From Mouth to Knee in HHT: an unusual complication of hereditary hemorrhagic telangiectasia

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LEARNING OBJECTIVES

- Recognize clinical features and complications of hereditary hemorrhagic telangiectasia (HHT)
- Recognize that pulmonary arteriovenous malformations (AVMs) are associated with paradoxical embolic spread of infection to remote sites

CASE

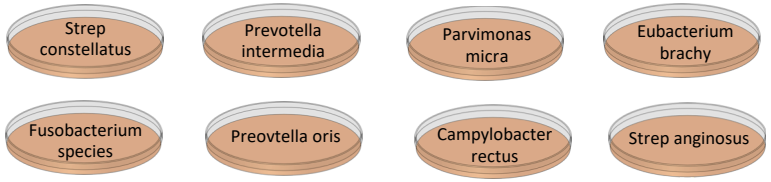
- 28-year-old female with no past medical history presents with right thigh pain after emigrating from Algeria a month prior to presentation. CT angiography demonstrates 3 large pulmonary AVMs, concerning for a new diagnosis of HHT

- Hospital course:
  - Becomes increasingly hypoxic, febrile, and develops swollen right knee
  - Joint aspiration yields frank pus → OR for irrigation & debridement
  - Blood cultures grow Strep constellatus
  - Continues to have fever → imaging showed large abscesses in popliteal and thigh compartments
  - Undergoes numerous surgical debridements, cultures from which grow multiple, new oropharyngeal flora
  - Formal dental evaluation, including CT Maxilla/Face, reveals no active infection
  - TTE negative for vegetations

IMPACT

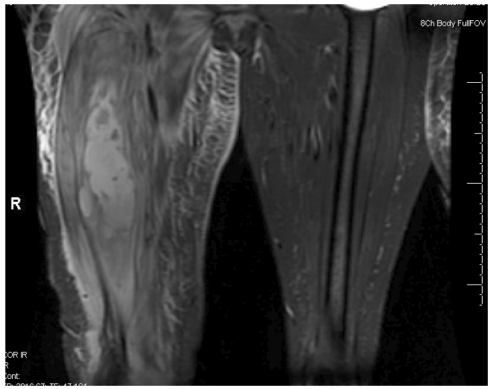
- This case represents an unusual presentation of a common autosomal dominant genetic disorder (HHT)
- It is difficult to determine the timing of embolization of AVMs in the setting of persistent bacteremia
- Demonstrates that transient bacteremia alone, without a primary source, can cause remote infection in patients with mechanisms to facilitate paradoxical emboli (HHT, AVMs)

MICROBIOLOGY



IMAGING

A) Posterior-anterior 3D imaging of pulmonary AVMs



B) Intramuscular abscess of right thigh

TREATMENT/MANAGEMENT

- Extensive discussions regarding timing of embolization of pulmonary AVMs, which is deferred in setting of persistent infection
- Started on ceftriaxone and metronidazole to complete a total 6 week course
- Ultimately requires an above-the-knee amputation of her right lower extremity due to recurrent infection
- Three months later, she undergoes embolization of her pulmonary AVMs

DISCUSSION

- HHT is autosomal dominant disorder characterized by abnormalities in vascular structure
- Common signs & symptoms include epistaxis, gastrointestinal bleeding, with mucocutaneous telangiectasias on exam
- Over 50% of HHT patients have pulmonary AVMs, which predisposes them to embolic strokes and cerebral abscesses from paradoxical emboli and/or bacteremia[1]
- There are case reports detailing brain abscesses from oropharyngeal flora, but no cases of septic arthritis
- Because there was no primary source of odontogenic infection, management weighed need for source control with embolization to stop transient bacteremia from seeding the knee
- With regards to infection prevention, the literature identifies the need antibiotic prophylaxis prior to dental procedures, but no change to usual vaccination recommendations[2]

CONCLUSION

- HHT is an autosomal dominant disorder affecting blood vessels that results in mucocutaneous telangiectasias and AVMs
- Patients with pulmonary AVMs are at risk for extra-cerebral infections such as septic arthritis
- This case highlights that transient bacteremia alone, without a primary infectious source, can cause remote infection

REFERENCES

[1] Musso M, et al. Extra-cerebral severe infections associated with haemorrhagic hereditary telangiectasia (Rendu-Osler-Weber Disease): five cases and a review of the literature. Infez Med. 2014 Mar;22(1):50-6.

[2] Shovlin C, et al. Prevention of serious infections in hereditary hemorrhagic telangiectasia: roles for prophylactic antibiotics, the pulmonary capillaries – but not vaccination. Haematologica, 2019.