European Reference Network
for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

VASCERN DO'S AND DON'TS FACTSHEETS FOR RARE VASCULAR DISEASE PATIENTS FACING FREQUENT SITUATIONS

Hereditary Haemorrhagic Telangiectasia

European Reference Network
for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

Co-funded by the Health Programme of the European Union

VASCERN

VASCERN, the European Reference Network on Rare Multisystemic Vascular Diseases, is dedicated to gathering the best expertise in Europe in order to provide accessible crossborder healthcare to patients with rare vascular diseases (an estimated 1.3 million concerned). These include arterial diseases (affecting aorta to small arteries), arterio-venous anomalies, venous malformations, and lymphatic diseases.

VASCERN currently gathers 48 expert teams from 39 highly specialized multidisciplinary HCPs, plus 6 additional Affiliated Partner centers, coming from 19 EU Member States, as well as various European Patient Organisations, and is coordinated in Paris, France.

Through our 6 Rare Disease Working Groups (RDWGs) as well as several thematic WGs and the ePAG (European Patient Advocacy Group), we aim to improve care, promote best practices and guidelines, reinforce research, empower patients, provide training for healthcare professionals and realise the full potential of European cooperation for specialised healthcare by exploiting the latest innovations in medical science and health technologies.

More information available at: https://vascern.eu
Follow us on Twitter, Facebook, YouTube and LinkedIn

Table of contents

Hereditary Haemorrhagic Telangiectasia

Abbreviations

HHT: Hereditary Haemorrhagic Telangiectasia

HHT-WG: Hereditary Haemorrhagic Telangiectasia Working Group

APA: Antiplatelet agent

ENT doctor: Ear, Nose, and Throat doctor

AVM: Arteriovenous malformation

VM: Vascular malformation

SaO2: Oxygen saturation

Introduction

These factsheets are based on existing French factsheets which have been reviewed and adjusted by the experts of the VASCERN HHT-WG.

The HHT-WG agrees with the recommendations but wishes to emphasize that these are recommendations made by consensus at expert level. We would recommend that these factsheets be used as a guide to implement locally agreed policies.

These factsheets are meant for patients as well as for caregivers. Implementing these recommendations should go hand in hand with strategies to educate patients about medical situations where specific care is required and about relevant symptoms and how to act when they occur.

Physical activity

WHAT IS RECOMMENDED

- There are no physical activity or sport restrictions, except in the event of acute hypoxia.

WHAT YOU SHOULD NOT DO

- Scuba diving with a diving tank in patients with pulmonary arteriovenous malformations, even if embolised (risk of air embolism).

Breast feeding

WHAT IS RECOMMENDED

- Breastfeeding is not contraindicated in women with hereditary haemorrhagic telangiectasia.

WHAT YOU SHOULD NOT DO

- No specific recommendations.

Contraindicated medications

No medication is formally contraindicated.

WHAT IS RECOMMENDED

- Always discuss the risks and benefits with the centre of reference or competence responsible for the care of the patient.
- Adapt the treatment to the patient's clinical condition (epistaxis, gastrointestinal bleeding).

WHAT YOU SHOULD NOT DO

- Prescribe antiplatelet agents (APAs) or anticoagulants without having weighed the potential risks and benefits.

Antiplatelet agents (APA) and anticoagulants

WHAT IS RECOMMENDED

- Discuss the risks and benefits with the centre of reference or expertise responsible for the patient.
- Adapt the treatment to the patient's clinical condition (epistaxis, gastrointestinal bleeding).
- After an ischemic stroke secondary to pulmonary arteriovenous malformations, there is no indication to continue this type of treatment (APA or anticoagulant) if all pulmonary arteriovenous malformations have been treated satisfactorily.

WHAT YOU SHOULD NOT DO

- Prescribe APAs or anticoagulants without having weighed the risks and benefits.

Deep-vein thrombosis, pulmonary embolism
(or venous thromboembolic disease)

No medication is formally contraindicated.

WHAT IS RECOMMENDED

- Follow the standard treatment for thrombosis and/or pulmonary embolism (anticoagulant treatment) having weighed the potential risks and benefits.
- Adapt the treatment to the patient's clinical condition (epistaxis, gastrointestinal bleeding, blood count).
- In the event of increased epistaxis while taking anticoagulation treatment, arrange for a consultation with an ENT doctor who has knowledge of the disease in order to consider appropriate treatment.
- Outside of an emergency situation, and depending on anti-coagulant tolerance, discuss alternative treatments (thrombectomy, cava filter) with the centre of reference or competence.

WHAT YOU SHOULD NOT DO

- Avoid treating vascular thromboembolic disease because of hereditary haemorrhagic telangiectasia.

Haemorrhagic stroke

WHAT IS RECOMMENDED

- Emergency management and treatment (as in non-HHT patients).
- Look for underlying brain AVMs to prevent recurrence.
- If the patient's clinical condition requires the insertion of a nasogastric tube, it should be soft, of small diameter (unless clinical circumstances demand a large bore tube), and put in place with extreme caution due to the risk of triggering a severe episode of epistaxis related to the presence of mucous telangiectases.

WHAT YOU SHOULD NOT DO

- No specific contraindications.

Brain abscesses

A brain abscess is a classic complication of hereditary haemorrhagic telangiectasia. It is related to the right-to-left shunt secondary to pulmonary arteriovenous malformations.

WHAT IS RECOMMENDED

- Emergency management and treatment (as in non-HHT patients).
- Perform a chest CT scan without injection, or contrast echocardiogram to identify pulmonary arteriovenous malformations (the most common cause of brain abscess in hereditary haemorrhagic telangiectasia), and treat the pulmonary arteriovenous malformations to reduce the risk of recurrence.
- If the patient's clinical condition requires the insertion of a nasogastric tube, this should be soft, of small diameter (unless clinical circumstances demand a large bore tube), and put in place with extreme caution due to the risk of triggering a severe episode of epistaxis related to the presence of mucous telangiectases.

WHAT YOU SHOULD NOT DO

- No specific contraindications.

Heart failure

Heart failure in Hereditary Haemorrhagic telangiectasia may be related to the evolution of hepatic AVMs that can entail cardiac chronic overload: both hepatic and cardiac hemodynamics must be investigated.

Medical treatment will be adapted to each particular case: treatment of heart failure, correction of anemia, management of arrhythmia.

WHAT IS RECOMMENDED

- Measure cardiac output and the cardiac index, the filling pressures and the presence or absence of pulmonary hypertension (often post-capillary).
- Search for hepatic arteriovenous malformations (Doppler ultrasound and/or hepatic scan).
- Refer the patient to a centre of reference.
- Correct the anaemia.

WHAT YOU SHOULD NOT DO

- Overlook cardiac evaluation (including echocardiography) if severe liver VMs are present.
- Treat pulmonary hypertension secondary to liver VMs with high output cardiac failure, with vasodilators.

Kidney failure

WHAT IS RECOMMENDED

- No contraindications to kidney biopsy puncture after exclusion of kidney AVMs by doppler sonography.

WHAT YOU SHOULD NOT DO

- No specific contraindications.

Care for patient with multiple traumatic injuries

It is always necessary to contraindicate nasal manipulations (nasal intubation, aspirations, etc.) due to the significant risk of triggering sometimes very severe episodes of epistaxis linked to mucous telangiectases.

Apart from the risk of bleeding related to the presence of mucous telangiectases (nasal, gastrointestinal), there are no coagulation anomalies associated with hereditary haemorrhagic telangiectasia and no surgical bleeding risk connected with this pathology.

WHAT IS RECOMMENDED

- Check that there is no low SaO2 that could be related to the presence of undiagnosed pulmonary AVMs, which would warrant treatment.

WHAT YOU SHOULD NOT DO

- Intubate or aspirate through the nose: risk of severe epistaxis.

Bronchoscopies

Be aware that coughing may be tolerated less well than in other patients due to the risk of haemoptysis from tube abrasion.

WHAT IS RECOMMENDED

- Follow the standard fibroscopy technique.
- In case of biopsy, perform with antibiotic prophylaxis if pulmonary AVMs are present, or if pulmonary status is unknown.

WHAT YOU SHOULD NOT DO

- Nasal manipulations during anaesthesia (nasal intubation, aspirations, etc.) due to the significant risk of triggering sometimes very severe episodes of epistaxis linked to mucous telangiectases.

Scheduled surgery / Anaesthesia

Before any surgery it is always necessary to contraindicate nasal manipulations (nasal intubation, aspirations, etc.) due to the significant risk of triggering sometimes very severe episodes of epistaxis linked to mucous telangiectases.

Apart from the risk of bleeding related to the presence of mucous telangiectases (nasal, gastrointestinal), there are no coagulation anomalies associated with hereditary haemorrhagic telangiectasia and no surgical bleeding risk connected with this pathology.

WHAT IS RECOMMENDED

- Emergency management and treatment (as in non-HHT patients).
- Follow standard treatment protocols for this pathology.
- If the patient's clinical condition requires the insertion of a nasogastric tube, this should be soft, of small diameter (unless clinical circumstances demand a large bore tube), and put in place with extreme caution due to the risk of triggering a severe episode of epistaxis related to the presence of mucous telangiectases.

WHAT YOU SHOULD NOT DO

- Intubate or aspirate through the nose: risk of severe epistaxis.

For a patient with pulmonary AVM(s) who has had an ischemic stroke

WHAT IS RECOMMENDED

- Recommend single antiplatelet.
- Treat iron deficiency.
- Reassess if further interventional PAVM treatment is possible. An effective embolotherapy might make an antiplatelet therapy redundant.
- Involve stroke neurologists on a case-by-case basis when there are untreated PAVMs and MR/CT signs of previous cerebral ischemic events.

WHAT YOU SHOULD NOT DO

- Give a prophylactic antiplatelet therapy in patients with PAVM if no stroke has occurred (except if there are other indications, such as atrial fibrillation).
- Prescribe dual antiplatelet therapy or combined antiplatelet and anticoagulant therapy.

Editorial Board/ Contributors

Based on the French original document produced by:
Fava-Multi (The French Network for Rare Vascular Diseases)

Editorial Board:

Dr Sophie DUPUIS-GIROD, HHT Center of Reference, CHU de Lyon HCL, France

Review Board from the French HHT Network:

Prof Brigitte GILBERT-DUSSARDIER (HHT Expert Centre, Poitiers), **Didier ERASME** (AMRO-HHT-France), **Prof Laurent LACCOUREYE** (HHT and Vascular Diseases Expert Centre, Angers), **Dr Christian LAVIGNE** (HHT and Vascular Diseases Expert Centre, Angers), **Dr Sophie RIVIÈRE** (HHT Expert Centre Montpellier), **Dr Geoffrey URBANSKI** (HHT and Vascular Diseases Expert Centre, Angers)

And other members of the French HHT Network:

Prof Marie-France CARETTE (HHT Expert Centre Paris – Hôpital Tenon), **Prof Thierry CHINET** (HHT Expert Centre Paris – Hôpital Ambroise Paré), **Dr Anne CONTIS** (HHT Expert Centre Bordeaux - Hôpital Jean Bernard), **Dr Romain CORRE** (HHT Expert Centre Rennes - Hôpital Pontchaillou), **Dr Pierre DUFFAU** (HHT Expert Centre Bordeaux - Hôpital Jean Bernard), **Xavier DUFOUR** (HHT Expert Centre Poitiers - Hôpital Jean Bernard), **Prof Vincent GROBOST** (HHT Expert Centre - CHU Estaing Clermont-Ferrand), **Pr Jean- Robert HARLE** (HHT Expert Centre Marseille - Hôpital de la Timone), **Prof Pierre-Yves HATRON** (HHT Expert Centre Lille -Hôpital Claude Huriez), **Dr Shirine MOHAMED** (HHT Expert Centre - CHRU Nancy), **Dr Pascal MAGRO** (HHT Expert Centre - Tours - Hôpital Bretonneau), **Dr Antoine PARROT** (HHT Expert Centre - Paris – Hôpital Tenon)

Hereditary Haemorrhagic Telangiectasia

English version translated by VASCERN and updated by members of the VASCERN HHT-WG:

Prof Elisabetta BUSCARINI (VASCERN HHT European Reference Centre, Maggiore Hospital, ASST Crema, Italy)

Claudia CROCIONE (HHT Europe/Associazione Italiana Teleangectasia Emorragica – HHT ONLUS)

Dr. Freya DROEGE (VASCERN HHT European Reference Centre, Essen Center for Rare Diseases, Essen
University Hospital, Germany)

Dr. Sophie DUPUIS-GIROD (VASCERN HHT European Reference Centre, CHU de Lyon HCL, France)

Prof. Urban GEISTHOFF (VASCERN HHT European Reference Centre, Odense University Hospital, Denmark)

Christina GRABOWSKI (Morbus Osler Selbsthilfe e.V.)

Dr. Ruben HERMANN (VASCERN HHT European Reference Centre, CHU de Lyon HCL, France)

Josefien HESSELS (VASCERN HHT European Reference Centre, St. Antonius Hospital Nieuwegein, The Netherlands)

Prof. Anette KJELDSEN (VASCERN HHT European Reference Centre, Odense University Hospital, Denmark)

Dr. Mikkel Seremet KOFOED (VASCERN HHT European Reference Centre, Odense University Hospital, Denmark)

Dr. Guido MANFREDI (VASCERN HHT European Reference Centre , Maggiore Hospital, ASST Crema, Italy)

Dr. Gennaro Mariano LENATO (VASCERN HHT European Reference Centre, Azienda Ospedaliero-Universitaria Consorziale di Bari Policlinico-Giovanni XXIII, Bari, Italy)

European Reference Network
for rare or low prevalence complex diseases
Network Vascular Diseases (VASCERN)

Co-funded by the Health Programme of the European Union

Hereditary Haemorrhagic Telangiectasia

English version translated by VASCERN and updated by members of the VASCERN HHT-WG:

Dr. Hans-Jurgen MAGER (VASCERN HHT European Reference Centre, St. Antonius Hospital Nieuwegein, The Netherlands)

Prof. Carlo SABBÀ (VASCERN HHT European Reference Centre, Azienda Ospedaliero-Universitaria Consorziale di Bari Policlinico-Giovanni XXIII, Bari, Italy)

Prof. Claire SHOVLIN (VASCERN HHT European Reference Centre, Hammersmith Hospital, Imperial College Healthcare NHS Trust London, UK)

Dr. Patricia SUPPRESSA (VASCERN HHT European Reference Centre, Azienda Ospedaliero-Universitaria Consorziale di Bari Policlinico-Giovanni XXIII, Bari, Italy)

Dr. Pernille M TØRRING (VASCERN HHT European Reference Centre, Odense University Hospital, Denmark)

European Reference Network
for rare or low prevalence complex diseases
Network Vascular Diseases (VASCERN)

Co-funded by the Health Programme of the European Union