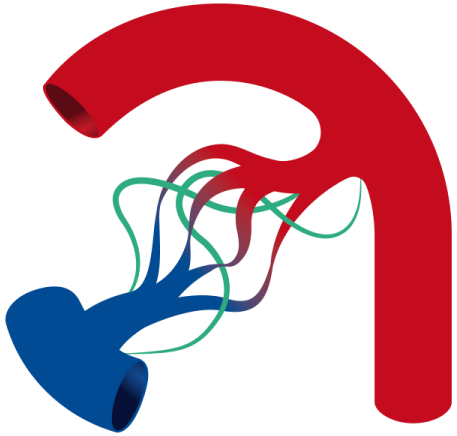
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

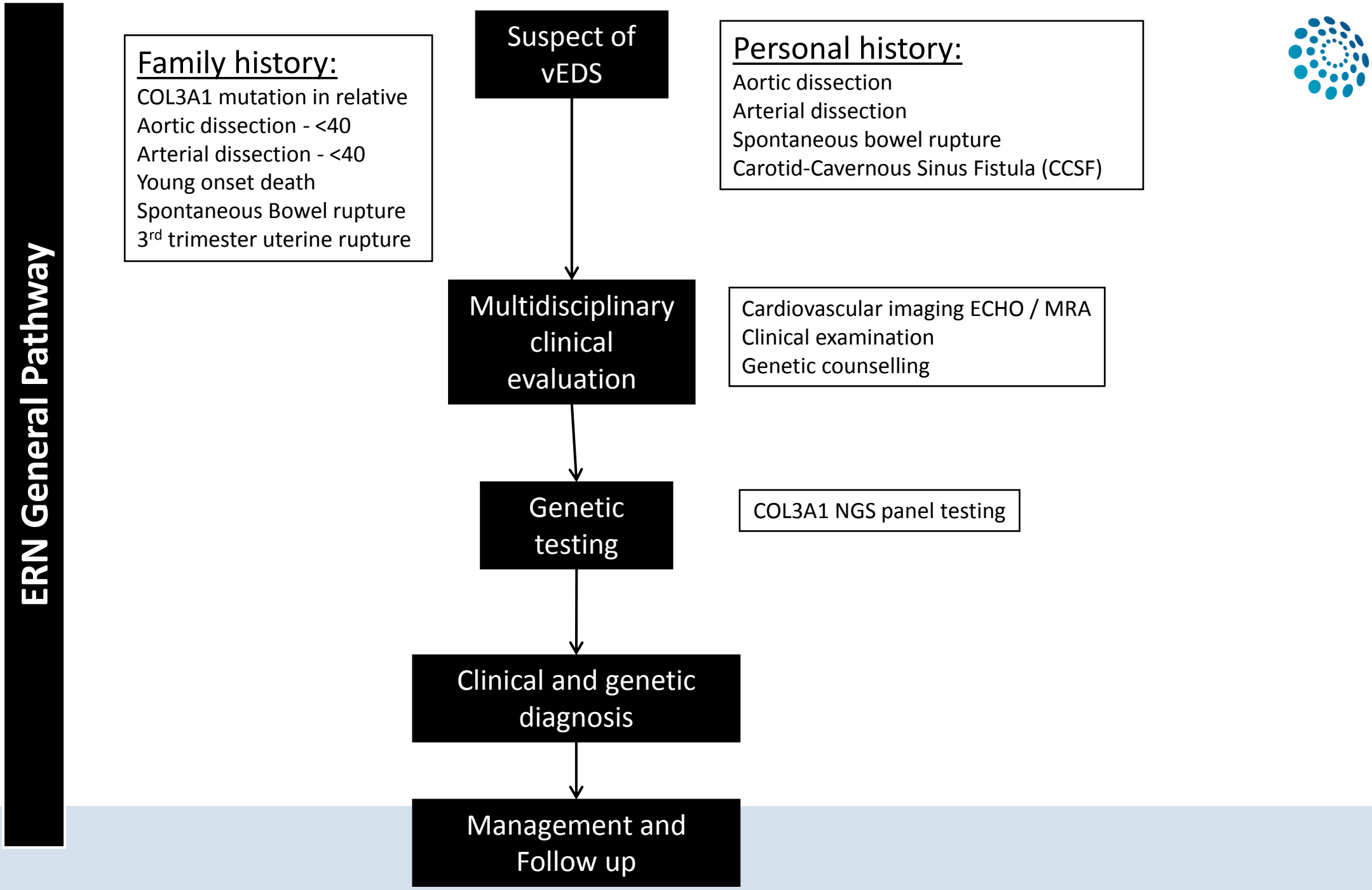


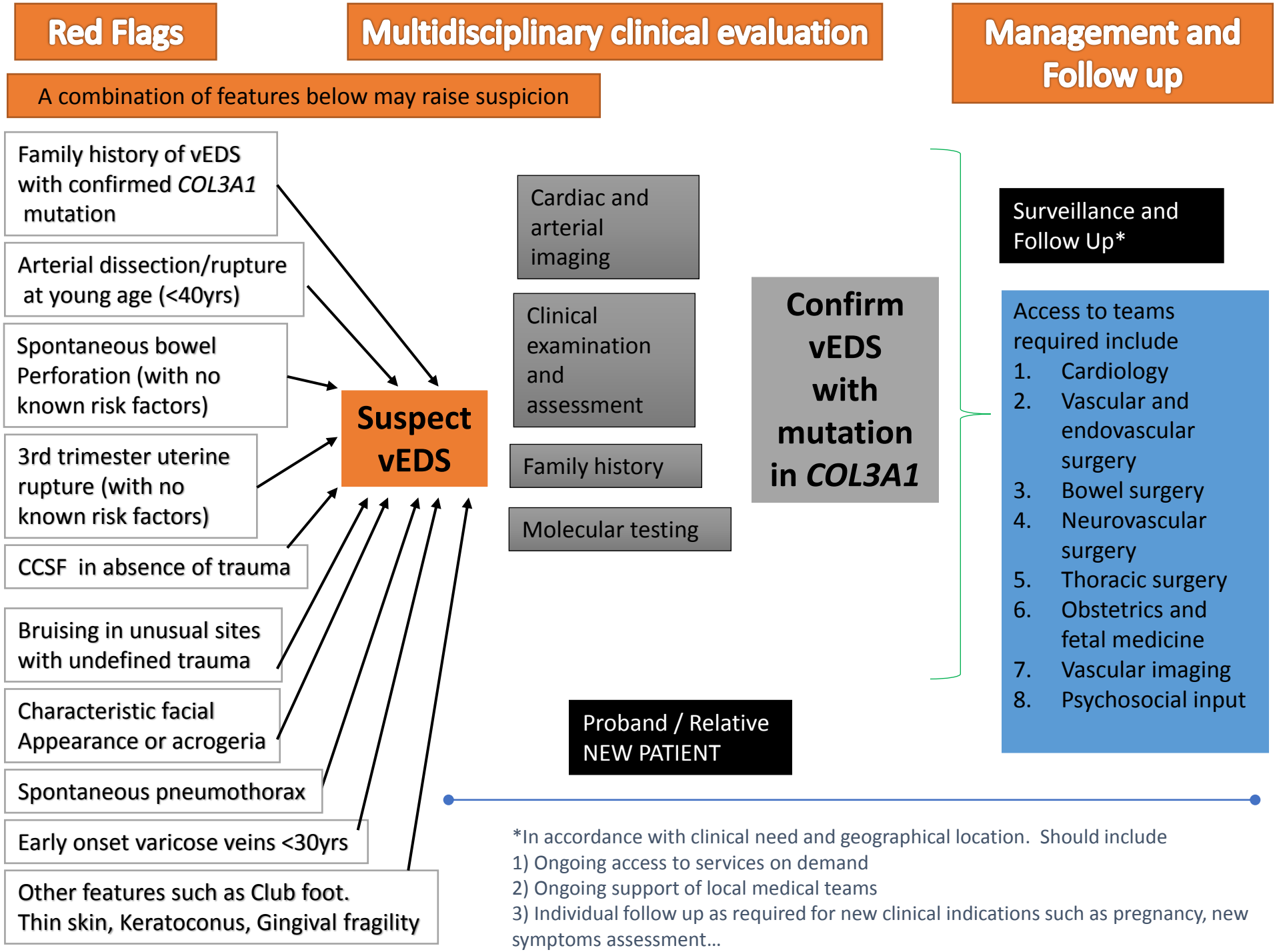
Vascular EDS

Final approved patient pathway
28/09/2018

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.

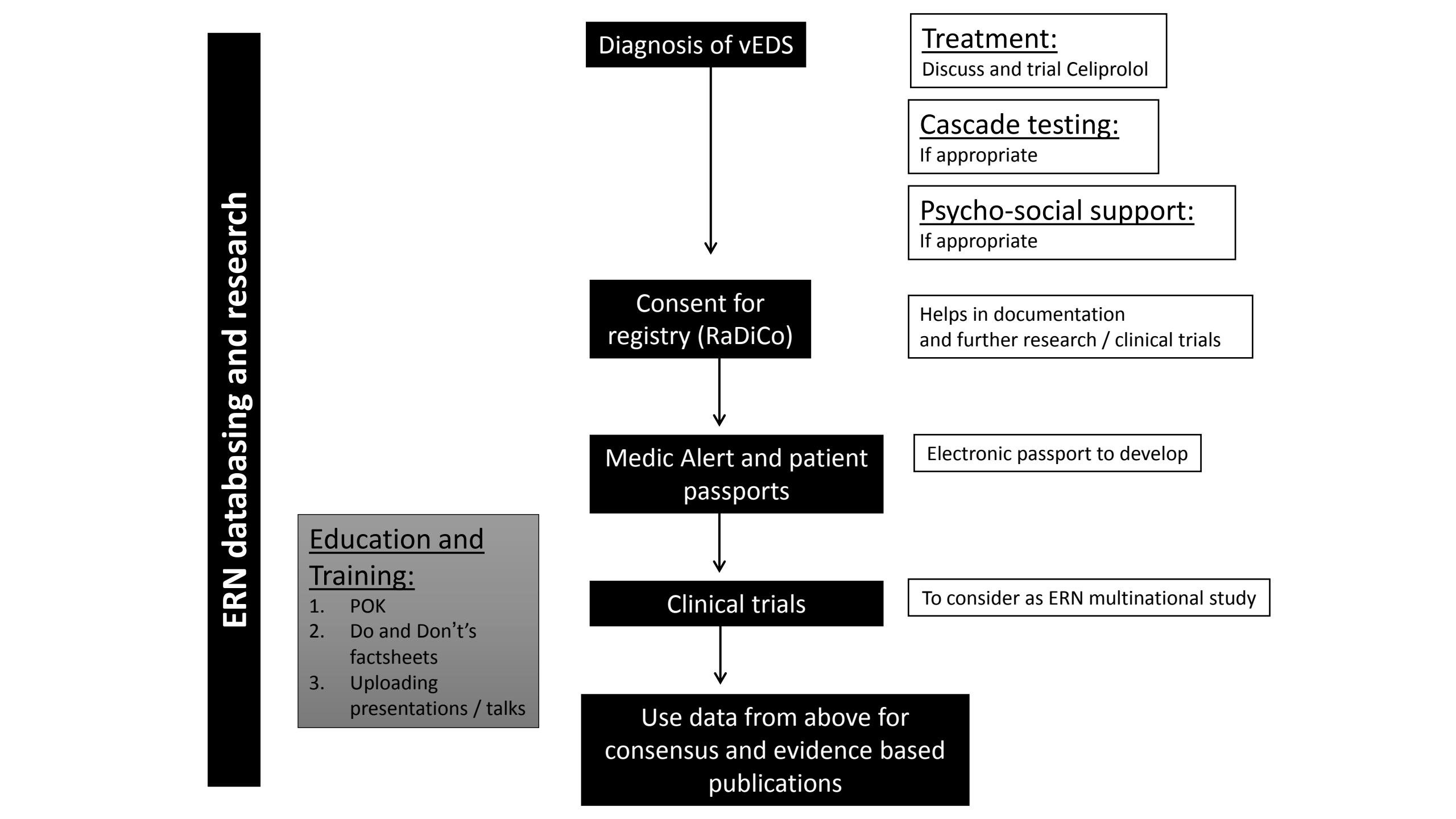

ERN General Pathway
Family history:
COL3A1 mutation in relative
Aortic dissection - <40
Arterial dissection - <40
Young onset death
Spontaneous Bowel rupture
3rd trimester uterine rupture
Suspect of vEDS
Personal history:
Aortic dissection
Arterial dissection
Spontaneous bowel rupture
Carotid-Cavernous Sinus Fistula (CCSF)
Multidisciplinary clinical evaluation
Cardiovascular imaging ECHO / MRA
Clinical examination
Genetic counselling
Genetic testing
COL3A1 NGS panel testing
Clinical and genetic diagnosis
Management and Follow up


Red Flags
Multidisciplinary clinical evaluation
Management and Follow up
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
Early onset varicose veins <30yrs
Other features such as Club foot. Thin skin, Keratoconus, Gingival fragility
Suspect vEDS
Cardiac and arterial imaging
Clinical examination and assessment
Family history
Molecular testing
Confirm vEDS with mutation in COL3A1
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…

Patient Presents @	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	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 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

For probands and relatives 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…


ERN databasing and research
Diagnosis of vEDS
Treatment:
Discuss and trial Celiprolol
Cascade testing:
If appropriate
Psycho-social support:
If appropriate
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
Education and Training:
1. POK
2. Do and Don't's factsheets
3. Uploading presentations / talks

About VASCERN

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

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

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

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

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