**The important role of Dentists in early diagnosis of HHT**

HHT is a hereditary life-threatening disease known. An early diagnosis and appropriate care can avoid its worst potential complications. Dentists can be the first to spot HHT manifestations and refer patients to a center of diagnosis and care, contributing to the overall well-being of the patient and their family members.

## Understanding HHT

HHT is a rare genetic vascular disease with autosomal dominant inheritance, with variable penetrance. Symptoms and signs may appear from a very early age and develop progressively. HHT prevalence is of approximately 1 in 6,000 people. HHT causes mucocutaneous telangiectases and visceral arteriovenous malformations, particularly in the lungs, liver, brain, gastrointestinal tract and central nervous system.

The visible signs are mucocutaneous telangiectases; visceral arteriovenous malformations responsible for haemorrhages, strokes and brain abcesses, remain more silent and hidden.

One of the consequences of mucocutaneous telangiectases is recurrent epistaxis. Epistaxis is common, and will not immediately lead a clinician to suspect a rare disorder. On the other hand, oral and facial mucocutaneous telangiectases, observed by an informed dentist, can lead to an early (potentially life-saving) diagnosis. For the sake of these patients, we are conducting this informative campaign and encourage you to read on.

## Clinical Diagnosis of HHT

Clinical Diagnosis is established by Curaçao clinical criteria:

- Spontaneous and recurrent epistaxis.

- Presence of characteristic multiple mucocutaneous telangiectases on the lips, tongue, oral cavity, fingers and facial skin.
- Presence of arteriovenous malformations (AVMs) in the lungs, liver, brain, digestive system, spine.
- Family history of a first-degree relative fulfilling at least two of the above criteria.

It is not infrequent to find a HHT patient with a *de novo* mutation and no relevant family history.

If two of these 4 criteria are met the diagnosis is uncertain and requires additional investigation. If 3 or 4 of the criteria are met the diagnosis is highly likely.

## Dentists and HHT: A Privileged point of view

HHT is often unrecognized by health care providers. Since the facial, labial and oral telangiectases are in the dentist's field of vision, he or she can easily detect their presence.

- Oral Telangiectases affect 75% of HHT patients. They usually appear from the second decade of life onwards and tend to increase in number with age.
- Intraoral lesions may be macular (flat) or papular (elevated) and are usually between 1-7mm in diameter. They may resemble mucocutaneous petechiae, but unlike petechiae, telangiectases blanch upon applied pressure.
- Most oral (and skin) telangiectases are asymptomatic, yet are prone to rupture and hemorrhage. Bleeding gums may result from brushing the teeth or other minor oral trauma.

Should a patient exhibit these lesions, it is advised to question them whether they or other family members suffer from epistaxis. This can further support a clinical suspicion of HHT and facilitate an early firm diagnosis.

## Risks and Cautionary measures when treating an HHT Patient

25-30% of patients with HHT have pulmonary arteriovenous malformations (PAVMs), while up to 90% of patients with PAVMs are HHT patients. These lesions cause direct communication between the pulmonary and systemic circulation and thus can lead to hypoxaemia, rupture of the PAVMs, haemothorax, strokes, brain and liver abscesses. Dental procedures on a HHT Patient with untreated PAVMs have the potential to result in **brain abscesses** due to paradoxical right to left shunting of oral bacteria. Some cases of liver abscesses have also been described. Effective identification and treatment of PAVMs is of paramount importance for HHT patients.

**Antibiotic Prophlylaxis in HHT Patients with Dental Procedures**

The International Guidelines for HHT recommend **antibiotic prophylaxis** before all invasive procedures in HHT patients with PAVMs or when PAVMs have not been ruled out by screening.

The prophylaxis should cover a broad spectrum of the usual bacterial flora of the mouth. Current recommended antibiotics for prophylaxis are co-amoxyclav (**amoxicillin/clavulanic acid** combination), or **clindamycin** in patients with penicillin allergy

**Where to refer patients you suspect have HHT**

Should a patient present signs of HHT that, in your opinion, require further investigation, we highly recommend you advise the patient to contact the National HHT Centre in Mercy University Hospital, Cork (margaretmurphyhht@gmail.com) for evaluation.

Patient Organisation – HHT Ireland
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**This document has been put together by HHT Ireland - Patient Organisation for HHT patients & their families - in kind association with Dr Adrian Brady, Consultant Radiologist from national HHT Centre, Mercy University Hospital, Cork.**

**Further Reading:**


Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia Marie E Faughnan https://pubmed.ncbi.nlm.nih.gov/32894695/

International guidelines for the diagnosis and management of hereditary haemorrhagic telangiectasia
M E Faughnan https://pubmed.ncbi.nlm.nih.gov/19553198/

Post-NICE 2008: antibiotic prophylaxis prior to dental procedures for patients with pulmonary arteriovenous malformations (PAVMs) and hereditary haemorrhagic telangiectasia - C.Shovlin - K. Bramford https://www.nature.com/articles/sj.bdj.2008.978

Detecting Hereditary Hemorrhagic Telangiectasia Sherri M. Lukes, RDH, MS, FAADH https://dimensionsofdentalhygiene.com/article/detecting-hereditary-hemorrhagic-telangiectasia/

https://www.cda-adc.ca/jcda/vol-75/issue-7/527.pdf

https://www.cdho.org/Advisories/CDHO_Factsheet_HHT.pdf