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Agata Orzechowska[1, A–D, F], Jadwiga Dwilewicz-Trojaczek[2, C, E–F],
Renata Górska[1, C, E–F]

Osler-Weber-Rendu Disease – Case Report

Choroba Rendu-Oslera-Webera – opis przypadku

[1] Department of Periodontology and Oral Diseases, Medical University of Warsaw, Warsaw, Poland
[2] Department of Hematology, Oncology and Internal Diseases, Medical University of Warsaw, Warsaw, Poland

A – research concept and design; **B** – collection and/or assembly of data; **C** – data analysis and interpretation; **D** – writing the article; **E** – critical revision of the article; **F** – final approval of article

Abstract

Osler-Weber-Rendu disease (HHT) is a rare, hereditary disease which is characterized by arteriovenous malformations – telangiectasias of the face, lips, oral cavity and the gastrointestinal tract. The lesions frequently bleed, leading to anemia and internal complications.

A 64-year-old patient presented due to numerous oral and gastrological lesions and a concomitant sideropenic anemia. She complained about frequent epistaxes and everyday bleeding of the lingual lesions for many years. The examination revealed numerous telangiectasias on the oral mucosa, as well as the lips, face and body skin. Similar lesions and epistaxes occur in the patient's first-degree relatives. According to the generally accepted Curacao criteria established in 1999, this patient may be confidently diagnosed with Osler-Weber-Rendu disease. The options of oral lesion treatment are limited to the use of coating agents and a delicate, non-harming diet. There are a few described cases of the use of Nd:Yag laser or local ethanolamine application next to the bleeding lesions. Apart from local treatment, internal diagnostics are necessary, as HHT may cause many general complications. Although HHT is not a contraindication to any dental procedures, patients need extra precautions to avoid complications (**Dent. Med. Probl. 2014, 51, 4, 537–540**).

Key words: hereditary hemorrhagic telangiectasia, telangiectasis, epistaxis.

Streszczenie

Choroba Rendu-Oslera-Webera (HHT) jest rzadką, genetyczną chorobą, która charakteryzuje się obecnością zmian naczyniowych w obrębie skóry oraz błony śluzowej jamy ustnej, przewodu pokarmowego i narządów wewnętrznych. Zmiany te prowadzą m.in. do częstych krwotoków, są przyczyną niedokrwistości oraz wielonarządowych powikłań.

Pacjentka lat 64 została skierowana w celu diagnostyki zmian na błonie śluzowej jamy ustnej z rozpoznanymi: niedokrwistością syderopeniczną oraz zmianami naczyniowymi żołądka. Pacjentka od wielu lat skarżyła się na częste krwawienia z nosa oraz codzienne krwawienia ze zmian na języku. Stwierdzono obecność licznych zmian o charakterze rozszerzeń naczyń na błonie śluzowej warg i języka, skóry twarzy i ciała. Podobne zmiany i krwawienie z nosa występują u krewnych I stopnia pacjentki. Na podstawie oficjalnie przyjętych kryteriów Curacao zdefiniowanych w 1999 roku można z całą pewnością rozpoznać u pacjentki chorobę Rendu-Oslera-Webera. Możliwości leczenia zmian w jamie ustnej w przebiegu HHT ograniczają się do stosowania preparatów powlekających i łagodnej diety niewywołującej urazów. Opisywane są pojedyncze przypadki doraźnego podawania etanolaminy w okolice silnie krwawiących zmian w jamie ustnej oraz zastosowania lasera Nd:YAG. Oprócz postępowania miejscowego jest konieczna diagnostyka internistyczna – HHT może również spowodować bardzo poważne powikłania ogólne. HHT nie stanowi przeciwwskazania do leczenia stomatologicznego, pacjenci wymagają jednak zastosowania pewnych środków ostrożności w celu uniknięcia ewentualnych powikłań (**Dent. Med. Probl. 2014, 51, 4, 537–540**).

Słowa kluczowe: naczyniakowatość krwotoczna wrodzona, teleangiektazje, krwawienie z nosa.

Osler-Weber-Rendu disease (hereditary hemorrhagic telangiectasia – HHT) is a rare, hereditary autosomal-dominant disease. It is considered as heterogeneous disorder, as there are at least two locations for HHT genes – OMIM#187300 (endoglin) for HHT1, localized on chromosome 9 and OMIM#600376 (activin receptor-like kinase 1) for HHT2, localized on chromosome 12 [1].

HHT is characterized by arteriovenous malformations – telangiectasias of the face, lips, oral cavity and the gastrointestinal tract [2]. The lesions bleed when harmed but they may also bleed spontaneously [3].

This gene mutation also leads to venous malformations and lesions in such internal organs as the lungs, brain and liver [2, 4].

The most common and characteristic manifestation is recurrent, spontaneous epistaxes, which occur in daytime as well as nighttime. It is the most common reason for patients' visit to the GP's office.

The recurrent epistaxes as well as internal organ lesions frequently lead to anemia manifesting with weakness and fainting [5].

Also, literature data indicates possible consequences of misdiagnosed HHT such as liver and spleen damage, brain hemangioma or even chronic DIC [6].

Case Report

A 64-year-old female was referred from an internal medicine ward because of oral mucosa lesions (Fig. 1). The patient was hospitalized due to recent loss of consciousness and general weakness. According to a blood test performed during the hospitalization, she had had diagnosed iron-deficiency anemia (the concentration of RBC: 4.53 T/L, HCT: 29.7%, MCV: 65.6 fl, MCH: 18.5 pg, MCHC: 27.5 g/dL, Hb: 8.4 g/dL, Fe: 18 µg/dL, with normal iron absorption evaluated directly, 1 and 3 h after iron overload: Fe0: 18 µg/dL, Fe1: 82.61 µg/dL, Fe3: 162.9 µg/dL). There was no information about TIBC and serum ferritin concentration, as the anemia diagnosis was made in the hospital.

The patient also had nodular goiter (confirmed by USG examination) with concomitant hypothyroidism and hyperadrenocorticism, pharmacologically controlled. The gastroscopy revealed stomach ulcers, polyps and vascular lesions. The *H. pylori* test was negative.

The patient had complained about frequent epistaxes and everyday bleeding of the lingual lesions for many years but had not been diagnosed before.

The examination of the oral cavity revealed numerous telangiectasias of the oral mucosa, as well as the lips, face and body skin (Fig. 2, 3). The diascopy test was positive, the lesions blanched under pressure.

Similar lesions and epistaxes occur in the patient's relatives – her brother and son.

Due to the numerous oral lesions, the patient had been referred for a laryngological examination to evaluate the nasopharynx mucosa.

She was administered an antibacterial and demulcent rinse to relieve oral discomfort. SRP therapy was performed (scaling and root planning) and she was given oral hygiene instructions.

After one month, the patient presented for a follow-up visit. She reported a slight subjective

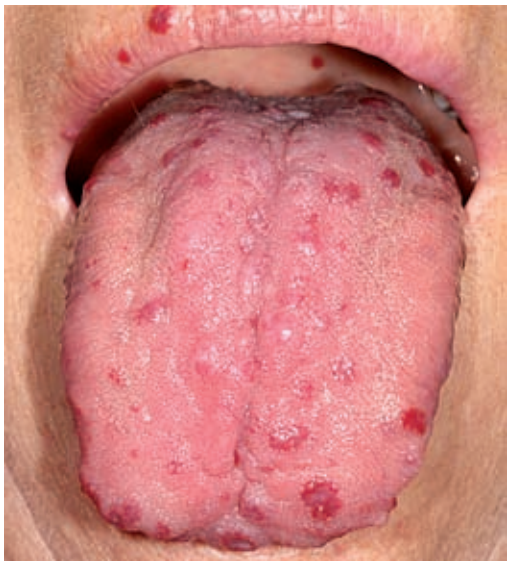

Fig. 1. *Telangiectasias* of the lingual mucosa

Ryc. 1. Zmiany o charakterze naczyniowym na błonie śluzowej języka

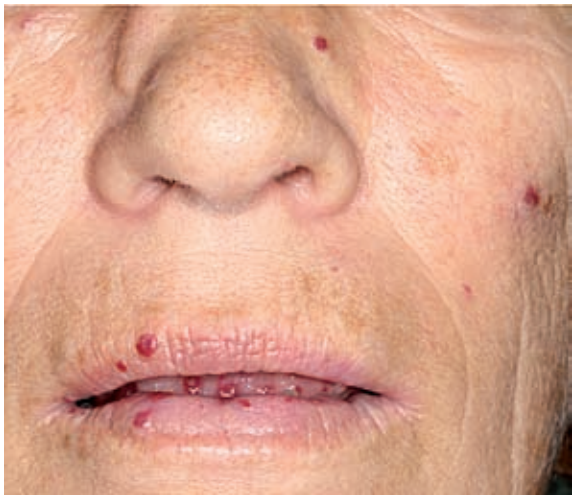

Fig. 2. *Telangiectasias* of the face skin and labial mucosa

Ryc. 2. Zmiany o charakterze naczyniowym na skórze twarzy oraz błonie śluzowej warg

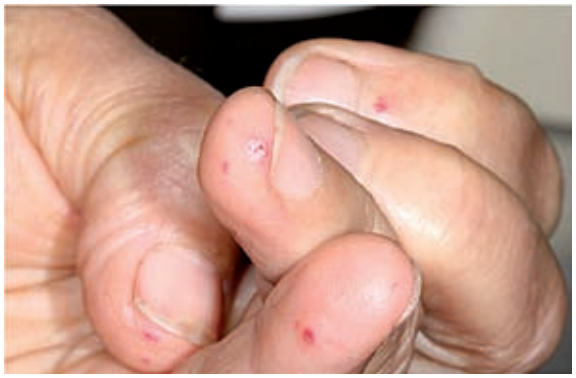

Fig. 3. *Telangiectasias* of the skin of the fingers

Ryc. 3. Zmiany o charakterze naczyniowym na skórze palców dłoni

improvement of her oral status. Clinically, there was no improvement or exacerbation of oral mucosa lesions. The patient had not undergone the laryngological examination.

After 3 weeks, the patient presented active bleeding of the tongue lesions and was referred to a hematologist.

Discussion

HHT can be conclusively diagnosed only when preceded by genetic examination. However, clinical practice relies on common criteria (1999 Curacao criteria) which quite precisely evaluate the likelihood of HHT occurrence. They are:

– recurrent, spontaneous epistaxes,

– numerous telangiectasia specifically localized (nasal and oral mucous, lips, skin on the fingers),

– family incidence of symptoms (HHT diagnosed in a I° relative),

– lesion occurrence in internal organs.

Incidence of 3 out of the 4 symptoms makes it possible to confidently diagnose HHT [7, 8].

According to these criteria, the presented patient was confidently diagnosed as HHT. The frequent epistaxis was the main cause of visiting the specialist, the lesions were observed in all characteristic locations and the family incidence was affirmed during the medical interview. There is no information about internal organ lesions, however they could not be excluded.

The iron-deficiency anemia has to be treated with iron agents. Due to the recurrent bleeding, the therapy may be chronic. In some cases, episodes of anemia require red blood cell transfusions.

The options for oral lesion treatment are limited to the use of demulcent, coating agents and a delicate, non-harming diet. Literature data describes a few cases of spreading ethanolamine around the lesions which led to their total healing (no new lesions in 2 year observations). Some authors [9, 10] describe possible use of a Nd:YAG laser in the intermedial parts of lips.

Although Osler-Weber-Rendu disease is not common, it is very important to know about, identify its symptoms and not understate them. Apart from local treatment, HHT patients need internal diagnostics. HHT is not only the cause of uncomfortable epistaxis, it can also lead to serious general complications. Literature data describes cases of misdiagnosed HHT which led to serious injuries of the liver and spleen as well as chronic DIC.

HHT is not a contraindication to dental treatment, however dentists must know that such patients require some special precautions to avoid possible complications.

First of all, when performing dental procedures in HHT patients, it is recommended to give up the ergonomic position and let patients stay seated as vertically as possible, to avoid an increase of lesion bleeding. Also, patients should have their blood pressure measured twice – before and after the procedure [5].

It is necessary to administer antibiotic prophylaxis prior to any procedure that would lead to bacteriemia, as one of the most common complications in HHT patients is brain abscess [11–13].

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Address for correspondence:

Agata Orzechowska
Department of Periodontology and Oral Diseases
Medical University of Warsaw
Miodowa 18
00-246 Warsaw
Poland
E-mail: agata.j.orzechowska@gmail.com

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