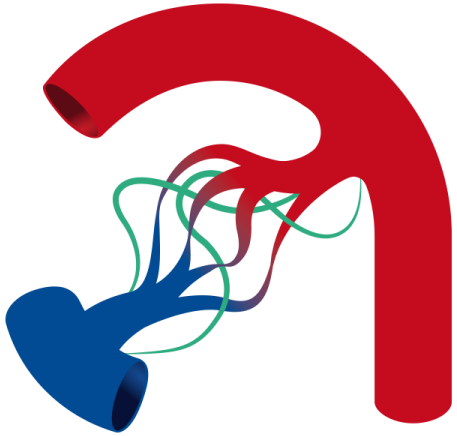
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



Vascular Ehlers-Danlos syndrome (vEDS)

Final Approved Patient Pathway by the MSA Working Group – 28/09/2018

VASCERN MSA working group Members:

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Disclaimer

- This document is an opinion statement reflecting strategies put forward by experts and patient representatives involved in the Medium Sized Arteries (MSA) Rare Disease Working Group of VASCERN.
- This pathway is issued on 28/09/2018 and will be further validated and adjusted as needed.
- Responsibility for care of individual patients remains with the treating physician.

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ERN General Pathway for Vascular Ehlers-Danlos syndrome (vEDS) (V1 - 28/09/2018)

Family history:
COL3A1 mutation in relative
Aortic dissection - <40
Arterial dissection - <40
Young onset death
Spontaneous Bowel rupture
3rd trimester uterine rupture

Suspect of vEDS

↓

Multidisciplinary clinical evaluation

↓

Genetic Testing

↓

Clinical and genetic Diagnosis of vEDS

↓

Management and Follow up

Personal history:
Aortic dissection
Arterial dissection
Spontaneous bowel rupture
Carotid-Cavernous Sinus Fistula (CCSF)

Cardiovascular imaging ECHO / MRA
Clinical examination
Genetic counselling

COL3A1 NGS panel testing

LEGEND: Clinical evaluation | Investigations | Treatment | Genes

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Patient Pathway for Vascular Ehlers-Danlos syndrome (vEDS) (V1 - 28/09/2018)

Red Flags

A combination of features below may raise suspicion

- Family history of vEDS with confirmed COL3A1 mutation
- Arterial dissection/rupture at young age (<40yrs)
- Spontaneous bowel Perforation (with no known risk factors)
- 3rd trimester uterine rupture (with no known risk factors)
- CCSF in absence of trauma
- Bruising in unusual sites with undefined trauma
- Characteristic facial Appearance or acrogeria
- Spontaneous pneumothorax
- Other features such as club foot. thin skin, keratoconus, gingival fragility
- Early onset varicose veins <30yrs

Multidisciplinary clinical evaluation

Suspect vEDS

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

Confirm vEDS with mutation in COL3A1

Management and follow-up

- Surveillance and Follow Up*
- Access to teams required include:
  1. Cardiology
  2. Vascular and endovascular surgery
  3. Bowel surgery
  4. Neurovascular surgery
  5. Thoracic surgery
  6. Obstetrics and fetal medicine
  7. Vascular imaging
  8. Psychosocial input

Proband/Relative NEW PATIENT

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

LEGEND: Clinical evaluation | Investigations | Treatment

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Patient Presents at:	Red Flags A combination of features below may raise suspicion	Suspect vEDS	Multidisciplinary Clinical Evaluation at ERN designated HCP or Centre of expertise	Confirm vEDS with COL3A1 mutation	*Surveillance and Follow-up
GP Emergency services Surgical team Vascular team Cardio-thoracic Team Genetics team **For probands and relatives New Patient**	Family history of vEDS with confirmed COL3A1 mutation Arterial dissection/rupture at young age (<40 yrs) Spontaneous bowel Perforation (with no known risk factors) 3rd trimester uterine rupture (with no known risk factors) CCSF in absence of trauma Bruising in unusual sites with undefined trauma Characteristic facial appearance or acrogeria Spontaneous pneumothorax Early onset varicose veins <30yrs Other features such as club foot, thin skin, keratoconus, gingival fragility		Cardiac and arterial imaging Clinical examination and assessment Molecular Testing Family history		Routinely led by cardiology in liaison with multidisciplinary teams Access to teams required include: • Cardiology • Vascular and endovascular surgery • Bowel surgery • Neurovascular surgery • Thoracic surgery • Obstetrics and fetal medicine • Vascular imaging • Psychosocial input • Clinical genetics

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

ERN Databasing and Research

Diagnosis of vEDS

Treatment:
Discuss and trial Celiprolol

Cascade testing:
If appropriate

Psycho-social support:
If appropriate

Education and Training:

1. POK
2. Do and Don'ts factsheets
3. Uploading presentations / talks

Consent for registry (RaDiCo) → **Helps in documentation and further research / clinical trials**

Medic Alert and patient passports → **Electronic passport to develop**

Clinical trials → **To consider as ERN multinational study**

Use data from above for consensus and evidence based publications

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to provide accessible cross-border healthcare
to patients with rare vascular diseases

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

VASCERN currently consists of 30 highly specialised multidisciplinary Healthcare Providers (HCPs) from 11 EU Member States and of various European Patient Organisations and is coordinated in Paris, France.

Through our 5 Rare Disease Working Groups (RDWGs) as well as several thematic WGs and the ePAG – European Patient Advocacy Group, we aim to improve care, promote best practices and guidelines, reinforce research, empower patients, provide training for healthcare professionals and realise the full potential of European cooperation for specialised healthcare by exploiting the latest innovations in medical science and health technologies.

More information available at: https://vascern.eu

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