European Reference Network

VASCERN

Do's and Don'ts Factsheets

for Rare Vascular Disease Care in Frequent Situations

Vascular Ehlers-Danlos Syndrome

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Do's and Don'ts for for Vascular Ehlers-Danlos (vEDS) Syndrome

Abbreviations

CTA: Computed tomography angiography

EDS: Ehlers-Danlos syndrome

GPIIβ/IIIα: Glycoprotein II beta/III alpha

LOVD: Leiden Open Variation Database

MD: Multidisciplinary

MDT: Multidisciplinary Team

MRA: Magnetic Resonance Angiography

MSA-WG: Medium-Sized Arteries Working Group

NSAID: Non-steroidal anti-inflammatory drug

SVI: Superficial venous insufficiency

vEDS: Vascular Ehlers-Danlos syndrome

Introduction

Vascular Ehlers-Danlos Syndrome (OMIM #130050) is a rare disorder that results from heterozygosity for mutations in COL3A1 which encodes the pro-alpha1 chains of the type III procollagen homotrimer. The condition is dominantly inherited but asymptomatic parental mosaicism can result in sib recurrence to normal parents. Arterial fragility, bowel rupture, and pregnancy complications are the major clinical complications and may limit survival to a median 50 years.

Molecular diagnostic testing is therefore a key factor in identifying patients for appropriate management. More than 1500 COL3A1 genetic variants are registered in the EDS database of the LOVD system, a large majority of them being private (http://www.lovd.nl/). This genetic testing requires having access to a genetic laboratory with experience to test and interpret COL3A1 variants, since not all variants are pathogenic and since those leading to haploinsufficiency result in a better prognosis than those with missense variants at a glycine residue or splice-site variants.

Clinical evaluation is usually undertaken in expert centres with the involvement of a multidisciplinary team familiar with the clinical features of this condition in probands and their close relatives. This rare disease suffers from lack of recognition since the main clinical diagnostic criteria for vEDS are nonspecific (hematomas, skin transparency, arterial events) and it is only their association that is suggestive of the diagnosis.

Major and minor clinical diagnostic criteria of the 2017 International classification of EDS illustrate the variety of physical signs that may constitute the clinical phenotype, adding to the diversity of arterial and visceral events during the natural course of the disease.

Do's and Don'ts for Vascular Ehlers-Danlos Syndrome

Because of the number of affected tissues, clinical evaluation often involves: cardiologists, clinical geneticists, vascular and interventional radiologists, ophthalmologists, orthopedic surgeons, physiotherapists, gastrointestinal surgeons, and vascular, endovascular and cardiac surgeons. Regular MDT meetings with multiprofessional team involvement will also aid in coordinating care of patients. Appropriate transition of patients from pediatric to adult care is also essential in management of patients.

Genetic counselling, pre-pregnancy counselling and management during pregnancy by specialist fetal maternal teams will also be an essential part of the care of pregnant women with this disorder. Provision of appropriate counselling for family planning will also be necessary as pregnancy is associated with death in about 5% of women.

Treatment is predominantly medical, surveillance is not universally adopted, and treatment with antihypertensive agents may provide a measure of protection from arterial rupture.

Medical therapy with celiprolol has shown its efficacy in reducing cardiovascular events in a small group of patients. Other betablockers may also provide protection from cardiovascular events, but there is a lack of appropriate drug trials. The need for detailed studies of natural history and mechanistic evaluation of arterial rupture are clear and probably the only way to identify helpful interventions.

Factsheets

These factsheets are based on existing French factsheets which have been reviewed and revised by the VASCERN MSA-WG members.

The MSA-WG wishes to emphasize that these are recommendations made by consensus at expert level. We would recommend that these factsheets be used as a guide to implement locally agreed policies.

These factsheets are meant for patients as well as for caregivers. Implementing these recommendations should go hand in hand with strategies to educate patients about medical situations where specific care is required and about relevant symptoms & how to act when they occur.

Complications in vEDS

Major life limiting complications are due to the increased risk of arterial ruptures. In these cases, urgent repair by any possible means is required. Locations that are commonly symptomatic are iliac and femoral arteries, mesenteric and celiac vessels, renal arteries, aorta (any location), and peripheral arteries of the limbs.

Peripheral Arterial Dissection

Major life limiting complications are due to the increased risk of arterial rupture. In these cases, urgent repair by any possible means is required. Locations that are commonly symptomatic are the iliac and femoral arteries, mesenteric and celiac vessels, renal arteries, aorta (any location), and peripheral arteries of the limbs.

WHAT IS RECOMMENDED

- Call the national expert centre or referring local expert to seek advice due to the seriousness of the risk of dissection.
- Quickly implement all necessary actions to confirm the diagnosis of vascular Ehlers-Danlos syndrome.
- Prioritise non-invasive examinations (such as MRA or CTA) and medical/conservative treatment whenever possible.
- When an arteriography is necessary (rupture, perforation), particular precaution should be taken when the guidewire is being moved upwards, due to risk of dissection/arterial perforation, and enhanced monitoring of the femoral insertion point is necessary
- Arterial ruptures should be treated in preference by embolization.
- A protocol of permissive hypotension is recommended without compromise to organ function.
- Caution with use of inotropes is recommended.
- Caution with use of indwelling catheters is advised.

WHAT YOU SHOULD NOT DO

- Insertion of stents (except in the case of a life-threatening emergency) in most cases.
- Consider complex surgical repair in an emergency situation.
- Initiate treatment with anticoagulation and/or antiplatelet agents without formal confirmation of its indication and expert advice.

Abdominal Emergencies

Vascular Ehlers-Danlos syndrome predisposes young adults to a high risk of digestive tract events that can be life-threatening. Bowel rupture is uncommon in early childhood, has been described in late childhood, and remains a risk into adulthood. Bowel rupture ultimately affects about 25–30% of people, but rarely leads to death.

The main abdominal emergencies are (in order of severity):

- spontaneous arterial rupture in the peritoneal and/or retroperitoneal cavity;
- spontaneous digestive tract perforation, most often of the sigmoid colon, but also of the rest of the colon, the small intestine, and even the stomach;
- spontaneous rupture of an intra-abdominal organ (spleen, liver).

WHAT IS RECOMMENDED

- Perform an emergency CT angiography in the presence of an acute abdominal syndrome.
- In the event of colonic perforation, a wide colonic resection is the preferred method of treatment (Hartmann type for sigmoid colon).
- In cases of arterial rupture, percutaneous embolization should be preferred over open surgery.

- The use of stents should be limited to life-threatening emergencies, and embolization preferred whenever possible.
- In cases of spontaneous perforation of the digestive tract in a young adult or a child, a diagnosis of vascular Ehlers-Danlos syndrome should be automatically considered.
- In case of an arterial event, a protocol of permissive hypotension is recommended without compromise to organ function.
- Caution with use of inotropes is recommended.
- Caution with use of indwelling catheters is advised.

WHAT YOU SHOULD NOT DO

- Attempt a minimal repair of a spontaneous perforation of the digestive tract.
- Perform a post-operative follow-up colonoscopy in the event of a spontaneous perforation of the digestive tract.
- Perform open surgery as first-line treatment in cases with arterial rupture (except for life-threatening emergencies).
- Any invasive or semi-invasive examination, particularly when the expected diagnostic rewards are minimal.

Haemoperitoneum

Haemoperitoneum is the cardinal indicator of acute bleeding in the abdominal cavity and requires emergency care.

WHAT IS RECOMMENDED

- Perform an emergency abdominal angiography scan to look for an arterial rupture, a digestive tract perforation or an intra-abdominal organ rupture.
- In the event of an arterial rupture, selective embolization should be preferred over open surgery.
- A protocol of permissive hypotension without compromise to organ function.
- Caution with use of inotropic drug.
- Caution with use of indwelling catheters.

WHAT YOU SHOULD NOT DO

- Prescribe long-term anticoagulant and antiplatelet treatments without expert or multidisciplinary expert advice/discussion.
- Arterial puncture/access for monitoring or sampling.

Arterial Revascularisation

Arterial events (dissections, aneurysms) are the most frequent complications of vascular Ehlers-Danlos syndrome. Most often, the arterial events are treated medically and therefore do not require intervention, such as surgery.

WHAT IS RECOMMENDED

- Prioritise medical management and treatment for arterial events whenever possible.
- In the event of arterial rupture, emergency angiography with embolization should be preferred, ideally in a specialised centre with prior experience in treating patients with vascular EDS.
- A protocol of permissive hypotension without compromise to organ function.
- In the event of an emergency surgical procedure, simple procedures (ligatures) are to be preferred over complex vascular reconstruction
- Caution with use of inotropic agents and indwelling catheters.

WHAT YOU SHOULD NOT DO

- Treat an arterial rupture with complex surgical reconstruction. In the case of a salvage procedure, special precautions with vascular clamping and sutures are required.

Aortic Dissection

Taking into account the arterial fragility which is characteristic of the disease, the occurrence of a dissection and, rarely, the rupture of the aorta are possible complications. These arterial accidents occur more often at the abdominal aorta than at the thoracic aorta.

WHAT IS RECOMMENDED

- If an acute chest and/or abdominal pain syndrome is present, it is imperative to urgently check for the presence of a dissection or rupture of the aorta.
- Give preference to non-invasive examinations (such as MRA or CTA) and medical treatment whenever possible.
- For type A aortic dissection, surgical treatment is indicated as the primary treatment option.
- A protocol of permissive hypotension is recommended without compromise to organ function.
- Caution with use of inotropic agents and indwelling catheters.

WHAT YOU SHOULD NOT DO

- Use a stent graft as a first option in the presence of a dissection of the descending aorta, unless there is no other therapeutic alternative.

Acute Coronary Syndrome

There appears to be a slightly increased risk of acute coronary syndrome in vascular Ehlers-Danlos syndrome. Given the arterial fragility, dissection of a coronary artery may result in an acute coronary syndrome (non-atheromatous).

WHAT IS RECOMMENDED

- In the presence of an acute coronary syndrome in a patient with vascular Ehlers-Danlos syndrome, rule out an aortic dissection.
- Prioritise non-invasive examinations (such as coronary CT scan) and medical treatment whenever possible.
- When a coronary angiography is necessary, particular caution should be taken when moving the guidewire upwards (due to the risk of dissection/arterial perforation) and enhanced monitoring of the femoral insertion point is necessary.

WHAT YOU SHOULD NOT DO

- Use of fibrinolytic or anti-GPIIβ/IIIα agents as first-line treatment.
- Insertion of stents requiring dual or prolonged antiplatelet therapy without MDT expert advice.
- Radial coronary angiography (risk of dissection and ischemia of the hand).

Carotid-Cavernous Fistulas (sCCF)

Carotid-cavernous fistula is a possible complication of vascular Ehlers-Danlos syndrome. It affects around 10-15% of patients with the syndrome and consists of a rupture of the intracranial carotid artery into the main venous blood collection site in the brain. Its spontaneous occurrence is pathognomonic of vascular EDS. The appearance of a pulsating murmur may suggest the presence of a carotid-cavernous fistula. Swelling around the eye and ocular protrusion may also be present.

WHAT IS RECOMMENDED

- If a carotid-cavernous fistula is suspected, a dedicated MR angiography and/or CT scan is recommended.
- Management and care in a neurovascular unit. Therapeutic occlusion of the fistula is often necessary, despite the inherent risk of the procedure. This procedure is carried out in a specialised centre with the highest possible level of experience.
- Blood pressure in particular should be closely monitored. Maintain a blood pressure target <130/80 mmHg (permissive hypotension).
- Caution with use of indwelling catheters and sheaths.

WHAT YOU SHOULD NOT DO

- Delay imaging procedures which alone determine the diagnosis.
- Delay embolization since sCCF may compromise ocular function.

Pneumothorax

Pneumothorax is associated with vascular Ehlers-Danlos syndrome and is the most frequent respiratory manifestation of the disease. It can occur from adolescence onwards and sometimes precedes genetic diagnosis. Treatment typically, but not always, involves the insertion of a needle or a chest tube.

WHAT IS RECOMMENDED

- In the event of a failure to remove the chest tube, mechanical pleurodesis should be discussed at the time of the first occurrence

WHAT YOU SHOULD NOT DO

- Pleurodesis for a small pneumothorax.
- Pleurodesis in a patient with asymptomatic bullous lesions without pneumothorax.

Stroke

Due to the vascular fragility which is characteristic of vascular Ehlers-Danlos syndrome, the cerebral arteries are often affected by complications, most commonly at the cervical level. These complications often present in the form of spontaneous dissections, which can obstruct the affected artery to varying degrees.

WHAT IS RECOMMENDED

- In the event of clinical signs that may suggest a stroke, urgently carry out an imaging scan of the brain and of the cerebral arteries (angiography scan or MRI).
- Seizure activity should be treated promptly to avoid further arterial complications.
- Search specifically for a carotid-cavernous fistula in the event of pulsatile tinnitus, associated or not with exophthalmos.

WHAT YOU SHOULD NOT DO

- Angioplasty with stenting of carotid and vertebral arteries except for salvage procedures.
- Revascularisation (thrombolysis, salvage arteriography) without vEDS expert physician advice.

Pregnancy, Delivery and Postpartum Care

Pregnancy

Pregnancy presents a particular risk for patients with vascular Ehlers-Danlos syndrome. It is associated with an increased risk of pregnancy-related complications, particularly arterial and uterine rupture. Obstetrical complications occur in about half of pregnancies and include premature rupture of membranes with preterm delivery, rare uterine rupture during labour, arterial rupture even after delivery, severe perineal tears, and antepartum and postpartum hemorrhage.

WHAT IS RECOMMENDED

- Inform the patient and partner about the maternal risks of pregnancy, the risk of disease transmission, and prenatal genetic testing and diagnostic possibilities (if applicable).
- Discuss any desire for pregnancy with the medical and obstetrical referral team prior to conception.
- Discuss and suggest the possibility of performing prenatal diagnosis.
- Perform a complete arterial lesion assessment before or in early pregnancy.
- Arrange for increased monitoring of the uterine cervix, especially from 28 weeks onwards.
- Continue treatment with celiprolol or other beta-blockers throughout pregnancy, or introduce treatment if the pregnancy started without treatment.
- Discuss preterm caesarean section at 35-37 weeks' gestation.

Delivery

There are no formal recommendations regarding the best method of delivery for patients with vascular Ehlers-Danlos syndrome. Caesarean section between 35 and 37 weeks of gestation is a commonly adopted approach in patients with dominant negative variants, especially for primiparous patients with a known diagnosis.

WHAT IS RECOMMENDED

- Schedule the delivery due to the elevated risk of maternal complications.
- Always discuss carrying out a caesarean section between 35 and 37 weeks of gestation.
- Plan for the birth to take place in a level 3 (or equivalent, according to country) maternity unit.

WHAT YOU SHOULD NOT DO

- Delivery at home or in a level 1-2 maternity unit.
- Vaginal delivery without prior multidisciplinary consultation.
- Plan delivery without advising the vascular surgery team.

Breastfeeding

Vascular Ehlers-Danlos syndrome is not in itself a contraindication to breastfeeding. Nonetheless, the beta-blockers prescribed to prevent arterial complications from occurring in the mother are present in breast milk.

WHAT IS RECOMMENDED

- Discuss breastfeeding with the attending obstetric and paediatric teams on a case-by-case basis.

WHAT YOU SHOULD NOT DO

- Interrupt the mother's beta-blocker treatment to allow for breastfeeeding.
- Consider breastfeeding if the newborn has displayed side-effects of beta-blocker use (including bradycardia, respiratory distress and/or hypoglycaemia).
- Consider breastfeeding in the event of acute arterial maternal complications in the peripartum period.
- Prescribe atenolol and nadolol during breastfeeding (highly excreted in breastmilk).

Invasive investigations and treatment of common medical emergencies in vEDS

Pulmonary Embolism and Deep Vein Thrombosis

There is no particular risk of pulmonary embolism associated with vascular Ehlers-Danlos syndrome, except for acute complications due to prolonged bed rest.

WHAT IS RECOMMENDED

- Follow standard treatment protocols for pulmonary embolisms (anticoagulant treatment).
- Prescribe anticoagulant treatment for the minimum recommended treatment period.

WHAT YOU SHOULD NOT DO

- Endovascular treatment should be avoided whenever possible, as should fibrinolysis.
- Mechanical thrombectomy and local thrombolysis of high deep vein thrombosis should be avoided.

Varicose Veins

Superficial venous insufficiency (SVI) of the lower limbs (varicose veins) is common in vEDS, which may be a risk factor for venous thrombosis. SVI in vEDS is characterised by its onset before the age of 20 years and the early presence of significant (>3 mm) varicose veins.

WHAT IS RECOMMENDED

- Carry out a venous assessment if there are functional or physical symptoms of superficial venous insufficiency of the lower limbs.
- Large and small saphenous vein insufficiency should be treated with endovenous laser or radiofrequency therapy when necessary.
- Endovenous foam therapy may be discussed, especially for redux varicose veins.

WHAT YOU SHOULD NOT DO

- Treat saphenous insufficiency by surgical vein stripping (risk of arterial or femoral venous rupture).
- Perform phlebectomy of superficial varicose veins (foam sclerotherapy should be preferred).

Antiplatelet Agents and Anticoagulants

The fragility of patients with vascular Ehlers-Danlos syndrome is not conducive to the prescription of antiplatelet and anticoagulant treatments.

WHAT IS RECOMMENDED

- Discuss the prescription of antiplatelet agents/anticoagulants on a case-by-case basis.
- In all cases, limit the prescription to the minimum possible duration.
- Limit the prescription of NSAIDs, and if they are necessary, use them only on an infrequent basis.

WHAT YOU SHOULD NOT DO

- Continue long-term antiplatelet/anticoagulant treatment unless otherwise indicated.
- Combine anticoagulants with antiplatelets and/or several antiplatelet agents.

Exercise

Most of the usual aspects of daily living and recreational activities are not restricted in vEDS. The major concerns associated with high level sports activities are the risk of vascular rupture with the trauma of collisions and the change in blood pressure during these activities.

WHAT IS RECOMMENDED

- Mild to moderate aerobic physical activity, where you can talk to your partner, is thought to be beneficial.
- The use of equipment such as stationary bicycles, elliptical trainers or well-cushioned treadmills could be encouraged.
- Light weights to maintain tone and strength and good breath control while using weights.

WHAT YOU SHOULD NOT DO

- Collision sports and isometric activities are generally discouraged.
- Running on hard surfaces and for long distances, which may exacerbate foot, ankle, knee, and hip pain.
- Activities involving rapid acceleration/deceleration should be discouraged as these may increase the risk of vessel dissection.

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Gathering the best expertise in Europe
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 affected). These include arterial diseases (affecting the aorta to small arteries), arterio-venous anomalies, vascular malformations, and lymphatic diseases.

VASCERN currently comprises 48 expert teams from 39 highly specialized multidisciplinary HCPs? coming from 19 EU Member States, as well as various European Patient Organisations, and is coordinated in Paris, France.

Through our 6 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 realize the full potential of European cooperation for specialised healthcare by exploiting the latest innovations in medical science and health technologies.

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