Telangiectasia Tales

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Patient Presentation

CC

Bloody tears OU

HPI

- 33 y/o M presents with ~1 month of episodic unprovoked bleeding OU. No known trauma. Associated with occasional tearing, conjunctival injection/irritation OU.
- Taking Vigamox, tobramycin, and fluorometholone (FML) gtts from outside provider for suspected conjunctivitis

History (Hx)

Past Ocular Hx: none

Past Medical Hx:

- seizure disorder
- anemia
- arthritis
- depression/anxiety

Fam Hx:

- glaucoma

Social Hx: non-contributory

Meds

- Vigamox, tobramycin, FML gtts
- Levetiracetam
- Hydroxyzine
- Cyclobenzaprine
- KCl
- B12

Allergies: no known

ROS: negative

External Exam

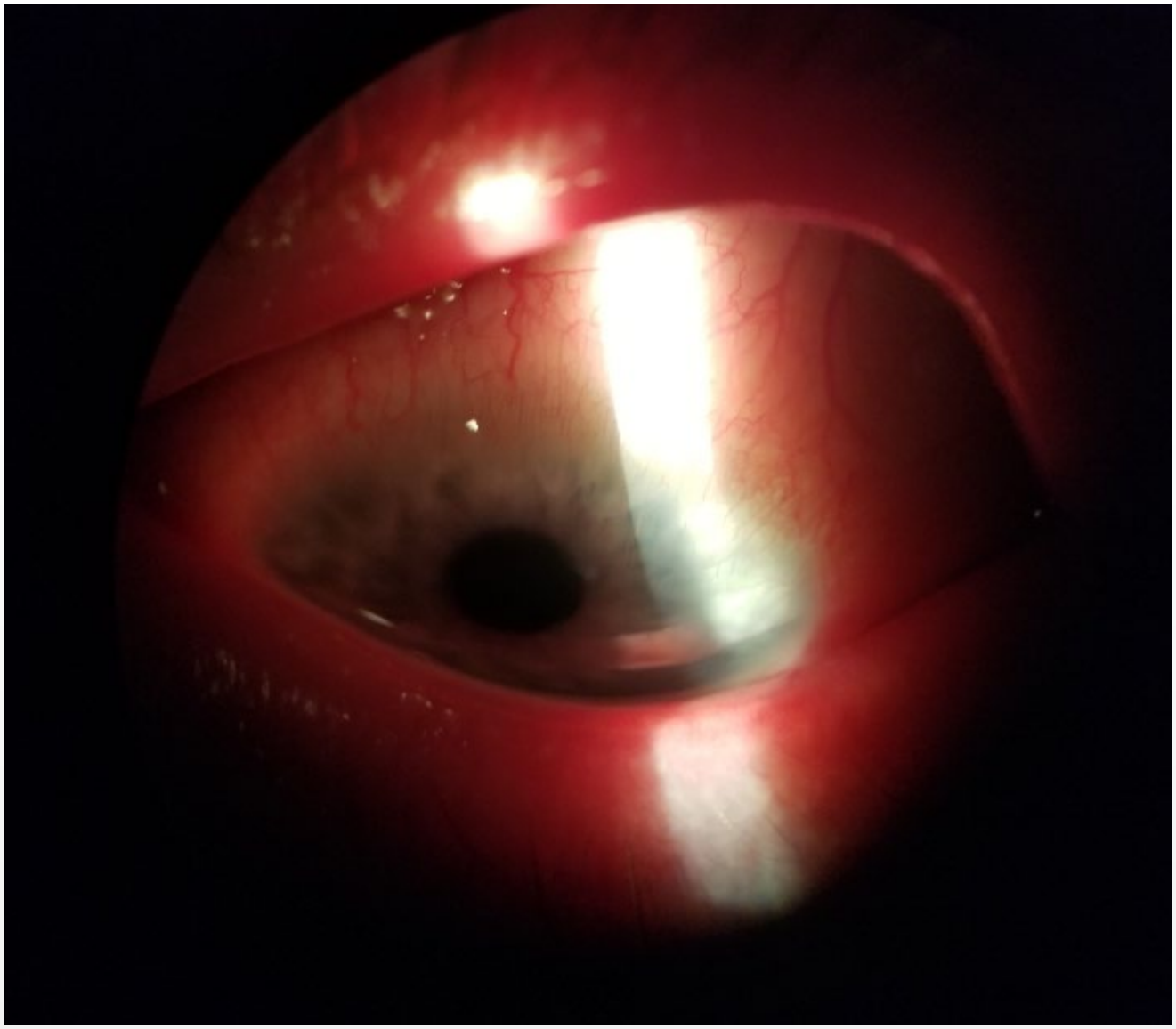
	OD		OS
BCVA	20/30-2 (Surface irregularities)		20/25
Pupils	Round, brisk	No rAPD	Round, brisk
IOP	9 mmHg		9 mmHg
EOM	full		full
CVF	full		full

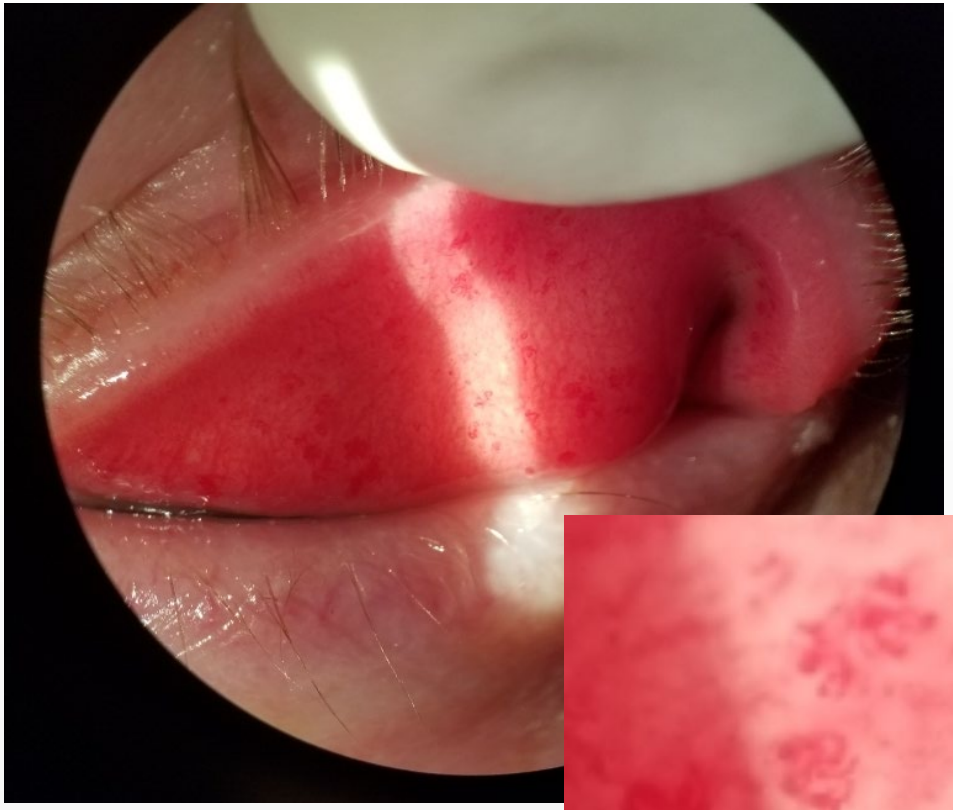
Anterior Segment Exam

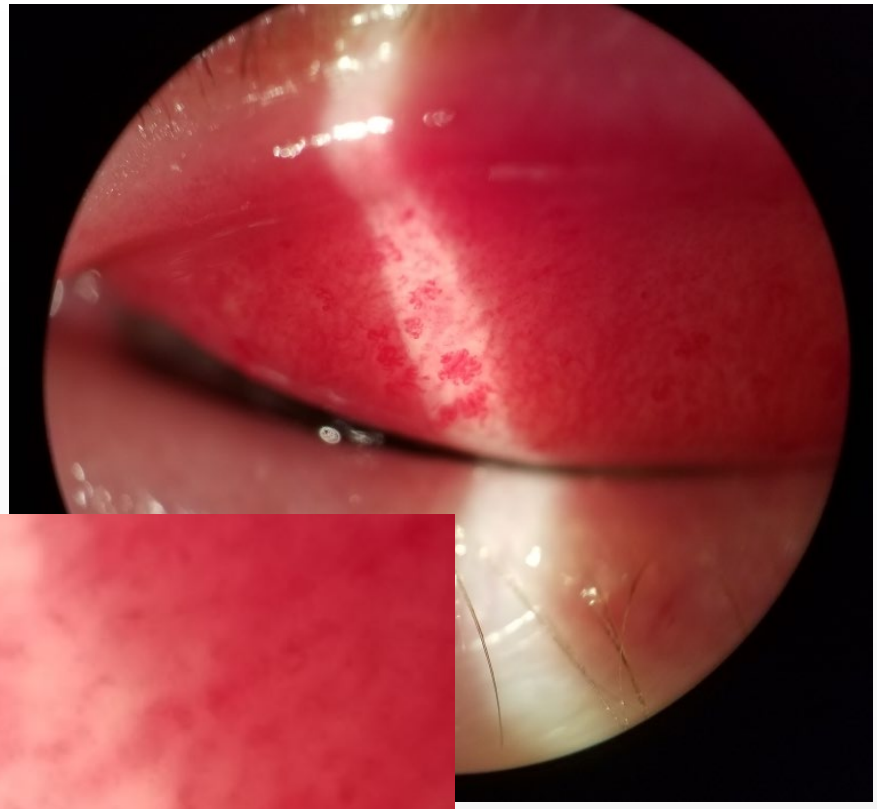
PLE or SLE	OD		OS
External/Lids	Blepharitis		Blepharitis
Conj/Sclera	Telangiectatic vessels on palpebral tarsal conjunctiva of upper and lower lid		Telangiectatic vessels on palpebral tarsal conjunctiva of upper and lower lid
Cornea	Pannus superior limbus		Pannus superior limbus
Ant Chamber	Deep & quiet		Deep & quiet
Iris	Flat		Flat
Lens	Clear		Clear

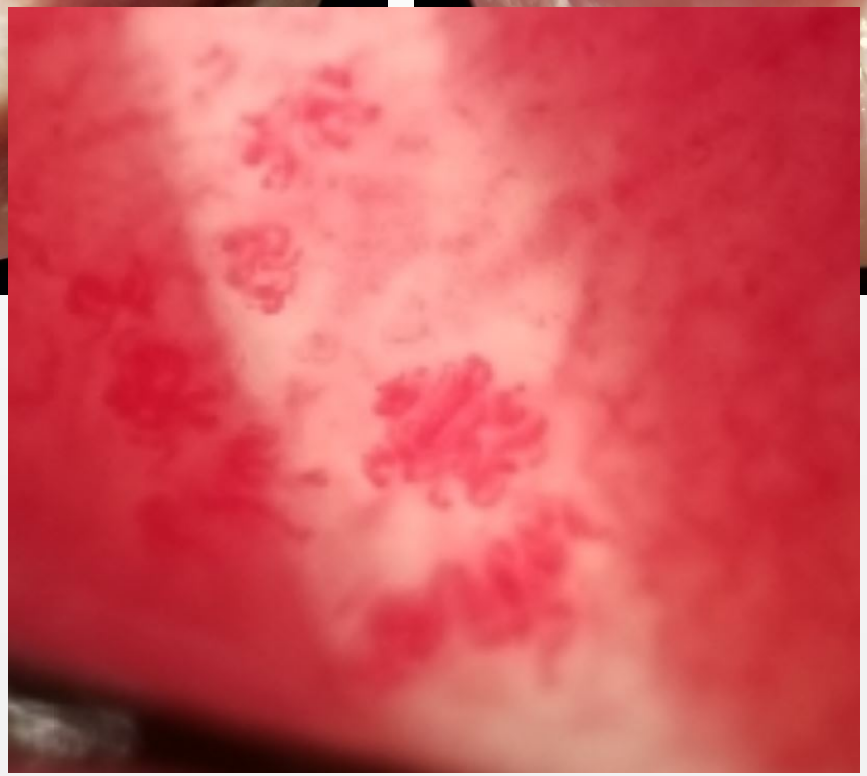
Posterior Segment Exam

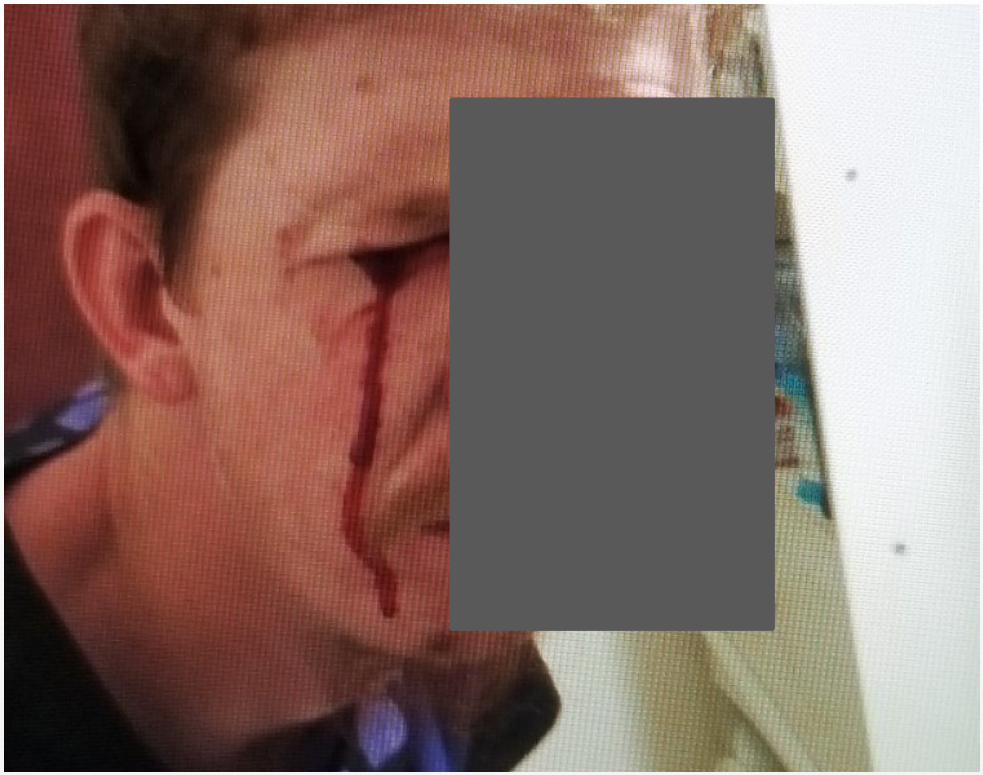
Fundus	OD		OS
Optic Nerve	No edema, pallor		No edema, pallor
Macula	No hemorrhage, sub-retinal fluid, or edema		No hemorrhage, sub-retinal fluid, or edema
Vessels	Normal caliber		Normal caliber
Periphery	No holes or tears 360		No holes or tears 360











Assessment

- 33 y/o M with chronic blepharitis and a 1 month Hx of episodic, unprovoked haemolacria OU. Exam notable for tarsal conjunctival telangiectasias and superior corneal pannus
- On further questioning, anemia 2/2 GI bleed associated with a “bleeding disorder”

Differential Diagnosis - Haemolacria

- Lacrimal gland pathology
  - Malignancy, infection
- Conjunctiva pathology
  - Malignancy, Stevens Johnson Syndrome, conjunctivitis, anticoagulation, trauma
- Canaliculitis
- Sinus pathology
  - Retrograde tear flow in epistaxis
- Factitious disorder
- **Hereditary hemorrhagic telangiectasia**

Plan

- Discontinue antibiotic gtts
- Continue fluorometholone (FML) gtt 0.1% qd for corneal pannus for 1 month
- Start artificial tears (ATs) prn, lid hygiene for chronic blepharitis
- Avoid eye rubbing, etc which could provoke bleeding
- Clinic follow up in 6 months

Discussion

- Hereditary Hemorrhagic Telangiectasia (HHT)
  - (Osler-Weber-Rendu syndrome/disease)
  - Prevalence: ~1 per 10,000
  - Autosomal dominant inheritance
  - TGFβ signaling genes: *endoglin, ACVRL1*
  - Systemic arteriovenous malformations (AVMs)
    - Absence of capillaries connecting arteries & veins
    - Tortuous, dilated, thin-walled vessels without vessel wall contractile elements prone to bleeding from minimal trauma

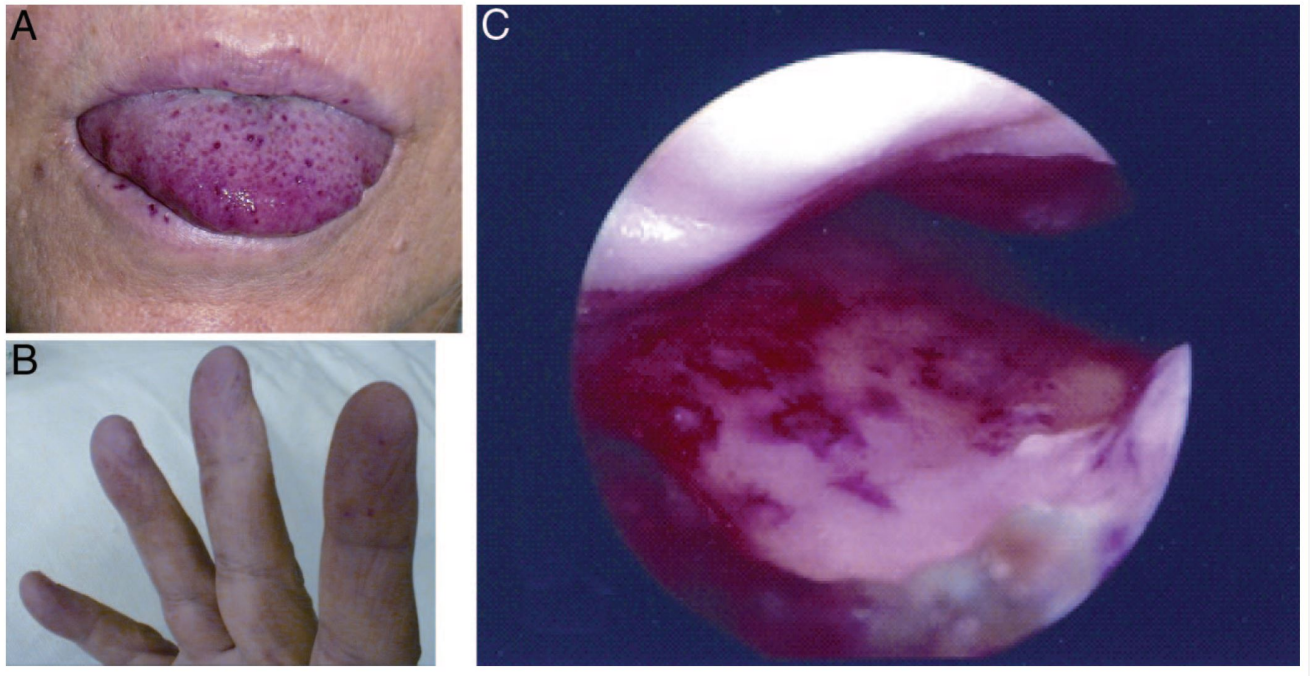
HHT Diagnosis

Table 1 The Curaçao criteria for the clinical diagnosis of HHT [6]

Curaçao criteria	
1. Epistaxis	
Spontaneous, recurrent nosebleeds	
2. Telangiectases (TAE)	
Multiple and at characteristic sites:	
Lips, oral cavity, fingers, nose	
3. Visceral manifestations	
– gastrointestinal (GIT) telangiectasia (with or without bleeding)	
– pulmonary arteriovenous malformations (PAVM)	
– hepatic arteriovenous malformations (HAVM)	
– cerebral arteriovenous malformations (CAVM)	
4. Family history	
A first grade relative with HHT according to these criteria	
The probability of the diagnosis depends on the number of positive criteria:	
Number of positive criteria	Probability of the diagnosis of HHT
3 or 4	Definite
2	Possible or suspected
1 or 0	unlikely

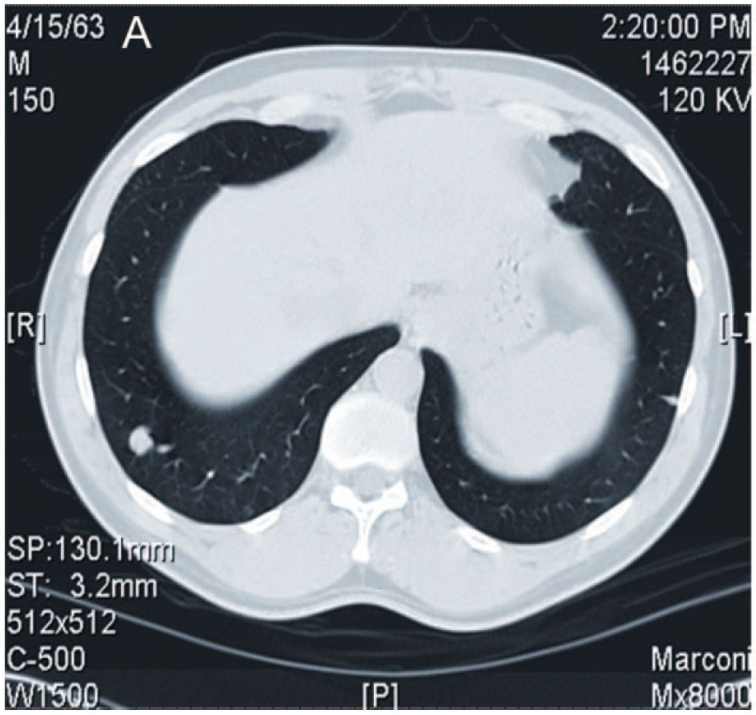
Clinical Manifestations

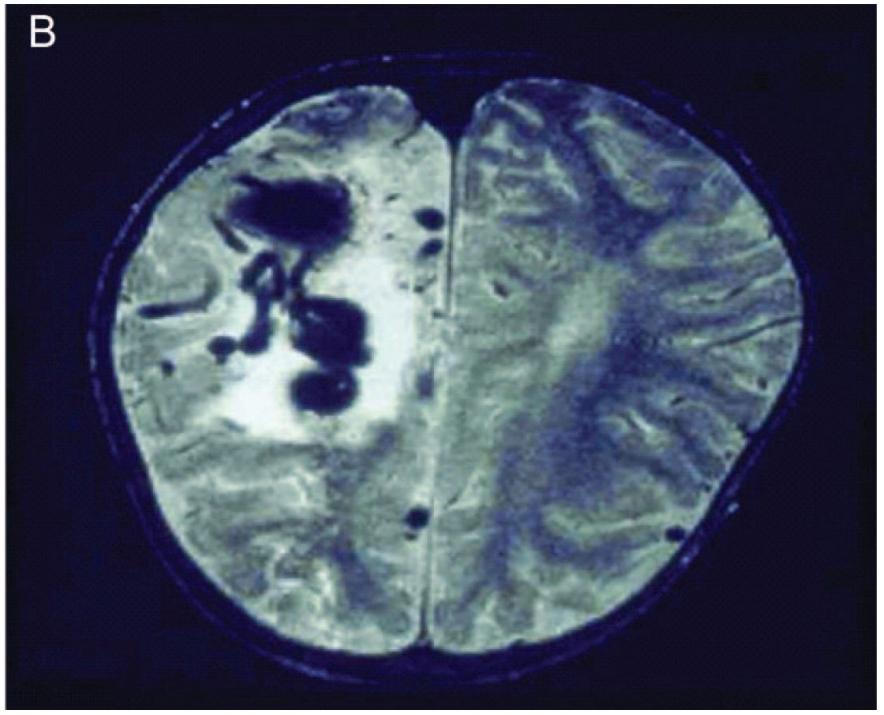
- Epistaxis most common by adolescence (80-90% by age 21)
- Other mucocutaneous telangiectasias present 5-10 years later



Clinical Manifestations

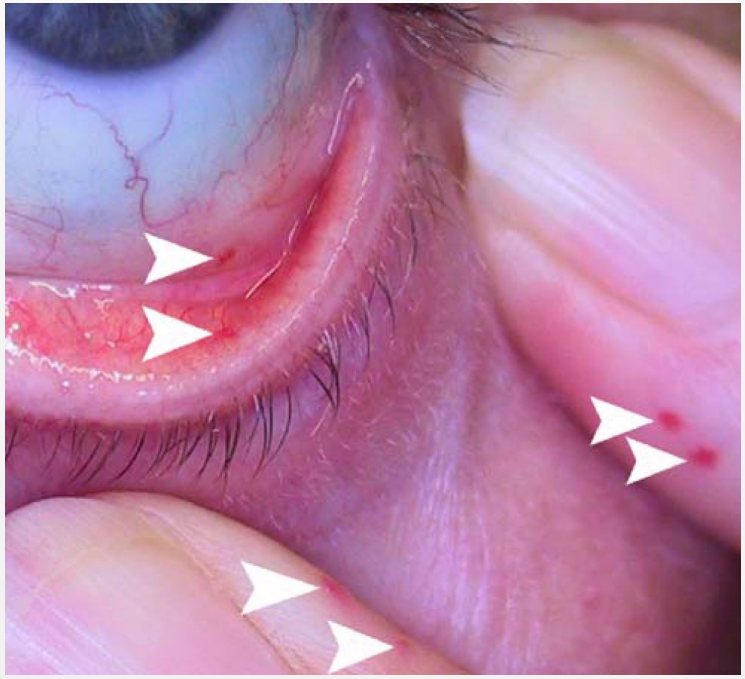
- Cerebral AVMs overwhelmingly congenital and remain unchanged
- Visceral AVMs may develop/progress slowly over time





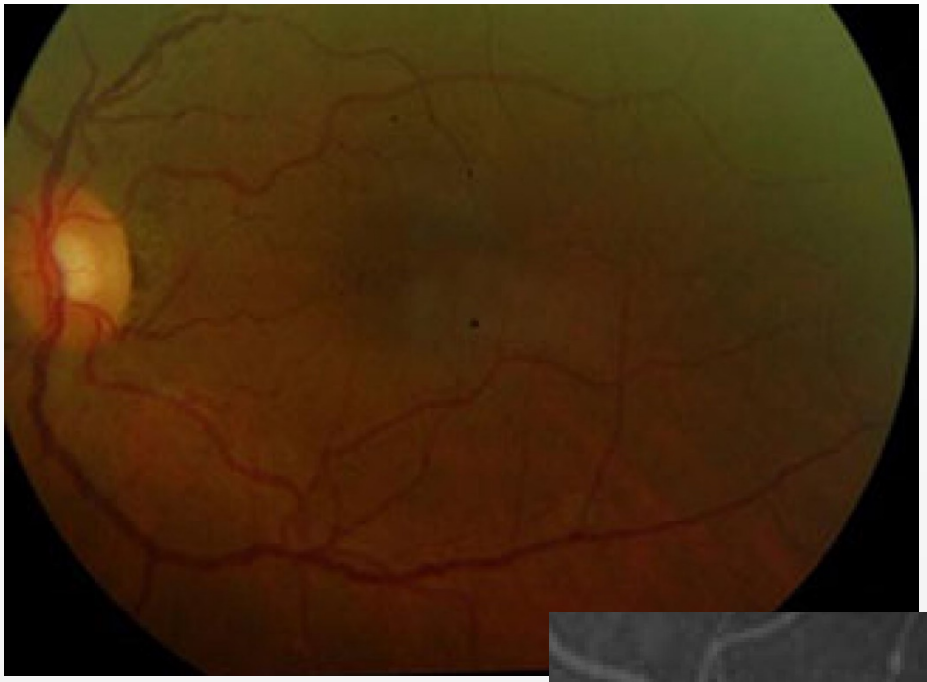
Ocular Manifestations

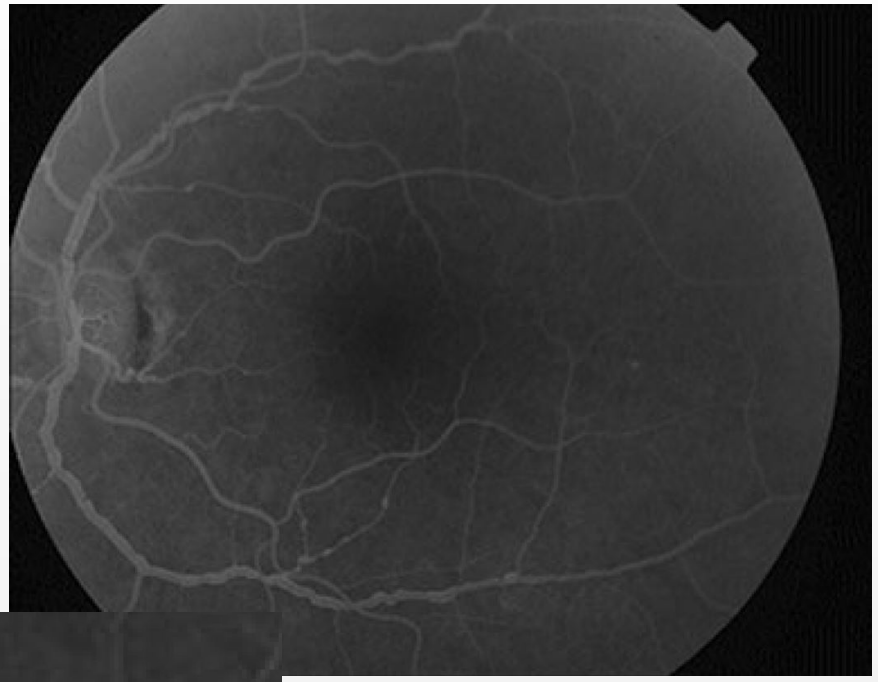
- Conjunctival telangiectasias
  - Conjunctival involvement most common
    - Palpebral > bulbar involvement
    - Haemolacria: disturbing to patients but clinically benign
  - Prevalence ~39%
  - No correlation to presence of specific visceral findings

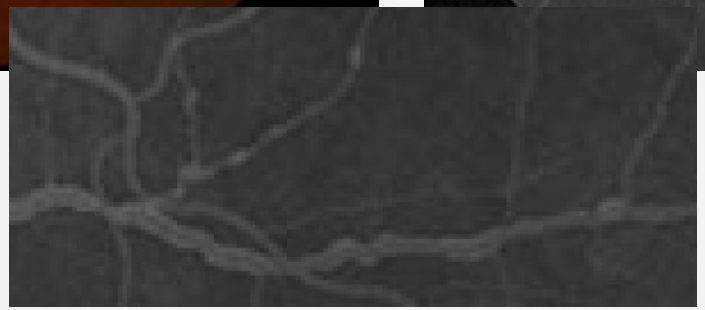


Ocular Manifestations

- Retinal and choroidal telangiectasias
  - Rare; ~1%; data limited
  - Case series of 8 family members showed 3 with chorioretinal atrophy/telangiectasia

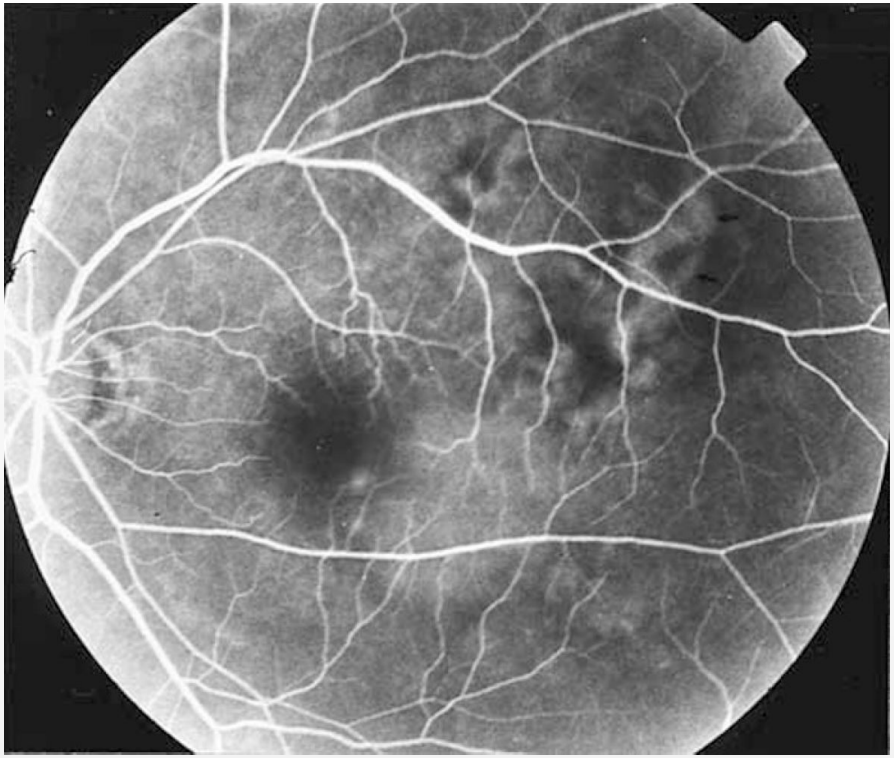


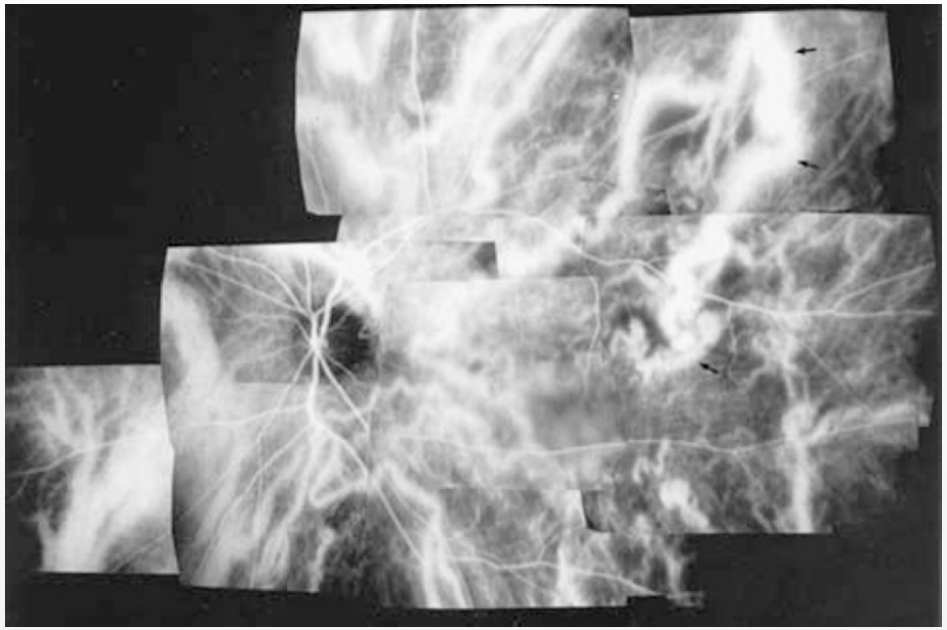




Retinal/Choroidal Telangiectasias

Fluorescein /indocyanine green angiography may reveal tortuous, dilated vasculature





Case Reports

Am J Ophthalmol. 2002 Feb;133(2):282-4.

Intraoperative choroidal hemorrhage in the Osler-Rendu-Weber syndrome.

Mahmoud TH[1], Deramo VA, Kim T, Fekrat S.

Author information

Abstract

PURPOSE: To describe a patient with Osler-Rendu-Weber syndrome who developed a nonsimultaneous intraoperative choroidal hemorrhage in each eye.

METHOD: Interventional case report. A 65-year-old Caucasian woman with Osler-Rendu-Weber syndrome developed a choroidal hemorrhage
developed a
phacoemuls
ophthalmos

RESULTS:

CONCLUSIO
possible ass

Clin Case Rep. 2015 Sep;3(9):725-7. doi: 10.1002/ccr3.324. Epub 2015 Jun 18.

Severe open angle glaucoma in hereditary hemorrhagic telangiectasia.

Kuchtey RW[1], Naratadam GT[1], Kuchtey J[1].

Author information

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Abstract

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant disease. Conjunctival telangiectasias and retinal vascular malformations are known ocular manifestations. We report here the first case of open angle glaucoma in a patient with HHT caused by a nonsense mutation, C471X in the ACVRL1 gene.

KEYWORDS: glaucoma; hereditary hemorrhagic telangiectasia; transforming growth factor beta

Conclusions

- Haemolacria may be a sign in the constellation of HHT findings
- Conjunctival telangiectasias most common ocular manifestation; chorioretinal involvement is rare or possibly under-recognized
- Conservative management is usually enough to stop bleeding from conjunctival telangiectatic vessels (in other organs surgical intervention or argon laser coagulation might be required)

Acknowledgement

- Special thanks to Dr. Asghari

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