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Case Report

Rendú Osler Weber Syndrome; case report^{☆,☆☆}

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ABSTRACT

Hereditary hemorrhagic telangiectasia (HHT), or Rendu-Osler-Weber disease, is a dominant autosomal disease characterized by the presence of multiple telangiectasia in skin and mucus, associated with arteriovenous malformations (AVM) of various organs, including the lungs, gastrointestinal system and brain. HHT is presented most frequently as recurrent, spontaneous epistaxis. Patients may also present digestive, pulmonary and intracranial hemorrhage, as well as secondary anemia.

This article reports the case of a female patient, 62 years old, with multiple episodes of epistaxis and vaginal bleeding, with diagnosis of complex HHT, which was managed with multiple embolizations, which improved symptoms and survival. In this kind of patient, it is possible, with timely diagnosis and treatment, to obtain a greater quality and expectation of life. Due to the fact that the severity and alterations in each patient are so variable, management should be individualized.

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Introduction

Hereditary hemorrhagic telangiectasia (HHT), also known as Rendu-Osler-Weber disease, is a rare dominant autosomal vascular disorder that occurs with an estimated frequency of

1–20 cases/100,000 [1,5]. It is characterized by epistaxis, mucocutaneous telangiectasia, and gastrointestinal arteriovenous malformations. A definitive clinical diagnosis requires the presence of at least three of the following symptoms: spontaneous recurrent epistaxis, mucocutaneous telangiectasia, visceral arteriovenous malformations, and evidence of

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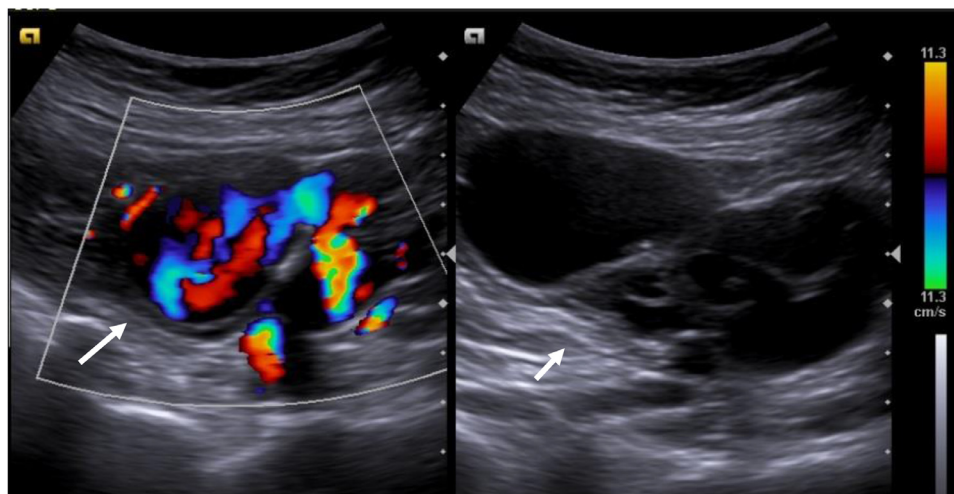


Fig. 1 – Ultrasound in grey tones and color Doppler, which shows an image with vascular characteristics at the level of the left annex topograph (white narrows).

dominant autosomal inheritance [2,3]. Although the daily symptoms of these patients are dominated by epistaxis that can notably alter their quality of life, the most severe manifestations of the disease are the consequences of pulmonary or gastrointestinal arteriovenous malformations [3–6].

Multimodal images, which include Doppler US, CT, and MRI, are used in the detection and initial characterization of vascular lesions in patients with HHT [7,8]. The diagnostic angiograph is of great importance in the correct characterization and determination of intervention treatments [8]. A multidisciplinary focus is a vital consideration for the correct early diagnosis and treatment; and treatment is recommended to be in centers with ample experience in management of this kind of case [8,9]. Intervention radiologists are essential in the treatment of this kind of alteration, due to their broad experience, not only in management, but also in complex embolization, which helps to improve the prognosis and quality of life for this kind of patient [10–12].

Method/presentation of the case

Female patient, 62 years old, Hispano-American, with weight of 69 kg, height of 165 cm, housewife, with history of recurrent epistaxis, hypertension under treatment with Enalapril 10mg daily, laparoscopic cholecystectomy in 2010, 3 pregnancies with eutocic births without complications, menopause at 45 years; family history was negative. Suffering began in April of 2019, with epistaxis and abundant transvaginal bleeding associated with hypogastric pain, which required a visit to emergency services, where the patient was found to be hypertensive (150/78 mmHg), tachycardial (105 beats per minute) and dehydrated (86%). Upon physical examination, there was evidence of mucocutaneous telangiectasia, as well as, in gynecological examination, abundant clotting in the vaginal canal (300 cc, approximately); management was begun with endovenous liquids and transfusion of red blood cells for the abrupt

drop in hemoglobin. Pelvic ultrasound was done, which reported an image in the left annex with a volume of 35 cc, which, from its characteristics, suggested a probable complex cyst, although not discarding a lesion of vascular origin (Fig. 1).

Image studies were complemented with thorax, abdomen and pelvis angiotomography, which showed relevant findings: solitary left pulmonary nodule with vascular behavior in regards to pulmonary arteriovenous malformation (Fig. 2), in addition to venous dilations that compromised the left renal vein, left ovarian vein, multiple collateral and confluent varicose veins, bilateral ovarian plexus veins, with dilation and severe twisting, as well as formation of parauterine venous sacculature with opacification by contrast medium in arterial phase, which occupied the pelvic cavity, with appearance of niche and extension to the vaginal plexus and iliac veins of left predominance (Fig. 3). Due to symptomology, clinical and radiological findings, a diagnosis of Rendu-Osler-Weber syndrome with associated uterine venous malformation was considered.

Said findings were evaluated by the intervention radiology service, which proceeded to perform embolizations; the first session acceded to the vascular system with right retrograde puncture in common femoral artery through vascular introducer 6 Fr passes pigtail catheter 5 Fr, with which aortogram was performed (Figs. 4 and 5). Then, a Cobra 1 caliber 5 Fr catheter with hydrophilic guide 0.035 × 260 mm, microguide with which the left renal artery was catheterized and Choice PT of 0.014 was passed with MAESTRO microcatheter, and segmentary and affluent artery was acceded to the MAV (Fig. 6), where embolization was performed with 500–700 mcm (1 Vial) microparticles. Later, platinum microcoils were placed in the proximal segment, as well as supra-selectively catheterizing affluent branches of the internal ilia artery, where 2 platinum microcoils were placed in each affluent artery towards the MAV. Right renal artery was selectively catheterized with Cobra 1 and Simmons 1 catheter, identifying the inferior segmentary renal artery, which gave arterial feed to the lesion. Attempts to channel the nutrient artery with microcatheter were unsuccessful. Exchange was performed with Envoy 6 Fr guide

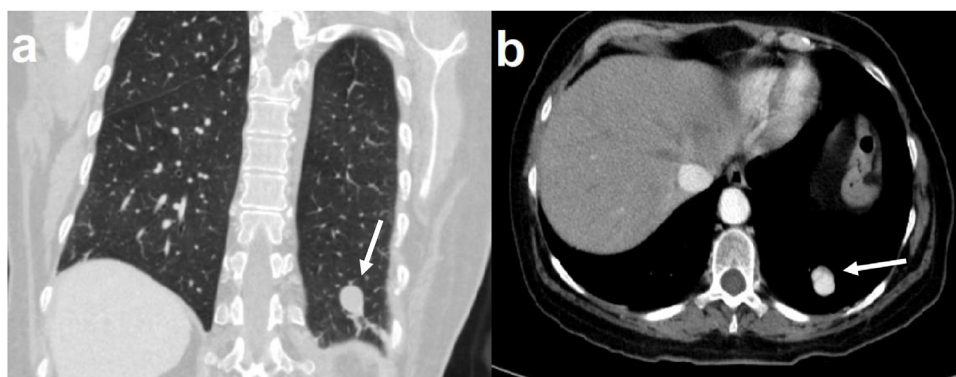


Fig. 2 – Computerized high-resolution tomography of thorax in coronal reconstruction (a), axial thorax tomography with window for mediastinum (b), images that showed pulmonary arteriovenous malformation in posterior left basal segment (white narrows).

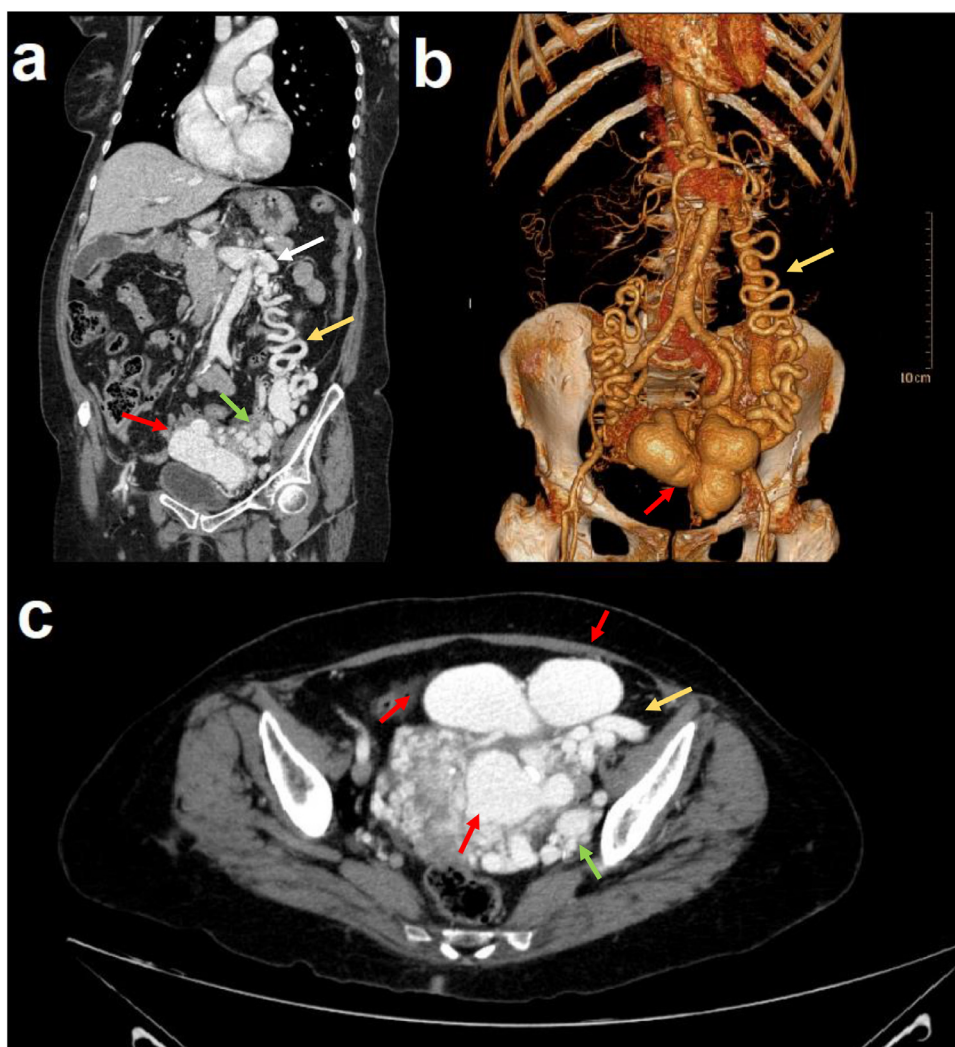


Fig. 3 – Angiotomography of thorax, abdomen and pelvis (a), angiotomography of abdominal aorta with 3D reconstruction (b), angiotomography of pelvis (c). Images that show venous dilations that compromise left renal vein (white narrow), left ovarian vein (yellow narrows), multiple collateral and confluent varicose veins, and bilateral ovarian plexus veins, with dilation and severe twisting (green narrows), as well as formation of paraarterine venous sacculations with opacification by contrast medium in arterial phase (red narrows).

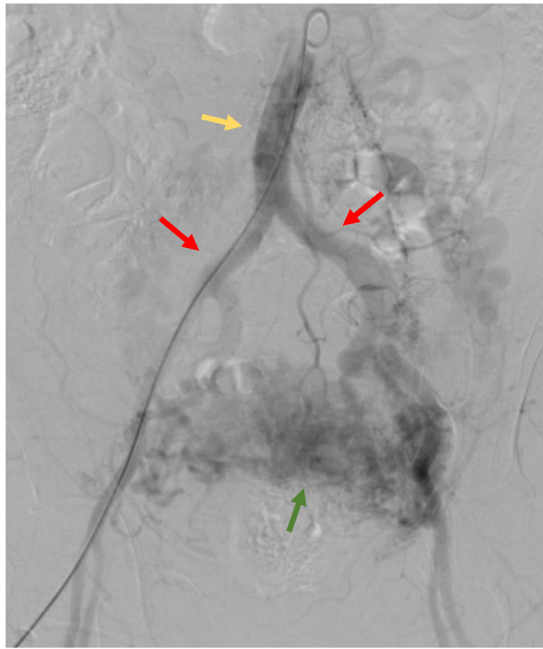


Fig. 4 – Arterial phase aortogram with opacification of aorta (yellow narrow), iliac vessels (red narrows) and vascular anomalies towards arteriovenous malformation.

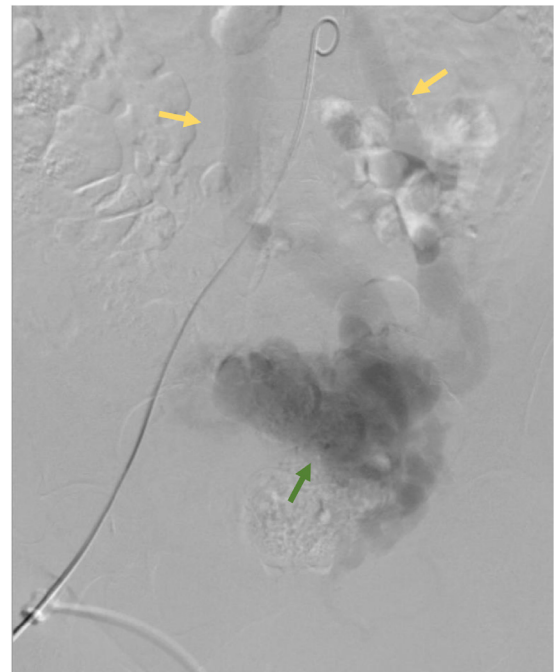


Fig. 5 – Venous phase aortogram visualizing opacification of gonadal venous vessels (yellow narrows) and strengthening of MAV (green narrow).

catheter, and a vascular plug 3 mm x 6 mm was placed before the forking of the subsegmentary arteries, where adequate devascularization was observed. Final control was achieved with reduction of arterial flow towards the lesion; however, the remaining arteries were visualized with arterial flow (Fig. 7). The procedure ended with use of limited contrast volume, and a second session was scheduled in 7 days, ending with partial embolization of MAV.

In the second session, embolization was performed with 355-500 mcm (1 vial) microparticles, then 4 platinum microcoils were placed in the proximal segment, then branches affluent to the right internal and external iliac artery were placed supra-selectively, placing 2 vials of spheres 900-1200 mcm, 3 platinum microcoils in each artery affluent towards the MAV, thereby observing adequate devascularization. Control with aortogram was performed, where the direct emer-

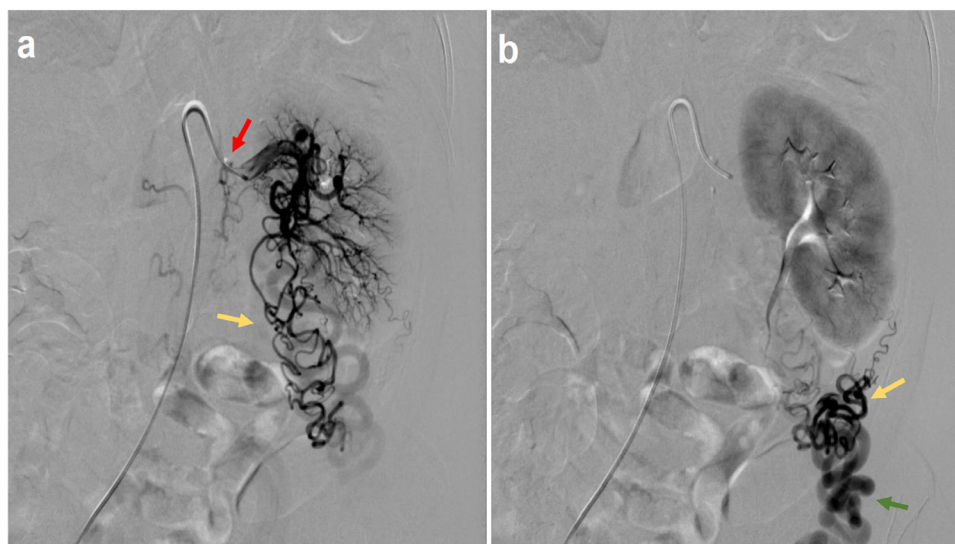


Fig. 6 – Catheterization of the left renal artery (red narrow); (a) one sees serpentine arterial vessels (yellow narrows), (b) veins and gonadals feeding MAV (green narrow).



Fig. 7 – Aortogram of post-embolization control with visualization of coils at the level of gonadal vessels (red narrow) and vascular plug before forking of subsegmentary arteries (green narrow); there is considerable reduction of vascular flow to MAV, but with persistence in some nutrient arteries and veins with regards to partial embolization (yellow narrow).

gent branch of the aorta which gave arterial flow to the lesion was identified. Attempts were made unsuccessfully to channel, due to stenosis of the ostium. The procedure was concluded with partial embolization, and a third session was proposed. Fifteen days later, a third embolization was performed, beginning with aortogram, where high-flow vascular malformation was observed, including the uterine artery with dilated and twisted afferent arteries from the right and left renal artery, inferior mesenteric, hypogastric arteries, gonadal and femoral artery and left external iliac, with precocious venous drainage towards the left gonadal vein. The right renal artery was catheterized and an attempt was made to pass a 0.014 microguide and Slim and Slim 130° microcatheter, without success. In addition, twisted anomaly vessels were identified, depending on the dorsal pancreatic artery, suggestive of vascular malformation. Selective cerebral angiogram was performed, observing internal carotid, vertebral, cerebral arteries and basilar artery of normal caliber and trajectory, with adequate passage of contrast medium, without noting extravasations, dilations or sacular images.

Considering the association of epistaxis and mucocutaneous telangiectasia with pulmonary arteriovenous malformation and visceral arteriovenous malformation, the patient met the diagnostic criteria for Rendu-Osler-Weber syndrome. The procedure was concluded with a complex pelvic arteriovenous malformation not susceptible to embolization. The patient showed posterior control of bleeding after partial embolizations and returned home. Given the pandemic, the patient did not attend controls and re-entered 2 years after

bleeding, in April, 2021, due to abundant transvaginal bleeding. Again, ultrasound and angiotomographic study was performed, showing worsening of the vascular dilations, with increased size of the MAV; four days after re-entry she presented massive secondary transvaginal bleeding with later hypovolemic shock, for which she was moved to emergency surgery by the gynecology and vascular surgery team. Exploratory laparotomy identified gigantic arteriovenous malformation with massive active bleeding. During the final surgery, she suffered heart attacks on 6 occasions, the last irreversible. The patient died during the surgical procedure.

Discussion

HHT is a rare dominant autosomal hereditary vascular disorder that occurs with an estimated frequency of 1-20 cases/100,000, which makes it difficult to diagnose and to find adequate treatment [1–4]. For its diagnosis, one should take into account the physical examination, as well as the clinical evaluation, pain, and a history of hemorrhages [5–7]. However, for a precise diagnosis, the use of images is necessary, such as angiotomography and angiography, to show the feeding vessels, the nest, and drainage veins, which is essential for embolization and adequate surgical incision or combined treatment [1,2,8,9]. In this case, angiotomography, as well as diagnostic angiogram, revealed and supported embolization. The high-flow arteriovenous malformation favors distension of the veins that transport a great part of the flow, causing venous hypertension, which allows the venous capillaries to break and cause hemorrhage, which can become slight or abundant and chronic, requiring hospitalization. This bleeding brings the patient to anemia, which should be treated with control of the hemorrhage and, if necessary, transfusion.

In regards to treatment, endovascular therapy guided by images, such as sclerotherapy, embolization, and surgical resection, are recommended as the best option, according to the kind of patient [9–12]. In our patient, there was epistaxis and recurrent vaginal bleeding, for which she was sent to the hospital center, where she was diagnosed with Rendu-Osler-Weber syndrome. This was correctly characterized by image assistance, for which the intervention radiologist performed embolizations, with great improvement in the symptoms and prognosis of the patient. Unfortunately, due to the COVID-19 pandemic, the patient could not perform the corresponding controls and follow-up, and her pathology became complicated, showing an increase in previously diagnosed arteriovenous malformations, situation which generated bleeding with later hemorrhage shock and death.

Sclerotherapy has the function of reducing the size of the venous nest; embolization was performed before surgical resection to facilitate the procedure with minimum risk of bleeding [8,11], although it also has been used as a permanent treatment [8–10]. Finally, surgery is the definitive recommended treatment [11]; however, it presents some complications, since the malformations tend to be extensive and poorly defined [1]. There is a risk of acute hemorrhage, and mismanagement of the procedure has grave consequences, including death [8,9,11].

In the case described, embolizations were performed, but due to the complexity of the pathology, the patient did not tolerate surgical resection, even though it improved the symptomatology, quality, and survival of the patient.

Conclusions

Timely diagnosis and classification help to find the correct, pertinent treatment. Treatment is highly complex due to the rarity of the disease; however, specialist physicians can offer conservative or invasive management in the case of high-flow arteriovenous malformations that compromise the life of the patient due to their location, size, or behavior.

For an adequate therapeutic plan, imaging methods are required, which offer important information on the organic implication and extension of the lesion, a vascular phase of great importance, since it gives us the first step in determining if the compromise is arterial and/or venous.

In these patients, adequate diagnosis and therapeutic choice, with multidisciplinary approach, may prevent severe complications and improve the prognosis and survival of the patient. Intervention radiologists are key members of this team and, at times, are at the vanguard of the management and treatment of these patients.

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Ethical responsibilities

The authors state that for this research no experiments were performed on humans or animals.

Data confidentiality

The authors state that this article does not include patient data.

Right to privacy and informed consent

The authors state that this article does not include patient data.

Protection of people and animals

The authors state that the procedures followed comply with the ethical standards of the responsible committee on experimentation and the World Medical Association and the Declaration of Helsinki.

Patient consent

Informed consent was obtained from the patient's family.

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