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Contact endoscopy of the nose in patients with Rendu–Osler–Weber syndrome

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Abstract

Objective: Teleangiectases are the source of hemorrhage in many HHT patients. Most frequent site of bleeding is the nose and more than 90% of all individuals with HHT suffer from recurrent epistaxis. Despite all efforts, treatment of epistaxis in HHT continues to be a problem for many otorhinolaryngologists, who can alleviate recurrent nosebleeds by Septodermoplasty or laser therapy, but rarely can stop nasal hemorrhages permanently. Recurrence is almost inevitable, but the mechanisms of recurrence are not fully understood.
Methods: Prior to routine Nd:YAG laser therapy of nasal telangiectases the nasal mucosa of 17 patients with HHT according to the clinical diagnostic criteria of the HHT Foundation International was examined with a 0° contact rhinoscope in areas with clinically visible telangiectases as well as in clinically normal mucosa. The digitally recorded images were compared to findings of a group of five healthy volunteers and the findings of five patients with polypoid sinusitis.
Results: Visualization of subepithelial vessels was feasible in all individuals of the study group as well as the control groups. Dilated vascular loops and tortuous vessels could be found in the study groups as well as in the control group, but the overall density of telangiectatic vessels was on an average higher in the HHT group. The process of vessel dilatation and tortuous configuration seemed to progress with age.
Conclusion: Contact endoscopy allows the investigation of the angioarchitecture of capillaries of the nasal mucosa in vivo. This observation may be of significance for studies of nasal diseases, which are accompanied by epistaxis. With this regard it seems to be of special interest for studies of HHT.

Keywords: Hereditary hemorrhagic telangiectasia; Epistaxis; Contact endoscopy; Vascular disorders; Rendu–Osler–Weber syndrome; Angioarchitecture

1. Introduction

Hereditary hemorrhagic telangiectasia (HHT, Osier's disease) is a systemic vasculopathy, which is inherited in an autosomal-dominant trait. A multitude of different mutations has been found on two gene loci so far. Endoglin mutations can be found on 9q34.1 (HHT-1) [1] and ACVRL-1 mutations are situated on 12q11–q14 (HHT-2) [2]. The most recent detection was recognized on 18q21.1 [3]. Mutations on this gene locus can lead to a combined form of juvenile colonic polyposis and HHT, the so-called JPHT syndrome [3]. Otolaryngologists play a key role in the medical management of HHT patients, because these patients are prone to seek medical advice because of recurrent epistaxis. More than 90% of all affected individuals develop epistaxis sooner or later in life, mostly between the 3rd and 4th decade. Recurrent epistaxis in HHT may be cumbersome to treat and generations of otolaryngologists have developed an armamentarium of different treatment options. Many methods can achieve a reduction of the nosebleeds, however it seems to be almost impossible to stop epistaxis permanently. The natural history of nasal telangiectases may play a key role in understanding HHT-related epistaxis. This study is about examination of the nasal mucosa in HHT patients with epistaxis by contact endoscopy. It was anticipated that contact endoscopy would provide new information through in vivo examination of the nasal mucosa, the predilection site of telangiectasia development.

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2. Patients and methods

Contact endoscopies were performed in 17 HHT patients (males: 9 p.; females: 8 p.; age range: 11–75 years, mean value: 53.8, median: 59.5, standard deviation 16.6) during routine endoscopic assessment of nasal teleangiectatic disease prior to Nd:YAG laser therapy sessions for treatment of recurrent epistaxis. The control group consisted of five healthy volunteers and five patients with polypoid sinusitis of similar age and gender. A detachable camera (Image1^{TM}, Karl Storz Company, Tuttlingen, Germany) was mounted to a contact rhinoscope (Karl Storz 7215 AA, 0°, length: 23 cm, diameter: 4 mm, Karl Storz Company) and all findings were recorded digitally with the Karl Storz-SCB^{TM} System (Karl Storz Company). All endoscopies and recordings were performed by one investigator. The digital images were evaluated in the media laboratory for dynamics of the blood flow, general morphology and density of the subepithelial vessels.

3. Results

Visualization of subepthelial vessels and studies of blood flow dynamics were feasible in all individuals through the observation of flowing erythrocytes. Thus, the identification of the afferent as well as the draining limbs of the capillary vessels was possible. The blood flow however could be reversed, if the endoscope was brought to bear on the nasal mucosa with increased pressure. If the endoscope was placed on the mucosa of the septum contact bleeding could be provoked easily in the HHT group. This effect was less pronounced if the mucosa of the lateral nasal wall and the turbinates was examined. All patients of the study group showed bright red telangiectases of the nasal mucosa (Fig. 1a). The absolute number and distribution of telangiectases showed strong individual differences. On an average, HHT patients exhibited nine dilated vascular loops per visual field, whereas in both control groups an average of four vascular loops could be identified. Numbers, however, were too small for statistic analysis. Telangiectases consisted of clusters of dilated vascular loops (Fig. 1b) and tortuous vessels (Fig. 1c). Dilated vascular loops and tortuous vessels were most pronounced in the draining limb of the vessels of the HHT group, but could also be found in the control group. The diameter of the draining limbs was twice the size of the afferent limb in the control group, in the HHT group the efferent limb exceeded the diameter of the afferent limb by the 3-fold at minimum. Dilatation and tortuous configuration of the draining limbs seemed to progress with age. Fig. 1d shows the vascular network of the septal mucosa in an 11-year-old male HHT patient, Fig. 1e shows the findings in a corresponding location of a 63-year-old female HHT patient. In both cases a high density of vessels can be seen, but tortuous and dilated vessels are almost exclusively evident in the 63-year-old patient. The overall density of vessels was higher in the HHT group than in the control group, even in normal nasal mucosa without macroscopically detectable telangiectases (see also, Fig. 2).

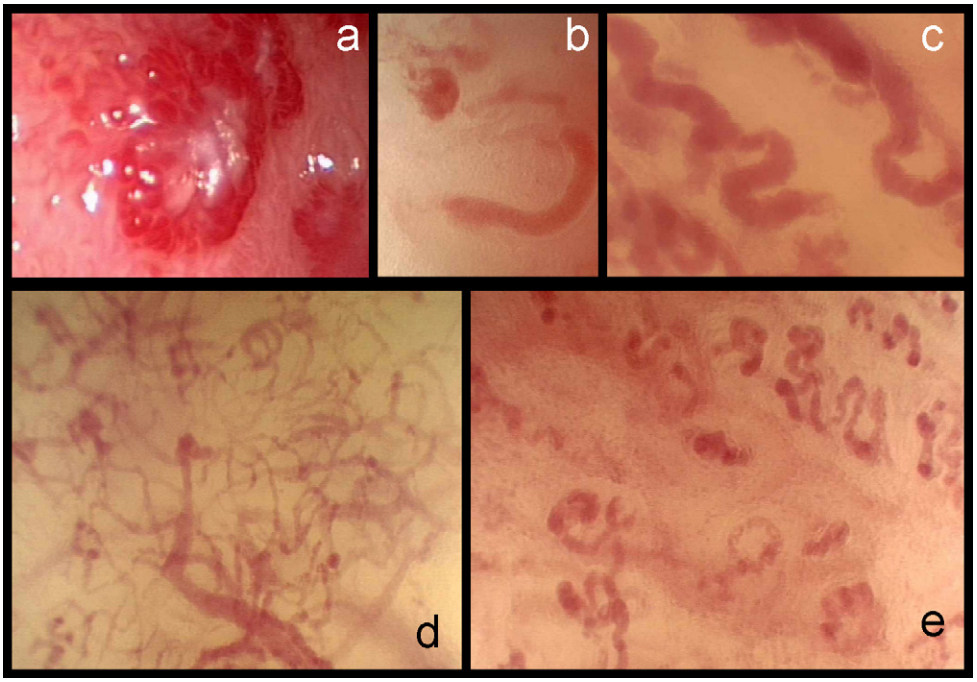

Fig. 1. A compilation of typical endoscopic ((a) 30°-angled Hopkins rod lense endoscope) and contact endoscopic ((b–e) 0°-angled endoscope, Karl Storz 7215 AA) aspects of endonasal telangiectases. The image shows that nasal telangiectases consist of accumulated loops of vessels, which are dilated and of tortuous configuration.