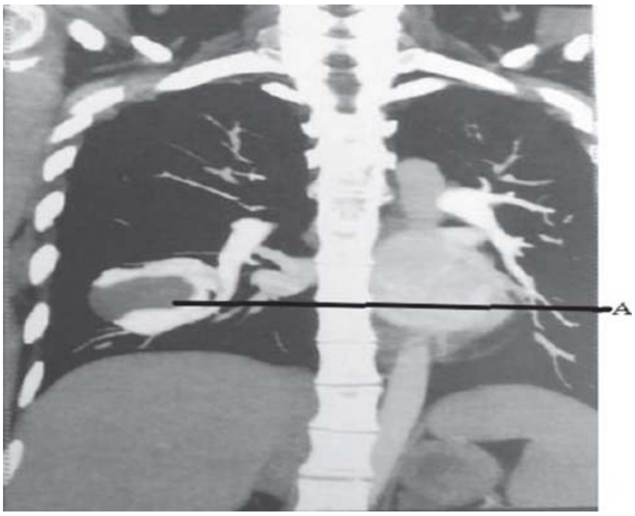

Figure 3. Coronal section of pulmonary angiography showing the presence of thrombus in pulmonary arteriovenous malformations.

On follow-up, the patient is stable on anticoagulants over one year. However, the patient decided not to undergo the embolisation procedure.

Discussion

Hereditary haemorrhagic telangiectasia, an autosomal dominant multi-organ vascular dysplasia, is characterised by the presence of multiple arteriovenous malformations (AVMs) deficient in intervening capillaries resulting in direct connections between arteries and veins.[1] The most common clinical manifestation is spontaneous and recurrent epistaxis beginning on average at the age 12 years.

The Curacao criteria published in 2000,[3] remain the mainstay of clinical diagnosis of HHT.[3] A definite diagnosis of HHT may be made in the presence of at least three separate manifestations: (1) spontaneous recurrent nose bleed, (2) muco-cutaneous telangiectasia (multiple at characteristic sites: fingertip pulps, lips, oral mucosa or tongue), (3) visceral involvement (gastrointestinal, pulmonary, hepatic, cerebral or spinal AVM) and (4) family history: a first-degree relative affected according to the above-mentioned criteria.

In our case, the patient presented with recurrent episodes of epistaxis with a positive family history of similar complaints and on routine chest radiograph, she had incidental finding of straight back syndrome. As the patient had hypoxaemia, arterial blood gas analysis was done which was suggestive of type 1 respiratory failure, refractory to oxygenation with orthodeoxia. Because of the repeated episodes of epistaxis and orthodeoxia and an opacity on chest radiography, AVM was suspected, and hence, CTPA was done which confirms the presence of AVM in the right lower lobe of the lung along with the presence of AVM in liver and pancreas.

A diagnosis of HHT was made as our patient fulfilled three criteria, as stated above. Incidentally, she also had straight back syndrome and mitral valve prolapse. Straight back syndrome, a developmental abnormality in which there is loss of normal thoracic kyphotic curvature of thoracic spine.[2,4] It is diagnosed as per De Leon's criteria as the AP diameter 'a' defined as the distance from anterior border of T8 to posterior border of sternum on lateral radiograph and the lateral diameter 'b' is defined at the level of diaphragm on frontal radiograph. Straight back syndrome is diagnosed when a/b ratio is 1/3 or less.[2]

As a consequence, the AP diameter of thorax becomes less and the heart and great vessels are compressed and shifted towards the left. Many of these patients have additional cardiac signs, and hence, earlier were postulated to have organic disease; but later it was conmfirmed that this is only a developmental abnormality and hence the name "pseudo heart disease".[5]

Subsequently, association with mitral valve prolapse was observed, as in our case.[6] The pathophysiology of this entity is idiopathic and autosomal dominance with incomplete penetrance has been proprosed as one of the aetiologies. The previous reports have always[2,6] stressed on the cardiac effects of the disease and data on respiratory involvement is limited. Pulmonary manifestations of straight back syndrome include vague chest pain, restrictive abnormality on spirometry and overlapping symptoms due to undiagnosed cardiac disease. Chest wall abnormalities like kyphoscoliosis are known causes of hypoventilation and type 2 respiratory failure.

It has already been reported that straight back syndrome can cause type 2 respiratory failure.[7] It was proposed that the ventilatory impairment due to the shape of chest and small size of the lungs were possible mechanisms.[7] The main modality of treatment for patients with HHT has been blood transfusion. In patients with recurrent epistaxis, multi-disciplinary approaches to be performed, including, electrocautery, laser, septodermoplasty, embolisation, arterial ligation, and, most recently sprayable fibrin sealant.[8] All approaches are largely palliative with variable results, requiring repeated interventions.

In recent years, the process of catheter embolisation for pulmonary AVMs was used. The enlarged vessels are closed using coils, sometimes supplemented by small balloons. Untreated AVMs may cause complications, *viz* stroke, brain abscess and many other neurological abnormalities. Hence, surgery and embolisation are often the mainstay of treatment in a case of pulmonary AVM.[9]

In conclusion, our patient presented with hereditary haemorrhagic telangiectasia characterised by the presence of multiple AVMs in various muco-cutaneous and visceral organs and incidentally was associated with straight back syndrome and mitral valve prolapse. *To the best of our knowledge, this trilogy association has not been reported so far*. Therefore, we suggest that a high index of suspicion regarding these co-morbidities should be kept in mind while approaching a case of hereditary haemorrhagic telangiectasia.

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