
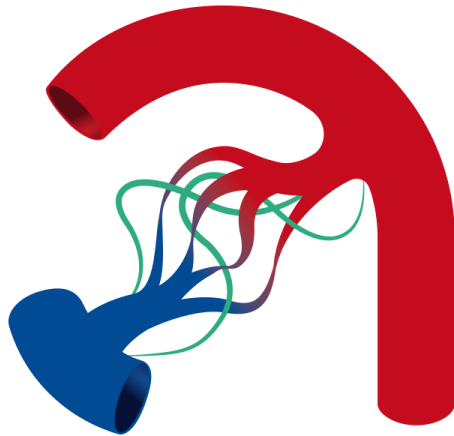




European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



#VASCERNdays2018



WiFi at Holiday Inn Express
Password: meeting1

VASCERN DAYS 2018 PLENARY SESSION

11-12 October, 2018
Paris, France

Welcoming words

- **72 Healthcare Professionals** from our **31 HCP Members'** expert teams





Welcoming words

- **15 Patient Advocates** members of our European Patient Advocacy Group



**European
Patient
Advocacy
Group**



Welcoming words

- The VASCERN **Coordination team**



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(VASCERN)

 **Coordinator**
Assistance Publique-
Hôpitaux de Paris, Hôpital
Bichat – France





Welcoming words

- **Guests** from several institutions



DIRECTION
GÉNÉRALE
DE L'OFFRE
DE SOINS





Program

Morning

- Plenary & Board session, including policy and project updates
- Feedback from WGs on Work Packages Progress

Afternoon

- rare disease working groups - parallel sessions
- epag

Networking evening

Tomorrow morning

- transversal working groups - parallel sessions
- rare disease working groups - parallel sessions

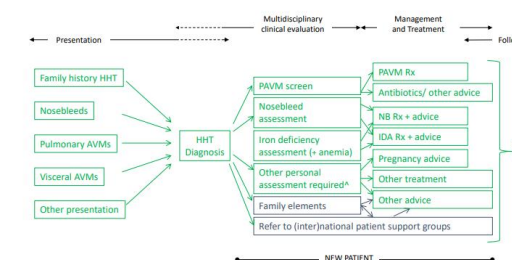
Afternoon

- debriefing in plenary session

After 19 months



Hereditary haemorrhagic telangiectasia
Patient Pathway v2, 11/07/2017



Vascular EDs

Final approved patient pathway
28/09/2018

IT'S SIMPLE! SEARCH FOR THE RARE DISEASE



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

Vascular Ehlers-Danlos



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

VASCERN Consensus Statement: ACTA2-Related Vasculopathy

VASCERN HTAD working group Members: Ingrid van de Lest*, Eloisa Arbustini, Kalman Benke, Erik Björck, Maarten Groenink, Marië Kempers, Viktor von Kodolitsch, Bart Loey, Barbara Mulder, Lisa Murphy, Guglielmina Pepe, Alessandro Pini, Leema Robert, Jolien Roos-Hesselink, Zoltan Szabolcs, Guillaume Jondeau, Julie De Backer.

Cooperating Guests: Arturo Evangelista, Ruxandra Jurcut, Celsa Teixeira

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Hereditary Haemorrhagic Telangiectasia

Red Flags

A combination of features below may raise suspicion

- Family history of vEDS with confirmed COL3A1 mutation
- Arterial dissection/rupture at young age (<40yrs)
- Spontaneous bowel perforation (with no known risk factors)
- 3rd trimester uterine rupture (with no known risk factors)
- CSSF in absence of trauma
- Bruising in unusual sites with undefined trauma
- Characteristic facial appearance or acrogeria
- Spontaneous pneumothorax
- Early onset varicose veins <30yrs
- Other features such as Club foot, Thin skin, Keratoconus, Gingival fragility

Multidisciplinary clinical evaluation

Cardiac and arterial imaging, Clinical examination and assessment, Family history, Molecular testing

Management and Follow up

Confirm vEDS with mutation in COL3A1

Access to teams required include:

1. Cardiology
2. Vascular and endovascular surgery
3. Bowel surgery
4. Neurovascular surgery
5. Thoracic surgery
6. Obstetrics and fetal medicine
7. Vascular imaging
8. Psychosocial input

Surveillance and Follow up*

Proband / Relative NEW PATIENT

*In accordance with clinical need and geographical location. Should include:
1) Ongoing access to services on demand
2) Ongoing support of local medical teams
3) Individual follow up as required for new clinical indications such as pregnancy, new symptoms assessment...

Thoracic Aortic Dissection

- Child: z-score ≥3
- Adult I: 2.5 > z-score < 3.5 | ≤60y.o. (No AHT¹) | >60y.o. (AHT¹)
- z-score ≥3.5

Root Aneurysm

m Size Artery Aneurysm/Dissection

e Family History²

ent Pneumothorax

- <18y.o. | ≤3⁴ | >3
- ≥18y.o. | ≤7 | >7

al Features³

a Lentis

id Aortic Valve⁵

POSITION STATEMENT

European Reference Network For Rare Vascular Diseases (VASCERN) Outcome Measures For Hereditary Haemorrhagic Telangiectasia (HHT)

Claire L. Shovlin^{1,2*}, Elisabetta Bucarini³, Anette D. Kjeldsen⁴, Hans Jurgen Mager⁵, Carlo Sabbà⁶, Freya Urban Gesthoff^{7,8}, Sara Ugolini⁹ and Sophie Dupuis-Grod^{10,11*}

Abstract
Hereditary haemorrhagic telangiectasia (HHT) is a multisystemic vascular dysplasia that leads to nosebleeds, anaemia to blood loss, and arteriovenous malformations (AVMs) in organs such as the lungs, liver and brain. HHT is estimated to affect 80,000 European citizens, but most health care providers have limited prior HHT exposure or training. Outcome Measures were developed and implemented by the HHT Working Group of the European Reference Network for Rare Vascular Diseases (VASCERN), in order to maximise the number of patients receiving good care. The measures specifically target areas where optimal management reduces morbidity and mortality in HHT patients, and were designed to be robust to emerging new evidence. Thresholds are the percentage of patients in particular settings who have recommended screening, or provided with written advice. The 5 Outcome Measures cover (1) pulmonary AVM screening; (2) written nosebleed advice; (3) assessment of iron deficiency; (4) antibiotic prophylaxis prior to dental and surgical procedures for patients with pulmonary AVMs; (5) detailed HHT management, but are suitable for summary. These 5 Outcome Measures promote encourage care improvement by all healthcare providers for patients with pulmonary AVMs.

Keywords: Anaemia, Antibiotic prophylaxis, Ear malformations, Pregnancy

Background
Development and implementation of Outcome Measures are an effective part of Quality and Safety Framework that lead to Service Improvements. More specific Outcome Measures are carefully selected for dissemination and implementation can directly impact patient care, including that from health care providers.

How can lymphedema be diagnosed?

Diagnosing test

ERN COLLABORATIVE PLATFORM [MEMBERS AREA] CLINICAL PATIENT MANAGEMENT SYSTEM [MEMBERS AREA]

VASCERN

Gathering the best expertise in Europe to provide accessible cross-border healthcare to patients with rare vascular diseases

HOME ABOUT VASCERN WHAT WE DO NEWS EDUCATIONAL MATERIAL CONTACT

ePAG (Patient Advocacy)

European Reference Network



Clinical Patient Management System (CPMS)

Latest News

Do's & Don'ts Factsheets for rare vascular disease patients

VASCERN Spotlights

Upcoming

TUE 09 OCT 2018
WED 10 OCT 2018
Clinical Practice Guidelines for Rare Diseases: development and quality assessment



After 1.5 year, what I think about VASCERN

- A lot of **outcomes** (deliverables)
 - For patients:
 - Ongoing translations (Bakumba, Petit Pierre)
 - Pills of Knowledge video on YouTube and subtitles in various EU languages
 - Start of CPMS cases discussion
 - Mobile app
 - For doctors:
 - Publication of patient pathways (homogeneisation), recommendations
 - For research: scientific projects research launched
 - A lot of **communications**:
 - Monthly virtual meetings, face to face meetings
 - Working together
 - Website and social media and communication in meetings
 - Rare Disease Working Groups visible from outside



After 1.5 year, what I think about VASCERN

- Questions to address within ERNs, VASCERN
 - Ambiguity: HCP expertise for ERN work or efficient clinical care network?

Medical coverage

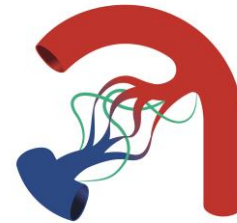


- **Rare disease:** 1 patient / 5000 inhabitants
 - means 200 patients / million inhabitants
 - 2000 patients / 10 millions inhabitants
 - 2000 visits/year/10 millions if lifelong disease and yearly visit
- 511 millions inhabitants in Europe
 - 1 expert centre / 10 millions means 51 expert centres
 - 5 RDWG within VASCERN : $5 \times 51 = 255$ HCPs each seeing 2000 patients an year
- Conclusion: medical coverage needs **at least 510 expert centres** for VASCERN pathologies



Medical coverage ≠ Expertise for ERN

- Conclusion: medical coverage needs at least 510 expert centres for VASCERN pathologies
- Solution: 1 HCP member for each country (or group of countries) per RDWG (140 HCPs either Full or Affiliated Partners)
 - ERN representative of the national network, and vice versa
 - Motivation for being the national representative
 - Motivation from the expert centres to be part of VASCERN (but not full member)
 - No dedicated funding
 - Expertise recognition (logo)
 - Re-reading of documents
 - Yskert Von Kodolitsch





After 1.5 year, what I think about VASCERN

- Questions to address within ERNs, VASCERN
 - Ambiguity: expertise for ERN work or efficient clinical care network ?
 - Solution : 1 HCP per country per RDWG ?
 - No funding for HCP: what is the benefit of being part of VASCERN ?

No funding for HCP: what is the benefit of being part of VASCERN ?



- Additional funding from the EU
 - No increase in funding for ERN coordination or HCP from the EU as of today
 - New EU calls for ERNs for which some HCP may be able to receive co-funding for action (ex: registries)
- New collaborative work & organisation, outcomes delivered
- VASCERN guarantee, to propose in the name of VASCERN is a +
 - Cover expertise centers in Europe : national network with representative
 - Guarantee for research
 - Number of patients reached
 - Translational and basic (including genetics) research
 - because both clinical teams and basic lab
- Proposals : maximal communication
 - National network of clinical centers
 - ERN: network of basic science labs (specific transversal WG ?)



After 1.5 year, what I think about VASCERN

- Questions to address within ERNs, VASCERN
 - Ambiguity: expertise for ERN work or efficient clinical care network ?
 - Solution : 1 HCP per country per RDWG ?
 - No funding for HCP: what is the benefit of being part of VASCERN ?
 - Solution: VASCERN a +: national network for clinical experts, European network for basic science, maximal communication
 - HCP and not expert teams are recognised : creating confusion



European
Reference
Network

VASCERN

Gathering the best expertise in Europe
to provide accessible cross-border healthcare
to patients with rare vascular diseases



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

VASCERN currently consists of 31 highly specialised multidisciplinary Healthcare Providers (HCPs) from 11 EU Member States and of various European Patient Organisations and is coordinated in Paris, France.

Through our 5 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>

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