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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Laboratoire	Inserm UMR S 970
Organisme	Institut National de la Santé et de la Recherche Médicale / French National Institute for Health and Medical Research (Inserm)
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).
Modalités	
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
Valorisation et accès	
Valorisation et accès	
Accès	
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 sedvasc@radico.fr
Accès aux données agrégées	Access on specific project only

Accès aux données individuelles Access on specific project only