RaDiCo-SEDVasc - National cohort on the vascular Ehlers-Danlos syndrome (SEDv)

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General

Identification

Detailed name	National cohort on the vascular Ehlers-Danlos syndrome (SEDv)
Sign or acronym	RaDiCo-SEDVasc
CNIL registration number, number and date of CPP agreement, AFSSAPS (French Health Products Safety Agency) authorisation	CCTIRS n° 15.955 - Decision CNIL n°DR-2016-265

General Aspects

Medical area	Cardiology Pediatrics Radiology and medical imaging Rare diseases
Study in connection with Covid-19	No
Pathology (details)	The inheritance of vEDS follows an autosomal dominant trait and is related to mutations in the COL3A1 gene, encoding the pro-? chain of type III procollagen. The mutation alters the assembly, stability and thus secretion and resistance to tensile stress of this fibrillar collagen, resulting in early spontaneous arterial, digestive and obstetrical accidents.
Health determinants	Genetic Healthcare system and access to health care services Lifestyle and behavior Medicine Occupation Social and psychosocial factors

Scientific investigator(s) (Contact)

Name of the director	JEUNEMAITRE

Surname	Xavier
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Unit	Inserm UMR S970
Organization	French National Institute for Health and Medical Research (Inserm)

Collaborations

Participation in projects, networks and consortia	Yes
Details	European Reference Network

Funding

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

Governance of the database

Sponsor(s) or organisation(s) responsible	Inserm
Organisation status	Public
Presence of scientific or steering committees	Yes
Labelling and database evaluation	Security audit certification of the database

Additional contact

Main features

Type of database

Type of database	Morbidity registers

Database recruitment is carried out as part of an interventional study	No

Database objective

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).
Inclusion criteria	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); - Patients (or his/her legal guardian) who are signed informed consent or does not oppose to his/her personal data collection.

Population type

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 covered	Sick population
Pathology	I73 - Other peripheral vascular diseases
Gender	Male Woman
Geography area	National
Detail of the geography area	European extension envisaged

Data collection

Dates

Date of first collection (YYYY or MM/YYYY)	2016
Date of last collection (YYYY or MM/YYYY)	2021

Size of the database

Size of the database (number of individuals)	< 500 individuals
Details of the number of individuals	340 subjects targeted
Data	
Database activity	Current data collection
Type of data collected	Clinical data Declarative data Paraclinical data Biological data Cost data
Clinical data (detail)	Direct physical measures Medical registration
Details of collected clinical data	Medical personal and family history; medical characteristics at baseline and throughout the study; type of genetic mutation; patient phenotype
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Paraclinical data (detail)	Laboratory features and imaging or surgical reports
Biological data (detail)	Measurements like hemoglobin, hematocrit, leucocytes and platelets, sodium, potassium, calcium, chloride, creatinine, fasting blood glucose, albumin, total protein, triglycerides, total cholesterol, HDL-cholesterol, LDL-cholesterol ASAT, ALAT, gamma GT
Cost data (detail)	Medico-economic cost of patient care
Presence of a biobank	No
Health parameters studied	Health event/morbidity Health event/mortality Health care consumption and services Quality of life/health perception
Care consumption (detail)	Hospitalization Medical/paramedical consultation Medicines consumption
Quality of life/perceived health (detail)	Short-Form Health Survey (SF-36 for adults, SF-10 for children), Hospital Anxiety and Depression Scale (HADS)

Procedures	
Data collection method	eCRF using REDCap; Cloud based, secure by design web accessible platform. Certified Health Data Hosting resource
Classifications used	HPO, ICD10, Snomed CT, Orpha Codes and ORDO, Drug dictionary (DCIs)
Quality procedure(s) used	Continuous data management; Data Management Plan and Data Validation Plan. Native controls and Query system
Participant monitoring	Yes
Monitoring procedures	Monitoring by convocation of the participant Monitoring by contact with the referring doctor
Followed pathology	I73 - Other peripheral vascular diseases
Links to administrative sources	Yes
Linked administrative sources (detail)	PMSI, AMELI, NABM, CCAM, NGAP, AMI, AMK
Promotion and access	
Promotion	
Access	
Presence of document that lists variables and coding procedures	Yes
Terms of data access (charter for data provision, format of data, availability delay)	Requests for access to RaDiCo-SEDVasc data (aggregated or individual) will be considered by the Scientific Committee following the submission of a summary of a specific research project, as defined in the Charter of access to resources. Requests should be sent to: sedvasc@radico.fr
Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only