ALGORITHMS FOR THE DIAGNOSIS, EVALUATION, AND MANAGEMENT OF HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)

Description:

The HHT Foundation International, Inc. (“Cure HHT”) has developed algorithms for the diagnosis, evaluation, and management of Hereditary Hemorrhagic Telangiectasia (HHT). These algorithms are designed to guide the screening and management of HHT according to the "Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia"[1]. HHT, a genetic disorder characterized by abnormal blood vessel formation, requires a systematic approach for effective patient care, encompassing diagnosis, screening, and ongoing management.

Key Components of the Algorithms:

1. Diagnosis

- **Clinical Criteria:** The algorithm incorporates the Curaçao criteria, which includes recurrent nosebleeds, mucocutaneous telangiectasia, visceral arteriovenous malformations (AVMs), and a family history of HHT.
- **Genetic Testing:** Recommended for patients meeting clinical criteria or with a family history suggestive of HHT.

2. Evaluation

- **For Patients:** Screening for visceral lesions, such as cerebral and pulmonary AVMs, is advised. This includes imaging modalities like MRI and echocardiography, based on individual risk factors and clinical presentation.
- **Family Members:** Screening for asymptomatic relatives is recommended, particularly in those with a known family history of HHT.

3. Management

- Symptomatic Treatment: Focuses on managing epistaxis and gastrointestinal bleeding through medical and procedural interventions.
- Preventative Treatment: Guidelines suggest specific approaches for the management of AVMs, including embolization and surgical options.

4. Follow-Up

- **Regular Monitoring:** Emphasis is placed on regular follow-up to monitor disease progression and manage complications.

These algorithms align with the comprehensive recommendations provided by Faughnan et al. (2020)[1], ensuring that patients with HHT receive consistent and evidence-based care. The guidelines emphasize a multidisciplinary approach, incorporating specialists from various fields to address the complex needs of HHT patients.

Diagnosis of HHT Using Curaçao Criteria

- **Epistaxis:** Spontaneous and recurrent
- **Telangiectases:** Multiple at characteristic sites (lips, oral cavity, fingers, nose)
- **Visceral lesions:** Gastrointestinal telangiectasia, pulmonary, hepatic, cerebral, spinal arteriovenous malformations
- **Family history:** A first-degree relative with HHT according to these criteria

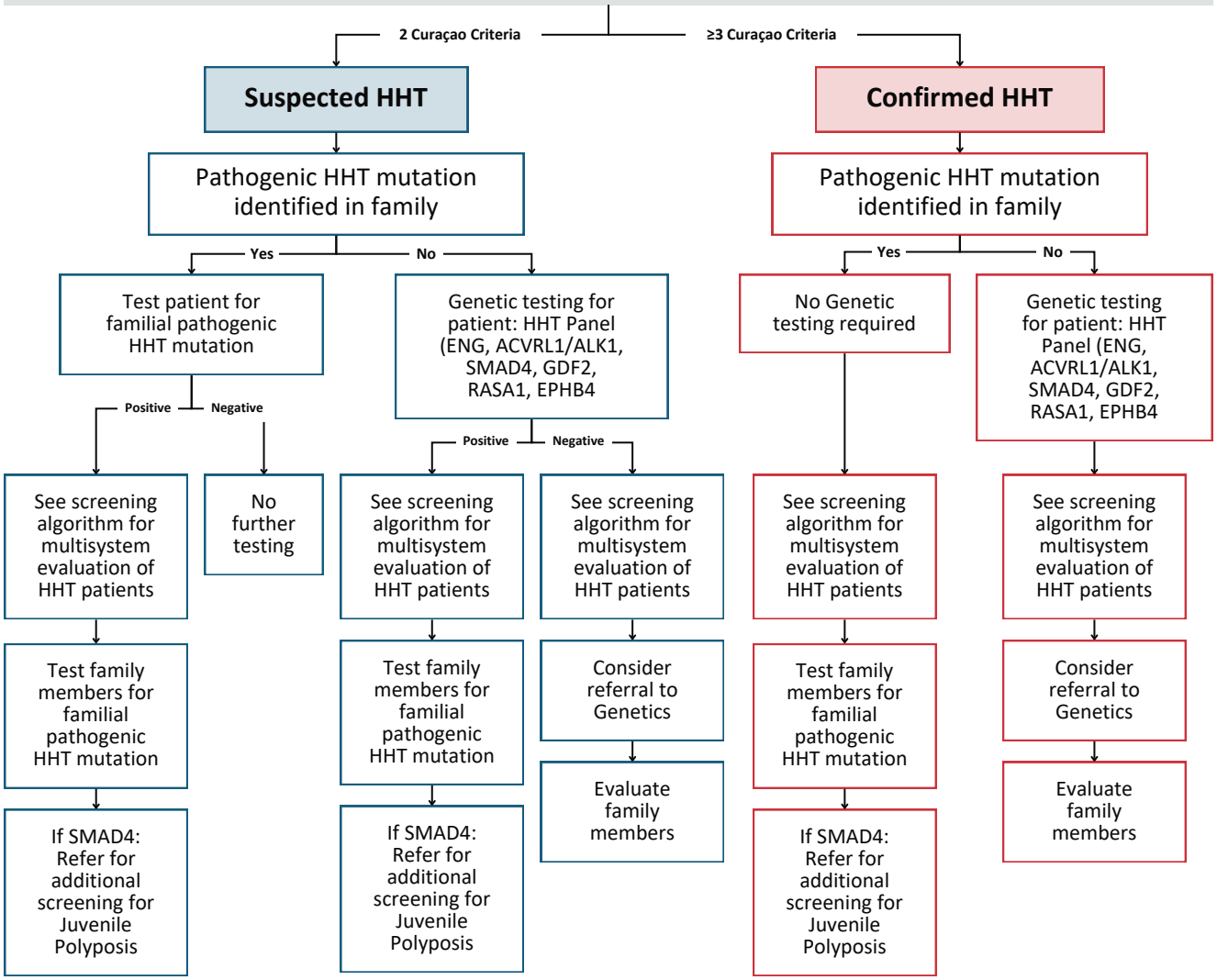

Figure 1. Algorithm for the diagnosis of HHT patients.

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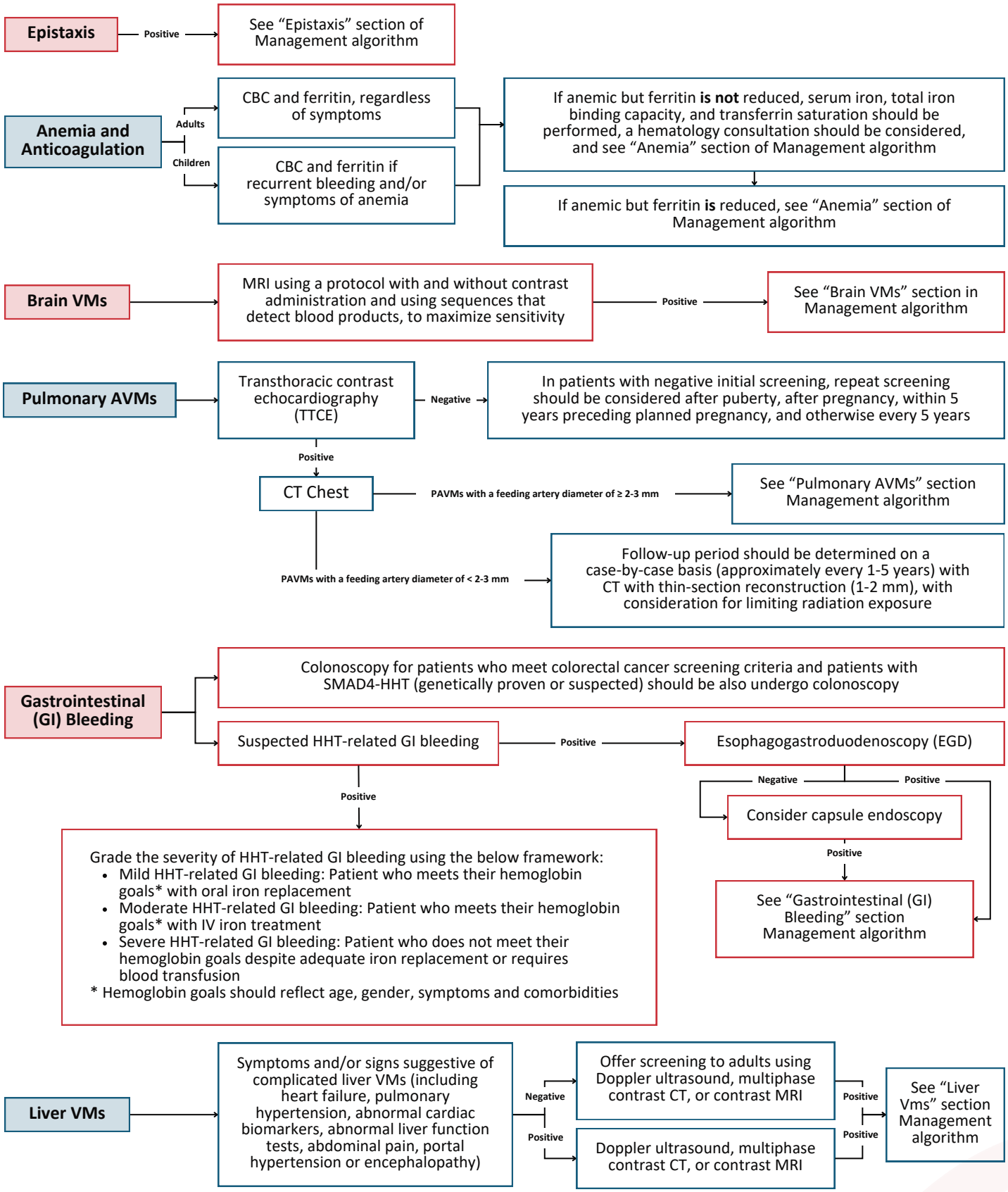

Figure 2. Screening algorithm for the multisystem evaluation of HHT patients.

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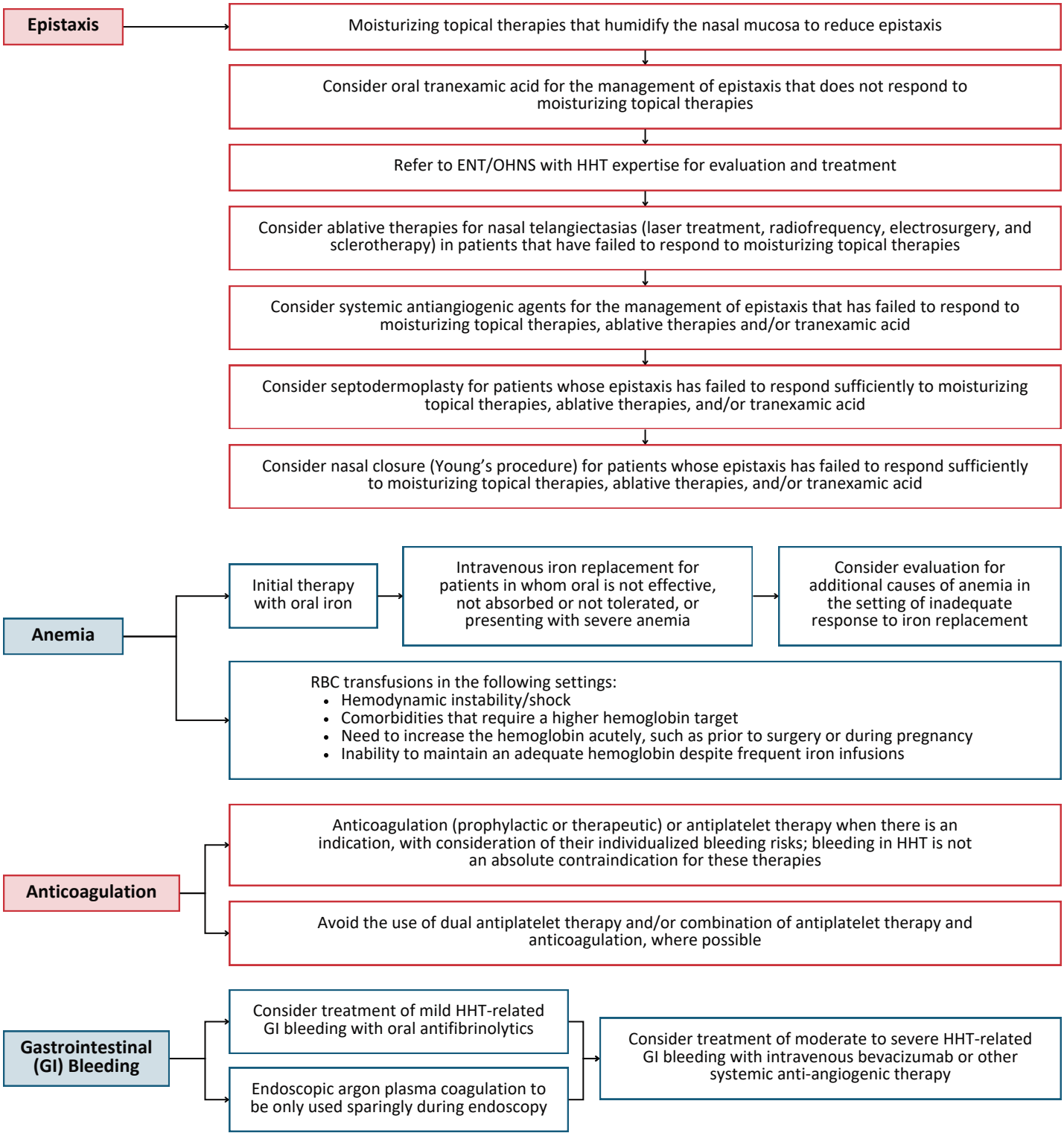

Figure 3. Management algorithm for epistaxis, anemia, anticoagulation, and gastrointestinal (GI) bleeding in HHT patients.

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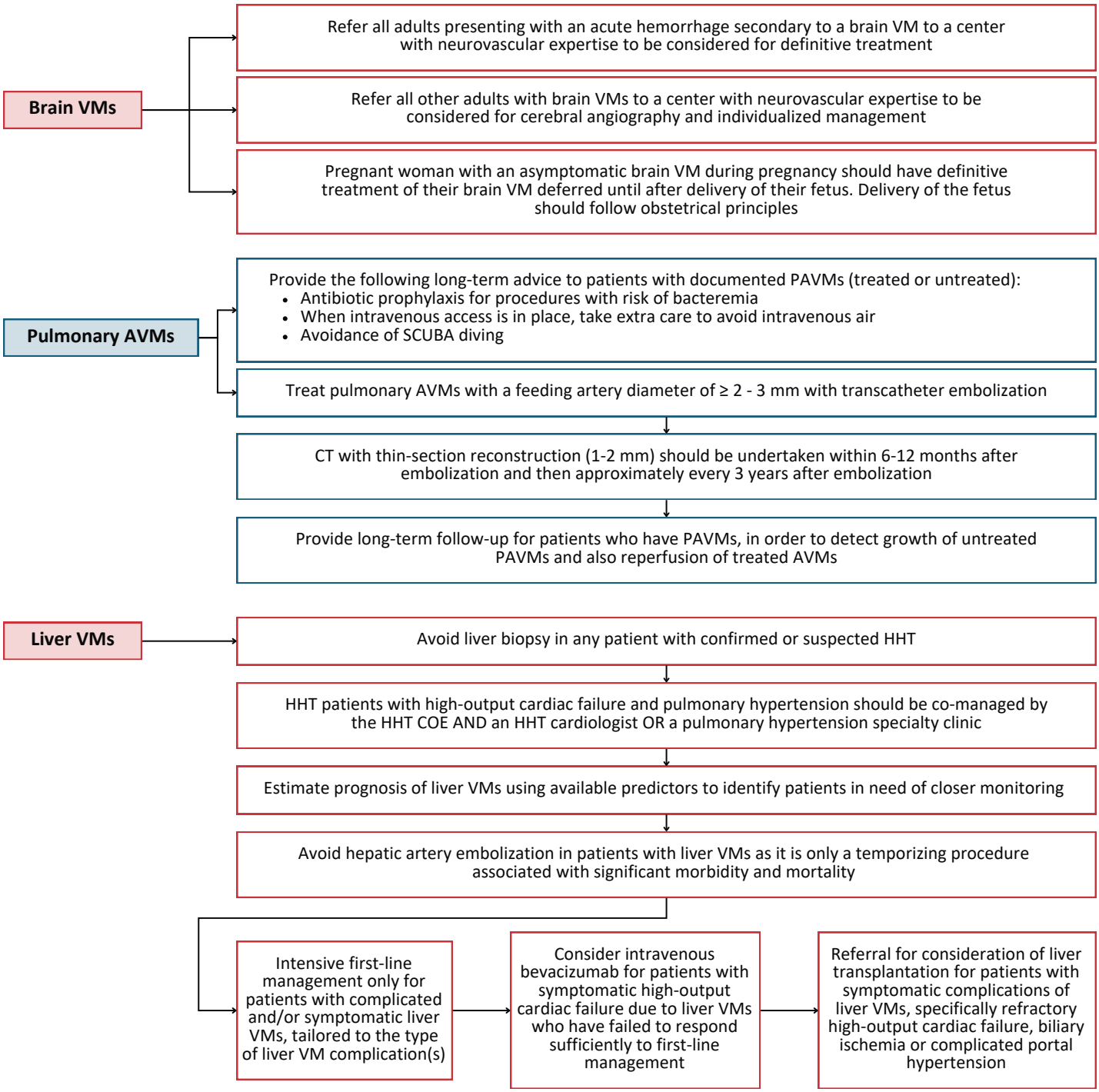

Figure 4. Management algorithm for brain VMs, pulmonary VMs, and liver VMs in HHT patients.

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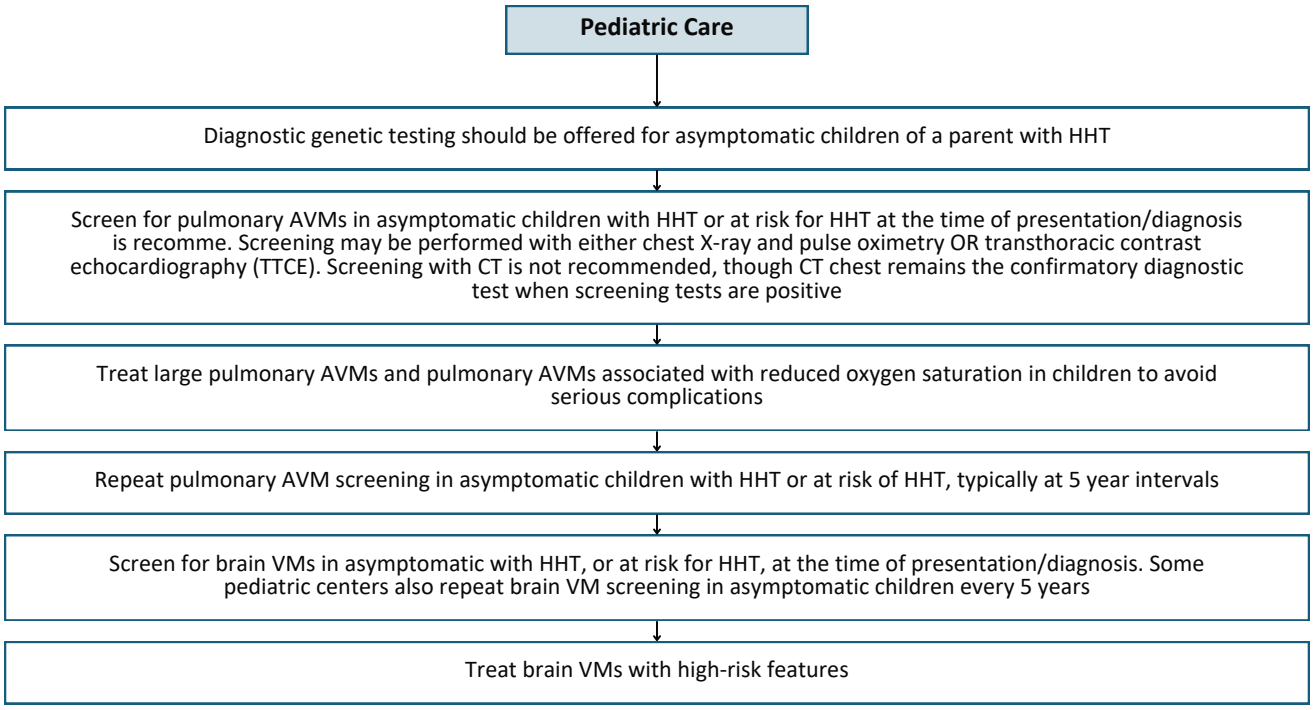

Figure 5. Algorithm for the evaluation and management of pediatric HHT patients.

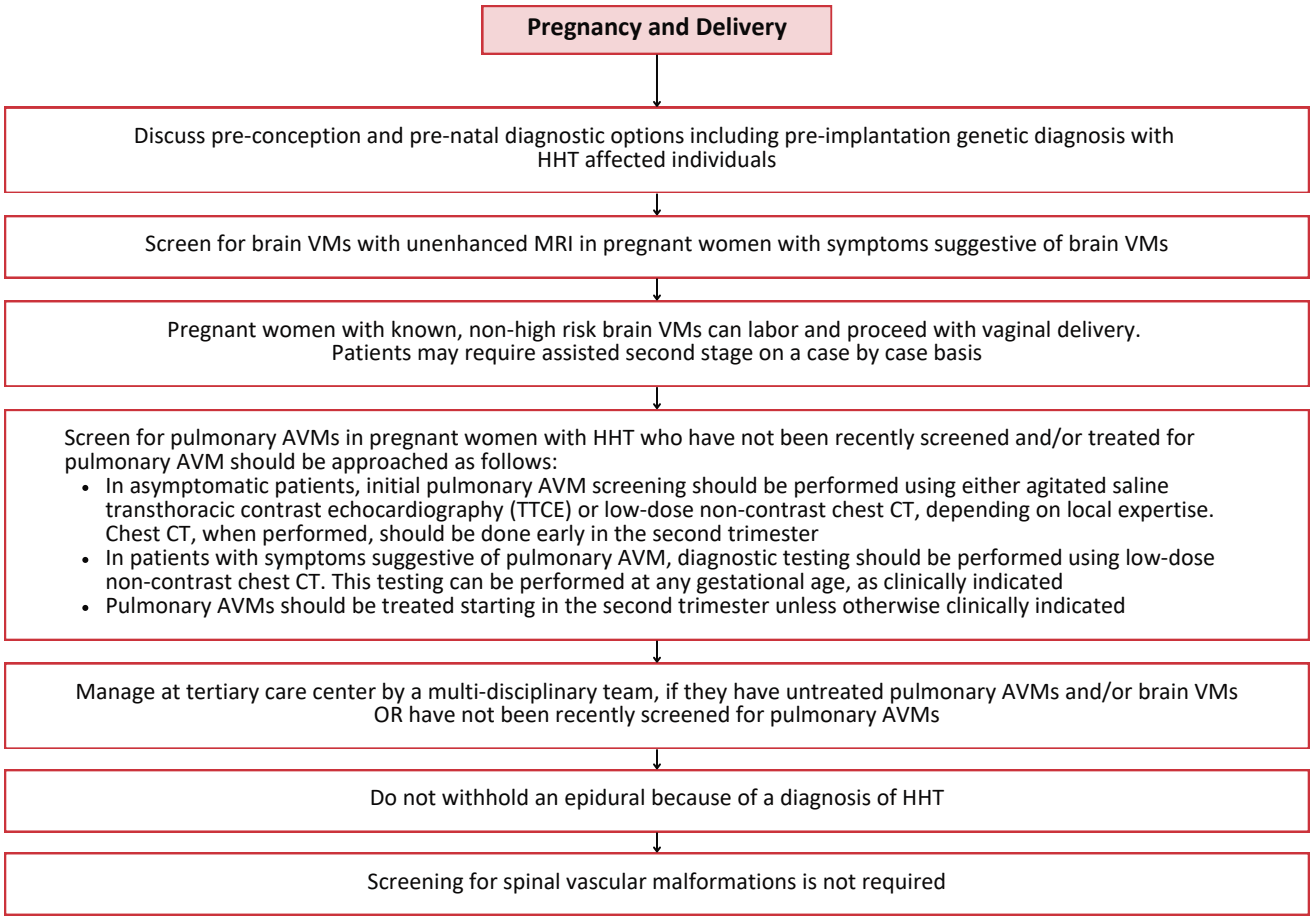

Figure 6. Algorithm for evaluation and management during pregnancy and delivery in HHT patients.

References:

1. Faughnan ME, Mager JJ, Hetts SW, et al. Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. *Ann Intern Med*. 2020 Dec 15;173(12):989-1001.
2. Faughnan ME, Palda VA, Garcia-Tsao G, et al. International guidelines for the diagnosis and management of hereditary haemorrhagic telangiectasia. *J Med Genet*. 2011 Feb;48(2):73-87.
3. Alkhalid Y, Darji Z, Shenkar R, Clancy M, et al. Multidisciplinary coordinated care of hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu disease). *Vasc Med*. 2023 Apr;28(2):153-165.
4. Beslow LA, White AJ, Krings T, et al. Current Practice: Rationale for Screening Children with Hereditary Hemorrhagic Telangiectasia for Brain Vascular Malformations. *AJNR Am J Neuroradiol*. 2024;45(9):1177-1184. Published 2024 Sep 9.

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