



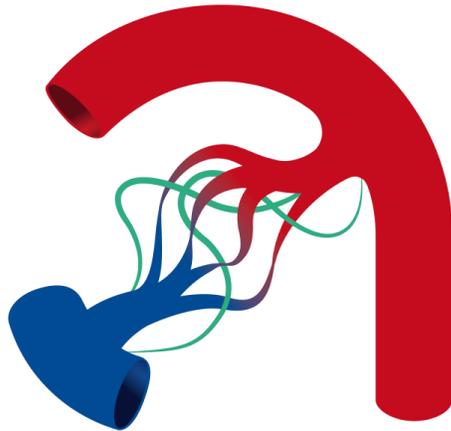
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Implementation of Work Packages

Discussion with the RDWG Chairs: Julie De Backer, Leema Robert, Robert Damstra, Miikka Vikkula, Ulrich Sure representing Claire Shovlin, moderated by Marine Hurard, and Q&A, sharing of best practices and interRDWG projects

<https://vascern.eu/what-we-do/>

Horizontal Work Packages

WP1 Coordination / Project management

M36 Report on coordination activities

WP2 Dissemination/ Communication

M1-36: website, newsletters, social media pages. M36 report

WP3 Evaluation of the action implementation

M36 report (based on ERN monitoring reporting)

Specific Work Packages related to the specific objectives of the action

WP4 [Sharing of experience: discussion of complex clinical cases](#)

M36 report on dissemination activities about case discussions

WP5 [Patient pathways:](#) improvements and updates

M18: mid-period improved pathways, **M36:** 5 or more published improved

WP6 [Mobile Application](#) (updates, improvements, V2, V3)

M18: V2. **M36:** V3

WP7 [Pills of Knowledge](#)

15 PoK / year (3*5 RDWGs)

WP8 [Registries](#)

M18: mid-term report. **M36** report

WP9 [Clinical trials & Research](#)

M18: mid-term report. **M36** report. Launch of 1 EU clinical trial/ year Research news all months

WP10 [Availability of videos on YouTube](#)

M36: 90 added (including 45 POK)

WP11 Multidisciplinary school for vascular disease (**Y5**)

M36: structured course by RDWG realised with the POK collection over 5y

WP12 [Definition of clinical outcomes](#)

M18: mid-term report. **M36** report

WP13 [Writing Clinical Practice Guidelines](#)

M36: 4 new CPGs (expert consensus, recommendations published)

WP14 [Do's and Don'ts factsheets](#)

M18: end of assessment for updates **M36** publication of new / revised

WP15 VASCERN Clinic days(**Y5 -tbc**)

Around **M32-33:** Event in 1 HCP



3 Horizontal Work Packages

Horizontal Work Packages	Milestones and Deliverables <i>M18: September 2020, M36: February 2022</i>
<p>WP1 Coordination / Project management 2019 (Y3) 5 RDWG F2F meetings held Annual 2-Day Board seminar ongoing Monthly virtual RDWG meetings monthly ePAG, eHealth 1 COM Advisory TF additional RDWG Task Force meetings :HTAD Clinical Outcomes (3), VASCA Orphanet Classification (1), lymphangiography (1)</p>	<p>M18 Milestone : Mid-term report state of play</p> <p>M36 Deliverable : Report on coordination activities including the number of meetings held (virtual and face to face) - <i>Yearly: organisation of the annual Board « VASCERN Days » including RDWG F2F, and 1 * 5 RDWG additional Face to Face meeting Council: 3 /year, WG meetings: 81 (average 6.75 monthly meetings)</i></p>
<p>WP2 Dissemination/ Communication Update done for 2019 (Y3)</p>	<p>M18 Milestone : Mid-term state of play report on activities conducted and communication, analysing impact, indicators, targets, results, analysis to implement corrective measures for M18-M36</p> <p>M1-36: Deliverables : tools, website active and regularly updated with new content, monthly newsletters, active social media pages (facebook, twitter, linkedin, youtube)</p> <p>M34 Deliverable report on dissemination activities</p>
<p>WP3 Evaluation Update done with regard to the ERN Monitoring indicators for 2018 and 2019 (Y3)</p>	<p>M18 Milestone : mid-term evaluation analysis (will also include a report with regard to targets fixed in the 3 year action plan)</p> <p>M36 Deliverable : evaluation report (based on ERN monitoring data submission report)</p>



All outputs are on the website

<https://vascern.eu/what-we-do/>

The screenshot shows the navigation menu of the VASCERN website. The menu is located at the top of the page and is divided into two main sections. The first section, highlighted in a darker blue, contains the following items: WHAT WE DO, NEWS (with a dropdown arrow), CONTACT, and REFERRAL OF PATIENTS TO CPMS. The second section, in a lighter blue, contains: CLINICAL GUIDELINES & EXPERT CONSENSUS STATEMENTS, CLINICAL OUTCOMES MEASURES, CLINICAL TRIALS & RESEARCH STUDIES, CPMS: DISCUSSION OF COMPLEX CLINICAL CASES, DO'S & DON'TS FACTSHEETS FOR RARE VASCULAR DISEASE PATIENTS, MOBILE APP (VASCERN APP), PATIENT PATHWAYS, PATIENT REGISTRIES, and PILLS OF KNOWLEDGE VIDEOS FOR PATIENTS AND HEALTHCARE PROFESSIONALS. To the right of the menu, there are three prominent buttons: a red button labeled 'Call for new HCP full members', a teal button labeled 'EU languages', and a larger teal button labeled 'Do's & Don'ts Factsheets for rare vascular disease patients'.

- WHAT WE DO
- NEWS ▾
- CONTACT
- REFERRAL OF PATIENTS TO CPMS
- CLINICAL GUIDELINES & EXPERT CONSENSUS STATEMENTS
- CLINICAL OUTCOMES MEASURES
- CLINICAL TRIALS & RESEARCH STUDIES
- CPMS: DISCUSSION OF COMPLEX CLINICAL CASES
- DO'S & DON'TS FACTSHEETS FOR RARE VASCULAR DISEASE PATIENTS
- MOBILE APP (VASCERN APP)
- PATIENT PATHWAYS
- PATIENT REGISTRIES
- PILLS OF KNOWLEDGE VIDEOS FOR PATIENTS AND HEALTHCARE PROFESSIONALS

Call for new HCP full members

EU languages

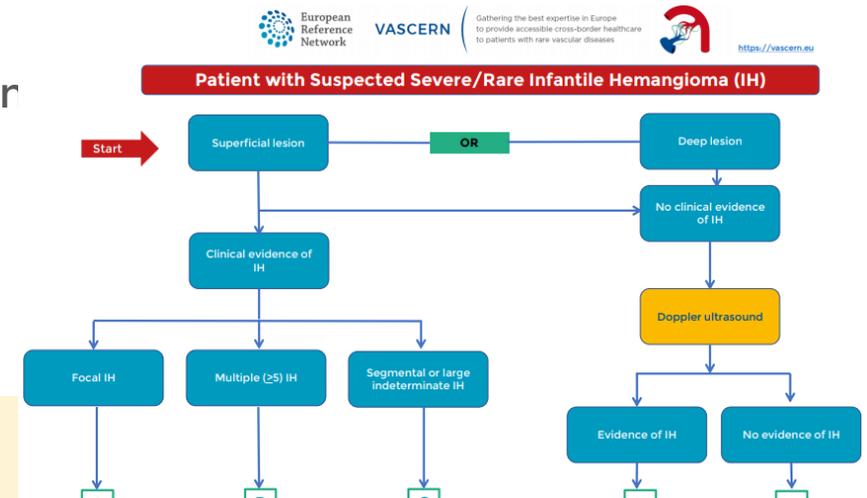
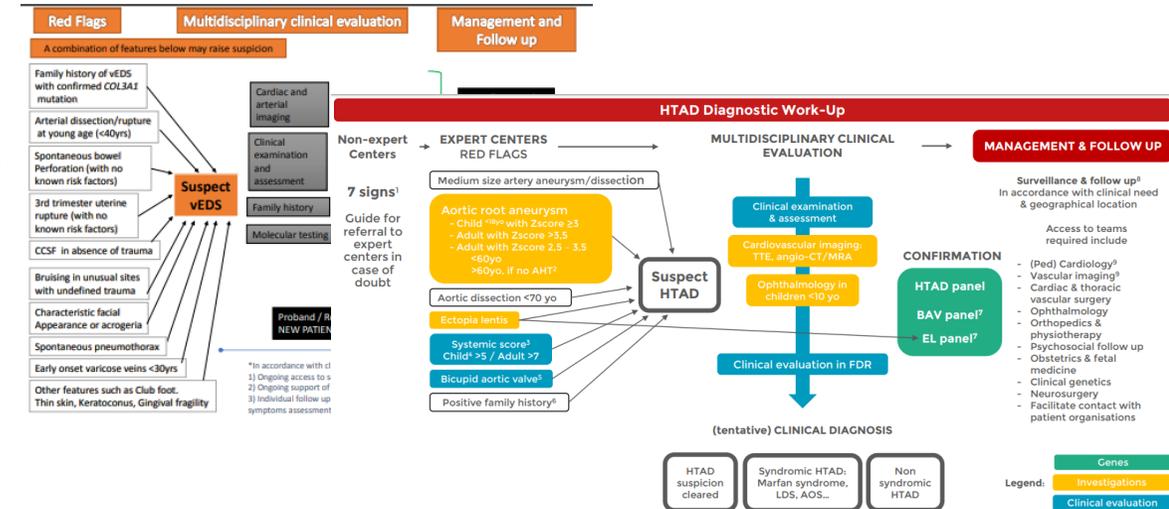
Do's & Don'ts Factsheets for rare vascular disease patients

WP5 Definition of (improved) patient pathways



<https://vascern.eu/what-we-do/patient-pathways/>

- HHT
 - 1st version published – translated in French
- HTAD
 - Improved version 2 published 08/2019– translation in several languages will be launched
- MSA
 - Discussions on v2 updates ongoing
 - **Translations** in 4 languages launched this summer, ongoing
- PPL
 - V1 tested within WG
 - Final improved version 2 under finalisation for publication
- VASCA
 - 1 patient pathways published and **translated** in 3 languages, 3 other ongoing
 - 3 patient pathways almost finalised
 - 3 other ones planned for 2020



WP6 VASCERN App Version 2.1

Will be presented by Alessandro Pini & Raffaella Gaetano

<https://vascern.eu/what-we-do/mobile-app/>



Version 2.1:

- Multilanguage: ongoing translation work
- Affiliated Partners will be added
- Increase of the number of ePAG
- Non ERN Members

WP7 Pills of Knowledge

<https://vascern.eu/what-we-do/pills-of-knowledge/>

- 14 new (VASCERN days videos = Y2 report)
 - 1 VASCERN with subtitles in all EU official languages
 - 3 MSA, 3 HTAD, 2 PPL, 4 VASCA, 1 HHT
 - Subtitles added in various EU languages of the RDWGs, thanks to the translation work of the members

• Y3 (as of March 2019) – foreseen

- 16 MSA new PoK videos
- 3 VASCA new PoK videos
- HTAD: 6 in the making
- PPL working on several PoK
- HHT ?

Espanol

Français

Hrvatski

Italiano

Latviešu

Lietuvių

Magyar



EU languages

All info on:
www.vascern.eu

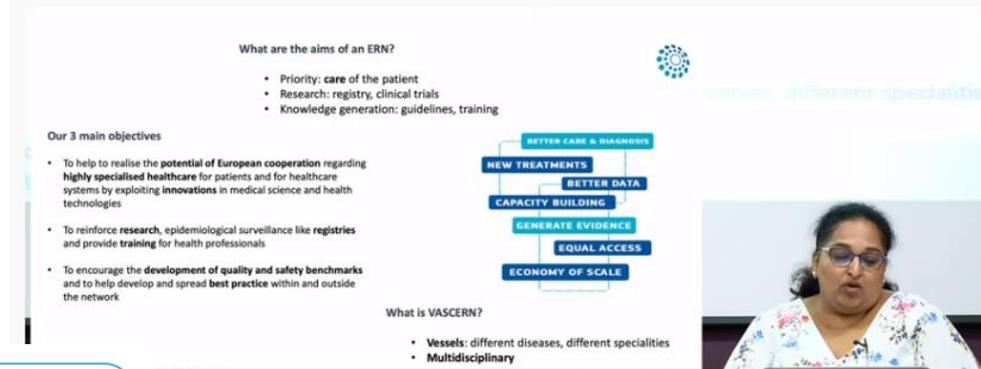
HTAD-WG Pills of Knowledge videos playlist



Heritable Thoracic Aortic Diseases (HTAD): Pills of Knowledge

- 1 Marfan Syndrome - Diagnosis by Prof Julie De Backer
- 2 3 a week - the importance of exercise in Marfan and HTAD
- 3 Le Sport et le Syndrome de Marfan

MSA-WG Pills of Knowledge Video Playlist



What are the aims of an ERN?

- Priority: care of the patient
- Research: registry, clinical trials
- Knowledge generation: guidelines, training

Our 3 main objectives

- To help to realise the potential of European cooperation regarding highly specialised healthcare for patients and for healthcare systems by exploiting innovations in medical science and health technologies
- To reinforce research, epidemiological surveillance like registries and provide training for health professionals
- To encourage the development of quality and safety benchmarks and to help develop and spread best practice within and outside the network

What is VASCERN?

- Vessels: different diseases, different specialities
- Multidisciplinary

BETTER CARE & DIAGNOSIS

NEW TREATMENTS

BETTER DATA

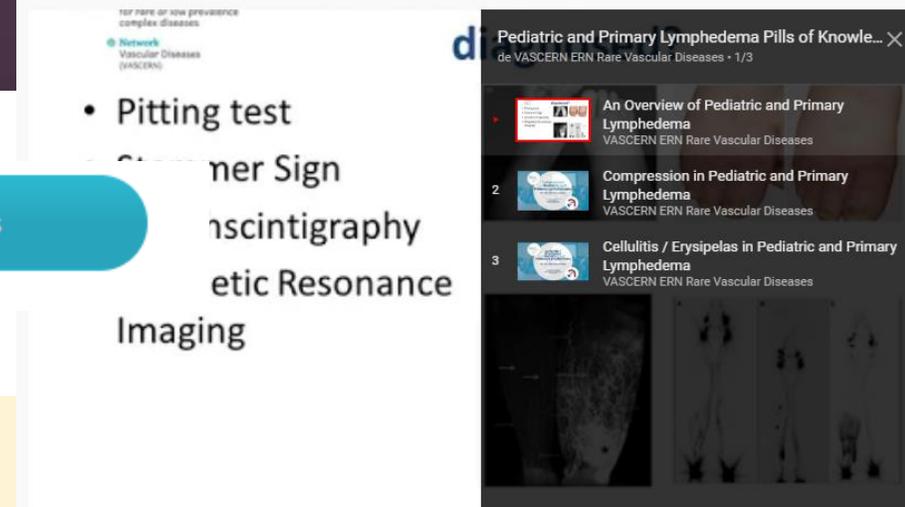
CAPACITY BUILDING

GENERATE EVIDENCE

EQUAL ACCESS

ECONOMY OF SCALE

PPL-WG Pills of Knowledge videos playlist



Pediatric and Primary Lymphedema Pills of Knowledge

- 1 An Overview of Pediatric and Primary Lymphedema
- 2 Compression in Pediatric and Primary Lymphedema
- 3 Cellulitis / Erysipelas in Pediatric and Primary Lymphedema

Pitting test

Ulcer Sign

Doppler Ultrasonography

MR Imaging

WP9 Clinical trials & Research



<https://vascern.eu/what-we-do/clinical-trials-research/>

- HHT
 - Research projects ongoing, multicentric clinical trial in set up (SAIPAN)
- HTAD
 - Several collaborative research projects, F101 Genomes; Myocardial function
- MSA
 - 2 clinical trial protocols being considered for replication in other countries
- PPL
 - Work on gathering and informing of national research projects
- VASCA
 - Several collaborative research projects and clinical trial (VASE) ongoing

Clinical Trials & Research Studies

VASCERN aims to promote European cooperation for scientific research and clinical trials. We collect information within the Rare Disease Working Groups (RDWGs) about clinical trials and research projects currently ongoing and perform regular bibliographical searches in order to report on collaborative research publications involving members from our 30 HCPs.

Read:

[Work Package 6 Deliverable - Clinical Trials and Research studies \(Year 1\)](#)

[Work Package 6 Deliverable - Clinical trials and Research \(Year 2\)](#)

We now also have a [Research News page](#) where you can access articles featuring the latest collaborative scientific publications from our members that have been reported in our monthly newsletter. Take a look [here](#).





WP10 Availability of videos on YouTube

<http://www.youtube.com/c/VASCERNERNRareVascularDiseases>



VASCERN ERN Rare Vascular Diseases
181 subscribers



- Currently **112 videos**
- Submit/ share with us if you are filmed

- Video collection, e.g:
 - Medical content: Pills of Knowledge + scientific meeting presentations
 - VASCERN related
 - Patient advocate/association videos, other meetings filmed

2018: 75 → **2019: 112**

WP12 Definition of clinical outcomes

<https://vascern.eu/what-we-do/clinical-outcomes-measures/>



- HHT
 - Shovlin, Buscarini et al, European Reference Network for Rare Vascular Diseases (VASCERN) Outcome Measures for Hereditary Haemorrhagic Telangiectasia (HHT)
- HTAD
 - Ongoing work in F2F meeting: 1 Clinical – 1 Patient driven
- MSA
 - Ongoing work
- PPL
 - Decided during F2F meeting in Sept
- VASCA
 - Ongoing

Shovlin et al. *Orphanet Journal of Rare Diseases* (2018) 13:136
<https://doi.org/10.1186/s13023-018-0850-2>

Orphanet Journal of Rare Diseases

POSITION STATEMENT Open Access

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

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Abstract
Hereditary haemorrhagic telangiectasia (HHT) is a multisystemic vascular dysplasia that leads to nosebleeds, anaemia due to blood loss, and arteriovenous malformations (AVMs) in organs such as the lungs, liver and brain. HHT is estimated to affect 85,000 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 been 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, and (5) written advice on pregnancy. They are not a blueprint for detailed HHT management, but are suitable for all clinicians to be aware of and implement. In summary, these 5 Outcome Measures provide metrics to identify healthcare providers of good care, and encourage care improvement by all healthcare providers.

Keywords: Anaemia, Antibiotic prophylaxis, Epistaxis, Iron deficiency, Nosebleeds, Pulmonary arteriovenous malformations, Pregnancy

Background
Development and implementation of Outcome Measures are an effective part of Quality and Safety Frameworks that lead to Service Improvements. More specifically, if the Outcome Measures are carefully selected, their dissemination and implementation can directly improve patient care, including that from health care providers with limited prior exposure or training on the specific disease. Therefore simple, clinical practice-based Outcome Measures are particularly important for rare multisystemic conditions. The current statement refers to one specific rare disease, hereditary haemorrhagic telangiectasia (HHT; Online Mendelian Inheritance in Man® #187300), which is a multi-systemic vascular dysplasia that leads to telangiectasia and arteriovenous malformations (AVMs) in visceral and mucocutaneous vascular beds [1]. Based on a conservative population prevalence of 1 in 6000 [2–4], HHT is estimated to affect approximately 85,000 European citizens. The main goal of management is to maximise the number of affected individuals receiving safe and effective preventative strategies in order to limit the number and severity of HHT complications. The reason this is

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BMC

WP13 Writing Clinical Practice Guidelines (expert consensus statements, recommendations)

<https://vascern.eu/what-we-do/clinical-guidelines/>



- HHT
 - 2 consensus statements published in scientific journal
- HTAD
 - ACTA2 finalised, submitted to OJRD
- MSA
 - Work on pregnancy in vEDS
- PPL
 - Project of consensus statement for compression in children
- VASCA
 - Not prioritised. Working first on VASCA classification and 7 patient pathways



VASCERN Consensus Statement: ACTA2-Related Vasculopathy

VASCERN HTAD working group Members:
Ingrid van de Laar*, Eloisa Arbustini, Kalman Benke, Erik Björck, Maarten Groenink, Marijes Kemper, Yskert von Kodolitsch, Bart Looys, Barbara Mulder, Libe Murphy, Guglielmina Pepe, Alessandro Pini, Leena Robert, Jolien Roos-Hesselink, Zoltan Szabolcs, Guillaume Zandevu, Julie De Backer

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VASCERN
The European Reference Network on Rare Vascular Diseases



WP14 Do's and Don'ts factsheets for rare vascular disease patients facing frequent situations

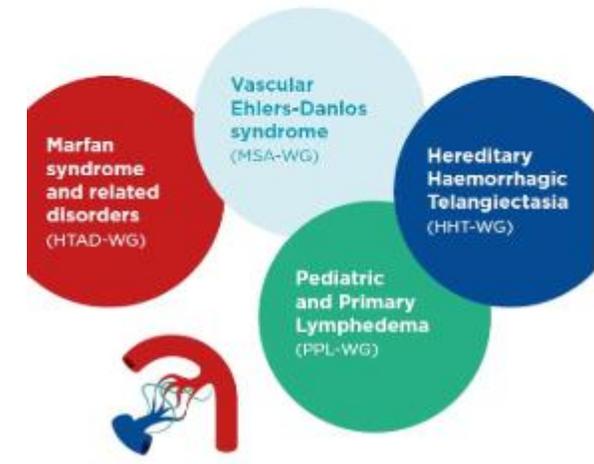


<https://vascern.eu/what-we-do/dos-donts-factsheets-for-rare-vascular-disease-patients/>

- HHT
 - Y1-2: 12 Factsheets [published](#)
 - Y3: Translated in [DA](#), [DE](#), [IT](#), [NL](#), [ES](#)
- HTAD
 - Y1-2: 23 Factsheets [published](#)
 - Y3: Translated in [HU](#), [IT](#), [NL](#), [ES](#) . Asap DE & SU
 - Y3: **New set of Factsheets!** LDS pre-final
- MSA
 - Y1-2: 17 Factsheets [published](#)
 - Y3: revision of English expected to published revised + start translations
- PPL
 - Y2: 13 Factsheets [published](#)
 - Translated in [DE](#), [NL](#), [ES](#). Further translation will be initiated with ePAG.
- VASCA
 - *Ongoing work*



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



https://vascern.eu/wp-content/uploads/2019/05/Fiches_do_dont-web-FINAL-2019.pdf

HTAD implementation of WP's



WP1	Coordination/ project management	NA
WP2	Dissemination/ communication	NA
WP3	Evaluation of the action implementation	NA
WP4	Sharing of experience: discussion of difficult clinical cases	Monthly
WP5	Patients Pathways: improvements and updates	Version 2.0 published
WP6	Mobile Application	NA
WP7	Pills of Knowledge	6 in the making
WP8	Registries	ROPAC; database
WP9	Clinical trials & Research	101 Genomes; Myocardial function
WP10	Availability of videos on YouTube	NA
WP11	Multidisciplinary school for multisystemic vascular disease (Year 5)	NA
WP12	Definition of clinical outcomes	1 Clinical – 1 Patient driven
WP13	Writing Clinical Practice Guidelines	ACTA2 paper in press OJRD
WP14	Do's and Don'ts factsheets	Marfan OK – LDS pre-final
WP15	VASCERN clinic days (year 5)	NA

RDWG F2F meetings : sharing best practices



- **HHT (1 day)**
 - **Educational** discussions in order to initiate expert consensus statements (e.g NOACs)
 - Clinician only part for clinical cases discussion
- **HTAD (1 day)**

Extensive expert **consensus** discussions

 - HTAD patient pathway v1 and v2
 - ACTA 2 expert consensus statement
- **MSA (1 day)**
 - vEDS **Study Day** – educational event. Open to external doctors.
 - Production of 16 MSA **Pills of Knowledge videos / webinars** (donation EDS Society)
- **PPL (1,5 day)**
 - 2018 and 2019: Lymphedema **clinic visit**
Clinician only part for clinical case discussion
 - Educational discussions to work on expert consensus statement
- **VASCA (1,5 day)**
 - Extensive work on several **patient pathways**
 - Discussions on Orphanet classification
 - Sharing best practices for new treatment, launching a new VASCA Lymphography Task Force with interventional radiologists