



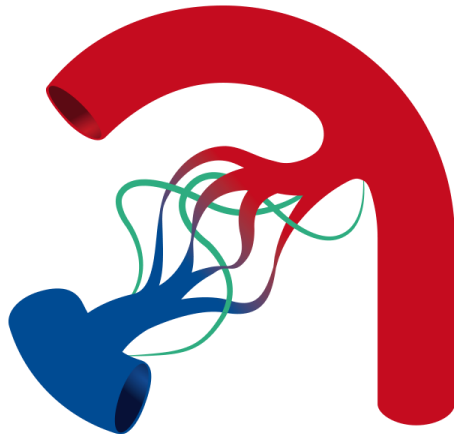
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



**WELCOME TO THE
VASCERN DAYS 2021
7, 8, 9 October**

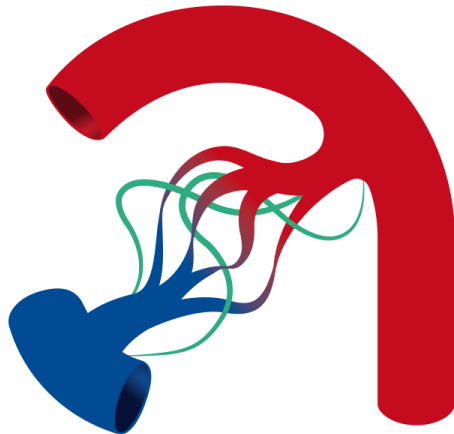
 **#VASCERNdays2021**



European Reference Network

for rare or low prevalence
complex diseases

🌐 **Network**
Vascular Diseases
(VASCERN)



8.30-12.05 VASCERN ANNUAL BOARD MEETING

8.30-9.00

VASCERN's Management & Coordination (Julie Hallac, Natasha Barr, Guillaume Jondeau)

- Welcoming words
- VASCERN Project Team
- VASCERN's EU co-funding projects
- Communication and dissemination

 [#VASCERNdays2021](https://twitter.com/VASCERNdays2021)

Welcoming words

Are present for the 2021 VASCERN days:

- **53 HCP full member representatives**
 - from 9 countries
 - 20 HCP
 - 26 Expert centers
- **4 Affiliated Partners representatives**
 - from 4 other EU Member States
 - 4 Affiliated partners centers
- **18 Patient Advocates**, members of our European Patient Advocacy Group, from 8 countries



Coordination & Project Management Team



Soon to
come!



Prof. Guillaume JONDEAU
VASCERN Coordinator
Cardiologist
+ 33 6 10 67 84 42
guillaume.jondeau@aphp.fr

Julie HALLAC
VASCERN Project Manager
+33 (0)1 40 25 80 66
julie.hallac@aphp.fr

Natasha BARR
VASCERN Project Officer
+33 (0)1 40 25 70 76
natasha.barr@aphp.fr

VASCERN Project Assistant
+33 (0)1 40 25 70 77
vascern.coordination@aphp.fr

Ibrahim DONMEZ
VASCERN IT Helpdesk &
End-User Support
Specialist
+33 (0)1 40 25 67 22
ibrahim.donmez@aphp.fr

Pim Kamerling
Data Steward
Radboud University Medical
Center, Nijmegen, Netherlands
Pim.Kamerling@radboudumc.nl

Working in cooperation with AP-HP departments & the Foundation of the AP-HP

European Unit, Clinical Research Unit for registry projects, HR, IT, Finance, Buying, Communication, etc.



DIRECTION
GÉNÉRALE
DE L'OFFRE
DE SOINS



Agenda



Thursday Morning

- Plenary: Board session, including policy and project updates
- Feedback from RDWGs on Work Packages Implementation
- ePag presentation

Thursday Afternoon

- RDWG
- CPMS & ePag meeting
- Council selection of brainstorming topic

Friday, Morning

- EJP-RD
- Registry Presentation
- Brainstorming session

Friday, Afternoon

- Pregnancy, brainstorming and Registry WG
- RDWG

Saturday, Morning

- RDWG
- interRDWG meetings
- Wrap-up session
- Council meeting



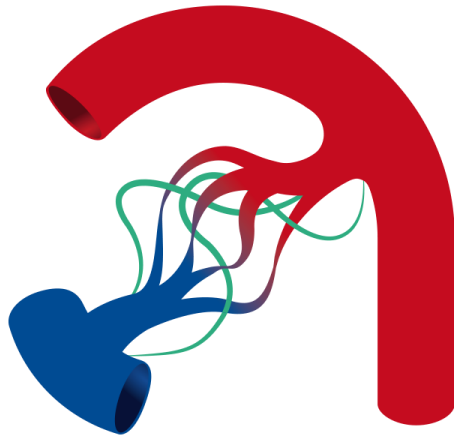
European Reference Network

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



VASCERN's EU co-funding projects

 [#VASCERNdays2021](https://twitter.com/VASCERNdays2021)

3rd EU Health Programme: since March 2017
Current: 3 Years Specific Grant Agreement (SGA)

March 2019-February 2022

ERN Management & Coordination:

Project Team (*manager, officer, assistant*), Meetings (*travel, hotel, rooms*), Communication (*webmaster, translations, scientific journal publications, app, etc.*)

600k EU co-funding (60% of budget)

400k co-funding France APHP & Health ministry (40%)



Co-funded by
the Health Programme
of the European Union

VASCERN's multi-EU's
co-Funding



Co-financed by the Connecting Europe
Facility of the European Union

Connecting Europe Facility Telecom – eHealth for ERNs
since January 2019

Current: Grant Agreement April 2021-September 2022

ERN eHealth Services

CPMS & IT helpdesk, 1 Data Steward, Mobile App,
eLearning: PoK videos, Webinars & Moodle, Monitoring:
data collection & validation, work on databases

210k EU co-funding (75% of budget)

70k co-funding (25%) partners APHP & RadboudUMC

3rd EU Health Programme :

Registry Grant Agreement May 2020-May 2023

5 registries project (*data stewards, licences, server hosting,
developers work on FAIRification*)


200k EU Co-funding (60% of budget)

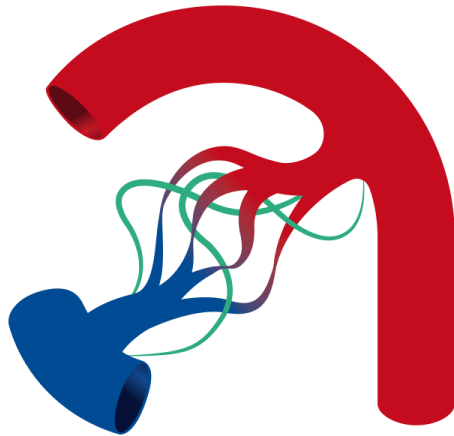
130k co-funding partners: APHP, RadboudUMC, INSERM, HCL



**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



#VASCERNdays2021



Communication and dissemination

Natasha Barr





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 CONTACT CLINICIANS ONLY - CPMS

Rare Diseases WGs

Transversal WGs

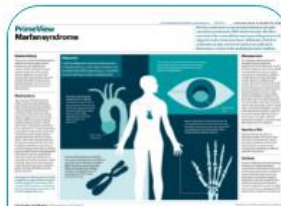
ePAG (Patient Advocacy)

VASCERN's COVID19 recommendations

VASCERN Resources in all EU languages

+ Subscribe to our newsletter

Latest News



New Nature Reviews Disease Primer on Marfan syndrome by



Guest case from Portugal discussed in the CPMS



VASCERN Registries project update

Jul 20, 2021

Upcoming

THU 30 SEP 2021	FRI 01 OCT 2021	ERN Workshop "Clinical Research: The Basics & Beyond Workshop In Clinical Epidemiological Research For ERNs"
SAT 02 OCT 2021	SUN 03 OCT 2021	LGDA/LMI International Conference on Complex Lymphatic Anomalies
MON 04 OCT		ERN Research Training Workshops funding

Website hits

Total page views (hits) in 2017: 20,312
Total page views in 2018: 32,509
Total page views in 2019: 58,404
Total page views in 2020 ; 58,431

Total page views 2021 (so far) :47903

- New:**
- VASCERN's COVID19 vaccination section updated
 - Updates to the content and structure of CPMS page
 - Updated member pages after Brexit
 - **New pregnancy WG page in transversal WG**
 - Changed headings of certain sections after feedback from ePAG so things are more clear



Website updates to do:

VASCERN Resources in all EU languages

Français

2. Select your language

3. VASCERN welcome text in your language and you can select the RDWG that interests you.

1. Click on “VASCERN Resources in all languages” button

Bienvenue sur le site internet de VASCERN !

Cette page contient toutes les ressources traduites en français.

La langue de travail dans toute l'Union Européenne (UE) est l'anglais, c'est pourquoi notre site internet et les documents produits par nos experts sont initialement produits dans cette langue.





Afin d'atteindre autant de patients et de professionnels de santé que possible, nous avons l'intention de continuer à traduire nos documents et vidéos dans les différentes langues de l'UE et d'autres ressources devraient bientôt arriver dans votre langue.

- ∨ Télangiectasie hémorragique héréditaire (maladie de Rendu-Osler)
- ∨ Maladies Héréditaires de l'Aorte Thoracique
- ∨ Maladies vasculaires rares des moyens et petits vaisseaux (syndrome d'Ehlers-Danlos vasculaire)
- ∨ Lymphoedème pédiatrique et primaire
- ∨ Anomalies Vasculaires
- ∨ VASCERN et les Réseaux européens de référence (ERNs)

- **Redesign of website in 2022:** not in the content but in the performance, improved plug-ins, analytics, improve ease of navigation, speed of website etc.

VASCERN Social Media channels



Social media channel	October 2020	October 2021
Twitter  https://twitter.com/vascern	866 followers →	1066 followers
Facebook  https://www.facebook.com/vascern.eu/	682 likes →	850 likes
Youtube  http://www.youtube.com/c/VASCERNE/RNRareVascularDiseases	505 subscribers →	824 subscribers
LinkedIn  https://www.linkedin.com/company/vascern/	266 followers →	382 followers



Continued success and growth of our social media channels



#VASCERNDays2021

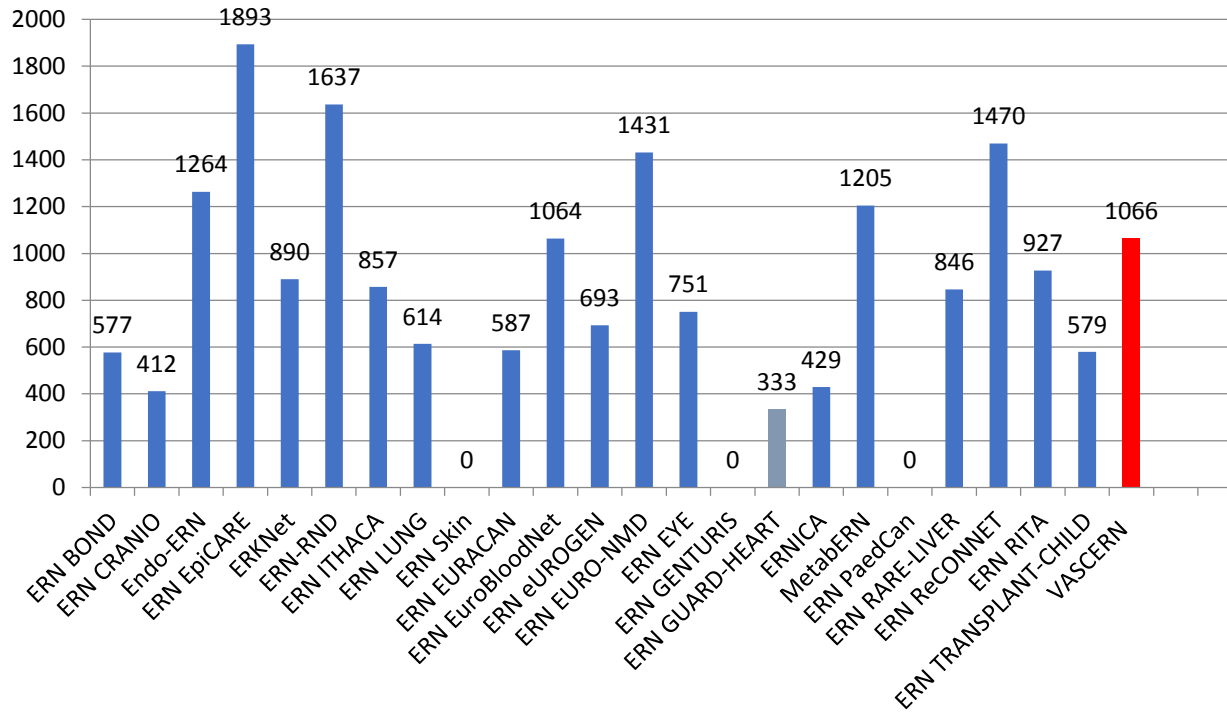


Benchmark with other ERNs – part 1

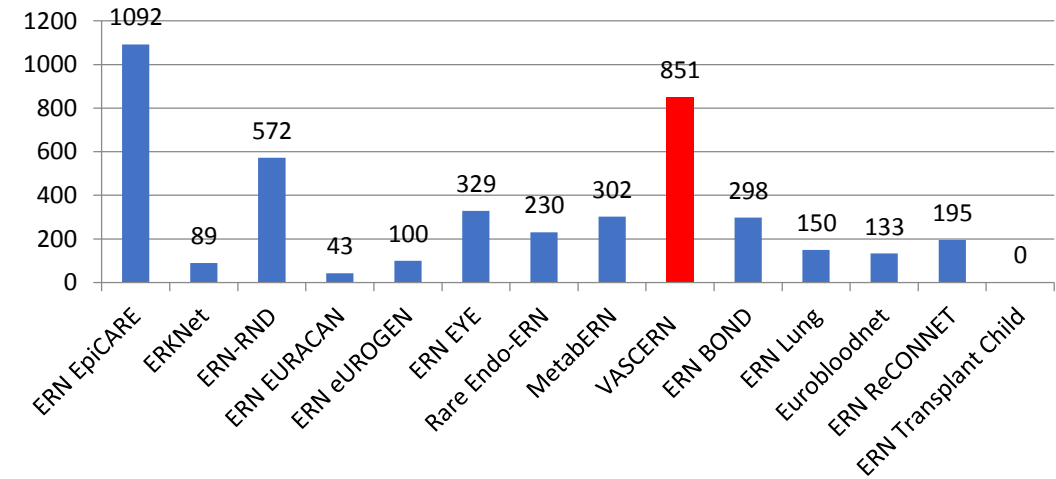
→ 21/24 ERNs have a Twitter account

→ 14/24 ERNs have a Facebook account

Twitter Followers (29/09/2021)



Number of FB likes for each network (24/09/2021)



- 79% of our followers are women, 19% men
- Top 5 countries where are followers are from: France, Italy, Netherlands, United States, Belgium
- Top 5 languages spoken by our followers: English, French, Dutch, Italian, German



Benchmark with other 24 ERNs – part 2

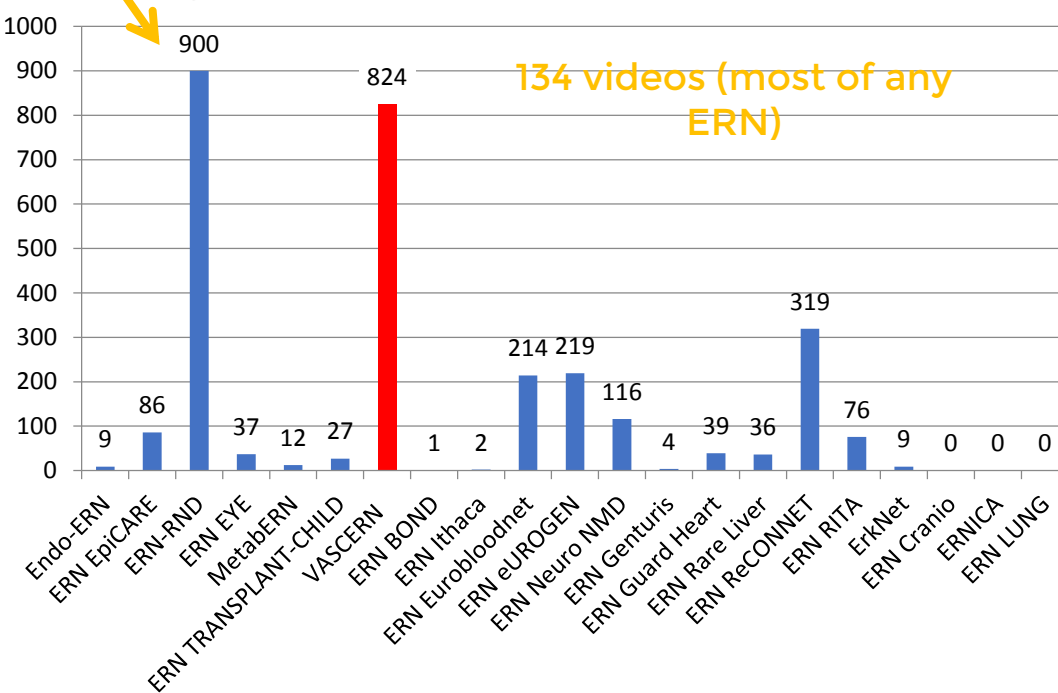
→ 21/24 ERNs have a YouTube account

→ 15/24 ERNs have a LinkedIn account

→ Only 5 had it in 2019

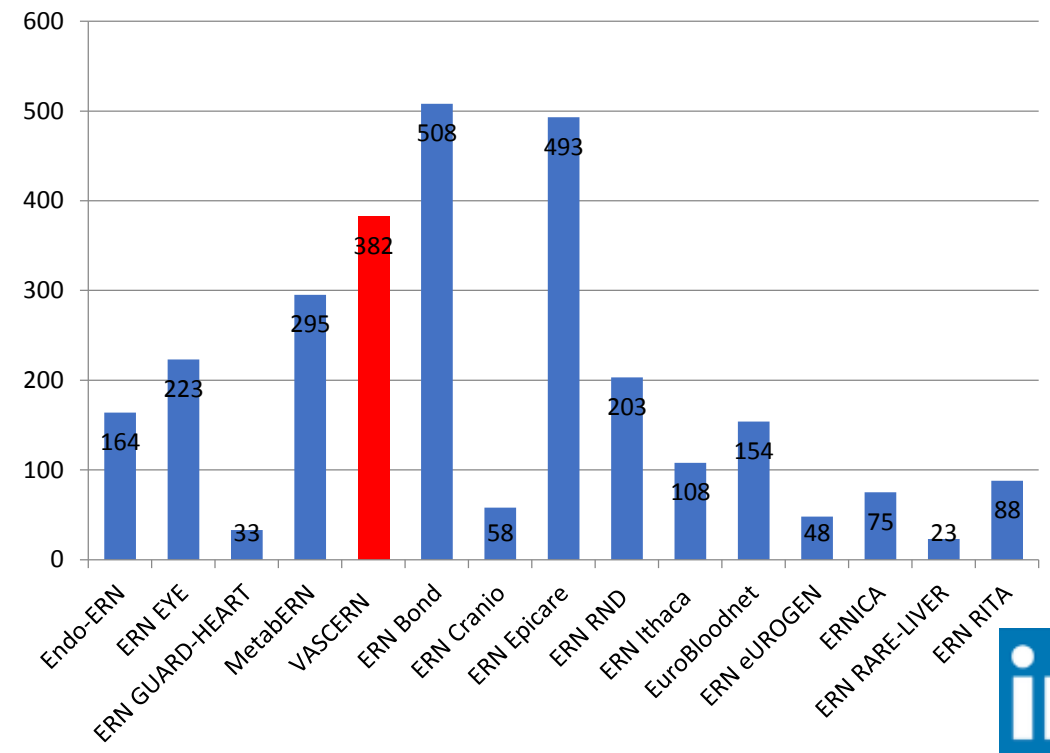
36 webinars done with EAN

Number of subscribers on existing youtube channels (24/09/2021)



134 videos (most of any ERN)



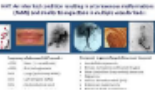


Number of LinkedIn subscribers/relationships per network (24/09/2021)



A few Youtube Statistics

Overall top 5 most viewed videos

Steady increase in numbers of hours viewed/day over time!

Vidéo		Durée moyenne d'une vue	Vues
1	 Marfan Syndrome - Diagnosis by Prof Julie De Backer 2 mars 2018	2:11 (26,3 %)	29 405
2	 Klippel -Trenaunay syndrome (KTS) 6 janv. 2020	0:43 (56,3 %)	19 635
3	 An Overview of Hereditary Haemorrhagic Telangiectasia (HHT) by Prof Claire Sho... 23 janv. 2018	3:12 (36,0 %)	13 260
4	 Vascular Ehlers-Danlos syndrome: Introduction and new criteria 31 oct. 2019	4:15 (32,5 %)	8 845
5	 Clinical Features and Diagnostic criteria of vascular Ehlers-Danlos syndrome (vE... 31 oct. 2019	2:48 (47,7 %)	4 208



Situation géographique	Vues ▲	Durée de visionnage (heures) ↓ ▲	Durée moyenne d'une vue
<input type="checkbox"/> Total	126 035	4 519,2	2:09
<input type="checkbox"/> États-Unis	28 214 22,4 %	1 229,2 27,2 %	2:36
<input type="checkbox"/> Royaume-Uni	4 373 3,5 %	200,1 4,4 %	2:44
<input type="checkbox"/> Inde	6 533 5,2 %	150,0 3,3 %	1:22
<input type="checkbox"/> France	2 878 2,3 %	77,1 1,7 %	1:36
<input type="checkbox"/> Allemagne	1 253 1,0 %	46,5 1,0 %	2:13

- 57,8 % of viewers are aged 25-34
- Subtitles viewed most in EN, followed by FR, DE, ES, NL
- 97% are not subscribed to our Youtube channel!



Webinars – so far lots of interest!

<u>Month</u>	<u>Presenter</u>	<u>RDWG</u>	<u>Topic</u>
March 2021	Dr. Kirsten VANDUINEN and Dr. Tanja PLANINŠEK RUČIGAJ	PPL	Infections and Lymphedema https://youtu.be/IJBzDwB_rAY
April 2021	Prof. Emir Q. Haxhija and Dr. Paolo GASPARELLA	VASCA	Classification of Vascular Anomalies https://youtu.be/ZID_ZkxZocU
May 2021	Dr. Hans-Jurgen Mager and Prof. Marco Post	HHT	HHT and the lungs https://youtu.be/sGFzDGJUgBY
<u>June 2021</u>	Dr. <u>Fransiska Malfait</u>	MSA	On collagen, the Ehlers-Danlos Syndromes and vascular fragility: what's in a name? https://youtu.be/ArGxB-MB4k

While we usually have around 100 people registered for the live event, the recordings are also popular



NOVEMBER

→VASCA webinar series covering their 3-4 patient pathways explaining the diagnostic and management pathway for different types of vascular anomalies.

→HTAD : Patient/Surgeon Interview/Q&A on aortic surgery

+++ in 2022 !

Collaborations



Notable collaborations since last VASCERN Days:

- Joint webinar with ERN reCONNET



- Joint webinar with ERN-Eye



- VASCA ePAG VASCA Magazine



PLUS: We have many partners in communicating our news including:

- EJP-RD
- FAVA-Multi
- Orphanet
- Marfan Foundation
- ePAG Patient Organisations
- ISSVA
- **YOU** (at conferences, on your social media networks etc.)



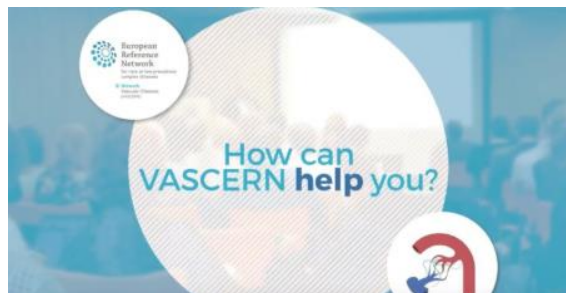
Communication tools at your disposal!

The return of in-person events = more opportunity to share the news from VASCERN Days

- We have Powerpoint templates to present VASCERN
- We can provide logos/brand guidelines
- The VASCERN Flyer (project to update with updated members/countries)
- The “How can VASCERN help you” video in all EU languages
- If you have events where VASCERN will be presented we will communicate – please let us know! (hopefully we can soon have a shared Excel where you can add events – MS Teams)



- [Flyer](#)
- [Web version here](#)
- [Print version here](#)



« [How can VASCERN Help You?](#) » Video: **2251** views to date (since January 2019)!



Reminder: Presentation of VASCERN at congresses/meetings is one of the monitoring indicators collected by the EC!
2019: 98
2020:6



Future Priorities and deliverables

- Increase newsletter subscribers! We are currently at 696 subscribers! We are close to our milestone target of 1000!
- Improve the design of the VASCERN website
- Continue to have regular update articles on CPMS and Registry Projects (part of CEF and Registry project deliverables)
- Continue with Research news and VASCERN Spotlights segments
- Make a simple tutorial video on how to access VASCERN resources in your language (once updates to website are made)
- Update VASCERN Flyer with APs and new HCPs
- Increase our collaboration with scientific societies/organisations/other ERNs/social media figures...
- Start a #WeareVASCERN or other social media campaigns to try and reach more people

→Always open to more ideas!





THANK YOU!

For contributing to VASCERN's communication actions!




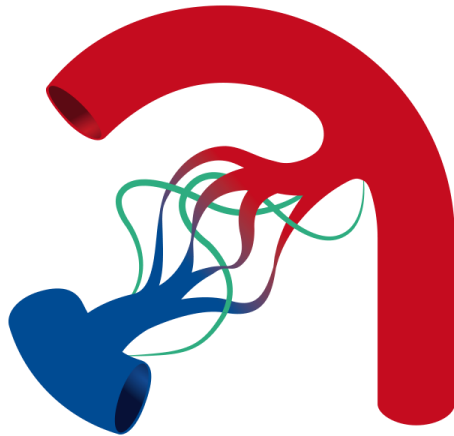
#VASCERNDays2021



European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



9.00-9.30 **VASCERN's developments** (Julie Hallac, Guillaume Jondeau, Ibrahim Donmez)

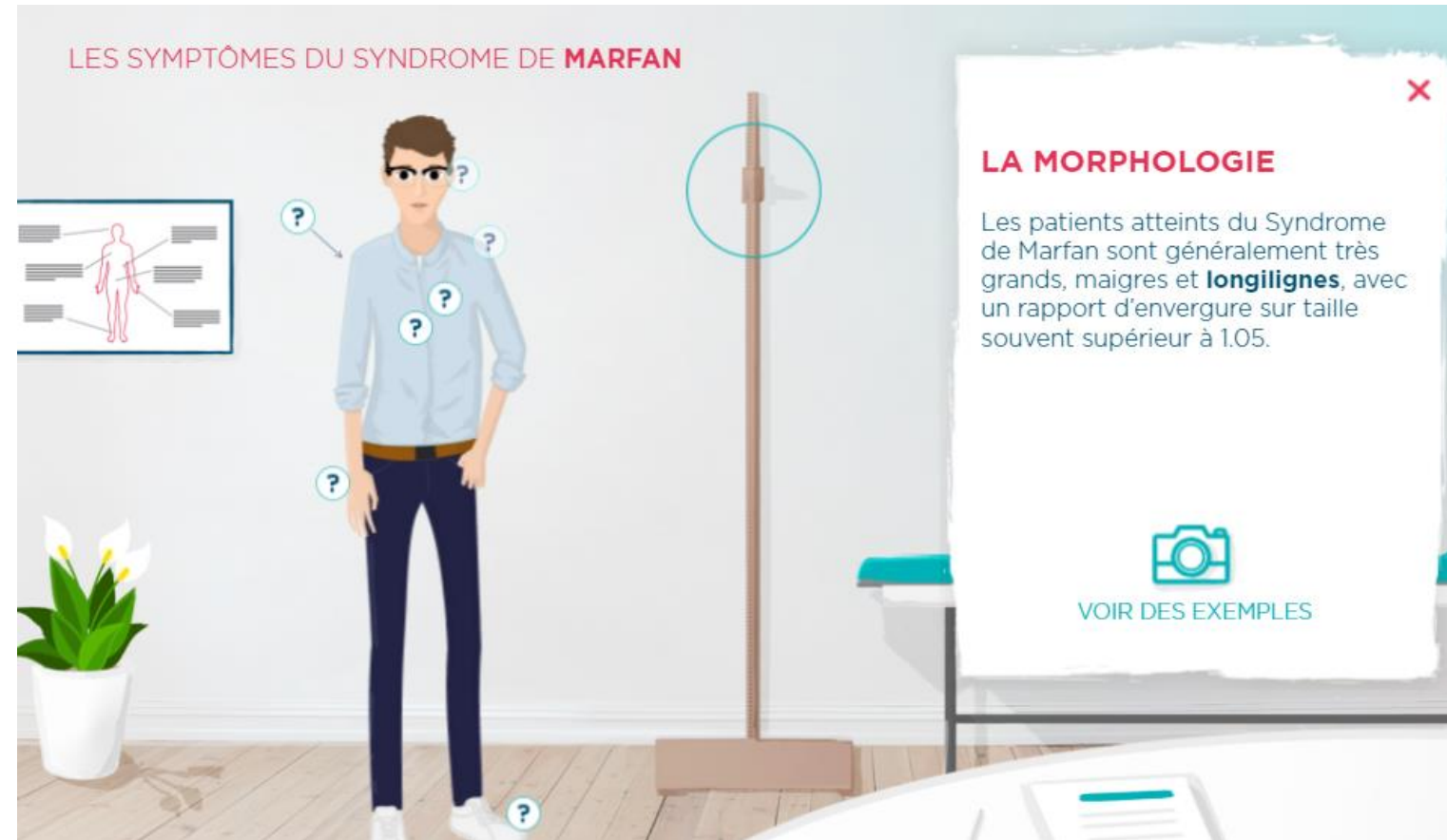
- eLearning, Moodle platform
- SubWG guidelines
- Consent forms
- Update on New Healthcare Providers Full Members
- New Patient Organisations
- UK former members' participation
- IT developments and means of communication

 **#VASCERNdays2021**

CEF 2020 & Moodle platform



- Looking for existing modules in our member institutions
- Translation and sending of the French content into English
- To be expanded with new diseases and new languages
- Exists for doctors → possible new content for patients
- Webinars and PoKs will be added to the platform



Webinars - up to 2 per month from 2022

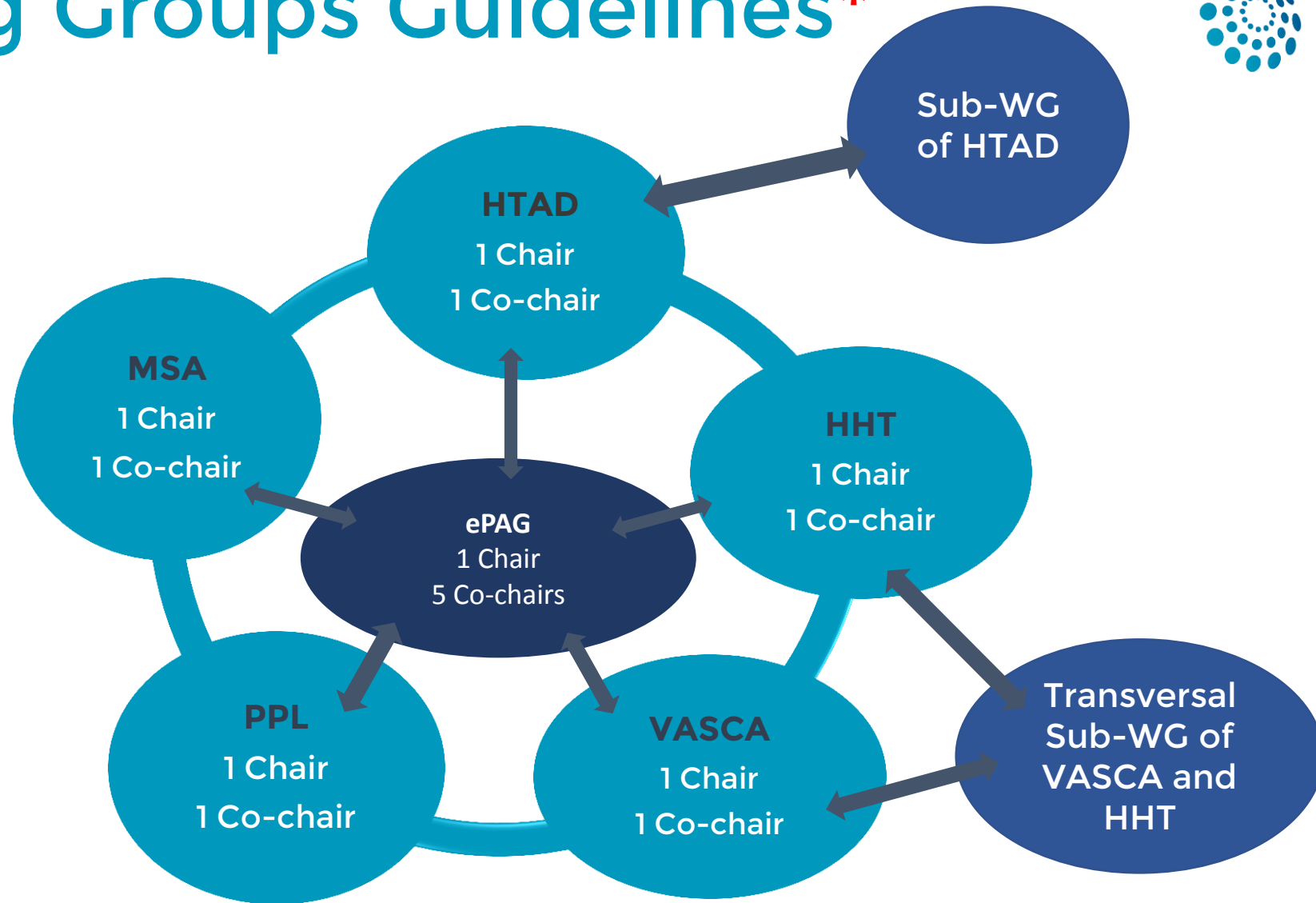


Month	Presenter	RDWG	Topic
March 2021	Dr. Kirsten VAN DUINEN and Dr. Tanja PLANINŠEK RUČIGAJ	PPL	Infections and Lymphedema https://youtu.be/IJBzDwB_rAY
April 2021	Prof. Emir Q. Haxhija and Dr. Paolo GASPARELLA	VASCA	Classification of Vascular Anomalies https://youtu.be/ZID_ZkxZocU
May 2021	Dr. Hans-Jurgen Mager and Prof. Marco Post	HHT	HHT and the lungs https://youtu.be/sGFzDGJUgBY
June 2021	Dr. Fransiska Malfait	MSA	On collagen, the Ehlers-Danlos Syndromes and vascular fragility: what's in a name?
Break			
November 2021	Dr. Andrea Diociaiuti	VASCA	Severe and rare infantile hemangioma Patient Pathway
December 2021	Dr. Nader Ghaffarpour	VASCA	Lymphatic malformation Patient Pathway
January 2022	Dr Eulalia Baselga TBC	VASCA	Capillary malformation Patient Pathway Venous malformation Patient Pathway
February 2022	Dr Klaus Kallenbach and ePags	HTAD	Aortic Surgery - Interview/Q&A

Sub-Working Groups Guidelines*



Sub-WG focus	Applies to
Patient management	<ul style="list-style-type: none"> • WP5 Patient pathways • WP7 Pills of Knowledge • WP14 Do's and Don'ts factsheets
Research	any sub-group with a project that involves a research output



*To be consulted on the USB key

SubWG guidelines validated by the Council*



Scope and time:

- SubWG only exists for a specific topic for the time of work
- Possibility of transversal sub-WGs

2 types of sub-WGs:

- Patient management sub-WG on “X topic” (WP5 Patient pathways, WP7 Pills of Knowledge, WP14 Do’s and Don’ts factsheets)
- Research sub-WG on “X topic” – applies to any sub-group with a project that involves a research output

Participants:

- HCPs & APs
- Members of an official national network within the EU
- Expert centres not members of VASCERN or an official national network (only research)
- VASCERN ePAGs (unless it involves case discussions)

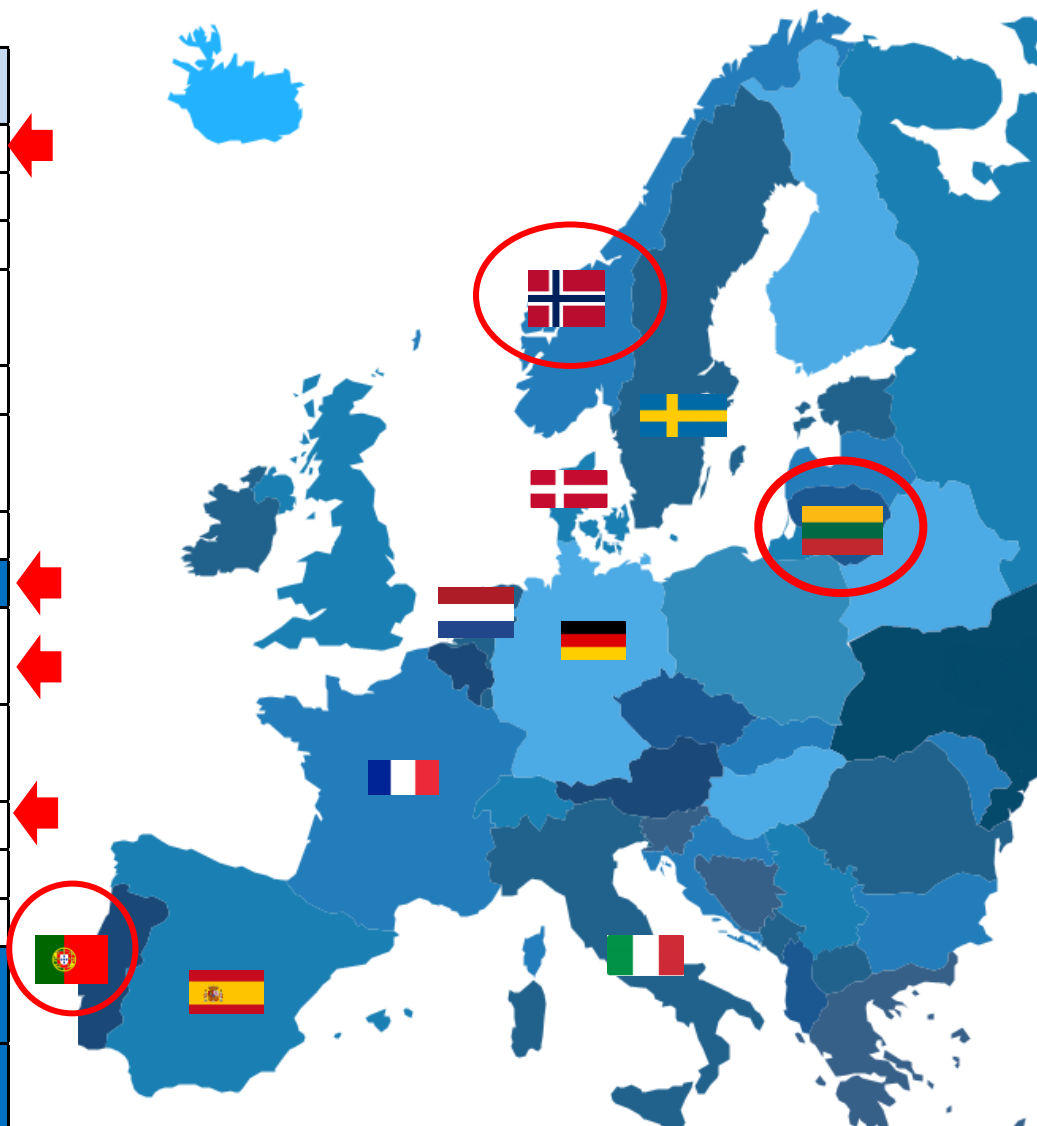
3 consent forms for PoKs and webinars



15 new HCPs and a 6th RDWG



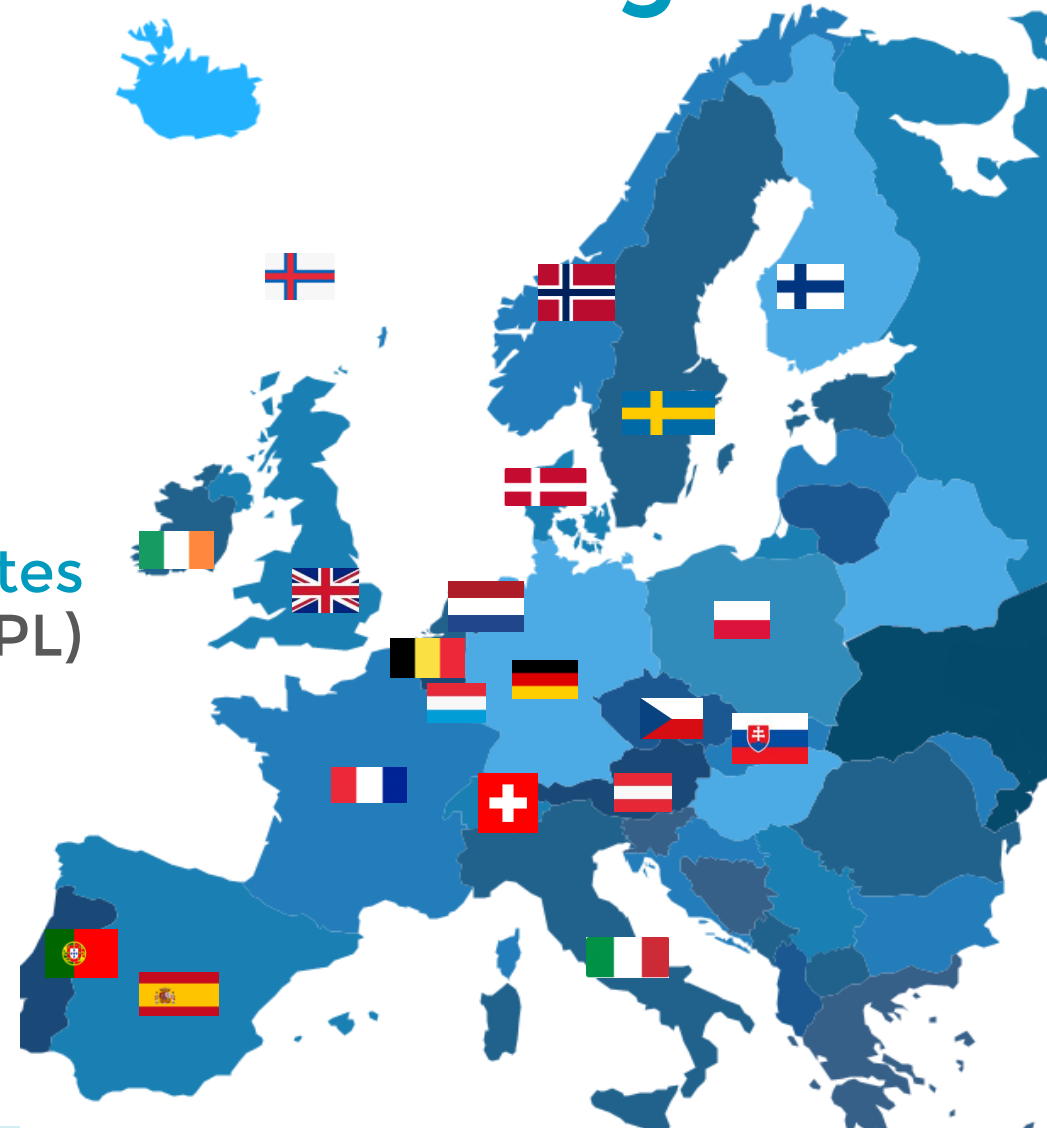
Healthcare Provider Name	City	Country	RDWGs
Alfried Krupp Krankenhaus	Essen	DE	VASCA - NEUROVASC
Charité Universitätsmedizin	Berlin	DE	VASCA
Universitätsklinikum	Marburg	DE	HHT
Rigshospitalet	Copenhagen	DK	PPL - VASCA
Aarhus Universitets Hospital	Aarhus	DK	HTAD - MSA
Hospital Universitari Vall d'Hebron	Barcelona	ES	HTAD
Hospital Sant Joan De Déu	Barcelona	ES	VASCA
Hôpital Lariboisiere	Paris	FR	VASCA - NEUROVASC
Fondazione IRCCS Istituto Neurologico Carlo Besta	Milano	IT	NEUROVASC
Vilniaus universiteto ligoninės Santariškių klinikos	Vilnius	LT	VASCA
Leiden University Medical	Leiden	NL	NEUROVASC
Oslo universitetssykehus	Oslo	NO	VASCA
Oslo universitetssykehus	Oslo	NO	HTAD - MSA
Centro Hospitalar de São João, EPE	Porto	PT	VASCA
Akademiska sjukhuset - Uppsala University Hospital	Uppsala	SE	MSA



New Patient Advocates in VASCERN ePag



- Aim of VASCERN is to have a broad representation of the patients accross Europe
→ No limitation in the number of patient organisation
- 3 new applications to become patient advocates from the Czech Republic (VASCA), Sweden (PPL) and France (PPL)
- Total number of Patient organisations: 72



British HCPs status



Update

Continuation of the work within VASCERN possible under a confidentiality agreement

4 British HCPs concerned

New status

Collaborating experts:

- Individual rather than representation of an HCP
- Only for centers who have gone through the EC evaluation

Prerogatives

Publications: Signature as invited/collaborating experts

Online meetings : participation allowed

Face-to-face meeting : participation allowed

CPMS: not allowed by the EC

Registries: possible if respect of the EU rules on Data Protection



Microsoft Teams



- Find all in one place in terms of online meetings, file sharing and file editing
- Multiple users can work on a file at the same time
- Able to access Teams across all devices (mobile version is really popular)
- Able to invite users outside of your organization

VASCERN Mobile App



- 31 HCPs Centres
- 7 Affiliated Partners
- 32 Referral Centres
- 72 Associations

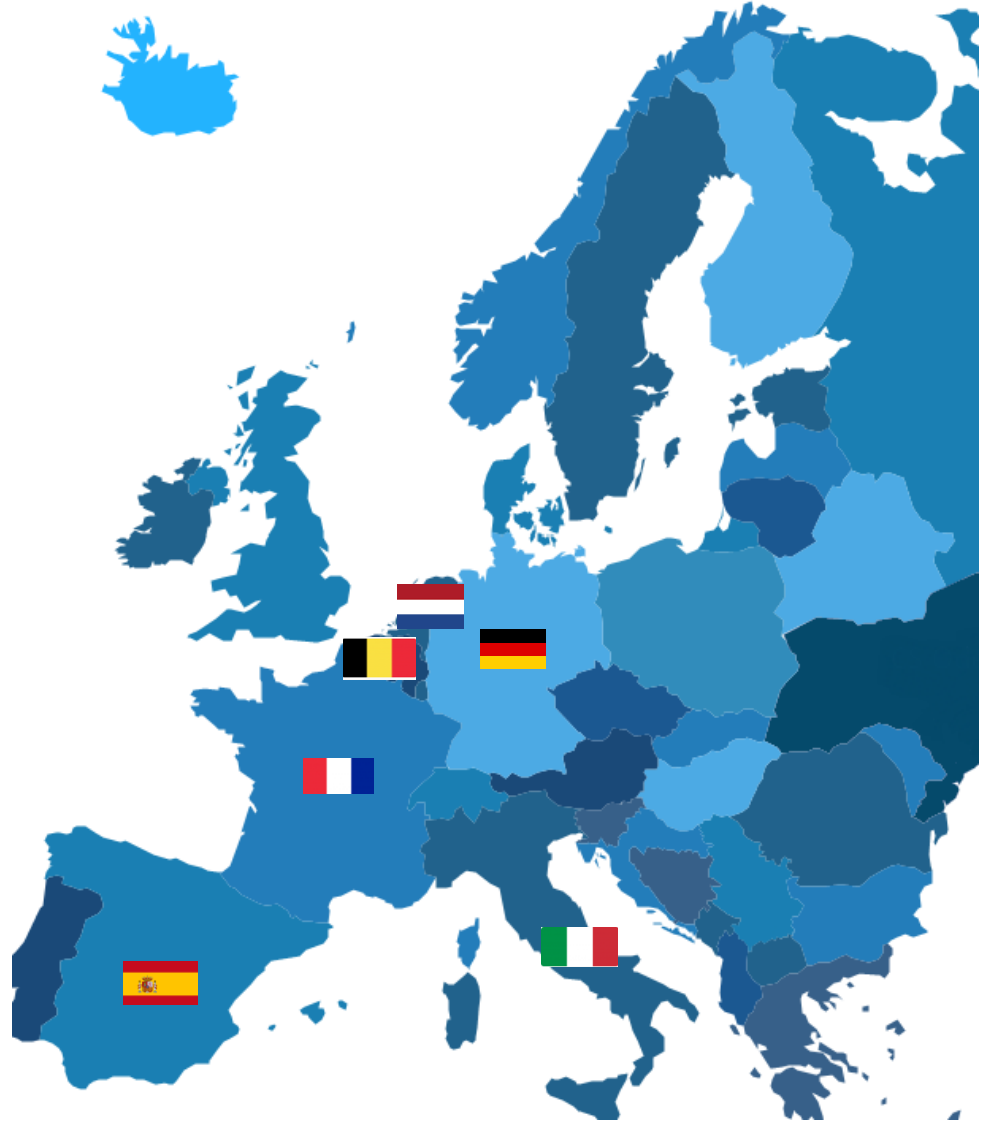
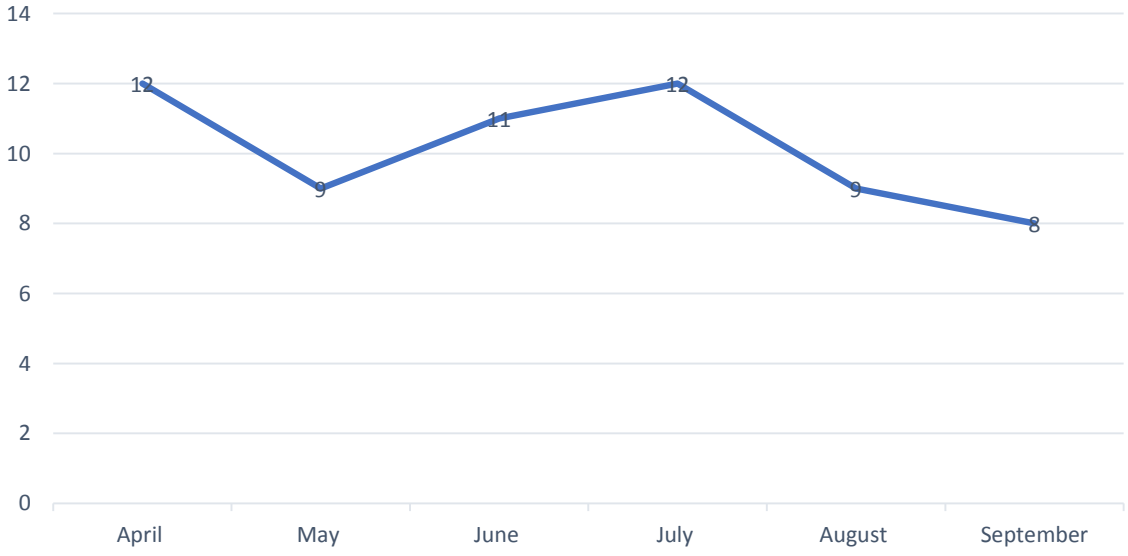
If you have any centres that are recognized by the national health ministry of your country, we can add these to the VASCERN mobile app



VASCERN Mobile App Usage last 6 months



Number of downloads each month

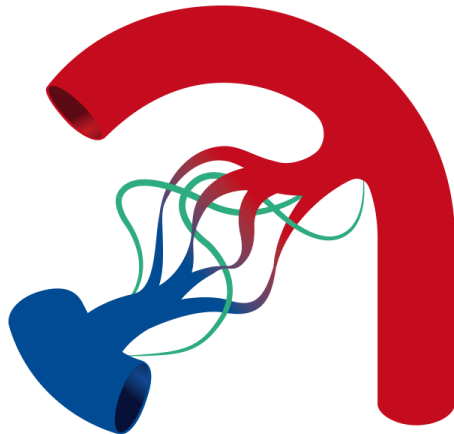




European Reference Network

for rare or low prevalence
complex diseases

• **Network**
Vascular Diseases
(VASCERN)



9.30-10.10 Inter ERN WG (Julie Hallac, Guillaume Jondeau)

- Monitoring (Julie or Guillaume) 5'
- EU initiatives (Guillaume) 5'
- Exchange (Natasha) 10'
- Pregnancy WG (Julie De Backer) 10'
- ERICA (Mari Murel) 10'

#VASCERNdays2021



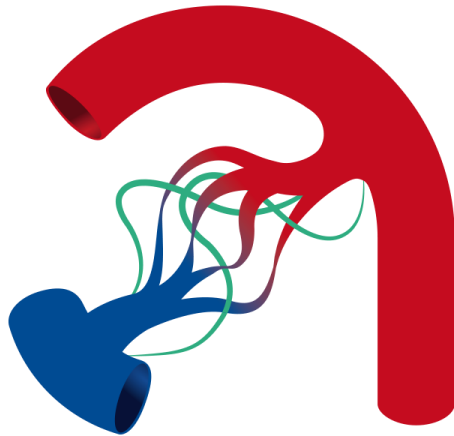
European Reference Network

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Monitoring, including meeting participation

 [#VASCERNdays2021](https://twitter.com/VASCERNdays2021)

The importance of monitoring



Purpose for the ERN

- evaluation of the ERN's relevance, outreach and sustainability
- → impact on the funding and longevity of the project

Importance for the centers

- Important for individual centers as evaluated on their answers
- Affiliated partners now participate to the data collection

Future

- CEF 2020: Demand for new indicators to improve the process for the ERNs
- Will be done through the registries → Importance of keeping the registries uptodate

Please give the number of new patients referred to your Health Care Providers & the number of congresses/meetings/conferences by **October 20th**

Continuous EU Monitoring: Data submission S2 2020 in March 2021



18 ERN Indicators	2018	2019	2020 S1 (January to June)	2020 S2 (June 2020- Feb 2021)
1. General organisation and coordination: to ensure that ERNs are operational				
1.1 Within an ERN, the number of Member States with Health Care Providers as full members or affiliated partners	11	14	17	17
1.2 Number of Health Care Providers (European Reference Centers) represented as full members in the ERN	31	31	35 (European Reference Centers) within 30 HCPs from 11 EU Member States	31 (Brexit)
1.3 Number of affiliated partners represented in the ERN	0	3	7 from 6 EU States	7 from 6 EU States
1.4 Number of patient organisations represented in the ERN → total number of patient associations represented by one or more persons actively involved in the ERN. "actively involved" can include participating in meetings, outputs, translations, mobile app datasets etc.	40	63	66	71 (current number of Patient Organisations in VASCERN App V2)
2. Patient Care: To improve access to clinical advice, diagnosis, treatment and follow up of patients within the ERNs				
2.1 Total number of new patients referred to the Health Care Providers participating in the ERN with the diagnosis of a disease or condition that falls within the scope of the ERN.	6071 missing 2 expert teams	6308 missing 1 expert teams	1862 29 expert teams replied. Less patients due to Covid19	3979
2.2 Number of patients entered into CPMS (total volume)	13	29	22	34



Continuous EU Monitoring: Data submission S2 2020 in March 2021



18 ERN Indicators	2018	2019	2020 S1 (January to June)	2020 S2 (June 2020- Feb 2021)
3. Multidisciplinary approach and sharing of knowledge within the ERN: To optimise patient outcomes by combining healthcare professionals' skills & resources used				
3.1 Number of panels reviewed by the ERN for which <u>an outcome report is produced</u> within the specified timeframe	13	12	4	11
3.2 Time taken to provide multidisciplinary clinical advice between referral to ERN and multidisciplinary clinical advice				
3.2a - non-urgent cases: days (median)	158	131	69	69
3.2b - urgent cases: days (median)	0	143	1	84
4. Education and Training: To increase capacity of professionals to recognize and manage cases of rare and complex conditions and diseases within the scope of the ERN				
4.1 Number of education/training activities not accruing higher education credits aimed at healthcare professionals delivered by the coordination teams or HCP members, ePAG advocates and Affiliated Partners of the ERN ➤ <i>Definition has evolved and includes: PoK, Webinars.</i>	4	35	1 webinar HTAD delivered by 2 HCPs and 2 ePAG patient organisations advocates	25
4.2 Number of formal educational activities (i.e. those accruing <u>higher educational credits</u>) aimed at healthcare professionals delivered by the coordination teams or HCP members of the ERN	0	0	0	0
	<i>To date, we do not have formal educational activities which are accruing higher educational credits.</i>			

Continuous EU Monitoring: Data submission S2 2020 in March 2021



18 ERN Indicators	2018	2019	2020 S1 (January to June)	2020 S2 (June 2020- Feb 2021)
5. Contribution to research and innovation: To reinforce clinical research in the field of rare and complex conditions and diseases by collecting data and carrying out research activities				
5.1.a Number of Clinical Trials (involving ERN members in at least two Member States) ➤ <i>2 HCPs from 2 Member States + acknowledgement of the ERN (recognized clinical trials registry like the ClinicalTrials.gov and published on the ERN Website)</i>	2	2	1 SAIPAN in HHT (5 HCPs involved)	1
5.1.b Number of Observational prospective studies (involving ERN members in at least two Member States) ➤ <i>At least two Health Care Providers from two different Member States within the ERN + acknowledgement of the ERN (published on the ERN Website)</i>	11	8	8	8
5.2 Number of accepted peer-reviewed publications in scientific journals regarding disease groups within the ERN and <u>which acknowledge the ERN</u> ➤ <i>PubMed, disease within the ERN, <u>at least 2 HCPs 2 different Member States + acknowledge the ERN (see guidelines) + published on the ERN website</u></i>	11	10	1 HHT	3

18 ERN Indicators

201
8

201
9

2020
(Semester 1:
January to
June)



6. Clinical guidelines: To ensure that patients referred to ERNs have equal access to high and quality health care services

6.1 Number of Clinical Practice Guidelines and other types of Clinical Decision Making Tools, adopted for diseases within the scope of the ERN (not developed by the ERN). Published on ERN website

30

1

2
PPL

6

6.2.a Number of new Clinical Practice Guidelines written by the ERN in the specified time period.

0
To date, VASCERN has not written any CPGs.

6.2.b Number of other types of new Clinical Decision Making Tools (clinical consensus statements or consensus recommendations), written by the ERN in the specified time period

10

5

8
2 HHT, 1 HTAD, 1 MSA, 4
VASCA

9

➤ *This includes our: Patient Pathways, Do's and Don'ts, Expert Consensus Statements*

7. Communication and dissemination within the scope of the ERN activities: To guarantee that knowledge is spread outside the ERN so that more people can benefit from the ERN activities

7.1 Number of congresses/ conferences/meetings at which the ERN activities and results were presented

74

98

5
Few ones due to Covid19

6

➤ *VASCERN's activities and results were presented via a dedicated slot in the programme/agenda, acknowledging VASCERN. The ERN and its activities should be the focus of the presentations. Please do not consider: presentations where the ERN was just mentioned.*

7.2 Number of individual ERN website hits

32 509

58 404

30 136
Provisional 2020: 59 098


58431

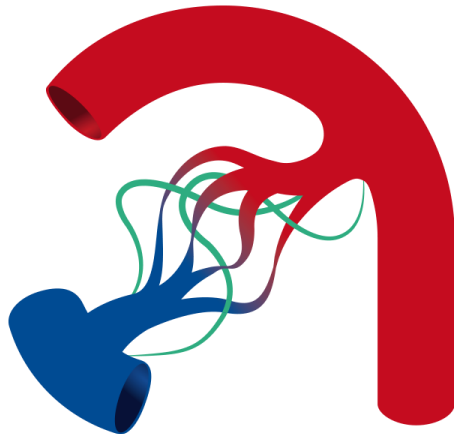




European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



EU Initiatives

 [#VASCERNdays2021](https://twitter.com/VASCERNdays2021)

Guidelines program



Who

Run by the Agency for Sanitary Quality and Evaluation of Catalonia in Barcelona

When

Meeting in June with the coordination team

5 ERNs at a time: VASCERN in the second half of 2022

How

6 months work on a new guideline or 2 tools from 2 WGs

- Consensus statement, Patient Pathways, Evidence report, Dos and Don'ts

Advise to check their Handbooks number 4 and 5 on tools and methodology

Provided by the EC:

- Microsoft Teams for 6 months
- Literature review
- Methodologist
- Training



AMEQUIS

- Integrated Assessment, Monitoring, Evaluation and Quality Improvement System
 - Launched in December 2020
 - Made by an external Foundation « Nivel » in the Netherlands
 - **Objectives:**
 - enable the ERNs to learn from experiences and identify potential pitfalls
 - An addition to the monitoring and help in interpreting the results of our activities
 - **Methods:** provide online surveys, interviews, toolkits, meetings, communication system...
 - Until 1st quarter of 2022
- VASCERN has not been contacted yet and waits for more information




CPMS

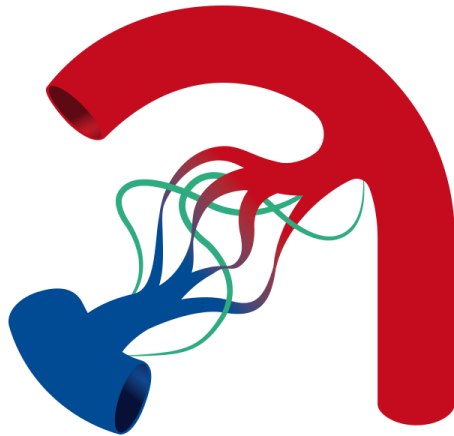
- Change of service provider : from Open App to Open source
- Uncertain phase of the CPMS
 - EU looking for improvements
 - COVID : less activity and less meetings held
 - Search for a way to connect to the registries and the Mobile App
- To be clarified by the EU with the question of the financing of the 2nd phase of the ERNs



European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



ERN Exchange Programme

[#VASCERNdays2021](#)

ERN Exchange Programme



European
Reference
Networks

Exchange Programme
2021-2022



Short-term Mobility and Exchange Programme for Health Professionals ERNs.

VASCERN will focus on the network priorities of:

- Improving medical knowledge through provision of trainings for healthcare professionals
- Sharing best practices, in particular involving organization of MDT, in order to improve medical care, management and efficiency in all of our HCPs.

→ **44 packages** (divided by 5 RDWG): **September 2021 until October 2022** (last visits must be done by **August 2022**)

→ 1 package = 5 days of exchange (daily allowance for professional that travels (200Euros/day), travel costs & travel insurance) arranged by Ecorys. Doesn't not include insurance for professional liability.

Currently: collecting exchange proposals to send to Ecorys/HaDEA for pre-approval! Name/country of Host HCP/visiting HCP, professional/their specialty visiting and aim of exchange.

14 sent so far: 3 for VASCA, 5 HHT, 3 HTAD, 3 PPL



Funded by
the Health Programme
of the European Union



Next steps:

- After getting pre-approval, we will send host and visitor the Mobility Agreement to specify the details of the exchange (date/detailed program, etc). Visitor must also sign the Declaration of Honour when going on a visit (standard procedure: 6 weeks has to be between the signed MA and date of exchange)

Mobility agreement concerning the short-term visit of a healthcare professional in the framework of the ERN Exchange Programme 2021-2022

I. DETAILS OF THE PARTIES INVOLVED

THE SENDING INSTITUTION (ERN coordinating office)
European Reference Network (ERN)
Coordinator of the European Reference Network:
- Name: _____ Surname: _____
- Function: _____
- Email address: _____

Coordinator of the European Reference Network (if coordinator and coordinator are the same)
- Name: _____ Surname: _____
- Function: _____
- Email address: _____

THE PARTICIPANT
Name: _____ Surname: _____
Specialisation: _____
Category (unilateral/): _____
Function: _____

Hospital where the participant is employed
- Name: _____
- Address: _____
- Country: _____
- Email address: _____
- ERN Member or affiliated Member: _____

II. DETAILS OF THE EXCHANGE PROGRAMME

THE HOSTING INSTITUTION
Healthcare provider hosting the exchange visit:
- Name: _____
- Address: _____
- ERN Member or affiliated Member: _____

Contact person at the hosting institution
- Name: _____ Surname: _____
- Function: _____
- Email address: _____

DETAILS OF THE EXCHANGE PROGRAMME
Start date of the exchange visit: _____
End date of the exchange visit: _____
Number of working days: _____

Detailed programme of the exchange visit:
- Objectives: _____

- Planned activities: _____

- Expected results: _____

III. COMMITMENT OF THE PARTIES INVOLVED

THE SENDING INSTITUTION (ERN coordinating office)
We approve the proposed visit and work plan.
The visit is within the following priorities of the Exchange Programme:

Coordinator's signature: _____
Date: _____

THE PARTICIPANT
I will undertake my tasks following the programme of the exchange visit as described above under the supervision of the representative of the host institution.
Participant's signature: _____
Date: _____

THE HOSTING INSTITUTION
We will host the participant and implement the programme of the exchange visit as described above.
Signature of the representative of the institution: _____
Date: _____

IV. Declaration of Honour
By the selected participant in the framework of the ERN Exchange Programme 2021-2022

I, undersigned hereby declare on my honour that:
- I understand that it is my responsibility to have a valid travel insurance in the framework of the European Reference Networks' Exchange Programme from the moment I receive a proof that my travel tickets have been booked following my written approval.
- I understand that the cost of the travel insurance will be covered by the daily allowance that I will receive for the exchange visit.
- Should I not have a travel insurance, I hereby commit to covering the costs of the travel ticket cancellation, if such cancellation is due to a reason depending on my personal unavailability (e.g. sickness, other commitments).
- If I cancel my trip following the purchase of my ticket, I confirm that I will transfer to Scorys the cost of the travel ticket without delay and within maximum 30 days.
- Should the visit be cancelled or shortened once I received the total amount of the daily allowance, I commit to reimbursing Scorys with the amount unspent for the exchange, in the amount of 600€ per overnight not used.
- I commit to promptly communicating to Scorys any change that may incur in my exchange visit depending on my personal availability / decision, in order to allow for a smooth coordination and adaptation of the plans.

Place and date: _____ Full name in capital letters and signature: _____



Next steps:

- Once approved by HaDEA: Visitor will then complete a dedicated travel form that he/she will receive from the logistics coordinators via email. In general, Ecorys will book the tickets 4 weeks before the exchange takes place.
- Visitor and host center will have to complete an evaluation form at the end of the exchange in order to give their feedback.
- Please note: We need to inform Ecorys if we plan to use all our packages so **please send proposals ASAP!**



Reimbursement Reminder



FONDATION DE L'AP-HP		SUMMARY OF TRAVEL EXPENSES										Fondation de l'AP-HP pour la Recherche		
Surname:		Project :				Date of request :								
First Name:		Purpose of the mission :				Date of mission :								
Date	Motive and place of stay Spending description	Accommodation	Meals	Receptions	€/km :		Transportation					Other	TOTAL	
					Nbre of km	Value according to the fiscal power	Kilometre allowance	Motorway fee Parking	Car rental	Train / Plane	Metro/bus/Taxi			
1														
2														
3														
4														
5														
6														
7														
8														
9														
10														
TOTAL														
Account number														
To imperatively join - Supporting documents <i>eg: train or plane ticket, boarding card</i> <i>Credit cards receipts cannot be used as a supporting document</i> - Bank account certificate														
NAME SIGNATURE		DATE		Direction Fondation Rodolphe GOUIN date : Signature :										



+Bank account details (IBAN)

+ Proof of travel/reservation

= successful reimbursement



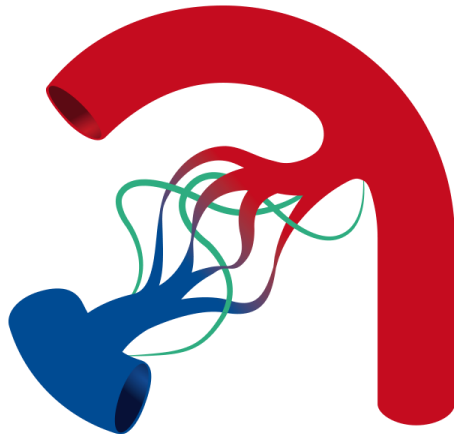
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Vascern Task force on Pregnancy and Family Planning

Julie de Backer



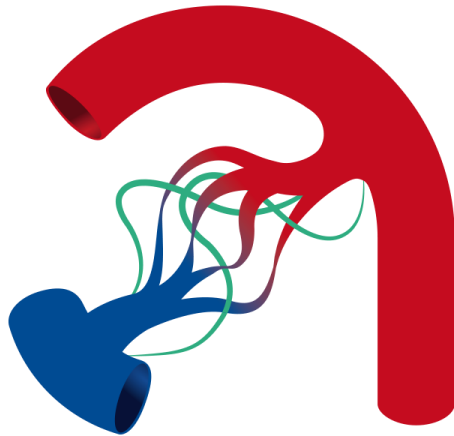
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



ERN Transversal Working Group on Pregnancy and Family Planning

Julie de Backer



Structure

Marta Mosca – ERN ReCONNET

Vascern

+18 other ERNs

Coordination: Petra Borgards, Julie De Backer, Guillaume Jondeau

HHT

HTAD

MSA

PPL

VASCA

ePAG

TF

TF

TF

TF

TF

TF



Aims

- stimulate research in this field
- deliver specific Clinical Practice Guidelines for pregnancy planning and management
- develop patients and healthcare professionals education and information
- collect European-wide evidence on the management of this specific condition



Structure – Vascern TFs

ePAG	HTAD	MSA	HHT	PPL	VASCA	Vascern
Petra Borgards	Jolien Roos Hesselink	Michael Frank Alexandra	Pernille Toerring	Janine Dickinson	Laurence Boon	Natasha Barr
	Guillaume Jondeau	Benachi (OBG Paris)	Olivier Dupuis (OBG Lyon)	Kirsten Vanduin	Corine Hubinont (OBG Brussels)	Julie Hallac
	Agnès Bourgeois-Moine (OBG)		Freya Droege (ENT)		Carine van der Vleuten	
	Bart Loeys					
	Aline Verstraeten					
	Julie De Backer					
	Edit Nagy					



Survey

- Proposal of an ERN-wide survey for the identification of unmet needs related to Pregnancy and Family Planning in rare and complex diseases
- ERN specific personalisation/ edits to the surveys
- VASCERN participation:
 - Petra Borgards (patient representative)
 - Corine Hubinont (obstetrician - VASCA and other rare vascular diseases)
 - Carine Van Der Vleuten (VASCA expert)
 - Bart Loeys (HTAD/MSA expert)

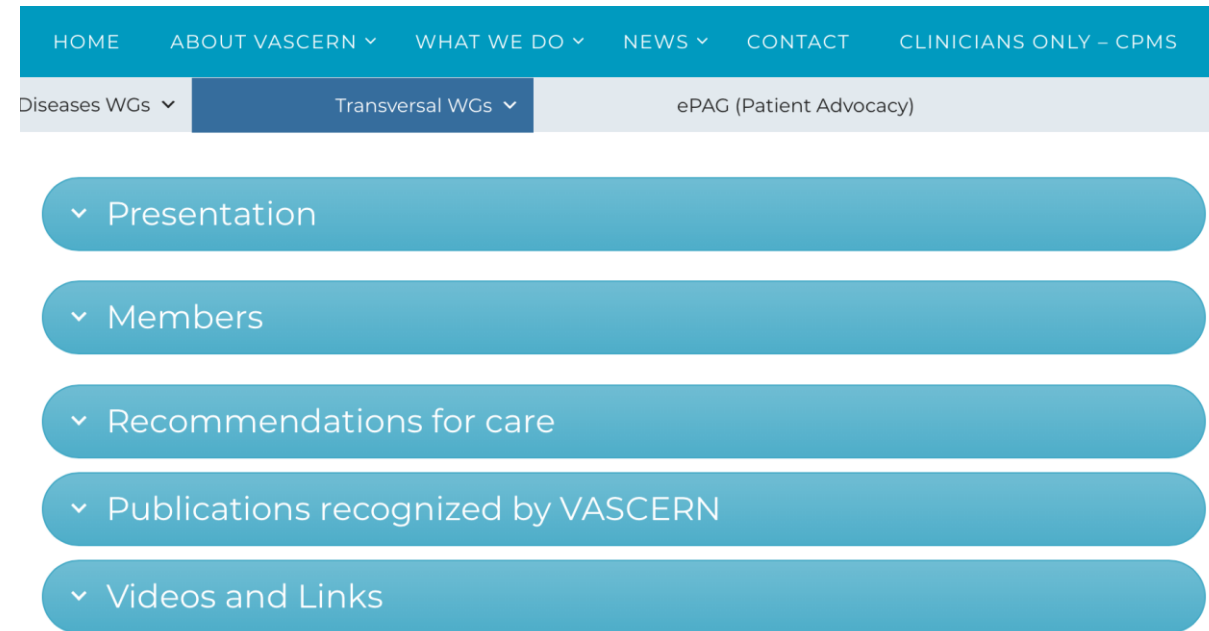
Work shop





Vascern pregnancy group - actions

- Website advertising
- **Collect available information** (CPGs, publications, Registries, patient leaflets)
- Define common interests/problems – organise a dedicated meeting





Common Interests

- Preconception genetic counselling
- Pregnancy Heart Team
- Drugs during pregnancy/lactation
- Safety of PGT
- Survey

Collaborating projects



EJP-RD

- European Joint Programme on Rare Diseases
- Presentation by Yanis Mimouni tomorrow at 8:45
- Request for an extension of 6 months to 1 year
- VASCERN is implicated in
 - Pillar 2 « Coordinated Access to Data and Services” through the Registry project
 - Pillar 3 « Training and Empowerment » as our members benefit from their services

ERICA

- Kick-off in May 2021
- European Rare Disease Research Coordination and Support Action
- All 24 ERNs
- build on the strength of the individual ERNs and create a platform that integrates all ERNs research and innovation capacity
- VASCERN’s implication:
 - WP2 Data collection: Professor SCHULTZE KOOL & Professor JONDEAU
 - WP4 Clinical trials : Prof Miikka VIKKULA



ERICA

European Rare Disease Research
Coordination and Support Action

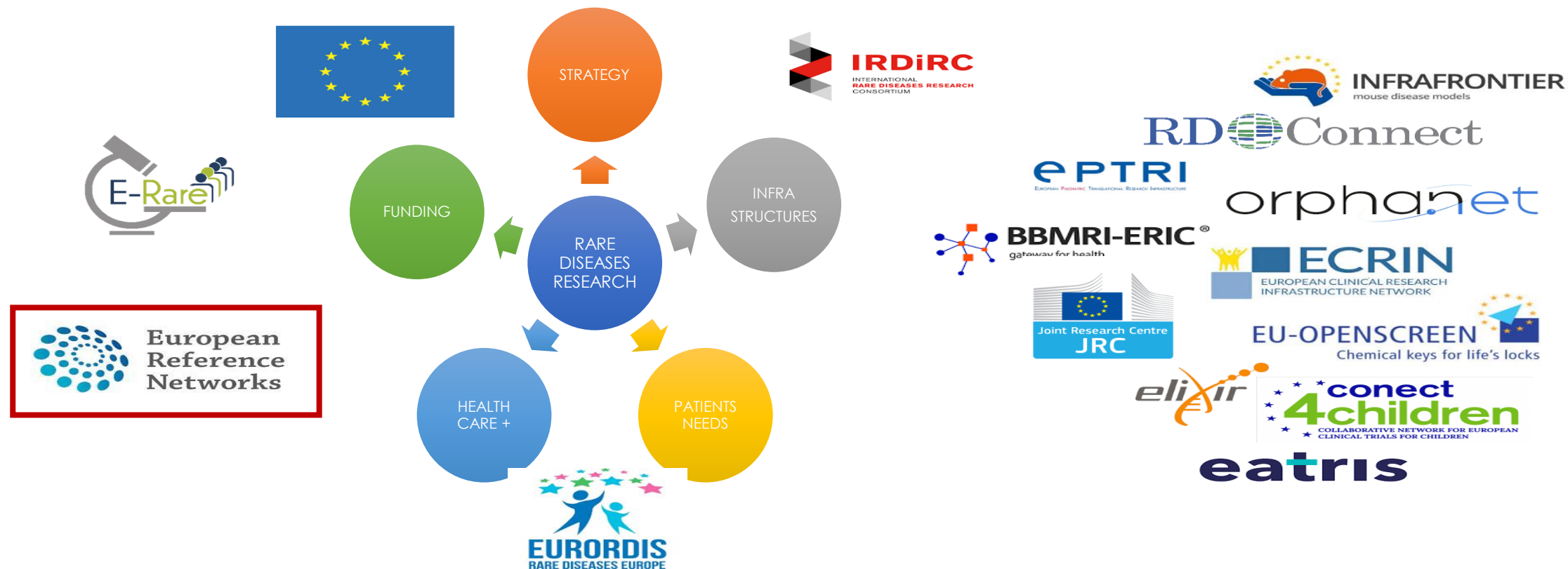
ERICA : the European Rare disease research Coordination and support Action

Mari Murel, Project Manager

VASCERN Days

Thursday 7th October

Rare Diseases Landscape in Europe



Courtesy of Dr. Daria Julkowska

There is an eminent **need for a structural framework** to facilitate collaborative research among ERNs as well as coordinated interaction with the EJP RD, IRDiRC, and the EU-based Research Infrastructures

Table 1.3a: Results of RWG Research Priorities Survey, in which three general items and the 10 highest ranked specific research topics are listed with the mean priority scores assigned for intra- and inter-ERN relevance and the desired support type.

Nr	Priority (scale 1-10)		Topic	Desired support type: N=Networking C= centralised structure I = IT support	ERICA WP to cover item
	Intra ERN	Inter ERN			
General					
A	8.7	8.6	Strengthening the ERN Research Working Group	N	WP1
B	8.3	7.9	International collaborations	N	WP1,6
C	7.6	7.5	Raising awareness of existing research infrastructures	N	WP1,6
Research topic specific					
1	9.0	8.3	Patient-centred outcome measures	N	WP3
2	9.0	8.3	Pragmatic (registry based) clinical trials	N	WP2,5
3	8.2	8.2	Omics / biomarker research expertise sharing	N	WP5
4	8.1	8.2	Legal issues (data protection, Informed Consent)	C	WP1,2,4
5	8.2	7.9	Harmonized data capture (incl. collection/storage and data FAIRification)	I	WP2
6	7.9	7.6	Genomic diagnostic expertise sharing	N	WP5
7	7.9	7.6	Patient involvement in clinical trials	N	WP3,4
8	7.9	7.5	Creation of biorepositories	N	WP2,5
9	8.1	7.1	Investigator initiated trial planning & execution	N	WP4
10	7.4	7.2	Advanced experimental therapies	N	WP4,5

Overall Objective

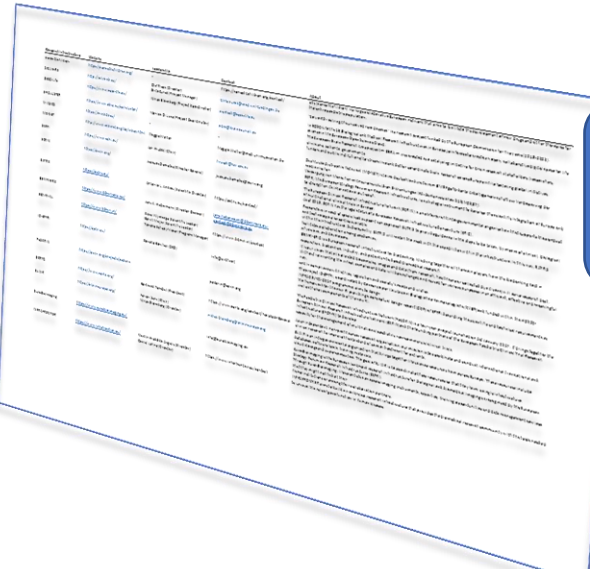
- To increase the research and innovation potential of ERNs, through:
 - facilitating inter-ERN collaboration
 - increase the visibility and impact of ERNs
(outreach and collaboration with multiple stakeholders)



1st March 2021

28th February 2025

Work Packages



Principal Strategies

1) **Central engagement of** all ERN Members in all project activities

(participation in the different WP specific **Expert Working Groups (EWGs)**)

- Ensures the identification of the research active ERN community
- Provides first-hand expertise of active clinical RD researchers
- Allows a continuous 'reality check' of the usefulness/ feasibility of research activities

2) **Seeking strategic partnerships** (all major European and intern RD research stakeholders).

- Information exchange, consensus building, and coordination of joint activities
- Steering (multi-stakeholder advisory board)
- Operational: inclusion of external stakeholders in WP specific EWGs

ERICA Consortium : 29 partners

Partner	Represented by
All 24 ERNs	Coordinators institutes WP1-7 co-chairs
EJP RD	WP2, WP4, and WP5 co-chairs
Orphanet	Ana Rath, INSERM, WP3 co-chair
Mapi Trust Research	Sonia Bothorel
Eurordis	Ines Hernando
EATRIS	Anton Ussi, WP5 co-chair

Executive Committee

Alberto Pereira	WP1, WP6, WP 7 leader
Franz Schaefer	WP2 leader
Eduardo Lopez Granados	WP2 leader
Mar Mañú Pereira	WP3 leader
Ana Rath	WP3 leader
Ralf-Dieter Hilgers	WP4 leader
Luca Sangiorgi	WP4 leader
Ruth Ladenstein	WP5 leader
Anton Ussi	WP5 leader
Maurizio Scarpa	WP6 leader



Alberto Pereira



Franz Schäfer



Eduardo Lopez Granados



Mar Mañú Pereira



Ana Rath



Ralf-Dieter Hilgers



Luca Sangiorgi



Ruth Ladenstein



Anton Ussi



Maurizio Scarpa

Strategic Advisory Board Members



Birute Tumiene



Helena Kääriainen



Eileen Treacy



Katarzyna Kotulska



Simona Martin



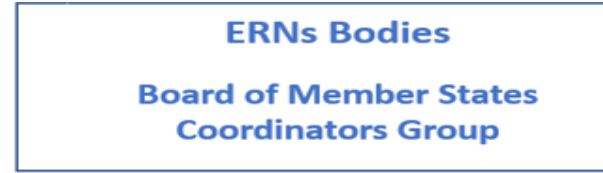
Lucia Monaco



Daria Julkowska



Rima Nabbout



ERNs Bodies
Board of Member States
Coordinators Group



**Rare Disease
Consortia**

EUROPEAN JOINT PROGRAMME
RARE DISEASES

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM



European Commission

Horizon Europe

Directorate-General for Health and Food Safety (DG SANTE)

RESEARCH & INNOVATION

Regulatory and Innovation Advisory Board Members



Kristina Larsson



Bruno Sepodes



Edith Frénoy



Bernard Grimm



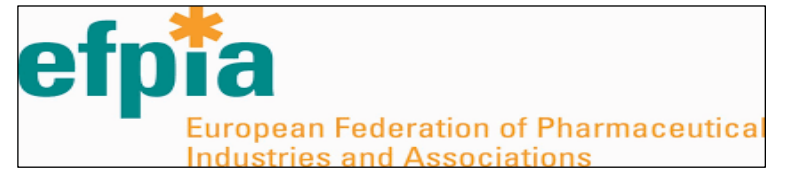
EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH



CHMP

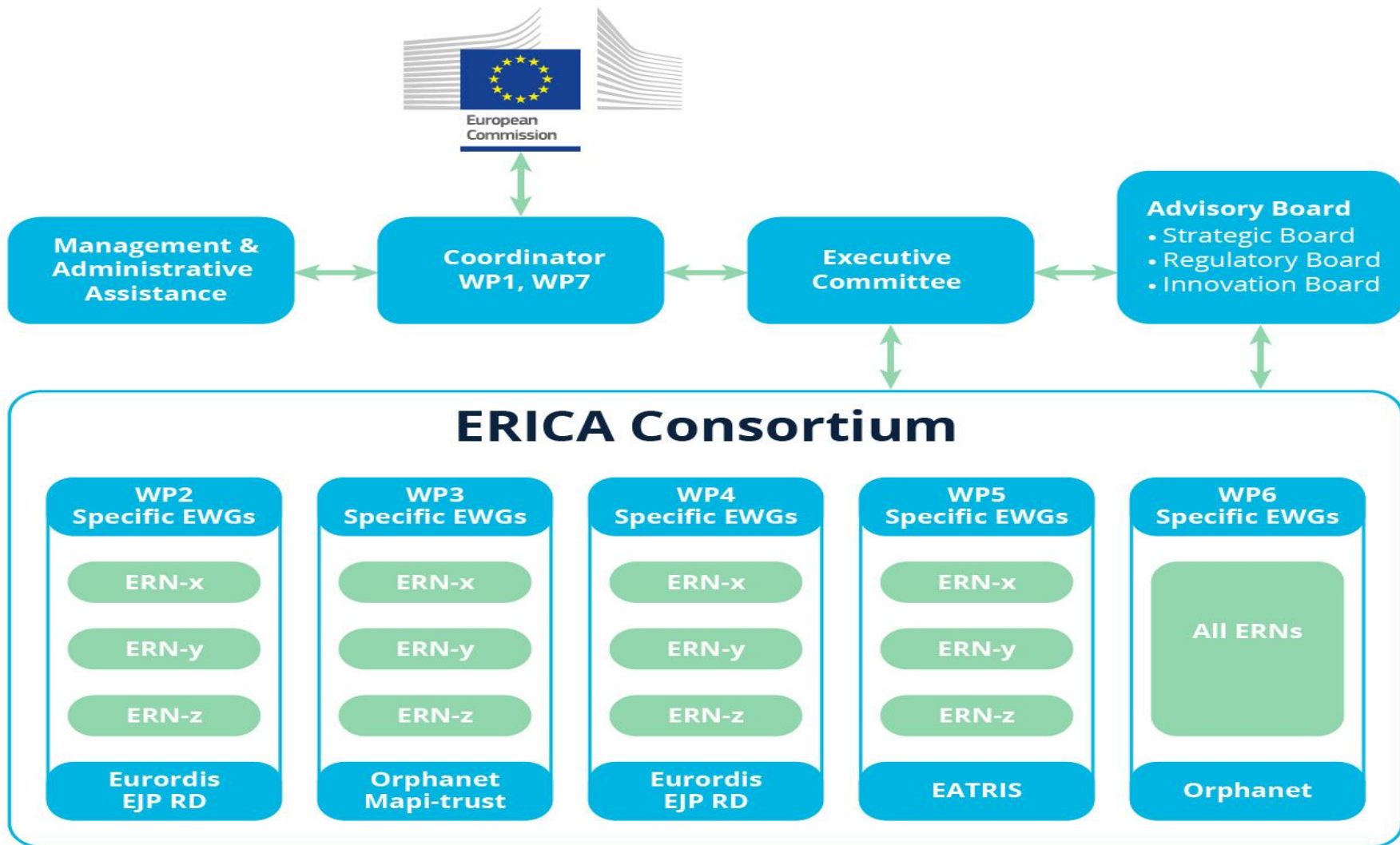
stands for

**Committee for Medicinal
Products for Human Use**



efpia

European Federation of Pharmaceutical
Industries and Associations



WP-Specific EWGs

WP2 Data Collection, Integration and Sharing:

- **Introduction**
- **ERN registries**
- **Expert Working Groups (EWG):**
 - **Legal & Ethical EWG**
 - **Data Collection EWG**
 - **Data Usage EWG**
 - **Research and Monitoring EWG**
 - **Biobanking EWG**

WP 3 Patient-Centred Research:

- **Introduction**
- **Expert Working Groups (EWG):**
 - **Patient-Centred EWG**
 - **Participants**

WP4 Clinical Trial Support:

- **Introduction**
- **Rare Disease Trial Workshops**
- **Expert Working Groups (EWG):**
 - **Clinical Trials Support EWG**

WP 5 Translation and Innovation:

- **Introduction**
- **Expert Working Groups (EWG):**
 - **Innovation EWG**

WP6 Integration, Outreach & Dissemination:

- **Introduction**
- **Workshops & Conferences**
- **Expert Working Groups (EWG):**
 - **Integration, Outreach & Dissemination EWG**

What did we do and accomplish since the start

- First Kick-off meeting and 1st GA 27-28th May

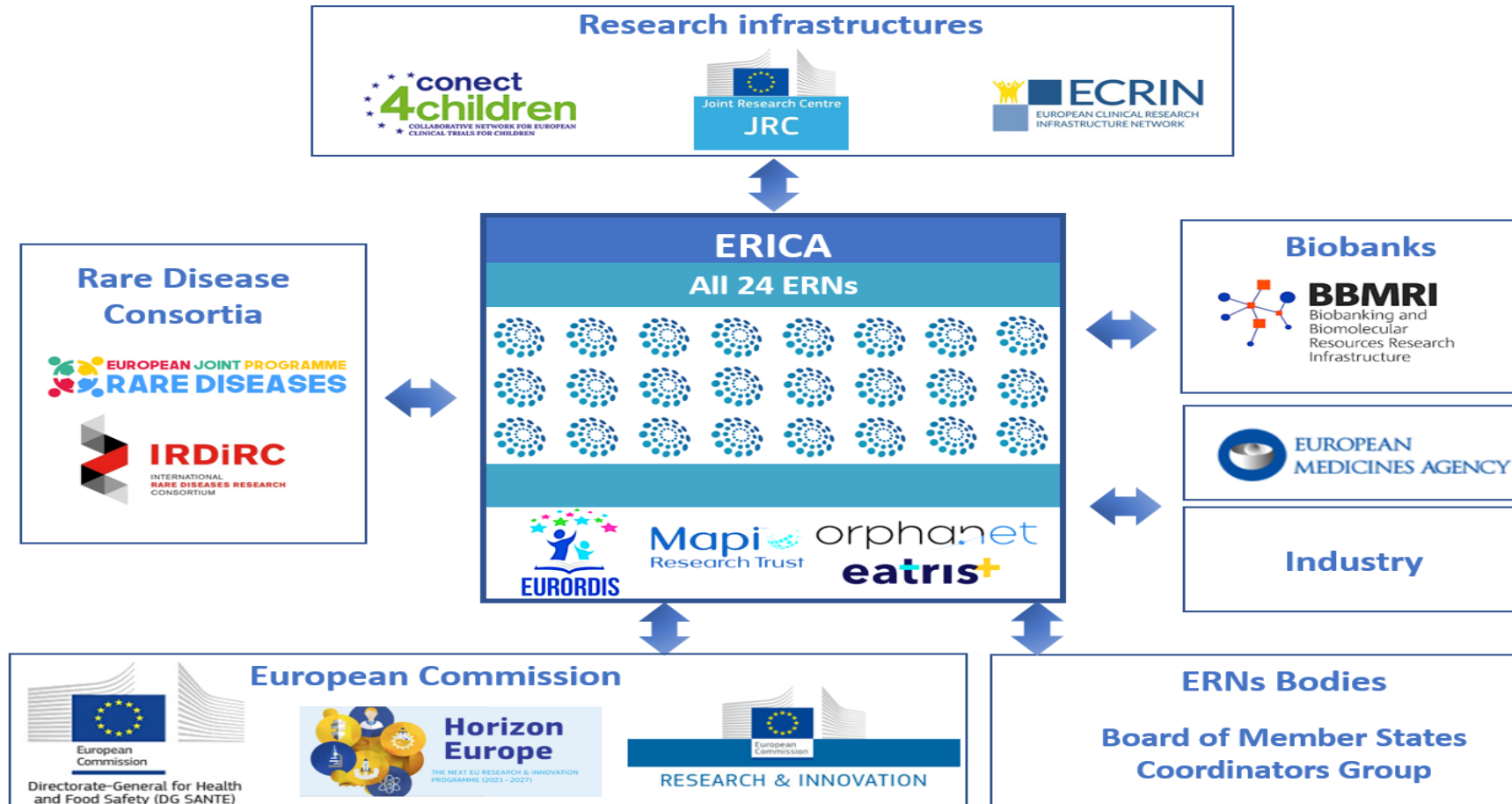


160 attendees

5 keynote Speakers (IRDiRC chair, EURORDIS CEO, EJP RD Coordinator, European Commission representatives).

6 WP-specific EWG meetings

Positioning of ERICA within the RD research ecosystem



Expected impact

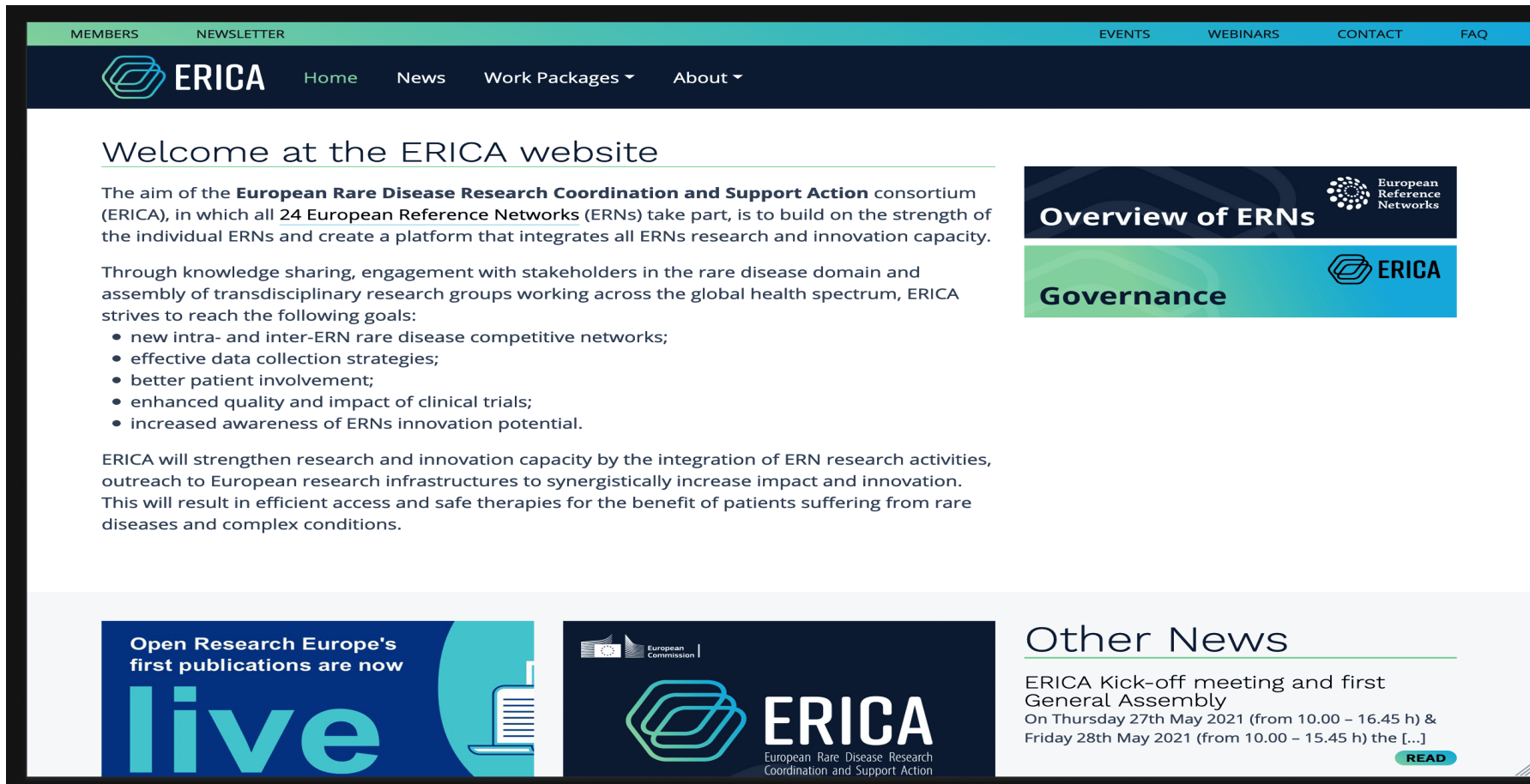
EJP RD:

- efficient communication of the specific needs of RD researchers to the EJP RD specialist workgroups to adjust working priorities
- facilitated application of EJP RD specialist guidance and newly developed tools by the ERN research community
- fostering of collaborative research projects that will take advantage of the power of the ERNs' large patient cohorts and the unique research expertise gathered in the EJP RD

ERN Generic:

- new intra- and inter-ERN rare disease competitive networks
- effective data collection strategies
- better patient involvement
- enhanced quality and impact of clinical trials
- increased awareness of ERN's innovation potential

ERICA website www.ERICA-rd.eu



The screenshot shows the ERICA website homepage. At the top, there is a navigation bar with links for MEMBERS, NEWSLETTER, EVENTS, WEBINARS, CONTACT, and FAQ. Below this is a dark blue header with the ERICA logo and navigation links: Home, News, Work Packages, and About. The main content area features a welcome message, a description of the ERICA consortium's aim, a list of goals, and information about strengthening research capacity. On the right, there are two featured sections: 'Overview of ERNs' with the European Reference Networks logo, and 'Governance' with the ERICA logo. At the bottom, there are three promotional banners: 'Open Research Europe's first publications are now live', a banner with the ERICA logo and European Commission logo, and 'Other News' with details about the ERICA Kick-off meeting and first General Assembly, including dates and times, and a 'READ' button.

MEMBERS NEWSLETTER EVENTS WEBINARS CONTACT FAQ

ERICA Home News Work Packages About

Welcome at the ERICA website

The aim of the **European Rare Disease Research Coordination and Support Action** consortium (ERICA), in which all 24 European Reference Networks (ERNs) take part, is to build on the strength of the individual ERNs and create a platform that integrates all ERNs research and innovation capacity.

Through knowledge sharing, engagement with stakeholders in the rare disease domain and assembly of transdisciplinary research groups working across the global health spectrum, ERICA strives to reach the following goals:

- new intra- and inter-ERN rare disease competitive networks;
- effective data collection strategies;
- better patient involvement;
- enhanced quality and impact of clinical trials;
- increased awareness of ERNs innovation potential.

ERICA will strengthen research and innovation capacity by the integration of ERN research activities, outreach to European research infrastructures to synergistically increase impact and innovation. This will result in efficient access and safe therapies for the benefit of patients suffering from rare diseases and complex conditions.

Overview of ERNs

European Reference Networks

Governance

ERICA

Open Research Europe's first publications are now live

European Commission

ERICA

European Rare Disease Research Coordination and Support Action

Other News

ERICA Kick-off meeting and first General Assembly
On Thursday 27th May 2021 (from 10.00 – 16.45 h) &
Friday 28th May 2021 (from 10.00 – 15.45 h) the [...]

[READ](#)


ERICA should be the first step towards having Clinical Research Networks covering all RDs, embedded in the ERNs, conducting collaborative research for RD that complies with the standards required by regulatory and HTA bodies while enabling systematic data collection, especially for neglected areas

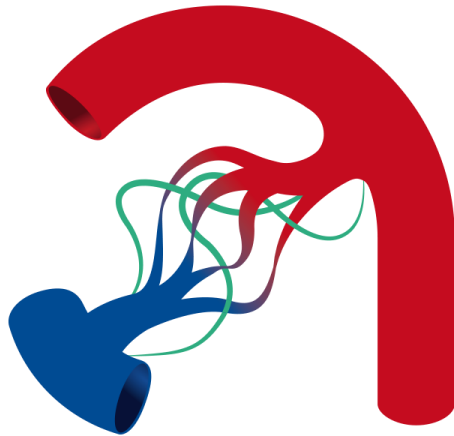




European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



10.10-10.30 coffee break

**10.30-11.30
Work Packages**

**10.30-11.05
Implementation (7' per RDWG Chair)**

HHT – Sophie Dupuis-Girod

HTAD – Julie De Backer

MSA – Michael Franck

PPL – Robert Damstra

VASCA – Miikka Vikkula

**11.05-11.30 Q&A and approval by the
Board**

 **#VASCERNdays2021**



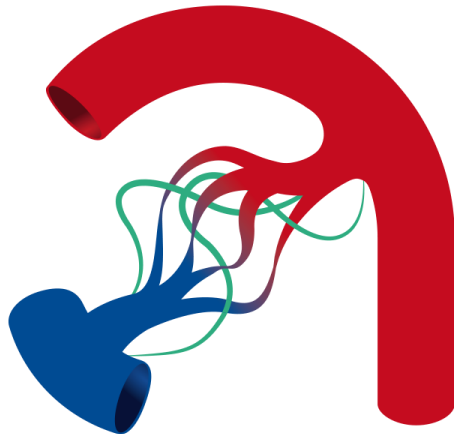
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Vascern HHT WG

Sophie Dupuis Girod HHT WG Chair

[#VASCERNdays2021](https://twitter.com/VASCERNdays2021)



The 2020-2021 context

- **Brexit, an unacceptable situation for our group**
 - ✓ **The HHT working group is built upon multidisciplinary centers of Excellence that have been collaborating since our first international meeting in 1996.**
 - ✓ **A major involvement of Claire Shovlin since 2017 in VASCERN;**
- **COVID**
 - ✓ **the devastation wrecked by COVID-19 on the HHT WG HCPs**



Sharing of experience (WP4)

- **Despite COVID, we managed a further 11 meetings from November 2020 to September 2021 (now at #50)**
- **Including 1 virtual FTF meeting in June 2021**
- **7 difficult clinical cases discussed**

COVID Monitoring and its impact on healthcare

The experience remains variable and some centres severely affected.

Around 60% reduction in number of new patients seen by HCPs in 2021:

1. Substantial reductions in new patient numbers for 2021 for the Italian (Bari/Crema) and UK (London) HCPs;

2. All reported even where services running again, patients were reluctant to travel and are asking to postpone elective procedures and screening;

3. Certain centres where staff at frontline of COVID care have significant tolls in terms of human resources- eg pulmonary teams at core.

number of new patients	2017	2018	2019	2020	2021 estimate	2021 Jan-june	Period with no new cases 2020	Now able to see new cases?	Usual patient willingness to be seen	Clinical research
Odense University Hospital	29	46	39	39	similar		Mar-early May	Yes- expect catch up by Sept/Oct	No	Yes
University Hospital Essen	52	40	66	66	~20% down		Mar-early May	Yes- expect catch up by early 2021	No	Yes
Azienda Ospedaliero-Universitaria	23	22	22	4	>80% down	Screening = 0 pts	March onwards	No	No	No
Maggiore Hospital, ASST Crema	102	126	107	18	>50% down	Screening 21 pts	Feb onwards	Yes	Yes	Yes
Hammersmith Hospital, Imperial College Healthcare NHS Trust London	159	118	217	~40	>80% down unless count TC		Late March onwards	(2 of 4- Y, 3rd to start Oct 2020)	No	No
St. Antonius Hospital	145	156	94	74	~20% down		Mid Mar-mid May	Yes -expect 2 month loss for year	No	Yes
Hospices Civils de Lyon	82	80	84	69	~20% down	53	Mid Mar-mid May	Yes -expect 2 month loss for year	No	Yes
OVERALL	679	686	726	<312	~60% DOWN					



Pills of knowledge - webinar (WP7)

- The HHT WG's first webinar:
- "HHT and the lungs" on May 31st, 2021
 - Dr. Hans-Jurgen Mager,
 - Prof. Marco Post,
 - Claudia Crocione
 - Christina Grabowski
- Over 100 participants registered
- 171 views

European Reference Network
for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

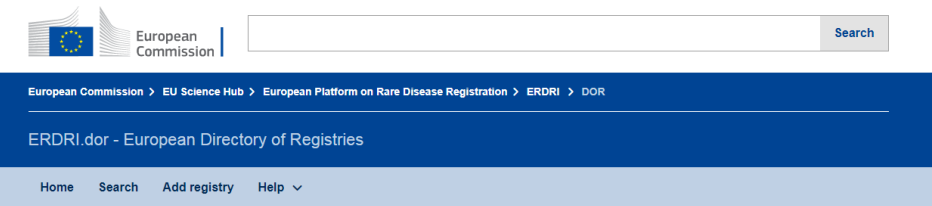
VASCERN Webinar:
HHT and the lungs

Dr. Hans-Jurgen MAGER
Dr. Marco POST

Claudia Crocione

CIROCO progress (WP8)

- The CIROCO database was selected to become the European registry.
- Project manager : Evelyne Decullier
- Developer : Jean-Philippe Allard
- CIROCO-HHT was registered on ERDRIdor
- The minimum dataset was implemented and now accessible on the internet.
- Monthly meeting of the registry WG with Pim Kamerling and Leo Schultze
- Now working on the Fair datapoint / Data transfer agreement
- Waiting for the pseudonymisation number



CIROCO-HHT	
General Information	
Acronym	CIROCO
Medical area	HHT
Type of Registry	Epidemiology, Clinical Registry, Patient Registry
Other type	
Description	CIROCO is the registry for HHT, in the scope of VASCERN. This database has been created in France for HHT patients and is now being modified for its European use. S. Dupuis-Girod and G. Jondeau are coordinating it, E. Decullier is data manager and JP Allard
Registry is member of Eurocat?	No
Registry is member of:	VASCERN
Website	https://vascern.eu/expertise/rare-diseases-wgs/hht-wg/
Sponsors	Radboudumc

CIROCO

EJP for RD | A.R.C. | sophie/9 | Malade | A146 F

SET OF COMMON DATA for RARE DISEASES REGISTRATION

1 PSEUDONYM EUPID

2 Personal Informations

Date of Birth optional if foetus: 19/06/1942 | If Foetus check:

Sex: M F Undetermined | NSP:

3 Patient Status

Alive Dead Lost Opted-out

4 Care Pathway

First contact with specialised centre: 01/01/2001 | dd/mm/yyyy

5 Disease History

At onset: | Postnatale | date: | years: | dd/mm/yyyy

At diagnosis: | Indetermined | date: | years: | dd/mm/yyyy

6 Diagnosis

Diagnosis of the rare disease - code:

Genetic diagnosis:

Undiagnosed case:

7 Research

AGREEMENT to be contacted for research: YES NO ND

CONSENT to the reuse of data: YES NO ND

Biological sample: YES NO ND

Link to a biobank: YES NO ND

Link:

8 Disability

Classification of functioning/disability:

Contact : Dr.Sophie DUPUIS-GIROD | sophie.dupuis-girod@chu-lyon.fr
 Hôpital Femme-Mère-Enfant - Bâtiment A11 - 1er étage
 Groupe Hospitalier Est
 59, boulevard Pinel
 69677 BRON

Clinical trial and research (WP9)

SAIPAN trial



- **Design: Multicenter, open-label RCT**
- **Start: 2019**
- **Intervention: 0.1mg Octreotide 2/day**
- **Sample size: N = 38**

Participating centers

- **Radboud UMC, Nijmegen - PI**
- **St. Antonius ziekenhuis, Nieuwegein**
- **Uniklinik Essen, Germany**
- **Hospices Civils de Lyon, France**
- **UMC Bari, Italy**
- **Maggiore Hospital Crema, Italy**
- **Hammersmith hospital, UK**



Videos on youtube (WP10)

- >2,000 views for each of the three HHT videos,
- 20,000 views combined



Hereditary Haemorrhagic Telangiectasia (HHT) is more than a bleeding nose.
What an ENT doctor need to know about HHT and why



European Reference Networks | Vascular Diseases (VASCERN) | Hereditary haemorrhagic telangiectasia

HHT: Hereditary haemorrhagic telangiectasia
An inherited blood vessel condition affecting ~ 1 in 6,000 people

Frequency of abnormal HHT vessels :

>95%	Nose (→ nosebleeds)
>90%	Skin (telangiectasia)
50%	Lungs (pulmonary AVMs)
50%	Liver (hepatic AVMs)
20%	Gastrointestinal tract
10%	Brain (cerebral AVMs)

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

HHT from VASCERN HHT
VASCERN HHT-WG
An Introduction to Hereditary Haemorrhagic Telangiectasia (HHT)

0:02 / 4:47



Clinical outcome measures (WP11)

- **Clinical outcome measures evaluation (AK) on going study**
- **Clinical outcome measures update discussed and postponed**



Clinical Practice guidelines (WP13)

COVID

VASCERN HHT Statement on COVID-19

A statement from the European Reference Network for Rare Multisystemic Vascular Diseases (VASCERN) for people with hereditary haemorrhagic telangiectasia (HHT) and their doctors:

VASCERN Hereditary Haemorrhagic Telangiectasia (HHT) Working Group – Statement concerning SARS-CoV-2 vaccination for patients with HHT

General statement

Follow the national safety advice for social distancing, mask wearing etc. Vaccination is important for the whole population but it remains your own choice.

Recommendation for vaccination

Please follow the national guidelines concerning vaccination.

HHT and SARS-CoV-2 vaccination

SARS-CoV-2 vaccination is encouraged in patients with HHT, particularly if patients require regular hospital attendance (e.g. for blood transfusions or iron), are older (>50 years old), or have comorbidities such as chronic respiratory disease, cardiovascular diseases, obesity or diabetes.

Vaccination during pregnancy should follow national guidelines for pregnancy.

COVID19 statement published at

<https://vascern.eu/home/vascerns-covid19-recommendations/> in March 2020

- 1. People with HHT should follow the standard Public Health Measures as recommended in their specific country.**
 - 1.1. These are directed at reducing the spread of infection, and strategies differ slightly between countries.
- 2. People with HHT should be no more and no less concerned about COVID-19 than the general population without HHT.**
 - 2.1. There is no reason to think people with HHT [1] will be at higher or lower risk of infection [2,3], or complications if they become infected.
 - 2.2. Some people with HHT, as for some people in the general population, may be less able to tolerate the extra demands placed on their bodies if they suffer infection with complications, but this will be a small group and should not be applied to all HHT patients.
- 3. The presence of HHT or AVMs in someone who currently has a normal or high exercise tolerance should not limit their access to medical treatment compared to someone without HHT or AVMs of the same age.**
 - 3.1. People with HHT have normal life expectancy managed in Europe, [4,5] likely attributed to the beneficial effects of reduced cancer, reduced rates of cancer [4,6,7] and fewer heart attacks [8].
 - 3.2. Anyone with normal or high exercise tolerance (able to walk uphill quite briskly without stopping) will have good cardiorespiratory reserve [9,10].
- 4. For those who are obliged to self-isolate because of the general situation**
 - 4.1. Maintain normal treatment regimes if possible. **It is very important that iron supplements for anaemia are continued.**
 - 4.2. Avoid sedentary states- exercise is recommended as discussed further for the general population [11].

AUTHORS:
Claire L. Shovlin, Carlo Sabha, Hans Jørgen Møger, Anette Kjeldsen, Ulrich Sure, Elisabetta Buscarini and Sophie Dupuis-Girod
VASCERN HHT Centre Leads in Denmark, France, Germany, Italy, the Netherlands and the UK, on behalf of VASCERN HHT
During the current epidemic, reference centres may be unable to perform elective diagnostic/therapeutic procedures for people with HHT but they continue to assist urgent/emergency presentations

COVID 19 VACCINATION STATEMENT was discussed, approved and published in jan 2021 and modified in march 2021
<https://vascern.eu/wp-content/uploads/2021/01/HHT-COVID-vaccination-statement.pdf>

Now working on Q&A with the ePag



Clinical Practice guidelines (WP13)

Special issue of EJMG

Paper 1 submitted by Claire Shovlin, under revision

How a European rare disease network for hereditary haemorrhagic telangiectasia impacted patients' care

¹Shovlin CL*, ²Buscarini E*, ³Sabbà C, ⁴Mager JJ, ⁵Kjeldsen AD, ⁶Pagella F, ⁷Sure U, ⁸Ugolini S, ⁹Toerring PM, ¹⁰Suppressa P, ¹¹Rennie C, ¹²Post MC, ¹³Patel MC, ¹⁴Nielsen TH, ¹⁵Manfredi G, ¹⁶Lenato GM, ¹⁷Lefroy D, ¹⁸Kariholu U, ¹⁹Jones B, ²⁰Fialla AD, ²¹Eker OF, ²²Dupuis O, ²³Droege F, ²⁴Coote N, ²⁵Boccardi E, ²⁶Alsafi A, ²⁷Alicante S, ²⁸Dupuis-Girod S*

* Corresponding authors

Paper 2 in progress

European Reference Network for Rare Vascular Diseases (VASCERN): When and how to use bevacizumab in Hereditary Haemorrhagic Telangiectasia (HHT) ?



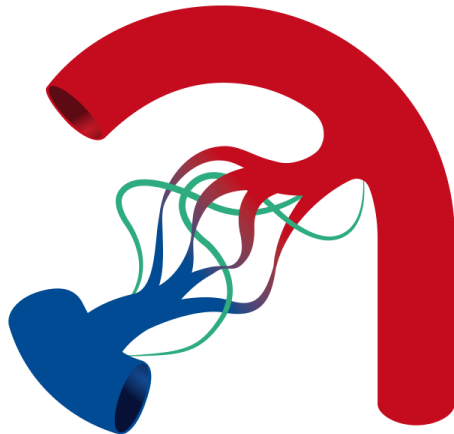
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



HTAD Working Group

WP implementation 2021

Julie de Backer

 #VASCERNdays2021



WP1	Coordination/ project management	Subgroups/projects
WP4	Sharing of experience: discussion of difficult clinical cases	Monthly Calls – 1-2 Cases + more requests external
WP5	Patients Pathways: improvements and updates	Manuscript submitted EJMG
WP7	Pills of Knowledge	4 completed by the end of this year
WP8	Registries	HTAD Database under in final stage - ROPAC
WP9	Clinical trials & Research	1 Research study completed - 1 ongoing – 1 proposal
WP12	Definition of clinical outcomes	Survey developed – analysis in progress
WP13	Writing Clinical Practice Guidelines	1 ongoing
WP14	Do's and Don'ts factsheets	2 available



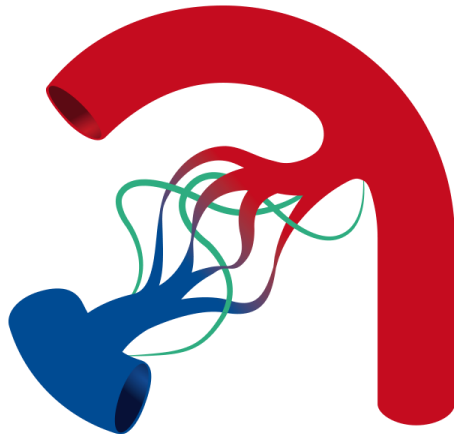
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



MSA-WG implementation

Chair : Michael Frank
Co-chair: Fransiska Malfait

 #VASCERNdays2021

MSA WG changes



 **FRANCE**

 AP-HP, CHU Paris IdF Ouest, HEGP Hôpital Européen Georges Pompidou, Paris, France

 VASCERN MSA European Reference Centre, Rare Vascular Diseases Reference Centre (CRMR)

 **Chair**
Dr. Michael FRANK
Consultant Cardiologist

 Prof. Xavier JEUNEMAIRE
Geneticist

 **BELGIUM**


 Ghent University Hospital, Ghent, Belgium


 VASCERN MSA European Reference Centre, Dept of Cardiology and Center for Medical Genetics

 Pr. Julie De BACKER
Cardiologist and clinical geneticist


 **Co-Chair**
Dr. Fransiska MALFAIT
Clinical geneticist


 **GERMANY**


 Ehlers-Danlos-Initiative e.V.


 European Patient Advocacy Group

 **ePAG Co-Chair**
Juergen GRUNERT

 Bindweefsel.be

 European Patient Advocacy Group

 **ePAG Deputy Co-Chair**
Charissa FRANK



**ePAG
patient
advocates**

 University Hospital of Antwerp, Antwerp, Belgium

 VASCERN MSA European Reference Centre, Center of Medical Genetics

 Pr. Bart LOEYS
Clinical cardiogeneticist

 Dr. Aline VERSTRAETEN
Clinical geneticist

 **NETHERLANDS**

 Erasmus Medical Center, Rotterdam, The Netherlands

 VASCERN MSA European Reference Centre, Department of Clinical Genetics and Cardiology

 Dr. Ingrid VAN DE LAAR
Clinical Geneticist

 Prof. Jolien ROOS-HESSELINK
Cardiologist


 **CYPRUS**


 The Cyprus Institute of Neurology and Genetics, Nicosia, Cyprus


 Department of Cardiovascular Genetics & Laboratory of Forensic Genetics

 Dr. George TANTELES
Consultant in Clinical Genetics

 Dr. Evy BASHIARDES
Molecular Biologist

 Mater Dei Hospital, Msida, Malta

  **MALTA**

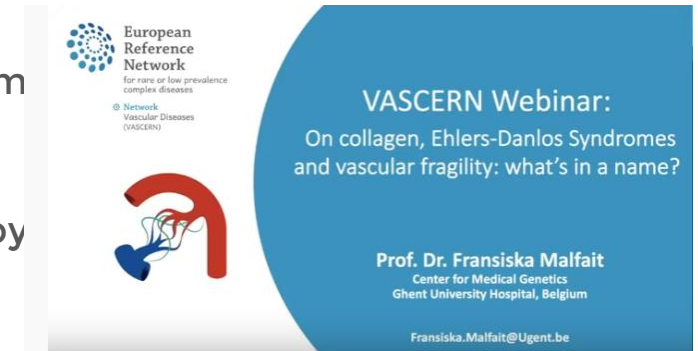
 Dr. Kevin CASSAR
Vascular Surgeon

**Affiliated
Partners**



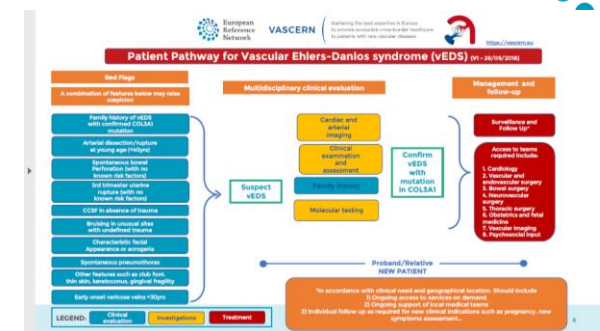
Work packages

- **WP2: dissemination /communication**
 - several participations in international online meetings on vEDS (Marfan Foundation, DEFY foundation) with reference to ERN
- **WP7/WP10: Pills of knowledge**
 - 19 active Pills of knowledge on vEDS published on YT, views ranging from
- **WP7/WP10: Webinars**
 - 2 webinars (1 joint with ERN Reconnet, further should be programmed by
- **WP14: do's and don'ts**
 - 17 available factsheets on most common complication of vEDS
- **WP13 Clinical Practice guidelines**
 - Online survey on patients monitoring as first step towards an expert consensus statement
 - Further guidelines to be discussed and agreed on worldwide





Work packages (II)



• WP5: patient pathway

- Improvement on preventive measures adapted last fall
- To come: implementation of patient monitoring following publication on practice in European expert Centres

• WP9: research/CT

- CT : multicenter RCT on ARB as add-on to betablocker in complement to the ARCADE trial held in France, awaiting to be financed
- Identification of modifying genes in vEDS:GWAS collaborative study in progress (study for colonic perforation)
- Survey on PGD and IVF
- Survey on Arterial monitoring in vEDS
- Papers to submit:
 - Common practice of arterial monitoring in vEDS patients (9 ERN expert centres totalling 331 patients)
 - Obstetrical complications of vascular EDS (retrospective cohort study, France) 40 patients 90 deliveries
 - COL3A1 deletion phenotype (Dr Marlies Kempers)

Assisted reproduction and PGD (Preimplantation Genetic Diagnosis) outcomes in Vascular Ehlers-Danlos syndrome (vEDS) patients

Vascular, abdominal and uterine complications during pregnancy in Ehlers-Danlos syndrome are common. The information and professional advice on assisted reproduction and PGD in vEDS patients is still limited. The ERN VASCERN MSA working group is looking at writing a consensus guideline for PGD in vEDS.

Your professional experience working with vEDS patients is highly valuable and we would be grateful if you could share it with us. Thank you very much for participating in our survey.

• WP4: sharing of experience (complex clinical cases)

7-9th of October 2021 • >6 cases per year



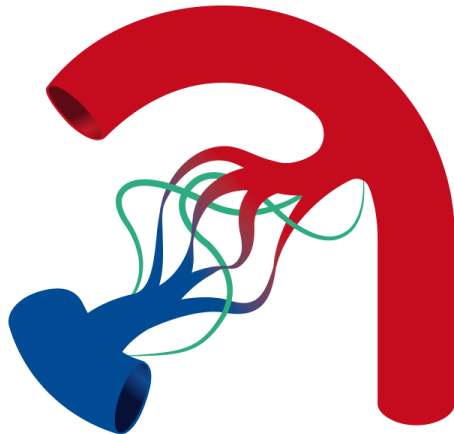
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Vascern PPL-WG

Robert Damstra PPL WG Chair

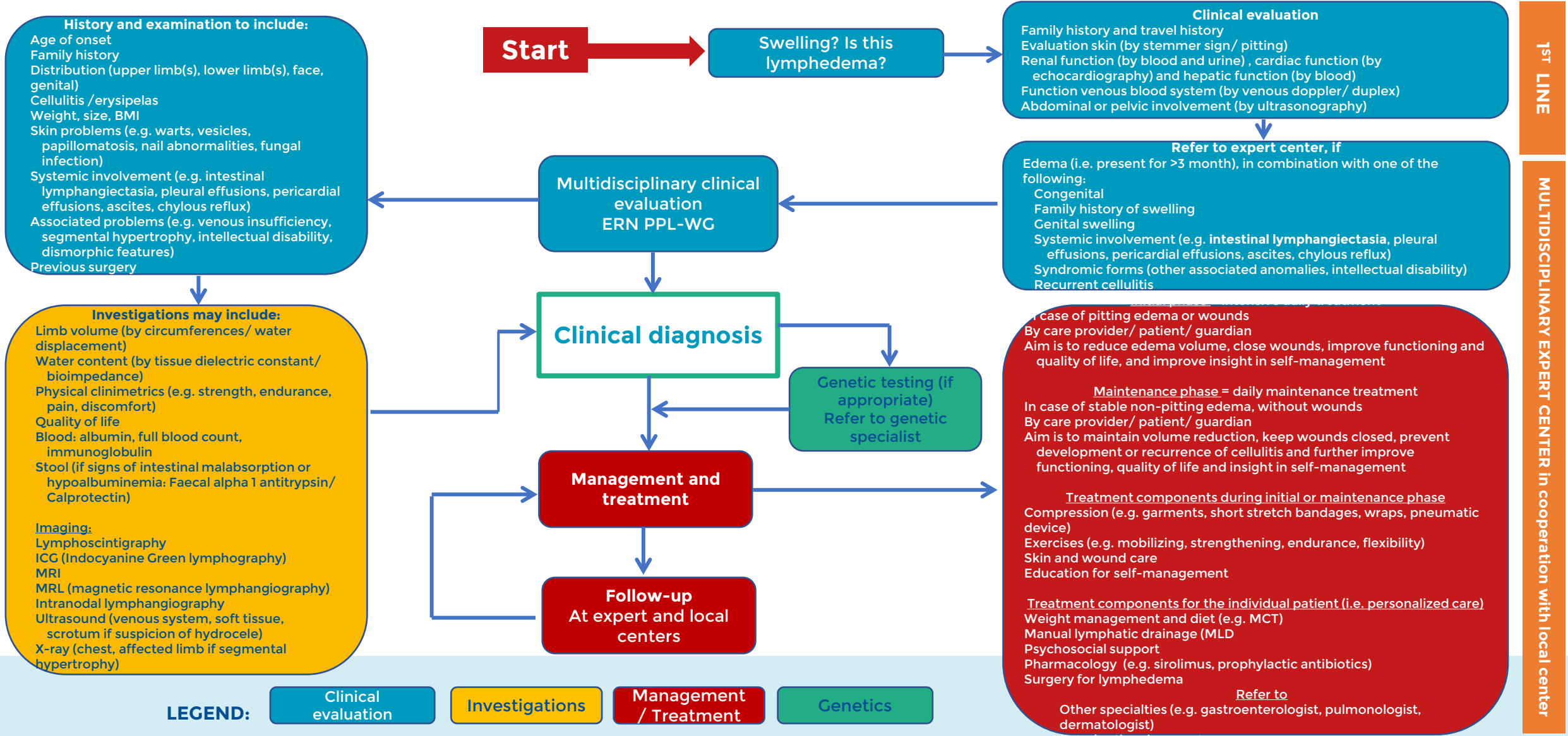
[#VASCERNdays2021](https://twitter.com/VASCERNdays2021)



WP 2: patient pathway

- We made a second version and finalized it
- Publication about it in preparation

General Patient Pathway for Pediatric and Primary Lymphedema (v2 - 12/05/2021)





Pills of Knowledge and webinars

- Compression in PPL
- Cellulitis in PPL
- Lymphedema from a patients perspective
- An overview of PPL



Do and don'ts factsheets

- 13 factsheets and did translation into German / Dutch / spanish
- Revisited some factsheets to better understanding by patients with thanks to ePAG members



Research and training

- Inventarisation among patients (ePAG members)
- Database patients 2019 in PPL centers; basis for publication
- Publication patients pathway in preparation
- Exchange program 2022: 9 candidates



FAIR registry and CPMS

- All centres are contacted
- ECL / Netherland starts with patient entry
- Virtual F2F and monthly patient discussions (n=25)
- We miss our English colleagues



New centers

- Danmark (become full member in 2022 ?)
- Spain / Hungary / Austria / Polen as affiliated centres



Plans 2022

- Run the exchange program
- Finalize publications and difine new research project
- Make FAIR registry operative and discuss many patients
- New webinars / do and don'ts



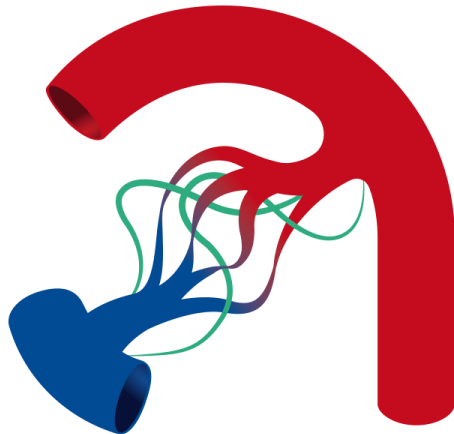
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



Vascern VASCA-WG

Miikka Vikkula VASCA WG Chair


[#VASCERNdays2021](#)

VASCERN - VASCA




BELGIUM

Chair




Pr. Laurence M. BOON
Pr. Miikka VIKKULA
Coordonator Center for Vascular Anomalies
Division of Plastic Surgery
Cliniques universitaires Saint-Luc
Human Molecular Genetics, de Duve Institute
Université catholique de Louvain
Brussels, Belgium

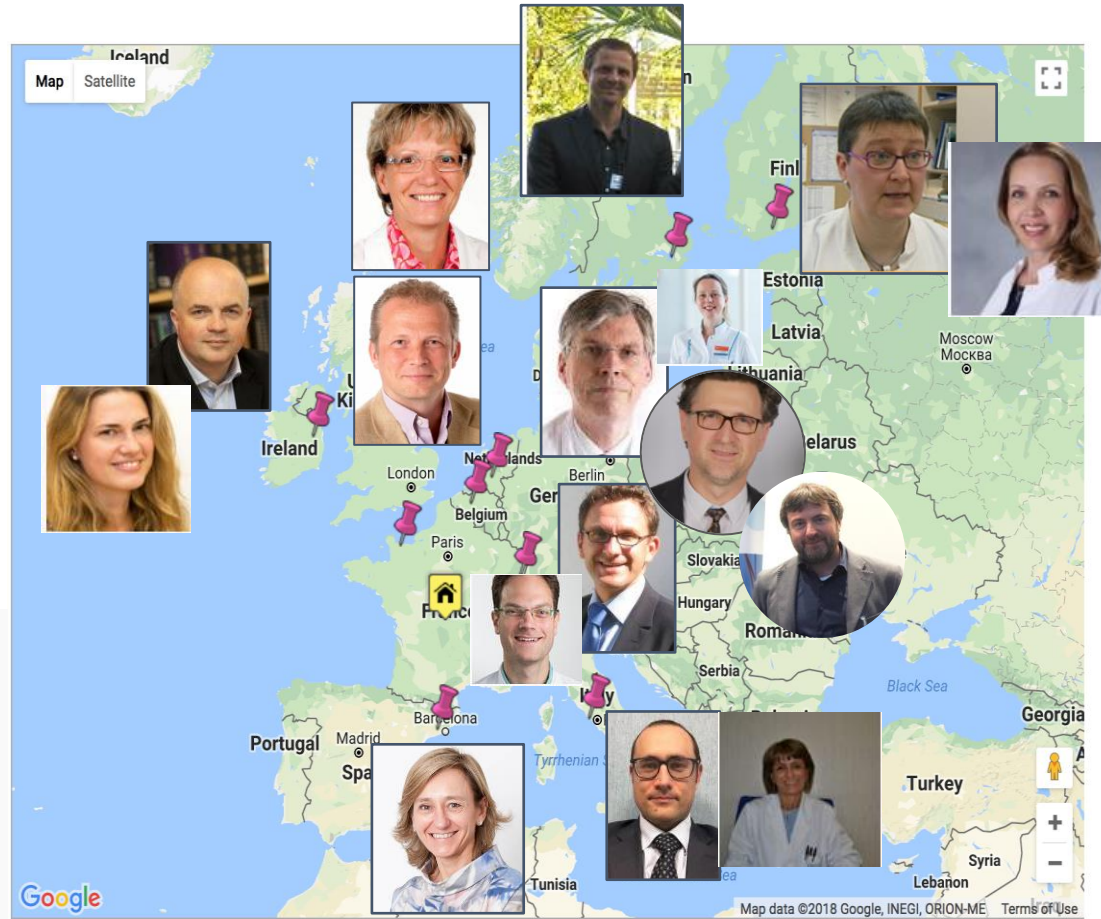


NETHERLANDS

Co-chair



Pr. Leo SCHULTZE KOOL
Expertcenter for Hemangioma and Vascular Anomalies (Hecovan)
Radboud university medical center
Nijmegen, Netherlands



- 7 HCP full member centers
- 1 Affiliated Partner center
- Applications from 5 new centers pending

VASCA-ePAG



<https://vascern.eu/>



1) « Patient pathways » (n=4)



- Presented at ISSVA 2020
- CM and VM at EADV 2021 (last Saturday) Lali and Anne D
- Each will be presented one at a time at VASCERN-VASCA seminars
- Manuscripts for EJMG
- IH almost finalised:

The VASCERN-VASCA Working Group Diagnostic and Management Pathways for Severe and/or Rare Infantile Hemangiomas

- LM and VM circulating for comments within VASCA
- CM drafting



2) Other VASCA publications

- Frontiers in Pediatrics, in press
- Prof L Boon as guest editor

frontiers
in Pediatrics

ORIGINAL RESEARCH
published: xx xx 2021
doi: 10.3389/fped.2021.697960

OPEN ACCESS

Edited by:
Udo Rolle,
University Hospital Frankfurt, Germany

Reviewed by:
Susanne Wiegand,
Leipzig University, Germany
Tobias Fall-Bock,
Helios Clinic Duisburg, Germany

***Correspondence:**
Friedrich G. Kapp
friedrich.kapp@uniklinik-freiburg.de

[†]These authors have contributed equally to this work

***CORRESPONDENCE:**
Friedrich G. Kapp
friedrich.kapp@uniklinik-freiburg.de

SPECIALTY SECTION:
This article was submitted to Children and Health, a section of the journal Frontiers in Pediatrics

RECEIVED: 20 April 2021
ACCEPTED: 26 August 2021
PUBLISHED: xx xx 2021

CITATION:
Holm A, te Loo M, Schultze Kool I, Salminen P, Passini VPC, Bassega Torres E, Duignan S, Dvorakova V, Irvine AD, Boon LM, Vikkula M, Ghaffarpour N, Niemyer CM, Rössler J and Kapp FG (2021) Efficacy of Sirolimus in Patients Requiring Tracheostomy for Life-Threatening Lymphatic Malformation of the Head and Neck: A Report From the European Reference Network. *Front. Pediatr.* 9:697960. doi: 10.3389/fped.2021.697960

Efficiency of Sirolimus in Patients Requiring Tracheostomy for Life-Threatening Lymphatic Malformation of the Head and Neck: A Report From the European Reference Network

Annegret Holm^{1,2}, Maroeska te Loo^{1,3}, Leo Schultze Kool^{2,3}, Päivi Salminen^{2,4}, Veronica Paola Celis Passini⁵, Eulalia Bassega Torres⁶, Sophie Duignan^{2,4,7,8}, Veronika Dvorakova^{9,10}, Alan D. Irvine^{2,4,7,8}, Laurence M. Boon¹¹, Mikka Vikkula^{2,8,10,12}, Nader Ghaffarpour¹¹, Charlotte M. Niemyer^{1,3}, Jochen Rössler^{1,2,13} and Friedrich G. Kapp^{1,2,14}

¹ Division of Pediatric Hematology and Oncology, Department of Pediatrics and Adolescent Medicine, Medical Center – University of Freiburg, Faculty of Medicine, University of Freiburg, Freiburg, Germany, ² VASCERN VASCA European Reference Centre, ³ Radboud University Medical Centre, Nijmegen, Netherlands, ⁴ Helsinki University Hospital, Helsinki, Finland, ⁵ Sant Joan de Déu Hospital, Barcelona, Spain, ⁶ Paediatric Dermatology, Our Lady's Children's Hospital Crumlin, Dublin, Ireland, ⁷ National Children's Research Centre, ⁸ Clinical Medicine, Trinity College Dublin, ⁹ Center for Vascular Anomalies, Division of Plastic Surgery, Saint-Luc University Hospital, Brussels, Belgium, ¹⁰ Human Molecular Genetics, de Duve Institute, University of Louvain, Brussels, Belgium, ¹¹ Department of Pediatric Surgery, Karolinska University Hospital, Stockholm, Sweden, ¹² Division of Pediatric Hematology and Oncology, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Bern, Switzerland

Extensive lymphatic malformations (LMs) of the head and neck region may require tracheostomy to secure the airway. Treatment of these life-threatening LMs is usually multimodal and includes sclerotherapy and surgery, among others. Recently, systemic therapy with sirolimus has been introduced as an effective treatment for venous and lymphatic malformations; its efficacy and safety profile in patients with extensive LM requiring tracheostomy are, however, as yet not fully known. We performed a retrospective, multicenter review and identified 13 patients with an extensive LM of the head and neck region, who previously underwent placement of tracheostomy and subsequently received sirolimus treatment with the aim to improve the local respiratory situation and remove the tracheostomy. Under sirolimus therapy, tracheostomy could be reversed in 8/13 (62%) patients, a further 2/13 (15%) patients improved markedly, and removal of the tracheostomy was planned at the time of writing, while 3/13 (23%) patients showed insufficient or absent response to sirolimus, rendering tracheostomy reversal not feasible. The median duration of sirolimus treatment until removal of tracheostomy was 18 months (range, 8 months to 5.6 years). Adverse events of sirolimus therapy were common [10/13 (77%) patients], yet the majority of these were mild [9/10 (90%) patients] and only one severe adverse event was recorded, with ulceration and necrosis at a catheter insertion site. In conclusion, sirolimus can be considered an effective and safe salvage treatment in patients with extensive LM even after placement of a



2) Other VASCA publications

- ePAG
- VASCA Magazine





3) EJP-RD workshop

- Willemijn Klein

📅 9 September 2021 – 09:00 📍 Online (due to the Covid-19 situation)

Organiser – Dr Willemijn M. Klein
9th and 10th Sept 2021

Will now be conducted online (due to the Covid-19 situation)

The poster is red with white and yellow text. It features a grid of white dots in the upper right and an illustration of a person in a blue shirt pointing at a whiteboard in the lower right. The text on the poster includes:

- WORKSHOP**
- ADVANCES IN DIAGNOSTIC AND THERAPEUTIC METHODS FOR LYMPH FLOW ABNORMALITIES**
- Willemijn M. Klein
Radboud University Medical Center Nijmegen
- 8-10 SEPTEMBER 2021**
Auditorium, Radboud University Medical Center, Nijmegen, the Netherlands
- ERN WORKSHOPS
THE EJP RD "ERN RESEARCH TRAINING WORKSHOPS CALL"

Logos for EJP RD, RARE DISEASES, and the European Union are at the bottom.



4) Guidelines for subWG work

Set-up of VASCERN-VASCA subWGs

- Involving more people than the leads only
- Thus, need of general working guidelines for the “new” participants
- Radiology: MR Intranodal lymphangiography (+ subsequent glue-procedures of complex lymphatic anomalies)
- Genetics; divided into 3: clinical genetics, lab germline; lab somatic
- Surgery
- Guidelines (with ESVS)
- Anesthesia
- Orphanet nomenclature
- Registry
- Clinical trials (divided on the basis of project)
-

5) CPMS



Leo as person in charge

Once a month on a Wednesday, separate from VASCA Webex

Several cases discussed



6) Other

- **Registry**
- **S4R**
- **Need for a Neuro WG within VASCERN**
- **PHTS foundation**
- **POKs by ePAG**

Thank you !




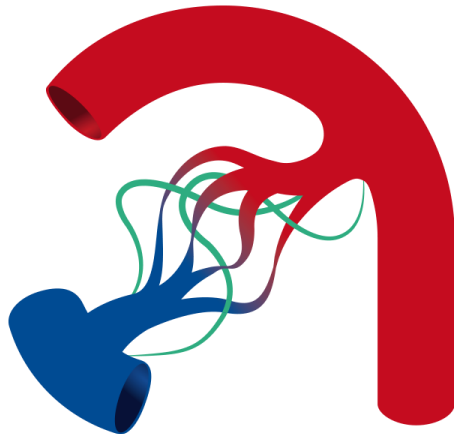
Nosologie Naturelle, Alibert 1817



**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



 **#VASCERNdays2021**

11.45-12.00

European Patient Advocacy Group (ePAG) views, projects & new developments, including Q&A

Juergen Grunert (Chair)
Caroline van den Bosch
Claudia Crocione
Pernille Henriksen
Elena de Moya Rubio (Co-Chairs)

<https://vascern.eu/expertise/epag/>

ePAG in the Rare Disease Working Groups



MSA
Medium Sized
Arteries

Co-Chair

Juergen
Grunert

*Deputy
Co-Chair*

Charissa
Frank



HHT
Hereditary
Haemorrhagic
Telangiectasia

Claudia
Crocione

Christina
Grabowski



HTAD
Heritable Theracic
Aortic Disease

Elena de
Moya
Rubio

Francoise
Steinbach



PPL
Pediatric and
Primary
Lymphedema

Pernille
Henriksen

Manuela
Lourenco
Marques



VASCA
Vascular Anomalies

Caroline
van den
Bosch

Maria
Barea



PPL ePAG and community members



**Pernille
Henriksen**

Dansk Lymfødem Forening (DALYFO)
Co-chair



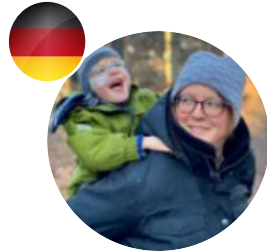
**Manuela
Lourenço Marques**

Associação Nacional de Doentes
Linfáticos (andLINFA)
Deputy co-chair



**Eline
Hoogstra**

Nederlands Netwerk voor
Lymfoedeem & Lipoedeem (NLNet)



**Carina
Mainka**

KIF11 Kids e.V



**Elena
Matta**

Lymphido Onlus



- Association Vivre Mieux le Lymphoedème (AVML)
- Lymphoedème Family



- Lega Italiana Lotta al Linfedema Aps (LILL)



- Lymph-what-oedema (LWO)
- Lymphoedema Support Network (LSN)



- Lymphoedema Ireland



- Svenska Ödemförbundet - SÖF



- Norsk lymfødem- og lipødemforbund (NLLF)



- Suomen lymfayhdistys



- Krabbameinsfélagið (KMF)



HTAD ePAG and community members



**Elena C.
DE MOYA RUBIO**
Marfan Hilfe (Deutschland) e.V.,
Co-chair



**Françoise
STEINBACH**
Association Marfans
Deputy co-chair



**Diana
MAAS**
De Contactgroep Marfan Nederland



**Romain
ALDERWEIRELDT**
Association Belge du
Syndrome de Marfan ABSM



**Carmen
QUIRÓS PAZ**
Asociación Española
Síndrome de Marfan (SIMA)



**Margit
ASCHENBRENNER**
Marfan Initiative



HTAD ePAG and community members



- Bindweefsel.be



- Slovak Marfan Association



- Landsforeningen for Marfan Syndrom



- Svenska Marfanföreningen



- Czech Association Of Marfan Syndrome



- Marfan Polska (Polish Marfan Association)



- Suomen Marfan - Marfan Syndrome Finland



- Den I Asbl Syndrome De Marfan



- Associazione J Peter Onlus
- Associazione Italiana Per La Lotta Alla Sindrome Di Marfan E Patologie Correlate
- Associazione Vittorio per la Sindrome di Marfan e malattie correlate
- MAGICA ONLUS



- Marfan Syndrome Support Group - Ireland



- Aorta Dissektion Föreningen Skandinavien



- Marfanforeningen - Norwegian Marfan Association



- Marfan Europe Network



- Marfan Stiftung Schweiz



VASCA ePAG and community members



Caroline van den Bosch

HEVAS, patient association for hemangioma and vascular anomalies, the Netherlands

Co-chair



Maria Barea

VASCAPA, VASCular Anomaly Patient Association, Belgium



Petra Borgards

Federal Association of Congenital Vascular Malformations e.V., Germany



Aaike van Oord

LGD Alliance Europe, the Netherlands



Lex van der Heijden

CMTC-OVM, Cutis Marmorata Telangiectatica Congenita and other VM, the Netherlands



Silvie Slívová

AVMinority, z.s., Czech Republic



VASCA

ePAG and community members (2)



- Interessengemeinschaft Sturge-Weber-Syndrom e.V.
- Naevus-Netzwerk
- Von Hippel-Lindau Verein
- Proteus-Syndrom e.V.
- LGDA Germany



- LGDA Belgium



- Nv WSWs (Portwine stain - Sturge Weber)



- Birthmark Support Group UK
- Segmental overgrowth conditions, especially Proteus and Klippel Trenaunay
- Sturge-Weber UK
- LGDA UK



- Finlands Klippel-Trenaunay Society
- Finnish Association for Ultra Rare Diseases
- HARSO umbrella organisation for patient associations supporting people with rare diseases and debilitating conditions



VASCA

ePAG and community members (3)



- Sturge-Weber Sverige
- Svenska KTS Nätverket
- Sturge Weber (SWW)



- Asociacion Española síndrome de Sturge-Weber (AESSW)
- AMCME (asociacion macrocefalia malformacion capilar españa) Macrocephaly-Capillary malformation and Cloves
- Asociacion española de cavernomas
- LGDA Spain



- L'Association Sturge-Weber
- Association Lymphangiomes
- Association Ollier Maffucci Europe
- Association Syndrome Cloves (FB Group)



- Cloves Poland (PB Group)



- ILA Associazione Italiana Angiodisplasie ed Emangiomi Infantili
- Girandola Onlus
- Macrodatt
- Fondazione Alessandra Bisceglia



HHT ePAG Team

ePAG and community members



Claudia Crocione

Co-chair

HHT Europe – Federation



Christina Grabowski

Deputy Co-chair

Morbus Osler Selfhelpgroup



Dara Woods

HHT Ireland



Didier Erasme

AMRO France HHT



Ria Blom

Harteraad



Torben Hellegaard

HHT Denmark



Luisa Maria Botella

HHT Espana



Paolo Federici

Fondazione Onilde Carini HHT



Fernando Brocca

HHT Italia



Mildred Lundgren

HHT Sverige



Karen Topaz Druckman

HHT Swiss



Tone Sodermand

Osler NO



Caroline Coxall

HHT UK



MSA ePAG and community members



**Juergen
Grunert**

Deutsche Ehlers-Danlos Initiative e.V.
Co-chair



Charissa Frank

Bindsweefsel.be
Deputy co-chair



- AFSED
- UNSED



- Associazione ARCASSED



- Ehlers-Danlos Support Group UK
- Annabelle's Challenge



- Bundesverband EDS Selbsthilfe e.V,



- EDS Riksförbund



- Norsk IForening for Ehlers-Danlos Syndrom



- ANSEDH



- VED Vereniging van ED Patienten (VED)



Representation across groups

- **Task force groups – EURORDIS**
 - **Guidelines**
 - Elena de Moya Rubio (HTAD)
 - **Steering committee**
 - Juergen Grunert (MSA) and Claudia Crocione, deputy (HHT)
 - **Education and Training**
 - Juergen Grunert (MSA)
 - **Research and Registries**
 - Maria Barea (VASCA) and Christina Grawobski (HHT)
 - **AMEQUIS**
 - Caroline van den Bosch (VASCA)
 - **Legal and ethical issues**
 - Romain Alderweireldt (HTAD)
- **Transversal working groups – VASCERN**
 - **Communication**
 - Carmen Quirós Paz (HTAD), Pernille Henriksen & Manuela Lourenço Marques (PPL)
 - **Pregnancy**
 - Petra Borgards (VASCA)
 - **Ethics**
 - Romain Alderweireldt (HTAD)
 - **Registries**
 - Maria Barea (VASCA) Christina Grawobski (HHT)



ePAG advocates and the community



- Expanding our footprint in Europe
- 19 countries
 - Still missing representatives in the Eastern European countries
 - Lack of PO
 - English skills



ePAG involvement in VASCERN

- Building a larger network within the patient community for the various diseases covered in VASCERN
- Integration into the National Healthcare Systems, connecting and networking between research, expert centres and patient organisations on a national level
- Development of patient pathways
- Development of Do's and Don'ts together
- Participation in the making of Pills of Knowledge (PoKs)
- Participation in the transversal working groups
- Regular online meetings of the RDWG, the Co-Chairs and the ePAG
- Translating the VASCERN APP; feeding with datasets



Attendance at webinar



VASCERN meeting, validation of patient PoK



ePAG Terms of reference

- Adapt to the new EURORDIS template elaborated in a Task Force
- Updated definitions and criteria to become a VASCERN advocate, adapting to the nature and way of working of our the ePAG



Through the year

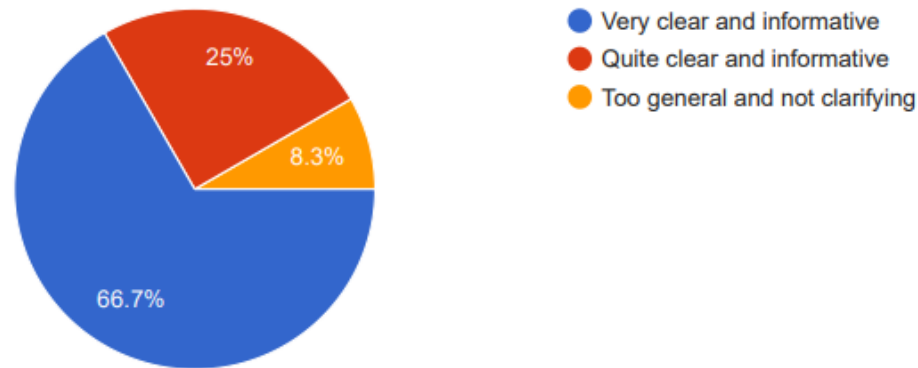
- Meet and Greet - January 2021
- Presenting VASCERN in annual meetings
- Participating in the national network developments
- Radio interview (ES)
- VASCA Magazine



Feedback from Meet & Greet

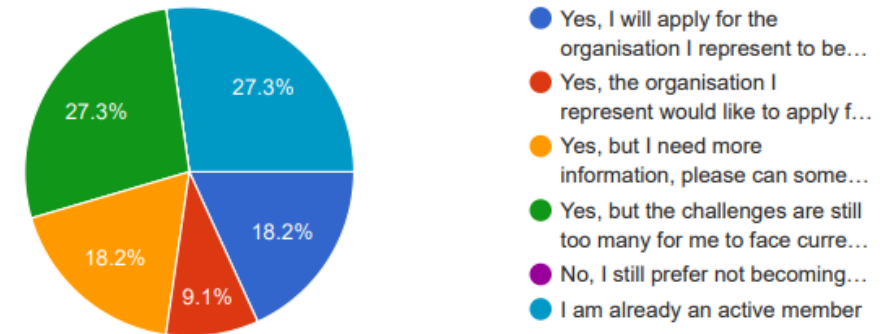
How did you find the presentation of the European Reference Network (ERN) and ePAG work?

12 responses



Did the meeting encourage you to be more involved?

11 responses



- 32 registered for the meeting
- New applications for ePAG Advocates and to join community
- Better understanding of the ERNs and roles of patient advocates
- Good platform for our further work and expansion



Insights, Outlook, Challenges for patients during COVID

- 2021
 - Growth
 - Contribution in ERN deliverables, excellent collaboration with clinicians
- Next year
 - Hope to see projects finished which have been long in the queue
- Challenges
 - Getting European Consensus Statements. (Potential reasons: national politics different, no evidence, etc)
 - Situation of Patients often unclear in the different countries
 - Conceptual not concrete, Consensus takes time
 - Translation: directly or agency (takes ages)




Any questions?

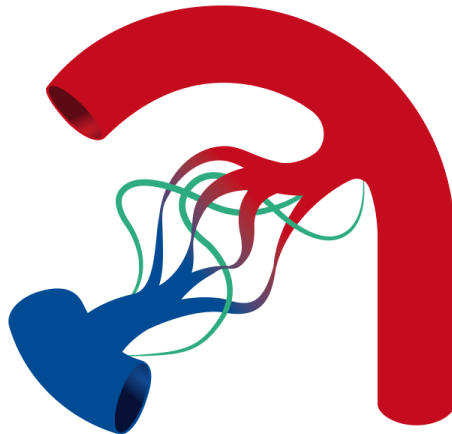
Thank you



European Reference Network

for rare or low prevalence complex diseases

 **Network**
Vascular Diseases
(VASCERN)








 [#VASCERNdays2021](https://twitter.com/VASCERNdays2021)

12.05-13.00 lunch break

13.00-17.00

RARE DISEASE WORKING GROUPS - PARALLEL SESSIONS

HHT-WG 	HTAD-WG 	MSA-WG 	PPL-WG 	VASCA-WG 
<i>13.00-15.15 Work Packages</i>	<i>13.00-15.15 Work Packages</i>	<i>13.00-15.15 Work Packages</i>	<i>13.00-15.15 Work Packages</i>	<i>13.00-15.15 Work Packages</i>
<i>15.15-15.30 coffee break</i>				
15.30-17.00: RDWGs CPMS clinical case discussions (clinicians only) & ePag meeting				

17.00-17.15

Council meeting (Chairs only) to select the 5 topics for the Day 2 Brainstorming plenary session



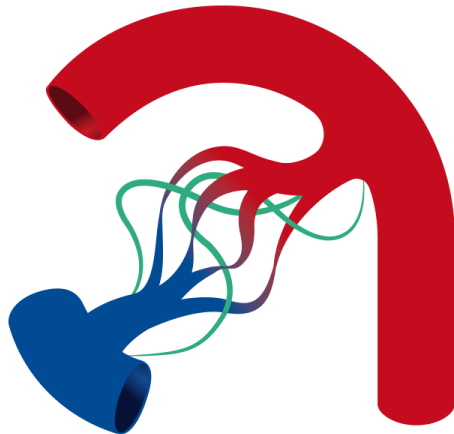
**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Vascular Diseases
(VASCERN)



**Thank you all for your
participation to this Board
session!**

Satisfaction survey

We would like to ask you please
to fill-in the **online satisfaction
survey** at the end of VASCERN
Days 2021