Registries for European Reference Networks (ERNs)

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European Reference Networks

Why the European Reference Networks (ERNs)?

- Many of those affected by a rare or complex condition do not have access to diagnosis and high-quality treatment
- Expertise and specialist knowledge may be scarce as patient numbers are low
- Important delay in diagnosis because of lack of care pathways, diagnostic capacity (no tests available) and treatments in many cases
- No country alone has the knowledge and capacity to treat all rare and low prevalence complex diseases

"The knowledge travels, not the patient"

Launch of the first ERNs in 2017

Legal basis:
Directive on patients' rights in cross-border healthcare
Chapter IV - Cooperation between MS
Article 12 - European Reference Networks

Application and evaluation process
from March 2016 to March 2017:

24 ERNs

European Reference Networks

› 300
HOSPITALS

› 900
HEALTHCARE UNITS

26 Countries

European Commission

European Reference Networks

https://ec.europa.eu/health/ern_en

ERN BOND	Bone Diseases
ERN CRANIO	Craniofacial anomalies and ENT disorders
Endo-ERN	Endocrine Conditions
ERN EpiCARE	Rare and Complex Epilepsies
ERKNet	Kidney Diseases
ERN GENTURIS	Genetic Tumour Risk Syndromes
ERN-EYE	Eye Diseases
ERNICA	inherited and congenital anomalies
ERN-LUNG	Respiratory Diseases
ERN-RND	Neurological Diseases
ERN-Skin	Skin Disorders
ERN EURACAN	Solid Adult Cancers

ERN EuroBloodNet	Onco-Hematological Diseases
ERN EUROGEN	Urogenital Diseases
ERN EURO-NMD	Neuromuscular Diseases
ERN GUARD-HEART	Diseases of the Heart
ERN ITHACA	Congenital Malformations and Intellectual Disability
MetabERN	Hereditary metabolic diseases
ERN PaedCan	Paediatric Cancer
ERN RARE-LIVER	Hepatological Diseases
ERN ReCONNET	Connective Tissue and Musculoskeletal Diseases
ERN RITA	Immunodeficiency, Auto-Inflammatory and Auto Immune Diseases
ERN TRANSPLANT-CHILD	Transplantation in Children
VASCERN	Multisystemic Vascular Diseases

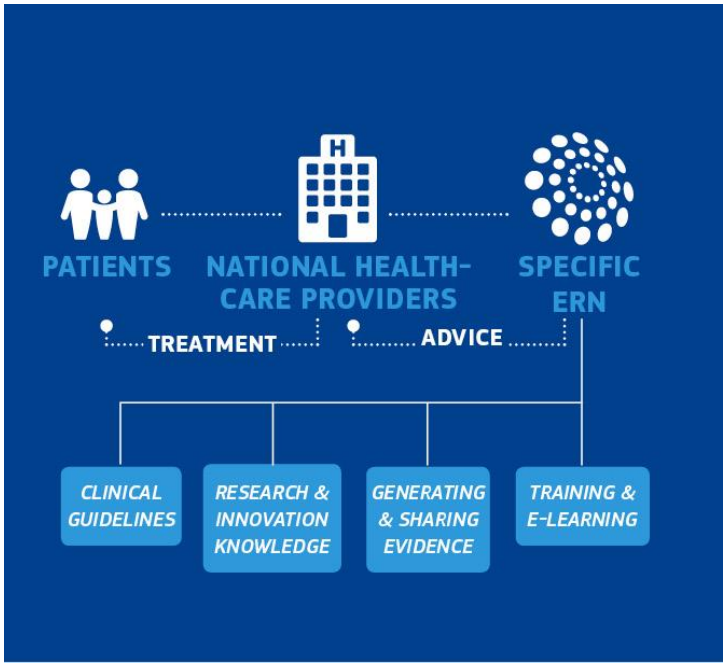
ERNs applied for specific (groups of) diseases

- **ERN EURACAN**: rare neoplasms of the: connective tissue (G1), female genital organs and placenta (G2), male genital organs & urinary tract (G3), neuroendocrine system (G4), digestive tract (G5), endocrine organs (G6), head & neck (G7), thorax (G8), skin & eye melanoma (G9), brain, spinal cords (G10)

- **ERN PaedCan**: leukemias, myeloproliferative and myelodysplastic diseases, lymphomas & reticuloendothelial neoplasms, CNS + miscell. intracranial & intraspinal neoplasms, neuroblastoma & other peripheral nervous cell tumors, retinoblastoma, renal tumors, hepatic tumors, malignant bone tumors, soft tissue and other extraosseous sarcomas, germ cell tumors, trophoblastic tumors and neoplasms of gonads, other malignant epithelial neoplasms & malignant melanomas, other unspecified malignant neoplasms (Hematopoietic and Lymphoid: LCH), hepatoblastoma, very rare tumours, nephroblastoma, neuroblastoma, brain tumours, Ewing sarcoma, soft tissue sarcoma, germ cell tumour, Langerhans cell histiocytosis

- **ERN EuroBloodNet**: 1) non-oncological rare hematological diseases: rare blood cell defect, bone marrow failures, bleeding and coagulation disorders, hereditary haemacrhomatosis and iron metabolism related disorders; 2) oncological rare hematological diseases: lymphoid and myeloid malignancies

What do the ERNs do?



- ✓ *Exchange of expertise and clinical data on **patient cases***
- ✓ ***Virtual remote consultations** through the IT tool developed by the EC: Clinical Patient Management System (CPMS)*
- ✓ ***Research** ←→ registries !*
- ✓ ***Clinical guidelines***
- ✓ ***Education & training***

Added value of the ERNs

Give patients and doctors across the EU access to the best expertise and timely exchange of knowledge, without having to travel to another country.

- ✓ increase the likelihood of early and accurate diagnosis and effective treatment
- ✓ improve public and professional awareness of rare and complex diseases
- ✓ platforms for the development of guidelines, training and knowledge-sharing
- ✓ facilitate large clinical studies to improve understanding and develop new drugs
- ✓ opportunity for networking with likeminded experts from across Europe — ending professional isolation

Governance of the ERN Initiative (1)

European Commission

- ✓ Co-definition of the legal framework for the establishment of the Networks / implementation of the procedures
- ✓ Provision of support to the governance bodies
- ✓ Funding of the initiative (the coordination of the Networks – the IT tools, other support activities …)

ERN Board of Member States

- ✓ Endorse the healthcare providers wishing to become members of the ERNs
- ✓ Approve the Networks and their membership
- ✓ Steer the initiative

Governance of the ERN Initiative (2)

European Reference Networks (ERNs)

- ✓ Coordinators (ERN Coordinators' Group)
- ✓ Members - in each ERN: A Board, Task Forces / Working Groups etc.
- ✓ Patients' representatives
- ✓ Hospital managers

Working groups, composed of representatives of the BoMS and of the ERN Coordinators

WG on Integration (in national healthcare systems)

WG on Research **+ established in 2019: Registries' Task Force (close cooperation with JRC Rare Disease Registration Platform)**

WG on Legal & Ethical issues, relations with Stakeholders

WG on Knowledge generation (education and training)

WG on Monitoring

Registries for ERNs

Five ERNs already funded – started in 2018

- ✓ ERKNet: ERKReg project - ERKNet Registry for Rare Kidney Diseases
- ✓ MetabERN: U-IMD project - Unified European Registry for Inherited Metabolic Disorders
- ✓ Endo-ERN: EuRRECa project - European Registries for rare Endocrine Conditions
- ✓ ERN-LUNG: RD Registry Data Warehouse project
- ✓ **ERN PaedCan: PARTNER (Paediatric Rare Tumours Network – European Registry)**

Ongoing call for the 19 other ERNs

- ✓ Call for grants under the EU Health Programme (10/10/2019) → to start mid 2020
- ✓ ERN Registries Task Force created under the ERN Research Working Group
- ✓ Strong cooperation with EU Rare Disease Platform and tools developed by Joint Research Centre (JRC) of the Commission with DG SANTE
- ✓ Synergies with EU-funded Research "European Joint Programme for Rare Diseases" (EJP RD)

For a common ground for all ERNs: EU Rare Disease Registration Platform and JRC "Common Data Elements"

European Commission | English | Search

European Commission > EU Science Hub > European Platform on Rare Disease Registration

European Platform on Rare Disease Registration (EU RD Platform)

Aim of the Platform

The EU RD Platform copes with the fragmentation of rare disease patients data contained in hundreds of registries across Europe. Read more >

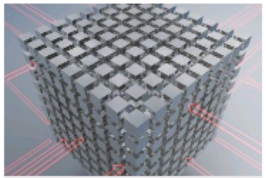
Searchable, findable rare disease registry data

European Rare Disease Registry Infrastructure (ERDRI)

European standards for data collection and data sharing

Trainings, Resources and Latest news

Data repository

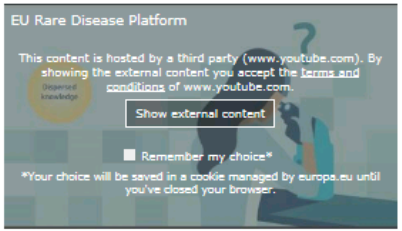


European RD Registry Data Warehouse

eurocat

Surveillance of Congenital Anomalies in Europe

Surveillance of Cerebral Palsy in Europe



In the EU about 30 million citizens in Europe are affected by more than 6000 different rare diseases. The information about these patients is spread between hundreds of registries across Europe, at national, regional and local levels. The EU Rare Disease Platform aims to provide researchers, healthcare providers, patients and policy-makers with a consistent instrument to improve knowledge, diagnosis and treatment of rare diseases. It will make registries' data searchable at EU level and will standardise data collection and data exchange; which will increase the value of each registry and its registration.

https://eu-rd-platform.jrc.ec.europa.eu/

For a common ground for all ERNs: JRC "Common Data Elements" (1)

EUROPEAN PLATFORM ON RARE DISEASE REGISTRATION (EU RD Platform)

SET OF COMMON DATA ELEMENTS FOR RARE DISEASES REGISTRATION

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
1. Pseudonym	1.1.	Pseudonym	Patient's pseudonym	• String	https://eu-rd-platform.jrc.ec.europa.eu/erdri/eupid-intro
2. Personal information	2.1.	Date of birth	Patient's date of birth	• Date (dd/mm/yyyy)	
	2.2.	Sex	Patient's sex at birth	• Female • Male • Undetermined • Foetus (Unknown)	
3. Patient Status	3.1.	Patient's status	Patient alive or dead	• Alive • Dead • Lost in follow-up • Opted-out	If dead then answer question 3.2
	3.2.	Date of death	Patient's date of death	• Date (dd/mm/yyyy)	
4. Care pathway	4.1.	First contact with specialised centre	Date of first contact with specialised centre	• Date (dd/mm/yyyy)	

https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements

For a common ground for all ERNs: JRC "Common Data Elements" (2)

5. Disease history	5.1.	Age at onset	Age at which symptoms/signs first appeared	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
	5.2.	Age at diagnosis	Age at which diagnosis was made	• Antenatal • At birth • Date (dd/mm/yyyy) • Undetermined	
6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case is defined	• Phenotype (HPO) • Genotype (HGVS)	
7. Research	7.1.	Agreement to be contacted for research purposes	Patient's permission exists for being contacted for research purposes	• YES • NO	
	7.2.	Consent to the reuse of data	Patient's consent exists for his/her data to be reused for other research purposes	• YES • NO	
	7.3.	Biological sample	Patient's biological sample available for research	• YES • NO	If YES answer question 7.4
	7.4.	Link to a biobank	Biological sample stored in a biobank	• YES (if appropriate use link) • NO	https://directory.bbmri-eric.eu
8.Disability	8.1.	Classification of functioning/disability	Patient's disability profile according to International Classification of Functioning and Disability (ICF)	• Disability profile / Score	http://www.who.int/classifications/icf/whodasii/en/

https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements

Cancer & rare diseases are 2 'use cases' under the "1 Million Genome Initiative"

European '1+ Million Genomes' Initiative

The Signatories of the declaration of cooperation “Towards access to at least 1 million sequenced genomes in the EU by 2022” are setting up a collaboration mechanism with the potential to improve disease prevention, allow for more personalised treatments and provide a sufficient scale for new clinically impactful research.

https://ec.europa.eu/digital-single-market/en/european-1-million-genomes-initiative

Cancer: a priority at EU level

- ✓ **Joint Action on rare cancers (JARC)**: Final meeting last Sept. 2019, close collaboration with ERNs
- ✓ **Health Commissioner Vytenis Andriukaitis** at ECCO Summit

President-Elect Ursula Von der Leyen:

- ✓ In her 'political guidelines' announced an 'Action Plan against Cancer'
- ✓ In the mission letter to Stella Kyriakides, Commissioner-designate for Health: "I want you to put forward **Europe's Beating Cancer Plan** to support Member States to improve cancer prevention and care. This should propose actions to strengthen our approach at every key stage of the disease: prevention, diagnosis, treatment, life as a cancer survivor and palliative care. There should be a close link with the **research mission on cancer in the future Horizon Europe programme**."

Cancer mission

- ✓ "the European research and innovation missions aim to deliver solutions to some of the greatest challenges facing our world, such as cancer"
- ✓ Chaired by Nobel Prize Winner Prof. Harald zur Hausen; Prof. Ruth Ladenstein (Coordinator ERN PaedCan) member of the Board

European Commission

Thank you!

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