Spontaneous Unilateral Epistaxis and Telangiectasia Presenting Similarly to Hereditary Hemorrhagic Telangiectasia

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Introduction

- Hereditary Hemorrhagic Telangiectasia (HHT) is a rare autosomal dominant disorder characterized by recurrent epistaxis, mucocutaneous telangiectasias, and visceral arteriovenous malformations
- The Curaçao criteria are a set of four observations used to diagnose HHT. A definitive diagnosis requires a patient to meet 3 of the 4 of the following criteria: Recurrent epistaxis, mucocutaneous telangiectasias, visceral arteriovenous malformations, and a family history of HHT.
- This case examines a rare presentation of unilateral recurrent epistaxis with intranasal telangiectasias, exploring its potential association with HHT

Patient Background

- 65 y/o male with a 10-year history of diffuse skin telangiectasias on the left cheek and nasal sidewall and severe left sided epistaxis
- Past medical history includes bleeding diathesis, abdominal aortic aneurysm, coronary artery disease, seasonal allergies, 20 pack year smoking history
- No family history of HHT or bleeding disorders
- Patient initially presented to emergency room for multiple syncopal episodes secondary to recurrent significant left-sided epistaxis

Patient Workup

- Upon follow up in the outpatient setting, found to have approximately 12 skin telangiectasias, with patient endorsing initial appearance 10 years prior
- After referral to oncology, it was concluded that the patient met the criteria for HHT but included a differential diagnosis of a primary bleeding disorder due to lack of clear AVMs and lack of family history of HHT.
- Patient was initially treated with nasal packing and humidification and referred to hematology/oncology for further workup
- Brain MRI and head MRA ruled out cerebral aneurysm
- Nasal endoscopy revealed deviated septum with multiple left-sided telangiectasias. Telangiectasias absent in right nare, no additional vascular lesions appreciated in the mouth, larynx, or extremities
- Positive for anemia, mild reduction in arachidonic acid release seen on platelet aggregation studies; no visceral arteriovenous malformations found.
- No pathogenic variant seen on HHT-related genetic mutations (ENG, ACVRL1, SMAD4), however unable to rule out HHT due to unidentified or undetected genetic causes
- Patient denied further intervention and followed up with persistent presentation of recurrent left-sided epistaxis with no change in skin or intranasal telangiectasias

Conclusion

- This case underscores the challenges of managing HHT-like conditions when genetic testing is inadequate to provide a definitive diagnosis
- Limitations exist within available genetic testing in distinguishing HHT from mimicking conditions or atypical cases
- Further investigation into HHT-like presentation is warranted to enhance diagnostic clarity and improve management strategies

References

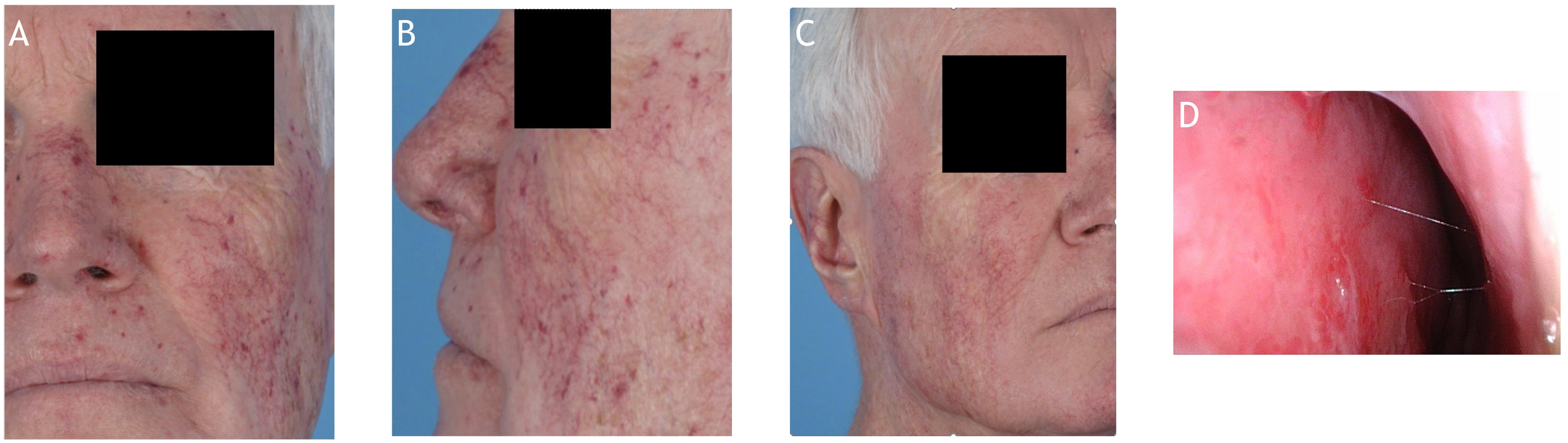

Figure 1. A) Anterior and B) lateral view of left-sided facial telangiectasias. C) Anterior view of right face without telangiectasias. D) Intranasal view of left-sided telangiectasias along septal mucosa.