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A case of Hereditary hemorrhagic telangiectasia/ Osler-Weber-Rendu syndrome diagnosed by CT Pulmonary Angiography

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ABSTRACT

Hereditary hemorrhagic telangiectasia, also known as Osler-Weber-Rendu syndrome is a rare inherited disorder which shows autosomal dominant inheritance pattern with variable sensitivity primarily affecting the blood vessels and is characterized by abnormal formation of blood vessels in skin, mucous membranes, and organs like lungs, liver and central nervous system. It is seen to involve the long arm of chromosome 9. It affects both sexes equally and is characterized by multiple telangiectasias and Arteriovenous malformation that causes direct connections between arteries and veins which bypasses intermediate capillaries. The most marked sites where telangiectasia occurs are lips, tongue, face, nasal mucosa, oral cavity and gastrointestinal tract. The main complications of telangiectasias are easily rupture and bleeding which occurs due to the thin walls, narrow tortuous course, and in close proximity to skin surface and mucus membrane. The symptoms are obvious and are classified according to the involvement of organ which includes repeated epistaxis, gastrointestinal hemorrhage, hemoptysis, skin lesions, and sometimes leads to dreadful condition like stroke. Complications do occur in Hereditary hemorrhagic telangiectasia and those includes bleeding from various sites, reduced hemoglobin, followed by stroke, pulmonary AVM, Transient Ischemic Attack (TIA), hypovolemic shock which occurs due to severe uncontrolled bleeding, which eventually leads to cardiac failure. Present case is of 40 year old male who presented with complaints of shortness of breath, epistaxis, non-productive, yellow mucoid cough sometime associated with hemoptysis, GERD, skin thickening with finger-tip ulcers and nail changes.

Keywords: Osler-Weber-Rendu syndrome, Finger-tip ulcers

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INTRODUCTION

A 40-year-old male presented to Outpatient Department (OPD) of Pulmonary Medicine with complaints of epistaxis two days back; he was having history of epistaxis for four years; shortness of breath, non-productive, yellow mucoid cough sometime associated with hemoptysis for 1 year. He also gives history of GERD. On physical examination, there was evidence of skin thickening with fingertip ulcers and nail changes. There was no any history of recent headache or trauma to head and face regions. The patient was a known case of scleroderma, and was having positive family history of epistaxis.

General examination of the patient showed telangiectatic macules in hands [Figure 1], skin thickening with fingertip ulcers and nail changes. Patient was conscious, oriented to time, place and person and was afebrile.

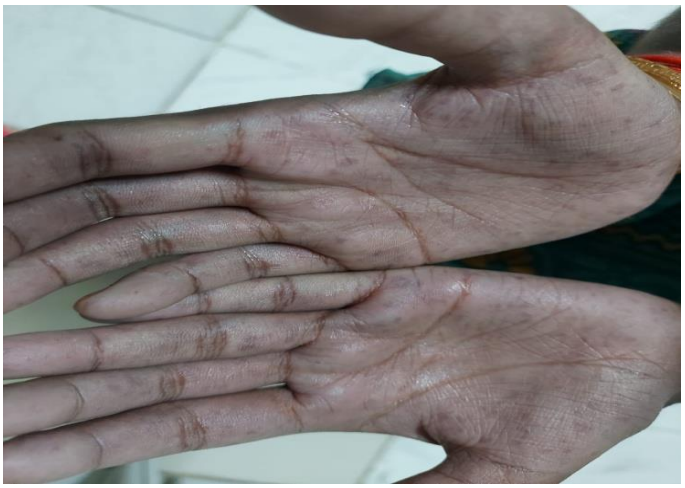

Figure 1: Shows telangiectatic macules in hands.

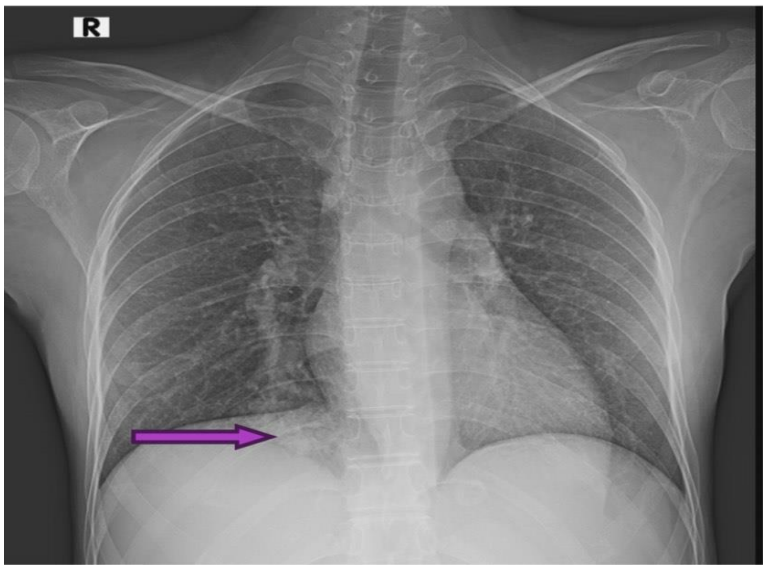

Figure 2: Chest X-Ray image showing a non-specific soft tissue mass likely a dilated pulmonary vessel (violet arrowhead).

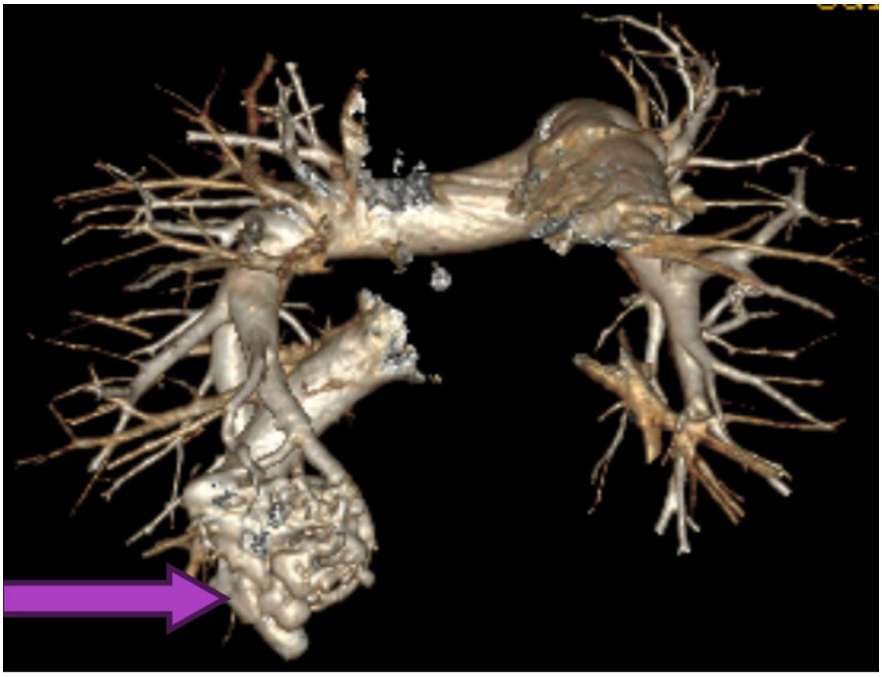

Figure 3: Computed Tomography 3-D Volume Pulmonary Angiography image showing Right lower lobe pulmonary Arterio-venous malformation and small left upper lobe pulmonary Arterio-venous fistula (violet arrowhead).

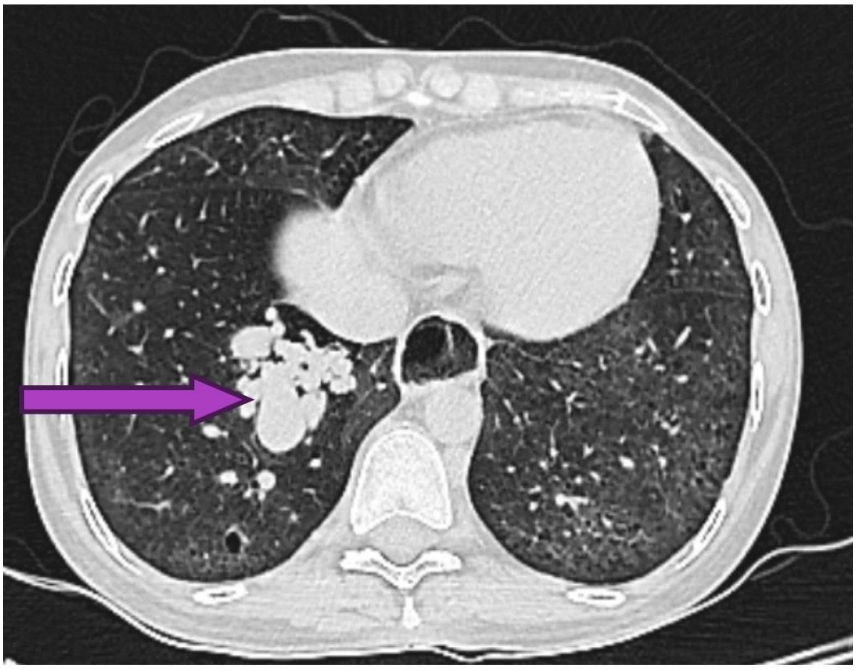

Figure 4: Axial Computed Tomography Contrast Pulmonary Angiography, Lung window image showing Right lower lobe pulmonary Arterio-venous malformation (violet arrowhead).

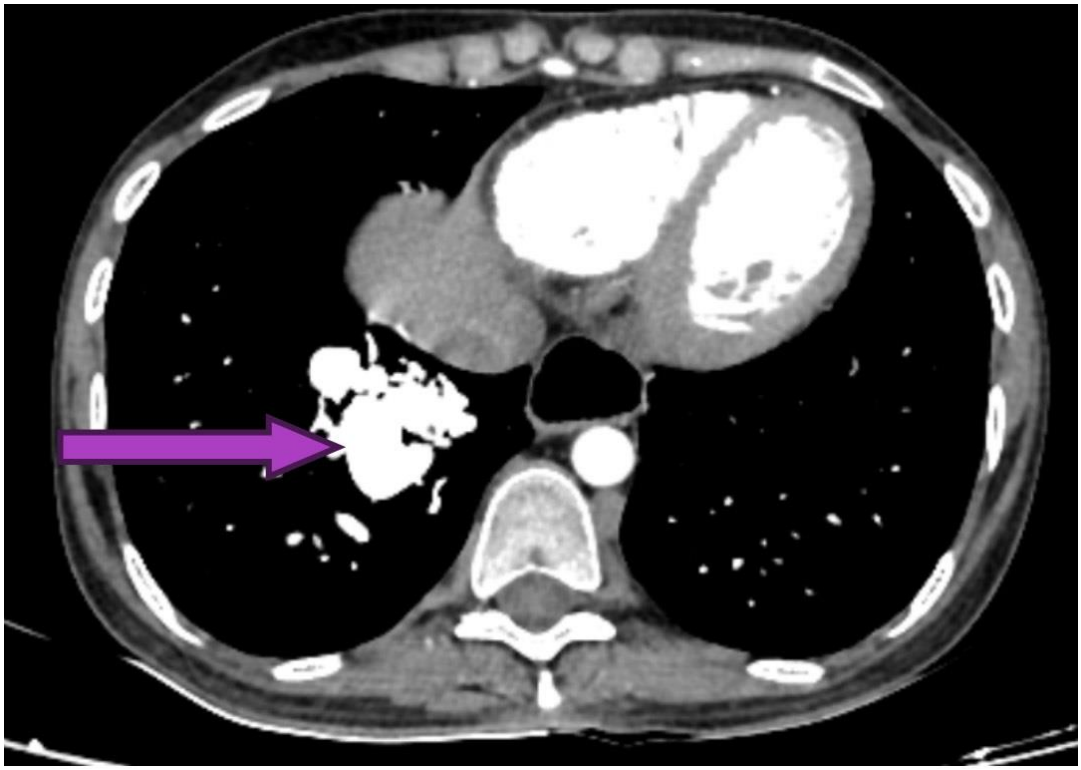

Figure 5: Axial Computed Tomography Contrast Pulmonary Angiography image showing Right lower lobe pulmonary Arterio-venous malformation (violet arrowhead).

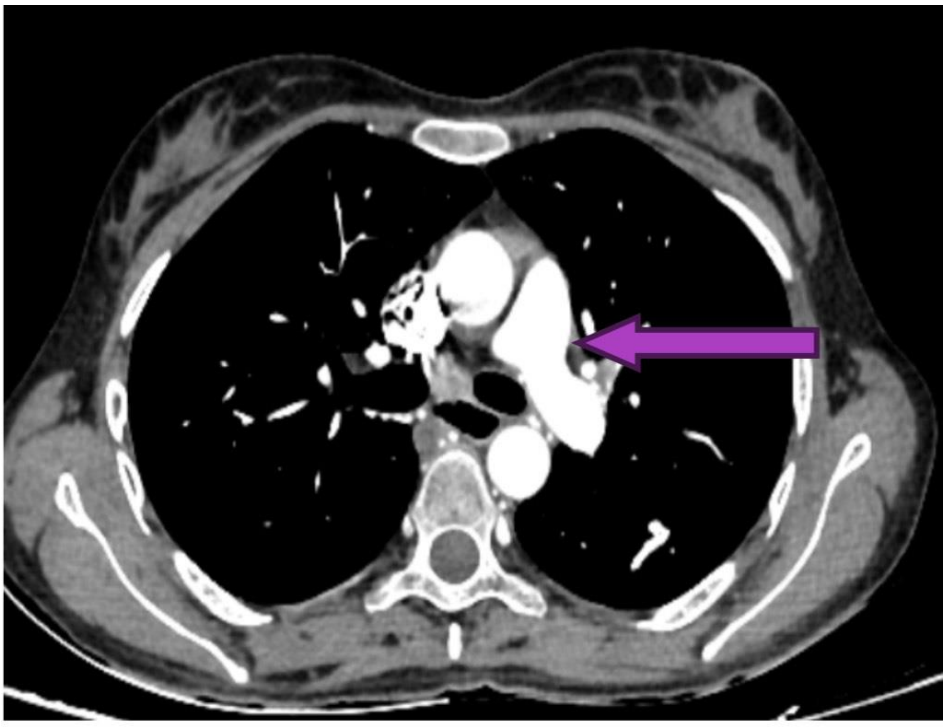

Figure 6: Axial Computed Tomography Contrast Pulmonary Angiography image showing small left upper lobe pulmonary Arterio-venous fistula (violet arrowhead).

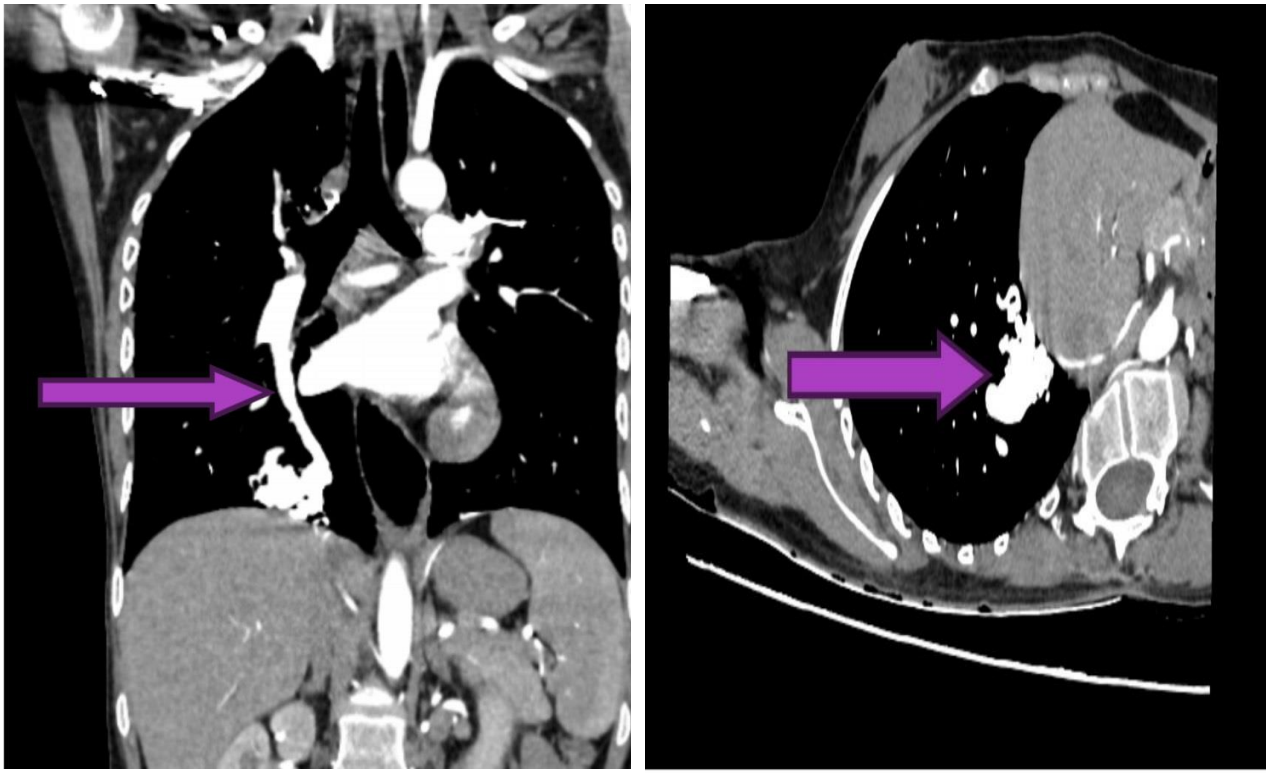

Figure 7 and 8: Computed Tomography Contrast Pulmonary Angiography coronal and axial image showing CELIAC ARTERY SUPPLY to Pulmonary AVM (violet arrowhead).

The final diagnosis of Osler-Weber-Rendu syndrome was made.

The most unique point of this case was Right pulmonary arterio-venous malformation (AVM) with systemic supply from celiac artery was made in a patient with Osler-Weber-Rendu syndrome.

DISCUSSION:

Although genetic analysis was not done in this case, but out of 4 CURACAO CLINICAL CRITERIA, this patient fulfilled all the 4 criteria for the diagnosis of Osler-Weber-Rendu syndrome.

- History of Recurrent nose bleeds.
- Telangiectatic macules in hands.
- Computed Tomography Contrast Pulmonary Angiography image showing Right lower lobe pulmonary Arterio-venous malformation.
- The patient was a known case of scleroderma, and was having positive family history of epistaxis.

Differential diagnosis includes Phakomatoses with cutaneous and visceral involvement, such as RASA1 mutation, Adams Oliver syndrome (scalp and limb defects with cutaneous

telangiectasias and visceral AVMs) and Wyburn Mason syndrome (Retino-Encephalo-Facial angiomatosis).

Management of these patients depends upon their clinical presentation. Most of them require supportive care in the form of oral iron supplements, whereas blood transfusion is given to patients with severe bleeding. Epistaxis is managed by local tamponade as in the present case. However, liver transplant is the treatment of choice in patients with hepatic HHT, with severe organ insufficiency.

CONCLUSION:

Hereditary hemorrhagic telangiectasia/ Osler-Weber-Rendu syndrome is clinico-radiological diagnosis, in which MDCT plays an essential diagnostic tool to detect pulmonary Arterio-venous malformation and pulmonary Arterio-venous fistula.

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