American Journal of Medical Case Reports, 2023, Vol. 11, No. 4, 71-73
Available online at http://pubs.sciepub.com/ajmcr/11/4/2
Published by Science and Education Publishing
DOI:10.12691/ajmcr-11-4-2

Case Report: Diagnosis of Hereditary Hemorrhagic Telangiectasia (Osler Weber Rendue Syndrome) in a 53-year-old Female Presented with Hypoxia

Lamees Khalil*, Layth Al-Karaja, Adham Itbaisha, Doaa Tarabieh, Nouraldin Hashlamon, Tumodir Abdallah, Rami J. Sweity

Al-Quds University, College of Medicine, Palestine
*Corresponding author: Lamees.ikhlayeel@gmail.com

Received February 22, 2023; Revised March 27, 2023; Accepted April 07, 2023

Abstract **Background**: Rendúe Osler Weber Syndrome is a vascular disease inherited as an autosomal dominant pattern. The most frequent complaints are epistaxis, gastrointestinal bleeding, iron deficiency anemia, and recognizable mucocutaneous telangiectasia. Also, may have Visceral arteriovenous malformations (AVMs) affect the Pulmonary, hepatic, and/or cerebral circulations; symptoms begin in childhood with an average age of 12 years. diagnosis is based on clinical presentation and confirmed by genetic testing. **Case presentation**: A 53-year-old female known case of polycythemia comes complaining of shortness of breath, severe hypoxemia (<70%), cyanosis in her lips, and epistaxis. On examination, she had telangiectasia in her face, lips, tongue, and neck. Chest CT showed pulmonary arteriovenous malformations at the lower lobes, and abdomen CT showed dilated and tortuous portal system. **Conclusion**: Clinicians need to have a low threshold of suspicion to diagnose ROW, after diagnosis, ROW needs a comprehensive and multidisciplinary team for proper management.

Keywords: *hereditary hemorrhagic telangiectasia, Osler Weber Rendue Syndrome, AV malformations*

Cite This Article: Lamees Khalil, Layth Al-Karaja, Adham Itbaisha, Doaa Tarabieh, Nouraldin Hashlamon, Tumodir Abdallah, and Rami J. Sweity, "Case Report: Diagnosis of Hereditary Hemorrhagic Telangiectasia (Osler Weber Rendue Syndrome) in a 53-year-old Female Presented with Hypoxia." *American Journal of Medical Case Reports*, vol. 11, no. 4 (2023): 71-73. doi: 10.12691/ajmcr-11-4-2.

1. Introduction

Hereditary hemorrhagic telangiectasia, commonly known as Osler Weber Rendu syndrome, is characterized by tiny clusters of dilated capillaries dispersed over the skin and mucous membranes. This condition is autosomal dominant and occurs with an estimated frequency of 1-20 cases/100,000 [1].

Hereditary hemorrhagic telangiectasia is characterized by multi-systemic vascular lesions, known as telangiectasias, and visceral arteriovenous malformations.

The patient experiences a great deal of distress on a daily basis as a result of these telangiectasias, which are small arterio-venous abnormalities that regularly bleed. Typical patient symptoms include nosebleeds, GI bleeding, and anemia due to iron deficiency.

HHT can be diagnosed by genetic testing with four Curaçao clinical criteria listed below [2] recurrent nosebleeds, telangiectasias on the skin and mucous membranes, visceral arteriovenous malformations, and first-degree relative.

Patients with this condition run the risk of developing potentially fatal consequences from arteriovenous malformations in several organ systems. These individuals must have the necessary diagnostic testing in order to avoid undesirable complications.

Worth to mention that the usual presenting age is during childhood. Here we reported a female patient 53 years old presented with hypoxia and AVM.

2. Presentation

A 53-year-old female patient with a known history of polycythemia for ten years, presented with shortness of breath and severe hypoxemia down to 70%. Over the past 10 years, she was complaining of cyanosis in her lips and a few episodes of epistaxis. There was no history of Diabetes Meletus, Hypertension, hemoptysis, chest pain, headache, liver disease, or infection. On examination, she had telangiectasia in her face, lips, tongue, and neck.

Radiological findings

Chest X-ray showed a normal-sized heart with partial consolidation of the left lower lobe of the lung and a 3.3 x 2.7 well-defined mass in the lateral aspect of the right lower lobe.

Doppler and m-mood echocardiography examinations were normal. Chest computerized tomography (CT) showed a large complex lesion composed of serpiginous tubular structures in both lungs' lower lobes (about four on

the right and six on the left). Their size range between 2 and 5 cm, suggestive of arteriovenous malformations as seen in Figure 1.

CT scan of the abdomen showed dilated and tortuous upper abdominal vessels (Portal vein, splenic vein, and hepatic artery) as seen in Figure 2.

Allele-specific quantitative real-time PCR is done to detect V617F mutation within the JAK2 gene, which gave negative results. As well as Philadelphia chromosome test was negative.

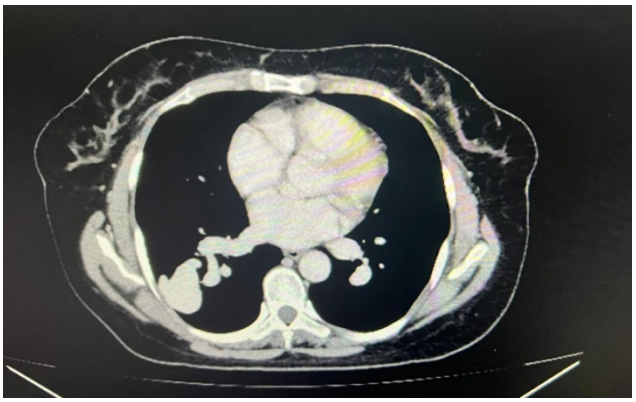

Figure 1. There is large complex lesion composed of serpiginous tubular structures in both lungs' lower lobes (about four on the right and six on the left). Their size range between 2 and 5 cm, suggestive of arteriovenous malformations

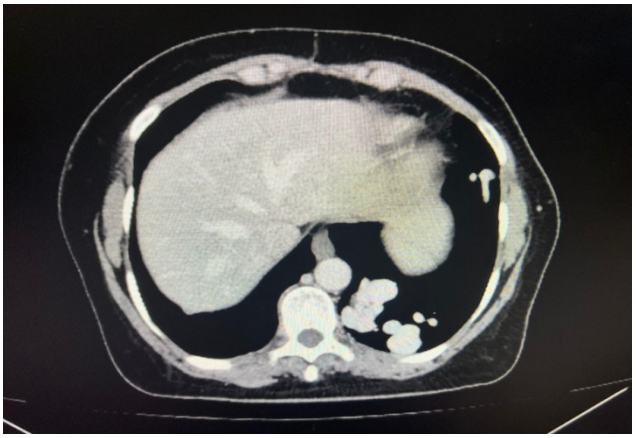

Figure 2. There are dilated and tortuous of upper abdominal vessels (Portal vein, splenic vein, and hepatic artery)

3. Discussion

Hereditary hemorrhagic telangiectasia is a vascular genetic condition with autosomal dominant and variable penetrance. Telangiectasia, which promotes mucocutaneous hemorrhage, is the most frequent pathological finding. AVMs seen in the internal organs are the second most typical finding. Ninety percent of patients experience bouts of epistaxis [3]. Three or more out of four criteria named (curaçao criteria) must be met to make the diagnosis include visceral telangiectasias/AVMs, HHT in a first-degree relative, spontaneous recurrent epistaxis, and distinctive mucocutaneous telangiectasias [5].

HHT has a variable age of onset, with an average of 12 years for epistaxis and 90% of patients before the age of 30. Telangiectasia tends to have a later onset, with initial signs appearing before the age of 20 and roughly two-thirds of patients before the age of 40, [7] although our patient appears to have a later onset than usual.

AV malformation affects many visceral organs throughout the body, frequently in the lung (50%), liver (30-70%), and brain (10%) which is an abnormal connection between arteries and vein bypass capillaries. AVM in the lung can be single or diffuse [1], as we described in our case which we found 4 AVM in the lower right lobe and 6 in the left lower lobe, it can cause a variety of clinical symptoms by bypassing the lung's filtering function and oxygen absorption capacity, resulting in right-to-left shunting. Even while PAVMs may stay asymptomatic, symptomatic people frequently exhibit respiratory symptoms, exercise intolerance, and cyanosis, the degree of the right-to-left shunt controls the severity of symptoms which may lead to clubbing and polycythemia [3]. Pulmonary hypertension is an uncommon complication of HHT, either due to high cardiac output as the result of Av shunting in the liver or due PAVMs. The gold standard for diagnosing is right cardiac catheterization. Additionally, echocardiography serves an essential role [5]. The significant and serious effects of PAVMs, which occur more in younger patients such as a transient ischemic attack, and embolic stroke, in addition to systemic severe infections, and infrequently substantial hemoptysis or hemothorax, may reveal the diagnosis [3]. However, early detection and treatment are crucial as the initial symptom may occasionally be fatal, and the characteristic findings on CT with contrast are mandatory for diagnosis. The likelihood of systemic issues and the severity of right-to-left shunting is significantly reduced with the treatment of PAVMs based on transcatheter embolization of the feeding artery. Long-term follow-up is indicated due to the tendency of treated PAVMs to recanalize and the potential for new or enlarged untreated PAVMs to develop HHT patients must be considered for routine PAVM screening via chest computed tomography or contrast echocardiography (followed by an ante-posterior chest radiograph) [4].

The symptoms of liver AVM depend on the location of the blood vessels connections, causing a significant amount of blood to bypass the body's organs, which can result in heart failure, however, if the connection occurs between the portal vein and blood vessels, portal hypertension can develop. Doppler ultrasonography of the liver is the most reliable first screening test and has a very good sensitivity for detecting lesions in the liver. A contrast-enhanced CT scan can be utilized if necessary to further describe AVMs [6].

4. Conclusion

In light of the Limited abilities and resources in developing countries, early detection of such rare diseases is essential. Although ROW is a rare condition, clinicians need to have a low threshold of suspicion to diagnose it, especially in the case of secondary polycythemia, after diagnosis, ROW needs a comprehensive and multidisciplinary team for proper management.

References

[1] Dendle C, Hill M, Fong K. Imaging of pulmonary arteriovenous malformations. Cardiovasc Intervent Radiol. 2012 Jun; 35(3): 584-93.

[2] Liu D, Li S, Chen Z, et al. Pulmonary arteriovenous malformations in a patient with hereditary hemorrhagic telangiectasia: a case report. Radiology. 2021 Dec; 298(3): e487.

[3] Ergul Y, Nisli K, Dindar A. Pulmonary arteriovenous malformation associated with Osler-Weber-Rendu syndrome. The Free Library. 2011 Sep 1. Available from: https://www.thefreelibrary.com/Pulmonary arteriovenous malformation associated with...-a0351949269 (accessed January 26, 20).

[4] Cottin V, Dupuis-Girod S, Lesca G, Cordier JF. Pulmonary vascular manifestations of hereditary hemorrhagic telangiectasia (rendu-osler disease). Respiration. 2007; 74(4): 361-78.

[5] Nikolaou I, Rafailidis V, Kartas A, Kouskouras K, Giannakoulas G. A case of pulmonary arteriovenous malformation in the setting of Rendu Osler Weber syndrome. Radiology Case Reports. 2020; 15: 12.024.

[6] Buscarini E, Plauchu H, Garcia Tsao G, et al. Liver involvement in hereditary hemorrhagic telangiectasia: consensus recommendations. Liver Int. 2006 Nov; 26(9): 1040-6.

[7] McDonald J, Bayrak-Toydemir P, Pyeritz R. Hereditary hemorrhagic telangiectasia: an overview of diagnosis, management, and pathogenesis. Genet Med. 2011; 13: 607-616.