A case of dyspnea: respiratory failure due to pulmonary arteriovenous malformation

Lucio Brugioni,1 Chiara Catena,2 Eugenio Ferraro,2 Serena Scarabottini,2 Francesca Mori,2 Enrico Clini3

1Department of Internal Medicine and Critical Area, Policlinico of Modena; 2Department of Internal Medicine and Critical Area, University of Modena and Reggio Emilia; 3Department of Pneumology, Policlinico of Modena, Italy

ABSTRACT

Pulmonary arteriovenous malformations (PAVMs) are abnormal communications between pulmonary arteries and veins. The clinical features suggestive of PAVMs are stigmata of right-to-left shunting (dyspnea, hypoxemia, cyanosis, cerebral embolism, brain abscess), unexplained hemoptyisis, or hemothorax. We present a case of a young man who presented to the Emergency Department complaining of dyspnea, polycythemia, and persistent hypoxemia. Anglo-computed tomographic scan of the chest detected multiple PAVMs. PAVMs are uncommon in the general population, but they represent an important consideration in the differential diagnosis of common pulmonary problems, including hypoxemia, pulmonary nodules, and hemoptyisis.

Introduction

Pulmonary arteriovenous malformations (PAVMs) allow a proportion of the right ventricular volume to bypass gas exchange, filtration, and other functions of the pulmonary capillary bed. The size of the right-to-left shunt determines the degree of hypoxemia and respiratory failure. PAVMs of any size allow paradoxical emboli that may cause ischemic strokes, myocardial infarction, cerebral (brain) and peripheral abscesses. Less frequently, PAVMs may cause hemoptyisis, hemothorax, and/or maternal death in pregnancy. Due to compensatory adaptations, respiratory symptoms are frequently absent or not recognized.

The differential diagnosis of PAVMs is wide and depends on the presenting complaint. Although PAVMs are unusual causes of pulmonary nodules, dyspnea, hemoptyisis, stroke, and cerebral abscess, they should be suspected when more common etiologies for these symptoms and signs are absent.

Case Report

A 19-year-old man presented to the Emergency Department after occasional finding of polycythemia (hemoglobin 19.8 gr/dL, hematocrit 59%) and persistent hypoxemia (SaO2 <85%) even though he underwent supplemental oxygen therapy. In his medical history, only autoimmune thyroiditis is reported. The clinical exam revealed clubbing fingers; also, his grandfather presented this sign. The arterial blood gas test showed hypoxic respiratory failure with respiratory alkalosis (pH 7.45, pO2 46 mmHg, pCO 2 29 mmHg, SpO 2 83%, HCO 3 22.5 mmol/L). Metabolic panel, thoracic X-rays, electrocardiogram and echocardiography were all normal. Anglo-computed tomographic (CT) scan of the chest detected multiple PAVMs in subsegmental arteries and veins of the left lung with marked dilatation of the left pulmonary vein. Saline bubble contrast echocardiography (TTCE) showed severe PAVM with right-to-left shunt. The patient was treated with injection of embolic material by selective angiographic catheterization of the biggest PAVMs (embolotherapy). The procedure was repeated after few months to complete the closure of remaining PAVMs. The patient subsequently was in good general conditions, asymptomatic for dyspnea, with persistent mild desaturation on exertion. Other tests were made for detecting other PAVMs associated with hereditary hemorrhagic telangiectasia (HHT), in particular in...
splanchnic and cerebral districts, which were normal.

Discussion

Pulmonary arteriovenous malformations are abnormal communications between pulmonary arteries and veins. Approximately 80-95% of PAVMs are associated with HHT, also known as Osler-Weber-Rendu syndrome. PAVMs may be present from birth and have usually completed major development by adult life, although they can enlarge later in life, for example, during pregnancy or other alterations in pulmonary hemodynamics. The clinical features suggestive of PAVMs are stigmata of right-to-left shunting (dyspnea, hypoxemia, cyanosis, cerebral embolism, brain abscess), unexplained hemoptysis or hemothorax.

These include life-threatening hemorrhage, symptoms and complications from paradoxical embolization, such as stroke and brain abscess. The primary determinants of stroke and abscess risks were unrelated to severity of PAVMs. There was a marginal association between brain abscess and low oxygen saturation. For ischemic stroke, there was no association with any marker of PAVM severity, or with conventional neurovascular risk factors. Surprisingly, low mean pulmonary artery pressure was strongly associated with ischemic stroke. Although the literature prior to 2000 suggested that most PAVMs were associated with pulmonary symptoms, studies since 2000 describe pulmonary symptoms in only 20 to 65 percent (approximately 40 percent) of patients, and the remainder are asymptomatic, and they are typically found incidentally upon chest imaging. For most patients with suspected PAVMs, the initial test of choice to evaluate the presence of a right-to-left shunt is TTCE (also known as bubble echocardiography). This test has high sensitivity and so it is the reason why TTCE was recommended by the international HHT guidelines committee as the initial PAVM screening test. Although PAVMs may be clearly visible on chest radiographs, many are not, and the chest radiograph has been reported as being normal in 10 to 40% of instances where clinically significant PAVMs were present. CT is generally considered the gold standard investigation for diagnosing PAVMs and demonstrating their size and extent before therapy. This imaging modality defines PAVM number, size, nature, and suitability for embolization. The feeding artery diameter, PAVM-related symptoms, and the patient’s ability to tolerate the procedure are the most important factors for selecting patients who are candidates for treatment. When indicated, most patients are treated with embolotherapy that is recommended for first-line treatment. PAVM treatment reduces risks from paradoxical emboli and improves oxygenation, other physiological parameters, symptoms exacerbated by right-to-left shunting and hemorrhage. PAVM embolization significantly reduced ischemic stroke rate. Surgery treatment is another option in case of embolization failure.

Conclusions

PAVMs are uncommon in the general population, but they need to be considered in differential diagnosis of common pulmonary signs and symptoms such as hypoxemia, pulmonary nodules and hemoptysis. PAVMs are often associated with malformations in other body districts, which need to be evaluated in case of suspected HHT syndrome. PAVMs morbidity and mortality are related to the development of serious complications including stroke, brain abscess, chronic hypoxic respiratory failure and life-threatening hemoptysis or hemothorax.

References

