The Rare Disease Cohorts (RaDiCo) program: Building and Monitoring National and International e-cohorts

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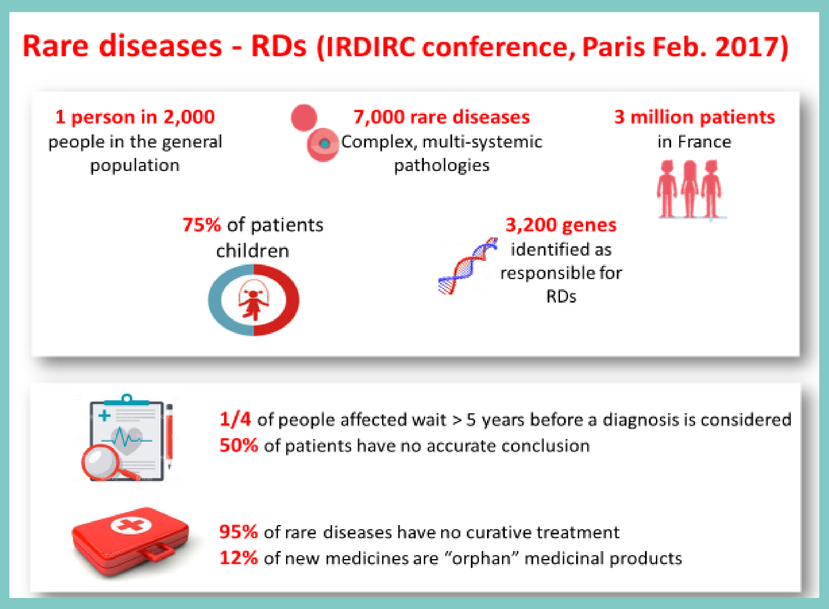
Background

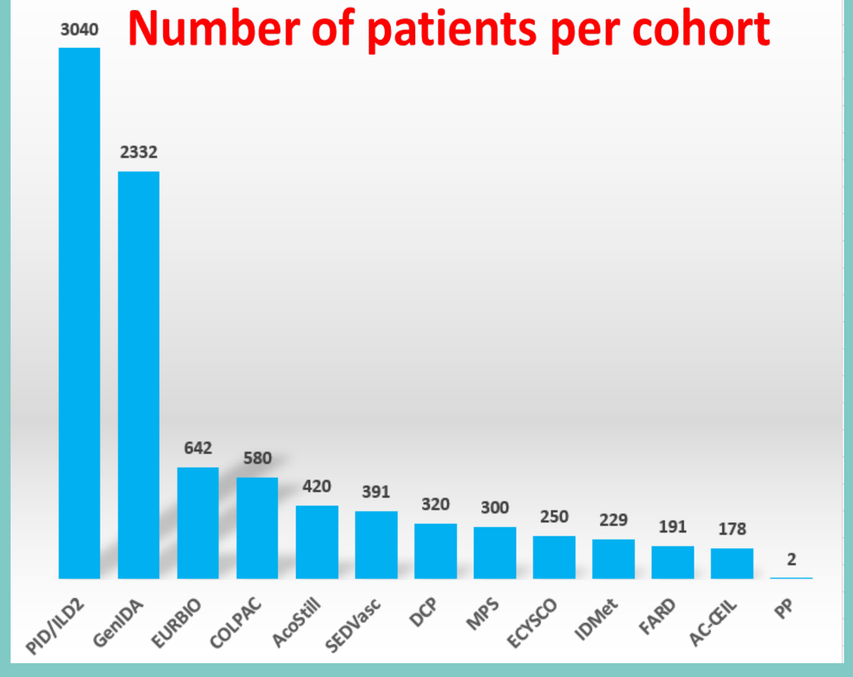
Rare Disease (RD) professionals have highlighted the critical need to implement national/international, multidisciplinary, high-quality cohort studies to address key scientific and medico-economic questions.



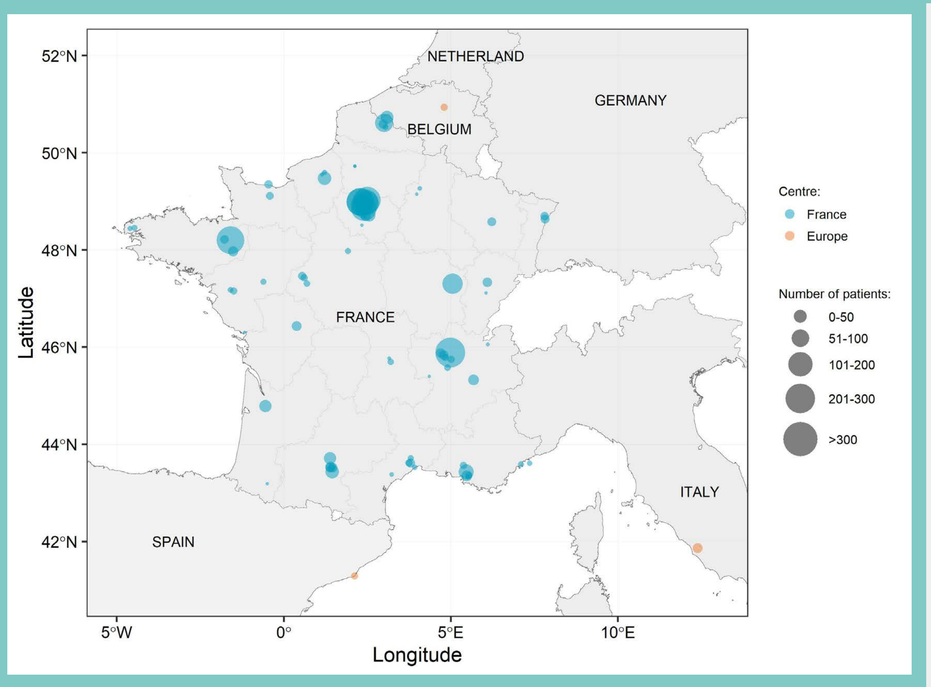
The regulatory authorizations from the different competent bodies have been obtained and the RD e-cohorts have been launched since 2016.

As of September 2025, **8875** patients have been included in **275** clinical centres.

The global achieved recruitment is **86% of the defined target** given the time since the 1st inclusion.



Geographical distribution of opened RD centres in France and Europe



10 specific research projects exploiting each cohort's resources have been established with 4 companies (Public-Private Partnerships) and 6 academic partners: acquired funds ~ 6M€

Objective

To implement RD e-cohorts within a research framework, supported by an interoperable platform of mutualised expertise, resources and services, with shared standardised processes and tools.

Material and Methods

The scientific objectives are: Describing RDs' natural history; Establishing phenotype-genotype correlations; Deciphering RDs' pathophysiology; Identifying new therapeutic avenues; Assessing RDs' societal and medico-economic impact; Identifying patients eligible for new therapeutic approaches.

Results

In 2014, RaDiCo platform launched a national call for RD cohort: 16 RD e-cohorts have been selected.

13 ongoing RaDiCo e-cohorts involving 67 RDs

Diseases	Cohorts	PI	RDHN	ERN
Eye congenital defects	**AC-ŒIL**	N. Chassaing P. Calvas	SENSGENE AnDDI-Rares	ERN-Eye
Still's disease	**ACOStill**	S. Georgin-Lavialle B. Fautrel	FAi2R	ERN-Rita
LPAC syndrome	**COLPAC**	C. Corpechot	FILFOIE	RARE-LIVER
Primary ciliary dyskinesia	**DCP**	E. Escudier B. Maitre	RESPIFIL	ERN-Lung
Cystinosis	**ECYSCO**	A. Servais	ORKiD	ERKnet
Alport syndrome	**EURBIO-Alport**	L. Heidet B. Knebelmann	ORKiD	ERKnet
Rare skin diseases burden	**FARD**	C. Bodemer	FIMARAD	ERN Skin
Intellectual deficiency and Autism spectrum disorders	**GenIDA**	J.L. Mandel	DéfiScience	AnDDi-Rares ERN ITHACA
Imprinting disorders	**IDMet**	A. Linglart I. Netchine	FIRENDO OSCAR DéfiScience	ENDO-ERN
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	**SEDVasc**	X. Jeunemaître	FAVA-Multi	VASCERN

RaDiCo contribution to national and EU RD actions

- European Joint Programme on RDs contribution to 9 tasks
- European Reference Networks contribution of the PIs to 11 ERNs
- Third National Plan for Rare Diseases (2018-2022) contribution to 4 research actions
- Appel à Manifestation d'intérêt (AMI) 2021: PNMR3 Action 3.2
- National Protocols for Diagnosis and Care 8 contributions
- RD National Databank ("BNDMR")

As of 2025, **65** publications in international peer-reviewed journals.

Discussion / Conclusion

The key benefits of this platform are its flexibility and ease of data sharing, which enable the creation of new cohorts. At the European level, two cohorts have been extended, with the cohort PIs involved in European Reference Networks.

RaDiCo has developed e-cohorts and standardized procedures for including new RDs. Industrial and academic partnerships have been established, and strong involvement in the Inserm 'France Cohortes' project will ensure its long-term future.

Remerciements

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