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A Very Rare Cause of Hemoptysis in an Adult Patient: Phaces Syndrome

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Introduction: Phaces syndrome is a rare neurocutaneous syndrome which is characterized by the large segmental hemangiomas of the face with one more of the following abnormalities including posterior fossa brain malformations, arterial abnormalities, cardiac defects, eye abnormalities, sternal defects and/or supraumblical raphe. Although the etiology is unknown, a strong female predominance and the deletions observed on 7q33 chromosome are reported as a possible region which provides genetic susceptibility to phenotipic expression with other confounding genetic or environmental factors. Endocrine dysfunction including pituitary abnormality and growth hormone deficiency have also been reported but pulmonary findings were not well defined in this syndrome. Herein, we report an adult patient presenting with hemoptysis which were diagnosed as PHACES syndrome.

Case Presentation: Thirty two year old female patient was admitted with 9 years of intermittan hemoptysis (daily volume of expectorated fresh blood was approximately 3-5 mL). Her complaint resolves spontaneously and quote a while after. She was otherwise asymptomatic. She had no history of smoking, occupational or environmental exposure to known organic, inorganic particles. No history of any drug use was also noted. Physical examination was normal. Large facial segmental hemangioma was observed at the right side of mandibula and oral mucosa, tongue, right brecut and arm on inspection. Pulmonary evaluation was normal. Laboratory investigations revealed normal whole blood cell count, serum biochemistry and hemostasis parameters. Chest radiography was also normal. Pulmonary function test and abdominal ultrasonography were also normal. Computerized tomography and high resolution CT indicated multifocal hemangiomatozis which begins from the right buccal localization and continues to thorax and upper anterior mediastium as well including right breast. Lung parenchymal investigation indicated a round well defined noduler lesion at the right lober lower segment which also indicated a possible vasculer lesion. After evaluating the clinical and radiological findings, the diagnosis of PHACES syndrome was confirmed by the coronal section of multidetector CT which showed sternal cleft in our patient. Then the patient was invetigated about systemic abnormalities. Cranial MRI only revealed venous angioma at right cerebellar hemisphere. No other systemic involvement including cardiac and oculer finding was present. Venous doppler ultrasonography confirmed hemangiomatosis which begins from the right side of mandibula and lies along the right shoulder, right arm and thorax.

Conclusion: Phaces syndrome is a rare cause of hemoptysis in adult patients. Cerebrovascular, cardiovascular or ocular involvement can be observed. Our patients is under routine follow up without massive hemoptysis since 7 years.

Keywords: Phaces syndrome, hemoptysis, hemanjiomatozis