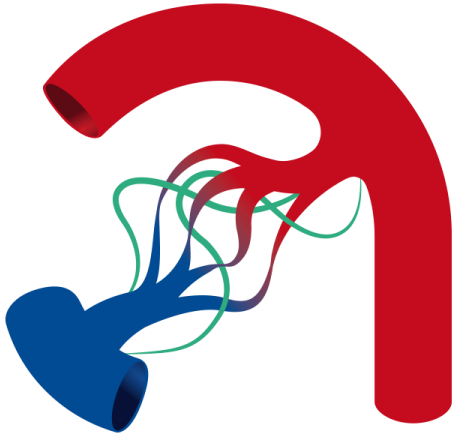
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



VASCERN Registry Update

Leo Schultze Kool

Introduction
registries

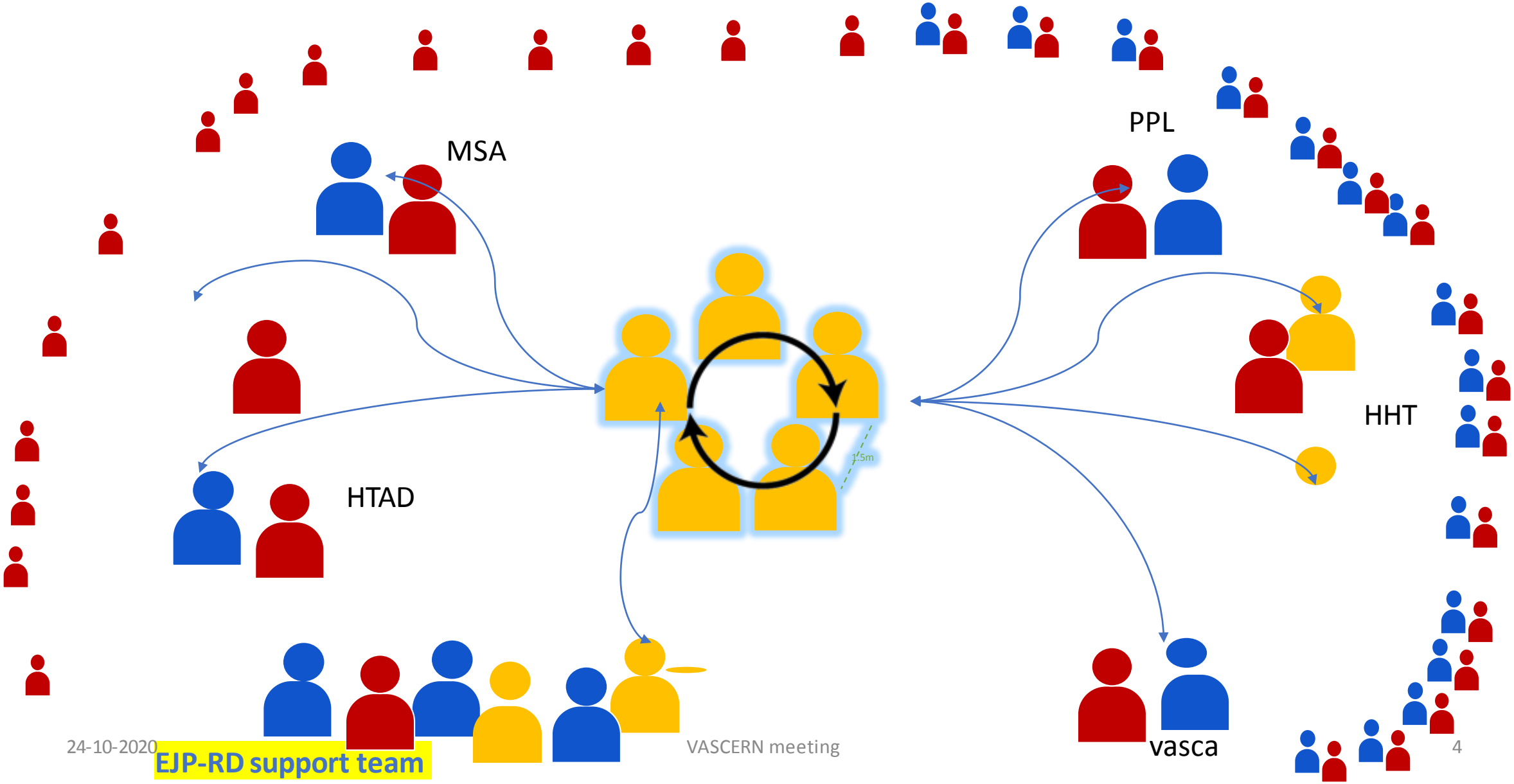
- Top priority
  - Horizon 2020
  - Mentioned in every grant proposal
  - Seen as a way to develop new treatments/evaluation/genes etc
  - Will be linked to all other existing EU platforms

  - EJP-RD linked to GA4GH (global alliance for genomics and health)
  - Go-FAIR initiative
  - Elixer RD (european infrastructure for life sciences)
  - IRDiRC (International Rare Disease Research Consortium)
  - BBMRI (biobanks infrastructure)

Registry kick-off 9th July 2020

- Introduction and welcome (*Leo Schultze Kool, Registry WG Chair*)
- time line and goals. (Leo)
- Indentification of contact persons in each HCPs who can take care of the registry work and liaise with VASCERN's datastewards (all)
- Present status registry project within vasca and PPL *(Pim, Leo Schultze Kool)*
- Progress of each Rare Disease Working Groups: definition of the specific diseases dataset items and state of play of registry building and FAIRification *(Registry leads: Guillaume HTAD, Leo/Pim VASCA PPL, Sophie HHT, Sonia/ Michael MSA vEDS)*
- International Summer School on Rare Disease Registries and FAIRification of Data
- WG Face 2 Face Meeting Paris, October, 23rd - invitations *(Marine)*
- Grant Agreement: Funding & Project Milestones/ Deliverables *(Marine)*

The team



EJP-RD support team

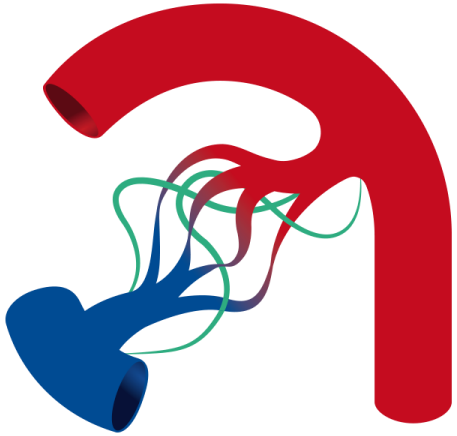
Data steward meeting 16-10-2020

- **Formation of the team (team meeting planned every three weeks)**
- **Introduction of the EJP-RD support**
- **Discussion of the Time line**
  - Formation of a team with links to the individual HCP's (08-2020)
  - Content datasets for the different WG (10-2020)
  - Fairification proces (01-2021)
    - Ontologies/semantic modelling
  - Implementation
    - At a local level
      - Informed consent/Medical ethics approval
      - EUPID

European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



VASCERN-Days 2020 registry update

VASCA

23/10/2020

Proces sofar

We have a working registry for VASCA. Built in Castor. For now it only holds the COMMON DATA ELEMENTS agreed on with the EU and being applied by all ERN's.
This data has been made FAIR :

Findable. (eg registered at the European directory for Rare Diseases ERDRI and good MetaData).

Accessible. (Castor made it possible to publish the data. Possibility to access by anyone. In the beginning only for those who entered the data. The HCP's stay owner of their data)

Interoperable. (Common Data Elements from EU and the data has been put in an universal and by computers understandable format)

Reusable. (data stays in one place and is identified by an universal identifier (Uniform Resource Identifier))

Current status

- In the Netherlands at the RadboudUMC we start today with entering the first live patients!
- Soon meetings with the other VASCA HCP's to talk about the project so far and the timeline ahead (work to be done and implementation at the other HCP's).

Now working on:

- Implementing a pseudonymized patient id.
- MetaData (describing the owner and content of the registry and its origin et cetera)
- Disease Specific Elements (specific VASCA data in the registry)

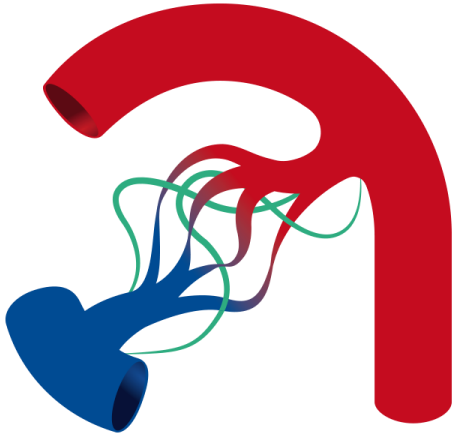
Future work:

- Authorization and Authentication. (for granting possible access to others)
- Automated upload from EPIC (at the Radboud) or other patient data system to Castor.

European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



VASCERN-Days 2020

Vascern PPL-WG 22-10-2020

FAIR registry: What is ready ?

- All PPL- members are contacted for technical data
- Inventarisation for connection with ERDRI: CASTOR Drachten
- PPL Member ERDF

- Start programming CASTOR for specific dataset PPL
- Informed consent EU is sufficient for this registry (NS)

Specific dataset

- Item 5.3: add a pedigree: up to the second degree
- Item 5.4: localisation of swelling: multiple items incl. central lymphatic disease
- Item 6.1: New ORPHANet codes (incl. genes found) / diagnosis
- Item 6.2: type of treatment (reductive / reconstructive / intervention radiology / others

Time table for PPL: get running in 2021

- Finalizing both datasets into CASTOR (1/2021)
- Data entry first by hand at the test side NS: All patients with PL included with IC
- Testing how to generate data from HIX into CASTOR (2/2021)
- Meantime: preparing other members PPL for connection to CASTOR or other system before loading ERDRI (2021)
- Obtain agreement on informed consent by all members PPL (2021)
- Great thanks to Pim Kamerling for his help and perseverance

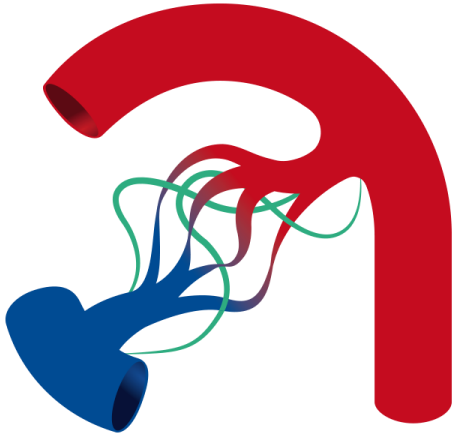
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)

HTAD

registry update



#VASCERNdays2020

Main choices

- **Commun registry for all the HCPs**
  - Development of **a secure web application** according to the needs of the registry
    - Centralisation of data on a health data host.
    - Use of laravel for development
      - A framework which gathers a set of basic functions
    - Allows us to integrate the genetics software and to link patient records within a family (thanks to Pedigree XP)
  - Each HCP (center)
    - Access only to its data
    - Share data when willing to participate in a study
    - Extraction of the data independantly

- **Based on existing registry**
  - Extensive: many items
    - Not all items required for all patients
  - Includes:
    - core dataset
    - Data included in MAC (US based registry for collaboration)
    - Evolution with specific studies
  - Prospective only (at least beginning)

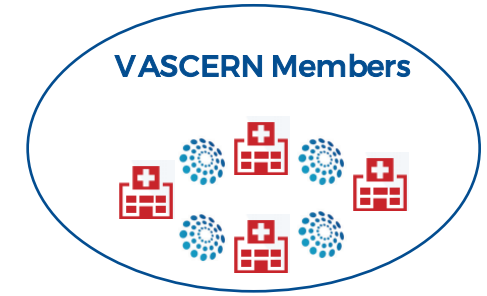
Registry status

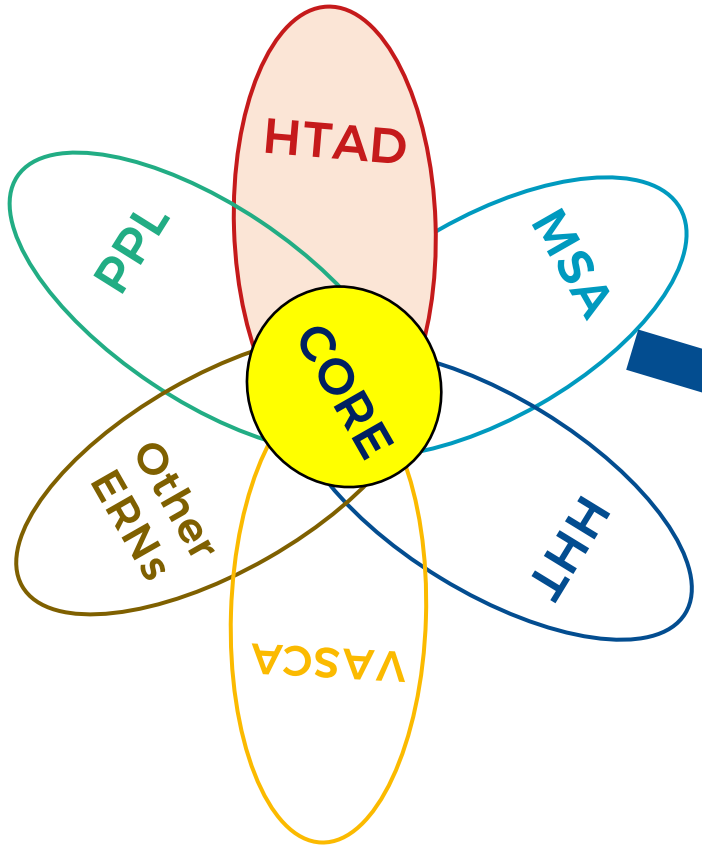
- **Work done**
  - Local version 1 being tested locally
    - All the items included
    - Intranet and not web
    - Creation of a patient file
    - Creation and addition of consultations for a patient file
    - Création of the profiles management pages

- **Improvement ongoing**
  - English translation in the program
  - A dynamic internet browsing between patient records via family trees (thanks to Pedigree XP)
  - Creation of management pages used by the administrator
  - Extraction of the data independantly for each HCP
  - Fairification

Based on the French registry
Discussions within the HTAD RDWG

Fondation 101 Génomes
FONDATION PRIVÉE
MAC
MONTALCINO AORTIC CONSORTIUM

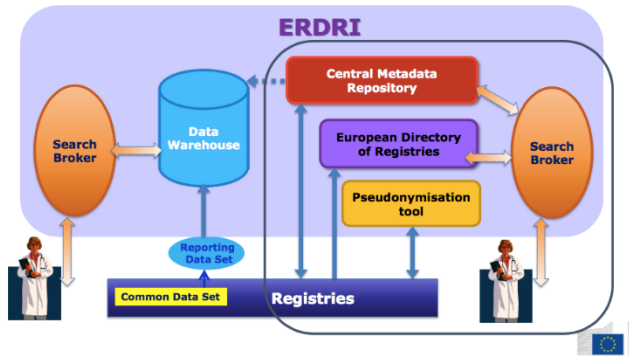




FAIR

Findable
Accessible
Interoperable
Reusable

European Platform on Rare Diseases Registration

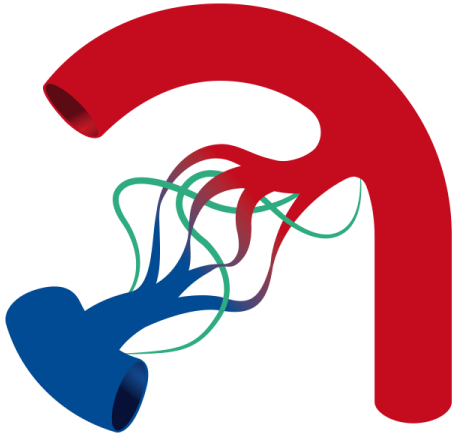


www.Vascern.eu

European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



VASCERN-Days 2020

Vascern HHT-WG 22-10-2020

HHT group

- Already existing registries used by our partners
  - Including many data

HCP	Approximate number of cases – on existing HCP database
Lyon	5-6,000
London	>2,000 HHT/PAVM
Crema	~1,300
Bari	800
Essen	>300
Odense	600
St Antonius	~3,000 including controls

  - Local database in most cases (Excel, custom made DB, SPSS)
  - Accessible on the internet (France and Denmark-Redcap)

- CIROCO database was selected
  - Accessible on the internet
  - Secure

HHT group

- Grant agreement: 20 k€ for the necessary modifications of CIROCO database to make it fair.
- Coordinator : G. Jondeau /Sophie Dupuis-Girod
- Project manager : Evelyne Decullier
- Developer : Jean-Philippe Allard

HHT group

- Now working on the minimum dataset

- Still many questions ….
  - How to adapt other databases
  - Consent form to obtain
  - Who will enter data ? Only for new patients ? Who will have time and authorization ?

VASCERN-MSA

RaDiCo-vEDS cohort

French National Cohort on the vascular Ehlers-Danlos Syndrome (vEDS)

Background information on the RaDiCo platform

Inserm national platform for implementing rare disease cohorts, that offers a centralized expertise in:

- **Clinical Research** (Clinical documentation, regulatory requirements, management of clinical study)
- **E-Health** (REDCap: Vanderbilt University open source Electronic Data Capture; process and interface between Clinical Research and IT)
- **Data-management** and **Statistics**
- **IT** (complete secured system and EDC improvement)
- **Legal environment, valorization, public-private partnerships and European collaborations**

Coordinated by the French Institute on Health and Medical Research (Inserm) and funded by the Ministry of Research via the Investments for the Future 'Cohorts' program managed by the National Research Agency (ANR-10-COHO-0003)

RaDiCo-vEDS - WP5 - Adaptation of existing registries

Tasks	Progress
Definition of the dataset:	☑ **Done** (Minimal dataset_EU RD Platform) ? Set the list of variables how to define?
Inclusion of the core elements (by RADICO)	☑ **Done** (Plan to mapping from vEDS data base to mini-dataset) ? Difference between core elements and mini-dataset? ? List of questions about mapping and missing variables
Encoding of the dataset (Orphanet, HPO, OMIM…)	☑ **Not applicable** (one disease, vEDS, ORPHA number 286)
Implementation of the program EDC (REDCap)	☑ **Done** (Database already available on the RADICO local server and accessible through web interface)
Exporting the registry address on the ERDRIdor	☑ **Done** (vEDS registry implemented)
Exporting the registry metadata on the ERDRImdr	☐ **Ongoing** (EU-RD-PLATFORM@ec.europa.eu)
Implementing the EUPID	☐ **Waiting**
Interoperability	☐ **To discuss** (EJP-RD Pillar 2 dedicated teams) ? Which application/Software/Tools and FAIRification guidelines