OSLER WEBER RENDU SYNDROME: CASE REPORT

HELAINE BUENO MORAES[1,2]; ROMULO BRAGA PIRES[1,3]; JULIANE HONDA GOMES[2,4]; CAROLINA RODRIGUES COSTA[1]; STÉPHANE LIMA RABAHI[1]

ABSTRACT

Osler Weber Rendu syndrome or Hereditary Hemorrhagic Telangiectasia is a rare autosomal dominant genetic disorder. The most common clinical manifestation is epistaxis, but it can also present with melena and dyspnea. The diagnosis can be made with the criteria of Curaçao, which are four, namely: recurrent noseblreds, telagiectasias, arteriovenous malformations and family history; three positive criteria confirm the diagnosis. Objective: To present a case of a female patient, diagnosed at age 53 with Osler Weber Rendu syndrome. Methodology: Collection of data from the medical records of the selected patient diagnosed with Osler Weber Rendu syndrome. Information was collected, clinical, exams performed, the therapy of choice and outcome. Articles were searched for bibliographic review in the Virtual Health Library (VHL), LILACS, PubMed. Conclusion: The case report contributed to the discussion and warning about the clinical manifestations of a rare syndrome, but which can be present in patients from different medical specialties, since its main signs and symptoms affect several systems of the human body.

KEYWORDS: RENDU-OSLER-WEBER SYNDROME, HEREDITARY HEMORRHAGIC TELANGIECTASIA, ARTERIOVENOUS MALFORMATIONS, EPISTAXIS, AUTOSOMA

INTRODUCTION

Osler Weber Rendu Syndrome or Hereditary Hemorrhagic Telangiectasia (HHT) is a rare systemic fibromuscular dysplasia, whose basic defect is an alteration of the elastic lamina and muscular layer of the blood vessel wall, which makes them more vulnerable to trauma and spontaneous ruptures[1,2].

The disease has an autosomal dominant transmission, although in about 20% of cases there is no family history. Its incidence in the population is 1-2/100,000 and it has a homogeneous distribution between race and sex.[3]

The diagnosis is made according to the Curaçao criteria: telangiectasias on the face, hands and oral cavity; recurrent epistaxis; arteriovenous malformations with visceral involvement; family history. The diagnosis is confirmed in the presence of at least 3 of these manifestations.[4]

Otorhinolaryngological manifestations are the most frequent, with recurrent epistaxis being the main one. Blood vessels in other regions may also be affected, especially the lungs, brain, skin and gastrointestinal tract1,[4-5]. The most common bleeding from this pathology is non-traumatic epistaxis, which affects about 50% of patients before twenty years of age and approximately all of them throughout their lives (78% to 96%)6; gastrointestinal bleeding, recurrent in 33% of patients, mainly after the fourth decade of life7; and pulmonary and cerebral bleeding, with a 0.5% chance of bleeding per year[8,9].

The present study presents a case of a 56-year-old patient who was diagnosed with Osler Weber Rendu syndrome at the age of 53, but who, since the age of 42, had presented signs and symptoms suggestive of the syndrome, but had not been diagnosed. Thus, this case report aims to alert physicians from different specialties about the importance of diagnosing this syndrome to contribute to the quality of life of patients by reducing and treating their decompensations.

METHODOLOGY

It consists of a case report, with data collection from the medical records of the selected patient diagnosed with Osler Weber Rendu syndrome. Information was collected, clinical examinations performed, the therapy of choice and outcome.

Articles were searched for bibliographic review in the Virtual Health Library (VHL), LILACS, PubMed and Scopus.

The study followed the ethical guidelines for the development of research with human beings, in particular what is recommended in resolution 466/2012 of the National Health Council. Obtaining the patient's informed consent form.

CASE REPORT

Female patient, 56 years old was admitted to the gastroenterology service of the Hospital Geral de Goiânia (HGG) in May 2023 to undergo an argon colonoscopy due to chronic melena and anemia with frequent need to re-

1. Universidade Federal do Tocantins
2. Universidade Evangélica de Goiás
3. Hospital de Urgências de Goiás
4. Centro Universitário Atenas- MG

ADDRESS
HELAINE BUENO MORAES
Rua 222, n 15, Setor Leste Universitário, Residencial Solar Botafogo, Goiânia- GO.
CEP: 74643090
E-mail: helainebuenodmoraes@gmail.com

ceive blood transfusions.

The patient reports that at the age of 42 she had a stroke that evolved with mild motor sequelae. At the age of 53, she presented dyspnea, orthopnea and edema of the lower limbs that had lasted for 6 months, accompanied by anemia and urinary tract infection.

The patient was diagnosed with Osler Weber Rendu syndrome, as she had telangiectasias in the gastrointestinal tract seen by upper digestive endoscopy and colonoscopy. In addition to this, the pulmonary arteriovenous malformations evidenced by angiotomography corroborated the diagnosis of the syndrome. The telangiectasias justified the episodes of melena and, consequently, the anemia. Arteriovenous malformations (AVM) justified the picture of decompensated heart failure secondary to pulmonary hypertension.

Chest angiotomography performed in 2021 showed: Arteriovenous malformation located predominantly in the upper left lobe, measuring approximately 63 x 38 mm, nourished by the left lower lobe arterial branch and drained by the ipsilateral superior pulmonary vein. Note also another lesion with similar characteristics in the periphery of the left upper lobe, measuring 10 x 9 mm, nourished by a segmental arterial branch and draining into the interior of the malformation described above. Pulmonary arterial trunk with preserved caliber. Heart dimensions and anatomical configuration. There were no signs of pleural or pericardial effusion. Lung parenchyma with usual attenuation, without evidence of consolidations. Trachea, main and lobar bronchi patent, without significant parietal thickening. Absence of mediastinal lymph node enlargement. Musculoskeletal structures of the rib cage with a preserved appearance. Since then, the patient has been followed up by the thoracic surgery team to perform a possible lobectomy or pneumonectomy to correct the arteriovenous malformation.

In May 2023, the patient was admitted to the Emergency Care Unit with syncope, melena and a hemoglobin of 3.5, at the time a blood transfusion was performed and the patient was referred to the HGG for an argon colonoscopy by the gastroenterology team.

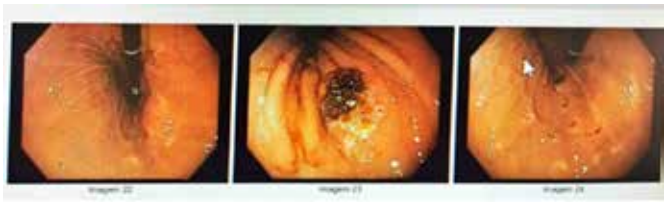
Colonoscopy and endoscopy were performed, both with argon application. The exams carried out during this hospitalization showed, colonoscopy: angiectasia in the cecum without signs of active bleeding, opting for ablative therapy with application of argon plasma. No active or recent bleeding was observed in the studied path. Bloody secretion with loose clots in the terminal ileum was visualized, suggestive of upper digestive bleeding.

At upper digestive endoscopy (figures 1 and 2) gastric and bulboduodenal vascular ectasias were seen. Hemostasis was performed by electrocoagulation with argon plasma, visualization of previous electrocoagulation scars in the gastric body

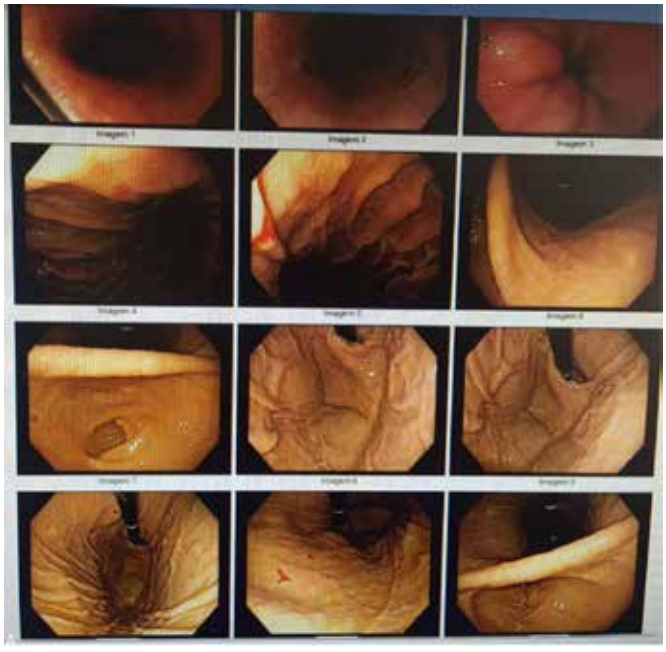
Cervicothoracic arteriography showed a patent left pulmonary artery and descending branch, with the formation of a large mass with rapid emptying of contrast, suggesting arteriovenous malformation. Malformation communication diameter ranging from 13 to 18 mm. Arteriovenous malformation in the left lung with pulmonary hypertension.

The chest angiotomography (figures 4 and 5) showed an arteriovenous malformation involving the apicoposterior and superior lingular segments of the left upper lobe, the arterial supply coming mainly from the left interlobar artery but also on a smaller scale from branches of the superior lingular segment. Venous drainage is through the left superior pulmonary vein. Lesion measuring approximately 70 x 49 mm. In the chest X-ray (figure 3), a radiopaque image can be seen in the left hemithorax, shown by the arrow.

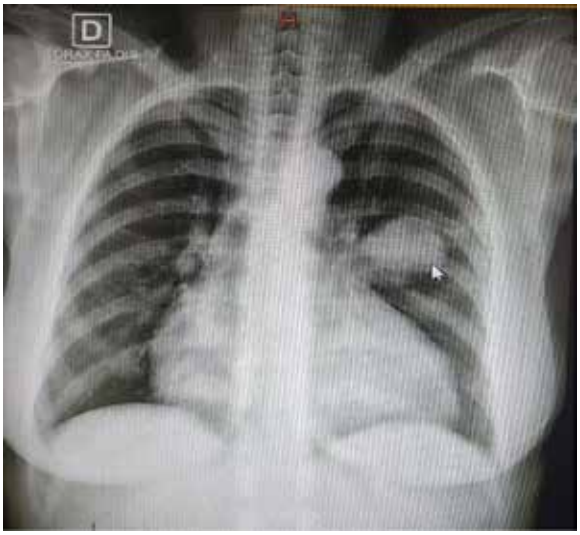
The patient evolved well and did not need new transfusions. During discussions with the multidisciplinary team, the conclusion was reached that pulmonary AVM should be evaluated during hospitalization by the vascular surgery and thoracic surgery team that opted for an elective approach to the arteriovenous malformation.



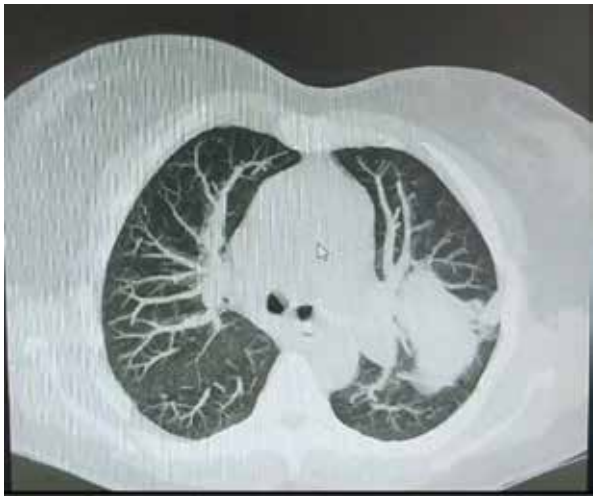
MORAES. FIGURE 1



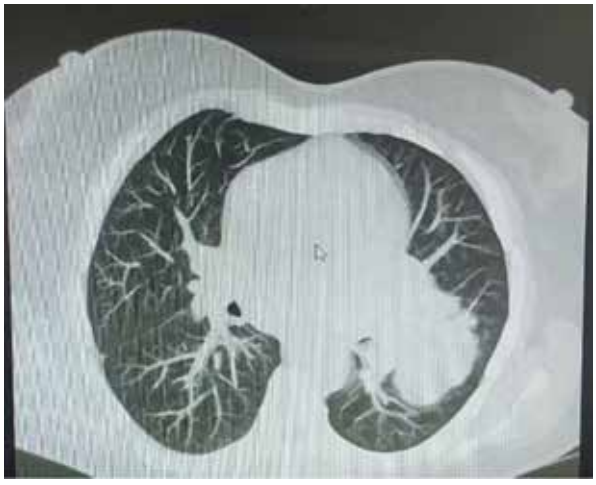
MORAES. FIGURE 2



MORAES. FIGURE 3



MORAES. FIGURE 4



MORAES. FIGURE 5

DISCUSSION

Osler Weber Rendu syndrome, or hereditary hemorrhagic telangiectasia, can be diagnosed using a probability score defined and presented in 2000 by the Scientific Advisory Board of the THH Foundation International, called the Curation criteria.[10] These criteria facilitate the recognition of clinical findings that are less common than epistaxis, which is the main manifestation of the disease in affected individuals,[11] and allow early recognition in individuals with less classic but potentially serious manifestations, such as pulmonary AVMs.

Based on these criteria,[10] the diagnosis can be definitive (when three criteria are present); possible, (if two criteria are present); or suspected (if less than two criteria are present). The criteria are: 1) presence of epistaxis (spontaneous and on more than one occasion); 2) presence of visceral lesions (gastrointestinal telangiectasia or pulmonary, hepatic, cerebral or spinal vascular malformation); 3) presence of mucocutaneous telangiectasia in a typical location; and 4) first-degree family history (or presence of the genetic mutation). In families with individuals with HHT, the diagnosis can be made from the findings of two sites with visceral lesions.[10]

The presence of chest X-ray findings compatible with pulmonary AVMs is the gold standard for diagnosing these malformations. The most common radiological presentation is the presence of well-defined peripheral nodules. The use of intravenous contrast is not mandatory, but it may allow a better definition of the angioarchitecture of the pulmonary AVM to plan endovascular therapy.[12]

Upper digestive endoscopic therapy is indicated in every patient with suspected upper digestive hemorrhage or in cases of patients with HHT when iron replacement is not enough to contain the anemia[13]. Endoscopy has potential for definitive treatment. In cases of intestinal angiectasia, therapy with argon plasma is indicated as the gold standard [13].

Embolization is the standard of care for pulmonary AVMs, 14,15 with substantial improvement in oxygenation and reduced risk of embolic events. [16.17]

Performing a lobectomy or segmentectomy is restricted to cases of complex or multiple pulmonary AVMs, when catheter embolization is not possible. [18]

FINAL CONSIDERATIONS

Osler Weber Rendu syndrome or hereditary hemorrhagic telangiectasia is rare, but must be part of the differential diagnosis of several signs and symptoms, including: epistaxis, melena, dyspnea, orthoodexia, platypnea, syncope and telangiectasia.

The patient in the present case already presented complications of the syndrome at the age of 42, however, she received the diagnosis only at the age of 53. Upon admission to the HGG service, the digestive hemorrhage was

approached through endoscopy and colonoscopy, both with argon injection. The pulmonary arteriovenous malformation was discussed by the vascular and thoracic surgery teams and the teams decided to study the best approach and schedule the procedure on an elective basis.

Thus, this case report alerts physicians from different specialties, whether clinical or surgical, about the importance of diagnosing this rare but serious syndrome that can lead to early mortality and morbidity.

REFERENCES

1. Rapoport PG, Uvo IP, Costa KS, Cecatto SB, Garcia RID. Síndrome de Rendu-Osler-Weber: tratamento clínico e cirúrgico. Rev Bras Otorrinolaringol 2003;694:577-80.
2. Maudonnet EN, Gomes CC, Sakano E. Telangiectasia Hemorrágica Hereditária (Doença de Rendu-Osler-Weber): um diagnóstico otorrinolaringológico. Rev Bras Otorrinolaringol 2000;662:172-80.
3. Pau H, Carney AS, Murty GE. Hereditary haemorrhagic telangiectasia (Osler-Weber-Rendu syndrome): otorhinolaryngological manifestation. Clin Otolaryngol 2001;26:93-8.
4. Fuchizaki U, Miyamori H, Kitagawa S, Kaneko S, Kobayashi K. Hereditary Haemorrhagic Telangiectasia (Rendu-Osler-Weber Disease) Lancet 2003;362:1490-4.
5. Haitjema T, Westermann CJJ, Overtoom TTC, Timmer R, Disch F, Mauser H, Lammers JWJ. Hereditary Hemorrhagic Telangiectasia (Osler-weber-Rendu Disease). Arch Intern Med 1996;56(8):714-9.
6. Pau H, Carney AS, Murty GE. Hereditary haemorrhagic telangiectasia (Osler-Weber-Rendu syndrome): otorhinolaryngological manifestations. Clin Otolaryngol Allied Sci. 2001;26(2):93-8.
7. Plauchu H, Chadarévian JP, Bideau A, Robert JM. Age-related clinical profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population. Am J Med Genet. 1989;32(3):291-7.
8. Kjeldsen AD, Kjeldsen J. Gastrointestinal bleeding in patients with hereditary hemorrhagic telangiectasia. Am J Gastroenterol. 2000;95(2):415-8.
9. Wong HH, Chan RP, Klatt R, Faughnan ME. Malformações arteriovenosas pulmonares idiopáticas: características clínicas e de imagem. Eur Respir J. 2011;38(2):368-375. https://doi.org/10.1183/09031936.00075110
10. Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, et al. Critérios diagnósticos para telangiectasia hemorrágica hereditária (síndrome de Rendu-Osler-Weber). Am J Med Genet. 2000;91(1):66-67. https://doi.org/10.1002/(SICI)1096-8628(20000306)91:1<66::AID-AJMG12>3.0.CO;2-P
11. dos Santos JW, Dalcin TC, Neves KR, Mann KC, Pretto GL, Bertolazi AN. Telangiectasia hemorrágica hereditária: uma causa rara de anemia grave. J Bras Pneumol. 2007;33(1):109-112. https://doi.org/10.1590/S1806-37132007000100020
12. Majumdar S, McWilliams JP. Abordagem das Malformações Arteriovenosas Pulmonares: Uma Atualização Abrangente. J Clin Med. 2020;9(6):1927. https://doi.org/10.3390/jcm9061927
13. Kwan V, Bourke MJ, Williams SJ, Gillespie PE, Murray MA, Kaffes AJ, et al. Argon plasma coagulation in the management of symptomatic gastrointestinal vascular lesions: experience in 100 consecutive patients with long-term follow-up. Am J Gastroenterol. 2006;101(1):58-63.
14. Majumdar S, McWilliams JP. Abordagem das Malformações Arteriovenosas Pulmonares: Uma Atualização Abrangente. J Clin Med. 2020;9(6):1927. https://doi.org/10.3390/jcm9061927
15. Terry PB, Barth KH, Kaufman SL, White RI Jr. Embolização por balão para tratamento de fístulas arteriovenosas pulmonares. N Engl J Med. 1980;302(21):1189-1190. https://doi.org/10.1056/NEJM1 98005223022107
16. Gupta P, Mordin C, Curtis J, Hughes JM, Shovlin CL, Jackson JE. Malformações arteriovenosas pulmonares: efeito da embolização no shunt direita-esquerda, hipoxemia e tolerância ao exercício em 66 pacientes. AJR Am J Roentgenol. 2002;179(2):347-355. https://doi.org/10.2214/ajr.179.2.1790347
17. Mason CG, Shovlin CL. Complicações relacionadas ao voo são infrequentes em pacientes com telangiectasia hemorrágica hereditária/malformações arteriovenosas pulmonares, apesar da baixa saturação de oxigênio e anemia. Tórax. 2012;67(1):80-81. https://doi.org/10.1136/thoraxjnl-2011-201027
18. Dupuis-Girod S, Cottin V, Shovlin CL. O Pulmão na Telangiectasia Hemorrágica Hereditária. Respiração. 2017;94(4):315-330. https://doi.org/10.1159/000479632

podem tolerar anticoagulação. Ana Hematol. 2012;91(12):1959-1968. https://doi.org/10.1007/s00277-012-1553-8