



**HAL**  
open science

## **RaDiCo (Rare Disease Cohorts): a platform for rare disease (RD) e-cohorts**

Sonia GUEGUEN, Serge Amselem, Annick Clement, Paul Landais

► **To cite this version:**

Sonia GUEGUEN, Serge Amselem, Annick Clement, Paul Landais. RaDiCo (Rare Disease Cohorts): a platform for rare disease (RD) e-cohorts. World Conference on Rare Diseases (RARE2021), Feb 2021, Virtuel Event, France. inserm-04059748

**HAL Id: inserm-04059748**

**<https://inserm.hal.science/inserm-04059748>**

Submitted on 5 Apr 2023

**HAL** is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

**Background** Implementing RD cohorts is a challenge: RDs are seldom, often underdiagnosed and spread over the national territories.

**Objective** To create a national platform for the development, within a research framework, of RD e-cohorts.

**Material and Methods** The platform, built on the "cloud computing" principle, oriented as an "Infrastructure as a Service"; Interoperable; In compliance with the GDPR; Within a Certified Health Data Host; Ensuring continuous monitoring of data quality and consistency; In line with the French Health Data Hub.

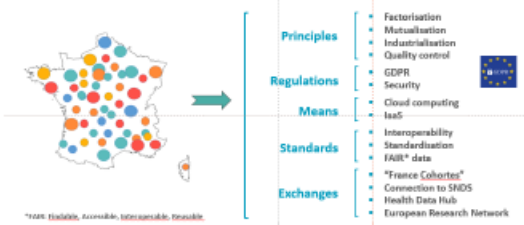
**Results**

**The RaDiCo Information System**



Cloud Microsoft AZURE; CHDI: Certified Health Data Host; CGM: CompuGroup Medical; Portal: Users authentication & selected access to the cohorts; Back Office: Management of access rights; PST: Patient Identification System Translator; REDCap: Research Electronic Data Capture; EGCS: Electronic data capture Gateway; Controller Service; MedDRA: Medical Dictionary for Regulatory Activities; Thélosque: Drugs databank

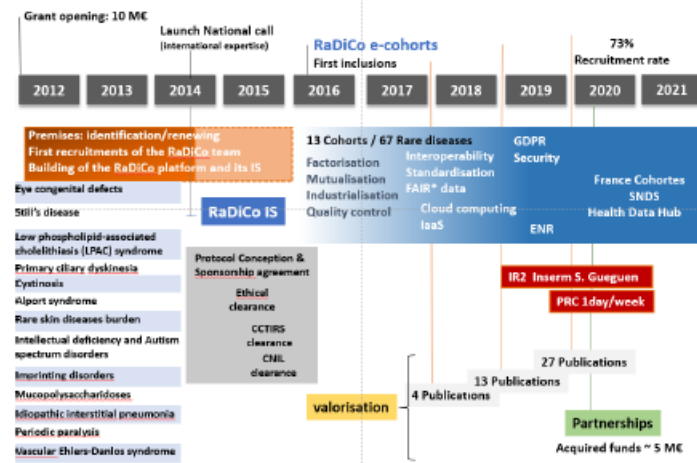
**The RaDiCo platform in brief: How did we proceed?**



**13 ongoing RaDiCo e-cohorts involving 67 RDs**

Diseases	Cohorts	PI	RDHN	ERN
Eye congenital defects	AC-CEIL	N. Chessex	SENSEGENE	ERN-Eye
		P. Calvas	AnDDI-Rares	
Still's disease	ACOSStill	S. Georgin-Laveille B. Fauret	FA2R	ERN-Rita
LPAC syndrome	COLPAC	C. Corpechot	FILFOIE	RARE-LIVER
Primary ciliary dyskinesia	DCP	E. Escudier	RESPIFIL	ERN-Lung
Cystinosis	ECYSKO	D. Maïtre A. Servalis	ORKID	ERKnet
Alport syndrome	EURBIO-Alport	L. Heidat	ORKID	ERKnet
		B. Knebelmann		
Rare skin diseases burden	FARD	C. Bodemer	FIMARAD	ERN Skin
Intellectual deficiency and Autism spectrum disorders	GenIDA	J.L. Mandel	DeScience	AnDDI-Rares ERN-ETHACA
Imprinting disorders	IDMet	A. Lingart	FISEND	ENDO-ERN
		I. Netchine	OSCAR DeScience	
Mucopolysaccharidoses	MPS	B. Héron	G2M	MetabERN
Idiopathic interstitial pneumonia	PID	A. Clement	RESPIFIL	ERN-Lung
Periodic paralysis	PP	S. Vicard	FILNEMUS	ERN-euro-NMD
Vascular Ehlers-Danlos syndrome	SEDVase	X. Jaunaultre	FAVA-Multi	VASCERN

**RaDiCo key milestones**



**Conclusion**

RaDiCo provides a flexible & easily sharable platform for the creation & follow-up of national/international RD e-cohorts.