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 Disability/handicap Gastroenterology et hepatology Neurology Pneumology Radiology and medical imaging Rare diseases
Pathology (details)	Thin translucent skin; Arterial/intestinal/uterine fragility or rupture; Extensive bruising
Health determinants	Genetic Healthcare system and access to health care services Lifestyle and behavior Medicine Occupation Social and psychosocial factors

Scientific investigator(s) (Contact)

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Unit	Inserm UMR S 970
Organization	Institut National de la Santé et de la Recherche Médicale / 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 objective	
Main 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). 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.
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); - 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.
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	? 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.
Declarative data (detail)	Paper self-questionnaire Internet self-questionnaire Face to face interview
Details of collected declarative data	SF-36 (adults) / SF-10 (children), Hamilton
Biological data (detail)	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).
Cost data (detail)	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.
Presence of a biobank	Yes
Contents of biobank	DNA

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)	quality of life (SF-36 (adults) / SF-10 (children), Hamilton).
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)	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 sedvasc@radico.fr

Access to aggregated data	Access on specific project only
Access to individual data	Access on specific project only