Supplementary File 1

Survey: “Epidemiological Study of Hereditary Hemorrhagic Telangiectasia in Uruguay, 2015–2023”

This structured questionnaire was specifically designed for the epidemiological study of hereditary hemorrhagic telangiectasia in Uruguay, based on international HHT screening and management recommendations and additional clinically relevant variables defined by the investigators.

Abbreviations: HHT, hereditary hemorrhagic telangiectasia; PAVMs, pulmonary arteriovenous malformations; CAVMs, cerebral arteriovenous malformations; CT, computed tomography; MRI, magnetic resonance imaging; TIA, transient ischemic attack; ESS, Epistaxis Severity Score.

Demographic information

1. Study identification number

2. Current age (years)

3. Age at first medical consultation for HHT (years)

4. Age at HHT diagnosis (years)

5. Biological sex

- Male
- Female

6. Place of residence

- Montevideo
- Other regions of Uruguay

Family history

7. Do you have a first-degree relative (mother, father, sibling, or child) diagnosed with HHT?

- Yes
- No

Epistaxis

8. Have you experienced recurrent nosebleeds?

- Yes
- No

9. At what age did your nosebleeds begin?

10. How often do you experience nosebleeds?

- Never
- Once per month
- Once per week
- Several times per week
- Once per day
- Several times per day

11. How long do your nosebleeds usually last?

- Less than 1 minute
- 1–5 minutes
- 6–15 minutes
- 16–30 minutes
- More than 30 minutes

12. Bleeding intensity

- Dripping
- Continuous/streaming

13. Have you ever sought medical attention because of nosebleeds?

- Yes
- No

Anemia

14. Do you currently have anemia (based on blood test results)?

- Yes
- No

15. Have you ever required a blood transfusion due to nosebleeds?

- Yes
- No

16. Lowest hemoglobin level ever recorded (g/dL)

17. Most recent hemoglobin level (g/dL)

Treatments

18. Have you ever received oral iron supplementation?

- Yes
- No

19. Have you ever received intravenous iron therapy?

- Yes
- No

20. Have you ever been treated with tranexamic acid?

- Yes
- No

21. Have you ever received propranolol?

- Yes
- No

22. Have you received any other treatment specifically for HHT?

- Yes
- No

Clinical manifestations

23. Do you have mucocutaneous telangiectases (skin, lips, nose, or oral cavity)?

- Yes
- No

Pulmonary involvement

24. Have you been diagnosed with pulmonary arteriovenous malformations (PAVMs)?

- Yes
- No

25. Have you undergone contrast-enhanced transthoracic echocardiography (bubble echocardiography)?

- Yes
- No

26. Have you undergone contrast-enhanced chest CT angiography?

- Yes
- No

Cerebral involvement

27. Have you been diagnosed with cerebral arteriovenous malformations (CAVMs)?

- Yes
- No

28. Have you ever experienced an ischemic stroke?

- Yes
- No

29. Have you ever experienced a transient ischemic attack (TIA)?

- Yes
- No

30. Have you ever been diagnosed with a brain abscess?

- Yes
- No

31. Have you undergone brain vascular imaging (magnetic resonance angiography and/or CT angiography)?

- Yes
- No

Gastrointestinal involvement

32. Have you undergone upper gastrointestinal endoscopy?

- Yes
- No

33. Were upper gastrointestinal telangiectases identified?

- Yes
- No

34. Have you undergone colonoscopy?

- Yes
- No

35. Were lower gastrointestinal telangiectases identified?

- Yes
- No

Hepatic involvement

36. Have you undergone hepatic Doppler ultrasound?

- Yes
- No

37. If hepatic Doppler ultrasound was performed, were hepatic arteriovenous malformations identified?

- Yes
- No

Epistaxis Severity Score

38. Epistaxis Severity Score (ESS)