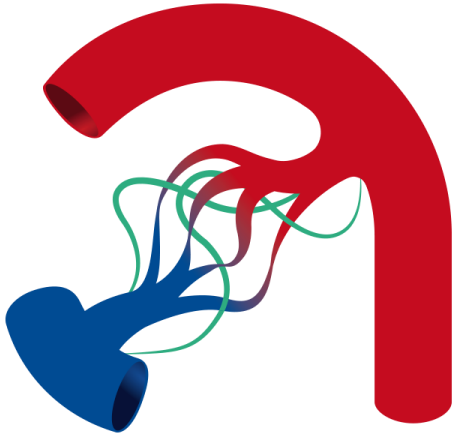
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

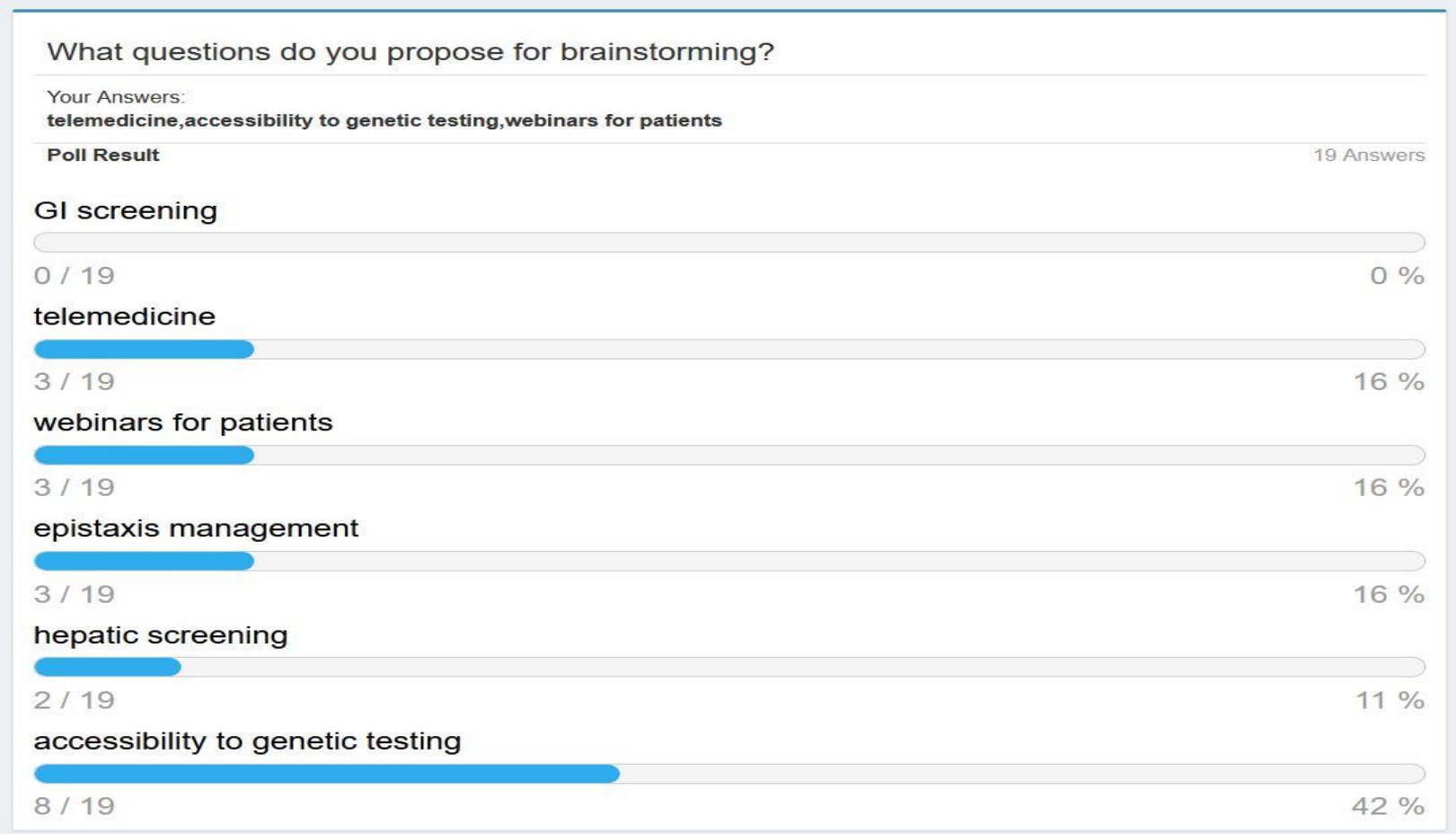


Brainstorming on 5 strategic topics selected by the RDWGs (12' by topic, each topic discussion moderated by the Chair of the RDWG)

#VASCERNdays2020

HHT WG SELECTED TOPIC FOR BRAINSTORMING SESSION

30 October 2020



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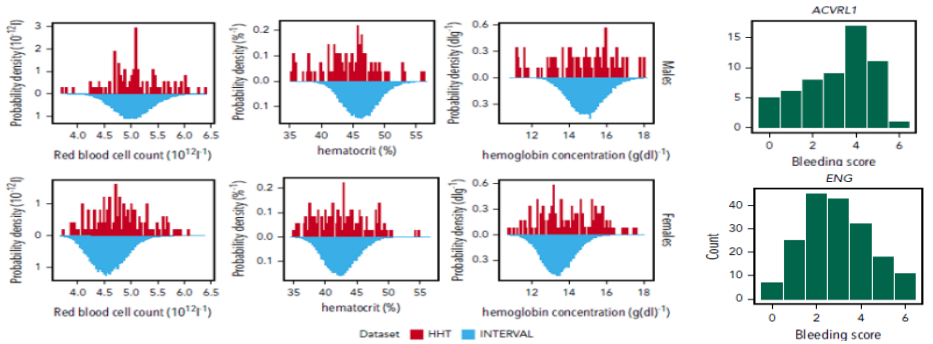
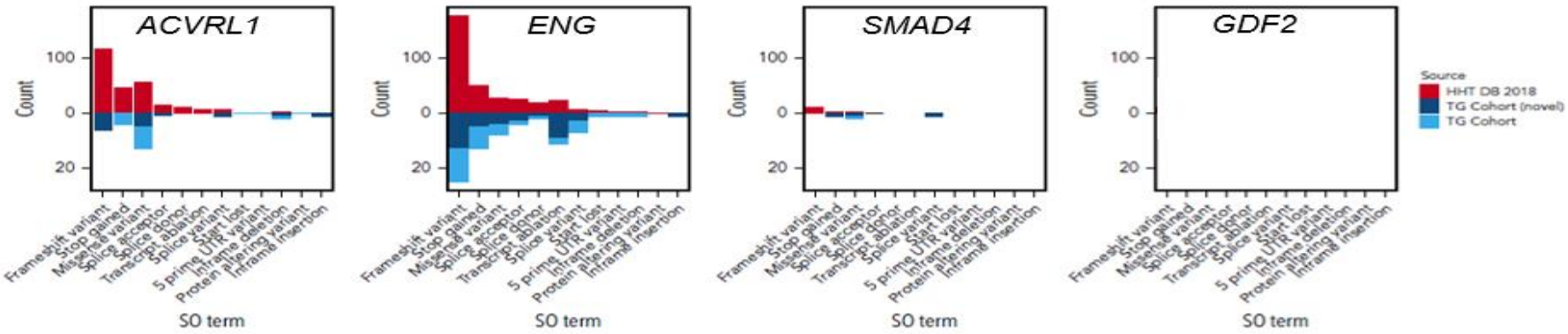


Shovlin *et al*, Blood **2020**



Patients with CONFIRMED Clinical Diagnosis

Sufficient clinical criteria

GENETIC TESTING QUESTIONS

1) Reproduction
2) Personalised care

Patients with SUSPECTED Clinical Diagnosis

Insufficient clinical criteria

GENETIC TESTING QUESTIONS

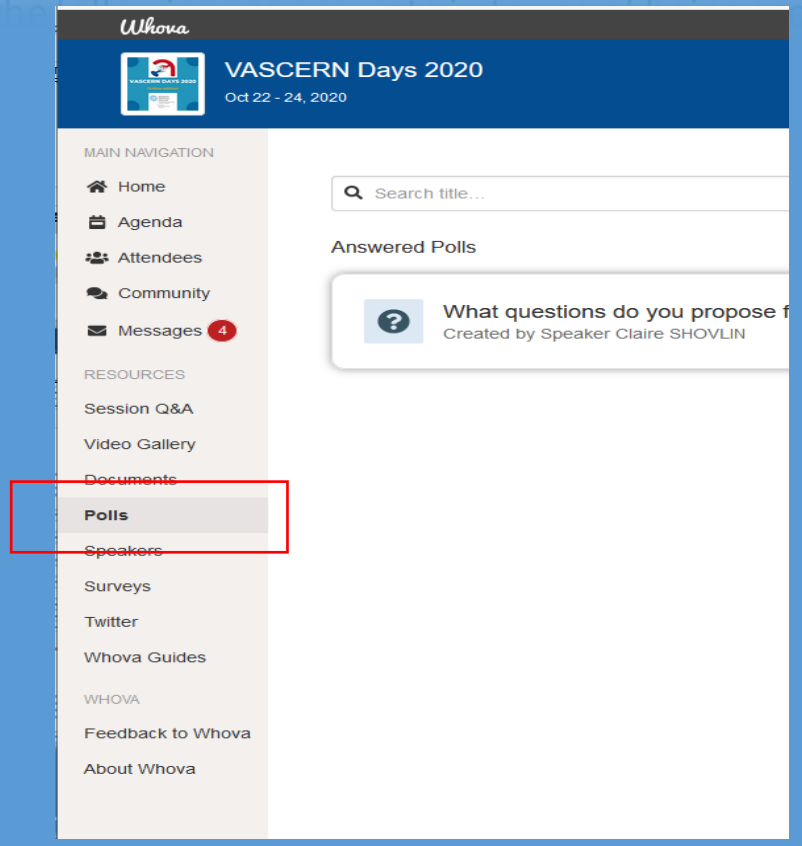
1) DO THEY HAVE HHT?
2) Reproduction
3) Personalised care

Thinking about genetic testing for your patients, which of the following are accurate in terms of being accessible to patients, at no financial cost to the patient?

- There are 4 "either/or" pairs, and 3 extra questions and .
- All answers are completely anonymous

0. My patients do not need genetic testing (do not answer questions 1-6)	STOP
1A. 100% of patients with a CONFIRMED clinical diagnosis have access to genetic te	
1B. NOT ALL patients with a CONFIRMED clinical diagnosis have access to genetic	
2A. 100% of patients with a SUSPECTED clinical diagnosis have access to genetic te	
2B. Not ALL patients with a SUSPECTED clinical diagnosis have access to genetic te	
3A. Available ONLY after pre-test genetic counseling	
3B. Available WITHOUT pre-test genetic counseling	
4A. There are NO negative consequences for mutation carriers in terms of accessibi	
4B. There may be SOME negative consequences for mutation carriers in terms of an	
5. I think patients have to pay for genetic tests	
6. I'm sorry, I do not know the answers to some of these questions	

You will find access to the poll on the website



Please complete now!

Your answers will be presented at the end of the session and we will see if this is an issue for VASCERN patients- or not!

Thinking about genetic testing for your patients, which of the following are accurate in terms of being accessible to patients, at no financial cost to the patient? There are 4 "either/or" pairs.

Total votes: 107

1A. 100% of patients with a CONFIRMED clinical diagnosis have access to genetic testing

23 Votes 21%

1B. NOT ALL patients with a CONFIRMED clinical diagnosis have access to genetic testing

7 Votes 6%

2A. 100% of patients with a SUSPECTED clinical diagnosis have access to genetic testing

18 Votes 16%

2B. Not ALL patients with a SUSPECTED clinical diagnosis have access to genetic testing

7 Votes 6%

Thinking about genetic testing for your patients, which of the following are accurate in terms of being accessible to patients, at no financial cost to the patient? There are 4 "either/or" pairs.

Total votes: 107

3A. Available ONLY after pre-test genetic counseling

13 Votes 12%

3B. Available WITHOUT pre-test genetic counseling

11 Votes 10%

4A. NO negative consequences for mutation carriers in terms of accessibility to healthcare/insurance

14 Votes 13%

4B. SOME neg consequences 4 mutation carriers in terms of any accessibility to healthcare/insurance

13 Votes 12%

Topic 1 (selected by HHT)
accessibility to genetic testing in the healthcare system

Ideas:

After Poll results are presented (107 votes), the results are highly dependent on country, who was replying (ePAG, HCP rep) and the RDWG anwering.

-Poll results show variation: this may be something that requires more formal survey (similar questions but with more info on who is responding, what country they are from)

- Poll to be done within VASCERN:

Including:

- Countries
- HCP or ePAGs/ patients
- HHT WG's Sophie, Claire, Claudia, Gennaro, Christina to start 1st draft of the survey to then circulate within VASCERN

Topic 2 (selected by HTAD)
How can we reach uniformity of treatment (medical/ surgical) in Europe

Ideas:

- Variability in the HTAD and VA teams : far from uniformity
- Criteria for diagnosis can vary from one country to another also
- Need to know what these differences are exactly

➢ First step is therefore to Map the differences!

- Collect simple items to show key differences in our centers (diagnosis, care, including genetic testing, interpretations of variants,…) via a survey
- Having databases / registry is also an important starting point

Topic 3 (selected by MSA):
limited participation to MSA from Europe : try to improve connexion with countries. How to make realise that there is a need for vEDS, FMD, SCAD centers.

Ideas:

- Missing big countries in the MSA WG (vEDS), no applications for 2021 new HCPs: Germany, Spain ? Should we have a strategy on how to approach this? (i.e: neither represented for EDS in ReCONNECT)

- FMD/SCAD: additional conditions in the MSA WG; needs expert represented in the WG. Difficulty is that these centers are already working together, and they might not see the point of joining ERN. This is true for FMD. Not for SCAD.

- What is our strategy for encouraging centers in larger countries to apply? How to favour the creation of new centers in smaller countries that do not have them? How to include them in the ERN?

- Problem of national networks and new diseases:
    - Expand the disease scope of HCP is possible: rule being discussed by the EU. Timeline unknown. Several cases in VASCERN.

Topic 4 (selected by PPL): Psychology in rare diseases of VASCERN: how to involve it more

Ideas:

- ICF method includes participation and activity, environmental and psychological aspects
- The psychological impact of rare diseases are sometimes underestimated
- QoL aspect:
  - PPL: have done research on this topic and have found a QoL questionnaire that is ICF based.
  - A holistic view should be applied, including the family members such as brothers/sisters
  - VA: questionnaire ready, developed with the patients.
  - France (COMPARE https://compare.aphp.fr/): patient driven registry. Including QoL, psychology aspects. It may be a good idea to have a registry completed by the patients.
  - NL eg the KLIK system
  - similar to SF-36
  - Develop a core set and then a specific disease set. ePAG could play important role.
  - Share4Rare
- https://www.cmtc.nl/en/quality-of-life-in-clinical-practice/
- https://qualitysafety.bmj.com/content/early/2020/08/23/bmjqs-2020-011219.abstract?ct

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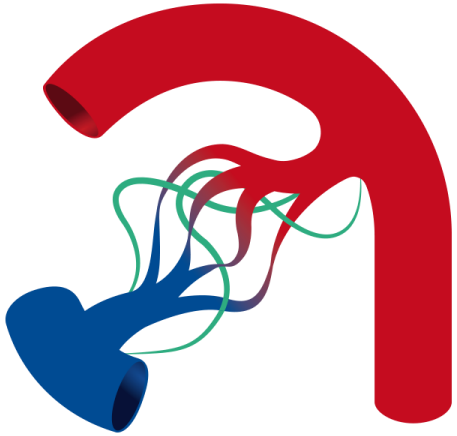

VASCERN National Networks

Some thoughts

23-10-2020

How can the Network be further extended?

- EU: 27 member states, 446M inhabitants (without the UK 26 and 400

To be decided by EU memberstates

1) 1 expert center per 10M inhabitants (40 x 5)

2) 1 expert center per country (1 representing 6 WG or 6 expert centers per country 1 for each WG (27 or 27 x 5)

3) All recognized centers are accepted and part of vascern. (>400)

National Network
How to set it up; pros and cons

Proposal:
1 (?) expert center per WG per country

- National network (to include other expert centers)
  - Criteria
    - recognition by the national government
    - If not available, criteria set by the WG

Benefits for the centers entering the network:

- National website for specific Rare disease(s)
- VASCERN APP
- Part of national subworking groups
- Possibility to
  - Review and comment VASCERN documents that are submitted for constation before publication
  - be partner in VASCERN research topics
- Registry
- Reimbursement (?)

Objections seen by centers only in the national network

- So far “competition” between University hospitals, (publications, grants etc)
- EGO
  - *Not part of the*
  - *We are the largest*
  - *No benefits*
  - *etc*

Proposals

- Start with (if already not in place) the formation of a national network in close collaboration with the national PO
- Start a website
- Develop guidelines for the national network ?
- National network decides who is the representing center in VASCERN (?) + rotative system of “chairing” in the ERN.

- For VASCERN:
- To National Network. Distributing of minutes, information about grants etc.

Topics 5

Ideas:

ePAG

- Patient Organisations can help in coordinating national networks in some countries. Interesting model to help HCPs to structure and organise these networks (as organisation/communication requires time, resources, etc.)
- Problem of mobility (jobs, travel, economy, COVID19 etc.) lead to a need for networks. So the networks provide a democracy of care.

- Need to know where are the centers and who are the leads in such networks, then the patient organisations can liaise with them
- Scientific societies are in many countries already and can be the forum for this networking

- In France, Netherlands: PO's were the ones that pushed for the national networks' creation. They have the power to lobby the government.
- Germany: are currently defining the expert centers criteria for certification first. Then once certified they can become a part of the network.

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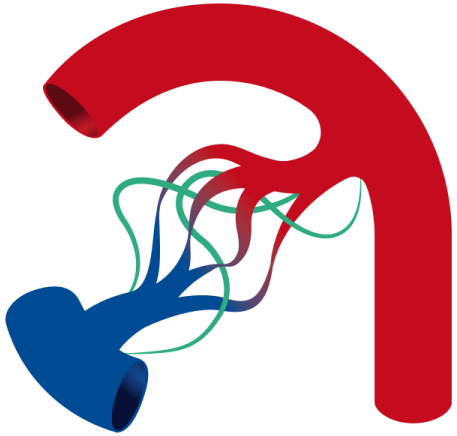

VASCERN CPMS

Some thoughts

23-10-2020

Purpose

- Offer a platform which allows secure exchange of patient data
- Offer a platform for consultations

User experiences

1. Steep Learning curve, which makes it user unfriendly for incidental consultations
2. CPMS mainly used for discussions within the ERN
3. Log in on EU platform needed
4. Only part of the database is used. Mostly uploading of pdf or powerpoint slide
5. Reminder emails disturbing
6. Case discussions with large group of attendees difficult
7. Time consuming

What is needed if the CPMS is used as the consultation platform for centers asking advice from the ERN?

- Logistic set up with an office which takes care of some of the administrative tasks. (contact with the advice asking center, informed consent, proper uploading etc)
- Within each ERN a team responsible for a timely respons to consultation requests.
- Legal issues.
- A form of consultation fee for the time reviewers spent on the case (in case of consultations from outside the ERN
  - Reimbursement from the Insurance companies?
  - EU level?

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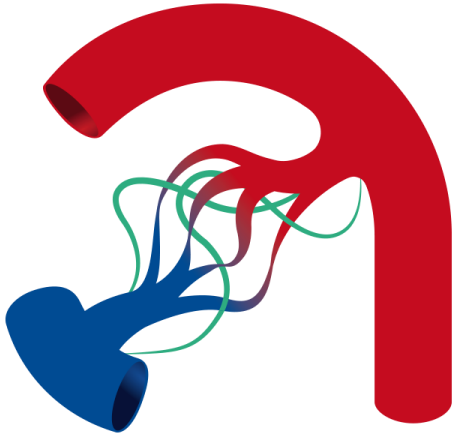

BREXIT

UK HCPs

- 4 HCPs in VASCEN
- What VASCERN can/ should do to help?

interERN level: actions taken including UK Parliament talk

Letter of support to be done.

National mouvement of the ERN HCPs.