

# RaDiCo-SEdVasc - National cohort on the vascular Ehlers-Danlos syndrome (SEdV)

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## Général

### Identification

Nom détaillé	National cohort on the vascular Ehlers-Danlos syndrome (SEdV)
Sigle ou acronyme	RaDiCo-SEdVasc
Numéro d'enregistrement (ID-RCB ou EUDRACT, CNIL, CPP, etc.)	CCTIRS n° 15.955 - Decision CNIL n°DR-2016-265

### Thématiques générales

Domaine médical	Cardiology Disability/handicap Gastroenterology et hepatology Neurology Pneumology Radiology and medical imaging Rare diseases
Pathologie, précisions	Thin translucent skin; Arterial/intestinal/uterine fragility or rupture; Extensive bruising
Déterminants de santé	Genetic Healthcare system and access to health care services Lifestyle and behavior Medicine Occupation Social and psychosocial factors

### Responsable(s) scientifique(s)

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## Collaborations

Participation à des projets, des réseaux, des consortiums

Yes

Précisions

European Reference Network

## Financements

Financements

Public

Précisions

Funded by the French « Investissements d'Avenir » cohorts programme, Grant « ANR » 10-COHO-0003.

## Gouvernance de la base de données

Organisation(s) responsable(s) ou promoteur

Inserm

Statut de l'organisation

Secteur Public

Existence de comités scientifique ou de pilotage

Yes

Labellisations et évaluations de la base de données

Security audit certification of the database

Contact(s) supplémentaire(s)

## Caractéristiques

Type de base de données

Type de base de données

Morbidity registers

Objectif de la base de données

Objectif principal

Main objective  
The main objective of this study is to describe the natural course of vascular Ehlers-Danlos syndrome,

in particular the order of appearance of different types of complications (arterial, digestive, pulmonary and uterine).

Secondary objectives are:

1. To study the prospective genotype-phenotype relationships;
2. To study the intra familial phenotypes relationships;
3. To assess the global cost of vEDS cares, including the standard pathway and the cares.
4. To assess the effect of different therapies on the occurrence of new sites of arterial dissection/rupture and the corresponding morbidity and mortality ;
5. To assess the correlation between the diffusion of arterial lesions and the occurrence of cardio-vascular complications (ie: identification of severity criterion);
6. To assess the quality of life of vEDS patients and the impact of the disease on professional activity.

#### Critères d'inclusion

Patients eligible for inclusion in this study have to fulfil all of the following criteria:

- Patients (adults and children) with genetically-proven vEDS (presence of a pathogenic mutation at the COL3A1 gene);
- With a signed informed consent for adults or signed informed consent of parents/guardians for minors/major protected.

There are no exclusion criteria for this study.

#### Type de population

##### Age

Childhood (6 to 13 years)  
Adolescence (13 to 18 years)  
Adulthood (19 to 24 years)  
Adulthood (25 to 44 years)  
Adulthood (45 to 64 years)  
Elderly (65 to 79 years)  
Great age (80 years and more)

##### Population concernée

Sick population

##### Pathologie

I73 - Other peripheral vascular diseases

##### Sexe

Male  
Woman

##### Champ géographique

National

##### Détail du champ géographique

European extension envisaged

## Collecte

### Dates

Année du premier recueil 2016

Année du dernier recueil 2021

### Taille de la base de données

Taille de la base de données (en nombre d'individus) < 500 individuals

Détail du nombre d'individus 340 subjects targeted

### Données

Activité de la base Current data collection

Type de données recueillies  
Clinical data  
Declarative data  
Paraclinical data  
Biological data  
Cost data

Données cliniques, précisions  
Direct physical measures  
Medical registration

Détail des données cliniques recueillies  
? Set 1: Patient's characteristics at inclusion: personal information, status within pedigree vital status, diagnosis, last follow-up, initiation of therapy, molecular diagnosis (type and group of mutation of COL3A1 gene), presence or not of diagnostic criteria (phenotype) and first arterial assessment (mandatory for all patients), the latter being the baseline comparator for the primary objective of this cohort study. Set 2: - age of the diagnosis with the use of biochemical or molecular genetic studies;- physical characteristics (characteristic facial features, thin skin with visible veins, easy bruising, and increased joint mobility of the hands);- causes of death : arterial rupture, organ rupture (uterus, heart, Liver or spleen), gastrointestinal rupture, other causes;- medical and surgical complications : arterial dissection or rupture, spontaneous bowel perforation, or organ rupture;- age at the time of a first complication;- arterial complications and surgical outcome : thoracic, abdominal, head, neck, limbs, central nervous system (fistulae involving the carotid artery and cavernous sinus, carotidartery dissection, aneurysm, and rupture);- gastrointestinal

complications and surgical outcome: sigmoid colon, perforation of the small, gastric perforation, rupture of the gastrointestinal tract, dehiscence of the wound, evisceration, haemorrhage of abdominal vessels, fistulas, and adhesions;- nature and location of mutations in the gene for type III procollagen (COL3A1);- outcome of pregnancy: abortion, death, live-born infants at term, complications of pregnancy, affected child;- lifestyle modification;- medication.

Données déclaratives, précisions

Paper self-questionnaire  
Internet self-questionnaire  
Face to face interview

Détail des données déclaratives recueillies

SF-36 (adults) / SF-10 (children), Hamilton

Données biologiques, précisions

Routine blood measurements are :- Hematology (hemoglobin, hematocrit, leucocytes and platelets);- Blood chemistry (sodium, potassium, calcium, chloride, creatinine, fasting blood glucose, albumin, total protein, triglycerides, total cholesterol, HDL-cholesterol, LDL-cholesterol ASAT, ALAT, gamma GT).

Données de coût, précisions

The economic analysis is about evaluating the global cost of vEDS, including the standard pathway and the cares. To avoid the risk of counting twice the same cares, the two levels of costs must be dissociated. Direct medical costs consist of monitoring and hospital costs. Hospital stays are valued according to the French decree (published yearly) for inpatient or outpatient stays. These data will be collected in collaboration with the French DRG manager for each participating center. Monitoring costs correspond to drugs, biological exams, radiology, consultations, physiotherapist care, home nursing care or any other form of care. They are valued according to the corresponding nomenclatures in force (NABM, NGAP, AMK ...). For external or private activity, the classifications in force will be used. Direct non-medical costs such as ambulance transport will be collected.

Existence d'une biothèque

Yes

Contenu de la biothèque

DNA

Paramètres de santé étudiés

Health event/morbidity  
Health event/mortality  
Health care consumption and services  
Quality of life/health perception

Consommation de soins, précisions	Hospitalization Medical/paramedical consultation Medicines consumption
Qualité de vie/santé perçue, précisions	quality of life (SF-36 (adults) / SF-10 (children), Hamilton).
<b>Modalités</b>	
Mode de recueil des données	eCRF using REDCap; Cloud based, secure by design web accessible platform. Certified Health Data Hosting resource
Nomenclatures employées	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, Drug dictionary (DCIs)
Procédures qualité utilisées	Continuous data management; Data Management Plan and Data Validation Plan. Native controls and Query system
Suivi des participants	Yes
Modalités de suivi des participants	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Pathologie suivies	I73 - Other peripheral vascular diseases
Appariement avec des sources administratives	Yes
Sources administratives appariées, précisions	PMSI, AMELI, NABM, CCAM, NGAP, AMI, AMK
<b>Valorisation et accès</b>	
<b>Valorisation et accès</b>	
<b>Accès</b>	
Existence d'un document qui répertorie les variables et les modalités de codage	Yes
Charte d'accès aux données (convention de mise à disposition, format de données et délais de mise à disposition)	Access requests to RaDiCo -SEdVasc data (rough / structured), biocollections or to analytic reports will be examined by the scientific committee following submission of a Specific Research Project (SRP) synopsis, as defined in the Resource Access Charter. Must be sent to <a href="mailto:sedvasc@radico.fr">sedvasc@radico.fr</a>
Accès aux données agrégées	Access on specific project only

Accès aux données individuelles    Access on specific project only