Work progress, achievements of:

Work Package 6
Integration, Outreach & Dissemination

programme

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WP6
Integration, Outreach & Dissemination

ERICA Office

orphanet

Objectives

- Disseminate knowledge and action results
- Set up a web-based ERN platform to increase ERN awareness
- Reach out to relevant stakeholders
- Coordination of workshop organisation

ERICA News & Seasonal Newsletter

SOP for concise update of news items

FREQUENCY: Seasonal

ERICA Office sends out a call for news items for the ERICA periodical Newsletter

ERNs/Partners offices forward news items for dissemination via ERICA Newsletter and other Social Media

ERICA

ERICA Partners

ERICA Office collects and disseminates the News items continuously via Social Media channels and periodically via Newsletter

www.erica-rd.eu

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Welcome at the ERICA website

The aim of the **European Rare Disease Research Coordination and Support Action** consortium (ERICA), in which all 24 European Reference Networks (ERNs) take part, is to build on the strength of the individual ERNs and create a platform that integrates all ERNs research and innovation capacity.

Through knowledge sharing, engagement with stakeholders in the rare disease domain and assembly of transdisciplinary research groups working across the global health spectrum, ERICA strives to reach the following goals:

- new intra- and inter-ERN rare disease competitive networks
- effective data collection strategies
- better patient involvement
- enhanced quality and impact of clinical trials
- increased awareness of ERNs innovation potential.

ERICA will strengthen research and innovation capacity by integration of ERN research activities, outreach to European research infrastructures to synergistically increase impact, and innovation. This will result in efficient access and safe therapies for the benefit of patients suffering from rare diseases and complex conditions.

ERICA ERN Research Conference

The ERICA ERN Research Conference will take place from **December 11th to December 13th, 2024** in Udine, Italy.

Visit the Conference page here

ERICA ERN Research Conference

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ERN input to ACT-EU priorities- Survey!

ACT-EU* – Accelerating Clinical Trials in the EU Please contribute to ACT-EU survey Target group: Academic

Overview of ERNs
PROMs Repository
Publications of ERNs
ERN Clinical Trials
Ukraine #ERNcare4Ua Rare Diseases Doctors

Other News

Apply Now for the EURORDIS 2025 Open Academy Trainings

Applications are now open for the 2025 edition of the Open Academy Schools, offering intensive, face-to-face training in Barcelona from 2-5 [...]

READ

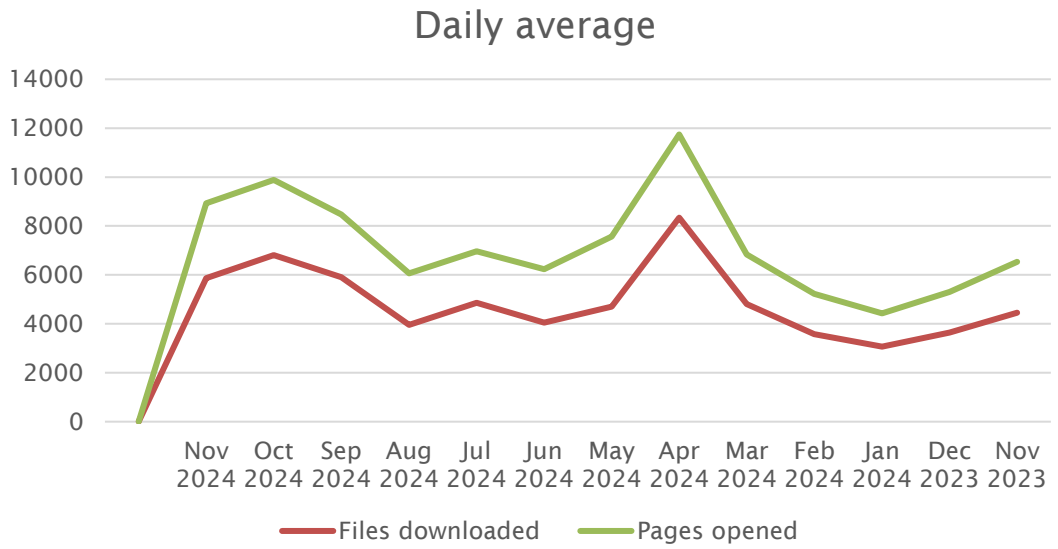
European Partnership Opens a New Era in Rare Disease Research

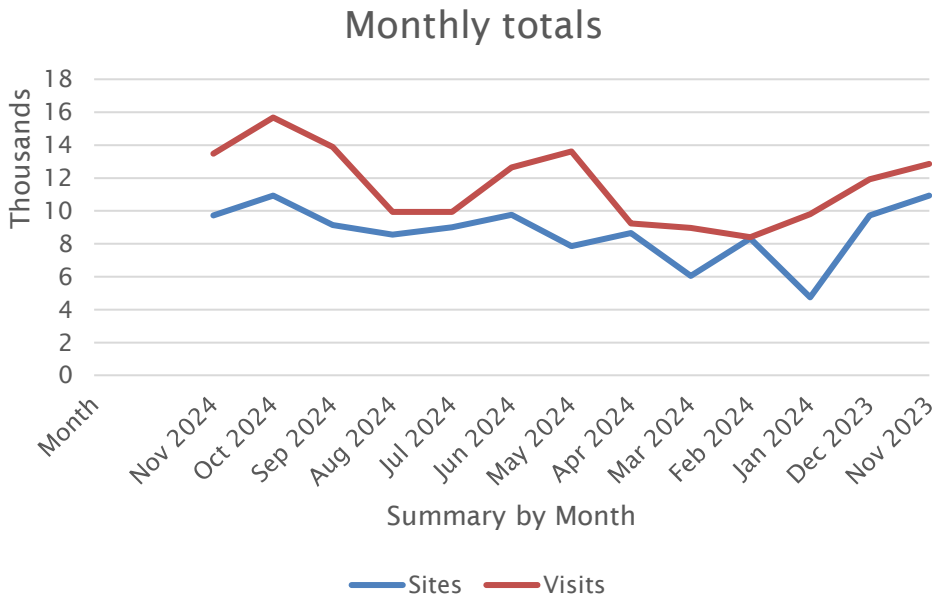
The European Rare Diseases Research Alliance (ERDERA) kicks off this September, with an estimated budget of 380 million euros and the [...]

READ

PLEASE CONTRIBUTE: Cross-Border Access to Paediatric Clinical Trials Survey

ERICA webplatform user statistics





All Results & Documents available under specific WP/Topics

gration and Sharing (WP2)

data capture, protection and access across ERNs keeping patient-centred lance will be provided in Patient-Centred Research WP3.

rking Group (EWG) within the ERN Coordinators' Research Workgroup will launch opment and integration of ERN-wide rare disease registries and their utilization for joint research initiatives. In addition, support for the creation of biorepositories within and across ERNs will be provided and the use of the EJP RD virtual platform for rare disease research will be promoted.

Generated documents

EJP RD and ERICA have developed a series a customisable template documents that can be used by the European Reference Networks (ERNs) to obtain the consent of the patients to get their data included in the registry (**Informed Consent Form**) and establish their governance structure (**Data Access Policy**). Additionally, legal contracts to be used when data is transferred from an HCP to a central registry (**Data Sharing Agreement**) or from a registry to an external stakeholder (**Data Transfer Agreement**) are also provided.

All templates are available under page Generated documents

Recommendations on registry data collection:

ERICA WP2 Recommendations on registry data collection (379 KB)

1st Monitoring Report on ERN Registry Data Collection (updated February 2024)

Monitoring Report v02.2024 (703 KB)

ERN-MATCH: Collaborative Research Wall

Overview of ERNs, their Websites & Registries Clinical Trials Publications

European Reference Networks (ERNs)

European Reference Networks (ERNs) are virtual networks involving Reference Centres across Europe. They aim to tackle complex or rare diseases and conditions, that require highly specialised treatment and concentrated knowledge and resources. Rare patient cases can be discussed by a 'virtual' advisory board of medical specialists across different countries and disciplines in a dedicated and secure IT platform, constructed by the EU Commission specifically for ERNs. This way it is the medical knowledge and expertise that travel, rather than the patients, who have the comfort of staying in their supportive home environments.

ERNs are based on directive 2011/24/EU of the European Parliament and the council European citizens have the right to cross-border healthcare. Here the European Union provides rules for facilitating the access to safe and high-quality cross-border healthcare and promotes cooperation on healthcare between Member States.

Health systems in the European Union aim to provide high-quality, cost-effective care, but this is particularly difficult with rare or low-prevalence complex diseases or conditions. Between 5,000 and 8,000 rare diseases affect the daily lives of around 30 million people in the EU. In order to provide this same quality of care for patients with rare conditions, ERNs were constructed.

More information about European Reference Networks (ERNs)

Overview of all Networks

The first ERNs were launched in March 2017, involving more than 900 highly-specialised healthcare units from over 300 hospitals in 26 Member States. 24 ERNs are working on a range of thematic issues including bone disorders, childhood cancer and immunodeficiency.

To obtain more information about a specific ERN, click on the "Network name" in the table below. This page also provides access to numerous scientific publications related to the ERN.

PubMed®

Network name	Description	Registries
MetabERN	European Reference Network on hereditary metabolic disorders	U-IMD

Below an overview is given of scientific publications related to MetabERN. Publications were obtained from the PubMed database. All publications where one of the authors has an affiliation link where "MetabERN" is mentioned, are added to the publication overview automatically. Updates are performed once a week.

PubMed

Filter list publications

Title | Authors

Date | Keywords

Issue

Search Reset | Column Visibility ▼ | Column Reset

Showing 253 publicatio

Title	Authors	Date	Keywords	Issue name
Simultaneous determination of cytosolic aminoacyl-tRNA synthetase activities by LC-MS/MS.	Mendes MI, Wolf NI, Rudinger-Thirion J, Lenz D, Frugier M, Verloo P, Mandel H, Manor J, Kassel R, Corpeleijn WE, van der Rijt S, Schroor EM, van Dooren SJM, Staufner C, Salomons GS, Smith DEC	2024-11-22		Nucleic acids research
Development of a novel tool for individual treatment trials in mucopolysaccharidosis.	Wiesinger AM, Bigger B, Giugliani R, Lampe C, Scarpa M, Moser T, Kampmann C, Zimmermann G, Lagler FB	2024-11-21	decision analysis framework, immunomodulation, individual treatment trials, mucopolysaccharidosis, personalized medicine, repurposing, risk–benefit assessment	Journal of inherited metabolic disease
Iron Skin Staining: A Rare but Permanent Complication Following IV Iron Infusion.	Dias da Costa T, Mateus-Pinheiro A, Moreira S	2024-11-18	Extravasation of Diagnostic and Therapeutic Materials, Ferric Compounds/adverse effects, Iron/adverse effects, Skin Pigmentation/drug effects	Acta medica portuguesa
Optimising 3D printed medications for rare diseases: In-line mass uniformity testing in direct powder extrusion 3D printing.	Mora-Castaño G, Rodríguez-Pombo L, Carou-Senra P, Januskaite P, Rial C, Bendicho-Lavilla C, Couce ML, Millán-Jiménez M, Caraballo I, Basit AW, Alvarez-Lorenzo C, Goyanes A	2024-11-16	3D printed pharmaceuticals, Additive manufacturing, Drug delivery systems, modified release formulations, Pediatric precision treatments, Pharma-inks, Rare metabolic disorders	International journal of pharmaceutics
Current global vitamin and cofactor prescribing practices for primary mitochondrial diseases: Results of a European reference network survey.	Neugebauer J, Reinson K, Bellusci M, Park JH, Hikmat O, Bertini E, Schiff M, Rahman S	2024-11-11	cofactors, cross sectional study, inherited metabolic disease, primary mitochondrial disease, survey, treatment, vitamins	Journal of inherited metabolic disease
The European reference network for metabolic diseases (MetabERN) clinical pathway recommendations for Pompe disease (acid maltase deficiency, glycogen storage disease type II).	Parenti G, Fecarotta S, Alagia M, Attaianese F, Verde A, Tarallo A, Gragnaniello V, Ziagaki A, Guimaraes MJ, Aguiar P, Hahn A, Azevedo O, Donati MA, Kiec-Wilk B, Scarpa M, van der Beek NAME, Del Toro Riera M, Germain DP, Huidekoper H, van den Hout JMP, van der Ploeg AT	2024-11-01	Acid alpha-glucosidase deficiency, Acid maltase deficiency, Glycogen storage disease (GSD) type II, Lysosomal storage disease, Pompe disease	Orphanet journal of rare diseases

ERN	Number of Publications
ern-rare-liver	541
ern-epicare	389
ern-guard-heart	379
ern-lung	289
metabern	255
endo-ern	184
ern-rnd	150
vascern	141
ern-ithaca	108
ern-reconnet	103
ern-rita	92
ern-euro-nmd	84
ern-skin	78
erknet	71
ern-bond	69
ern-genturis	54
ern-eye	46
ern-eurogen	42
ern-cranio	33
ern-eurobloodnet	28
ernica	17
ern-paedcan	8
ern-transplant-child	8
ern-euracan	2
Total	3171

Updated with additional ERNs

ERN Clinical Trials Repository

Linked to orphanet

NIH U.S. National Library of Medicine
ClinicalTrials.gov

UNLOCKED: A Phase 2, Open-label Study to Evaluate the Efficacy and Safety of KB195 in Subjects with a Urea Cycle Disorder with Inadequate Control on Standard of Care	Endo-ERN	Clinical trial	Terminated	35878	- Hyperinsulinism-hyperammonemia syndrome	2	2019-11-13	Link
SPINET: A Phase 3, Prospective, Randomized, Double-blind, Multi-center Study of the Efficacy and Safety of Lanreotide Autogel/Depot 120 mg Plus BSC vs. Placebo Plus BSC for Tumor Control in Subjects With Well Differentiated, Metastatic and/or Unresectable, Typical or Atypical, Lung Neuroendocrine Tumors	Endo-ERN	Clinical trial	Ongoing	97287	- Bronchial neuroendocrine tumor	3	2017-03-06	Link
MOSAIC: A Phase 1/2 Study of ARQ 092 (Miransertib) in Subjects with PIK3CA-related Overgrowth Spectrum and Proteus Syndrome	VASCERN	Clinical trial	Terminated	744 530313	- Proteus syndrome - PIK3CA-related overgrowth syndrome	1 2	2017-05-16	Link
VASE: Phase III multicentric study evaluating the efficacy and safety of sirolimus in Vascular Anomalies that are refractory to standard care	VASCERN	Clinical trial	Ongoing	211277	- Complex vascular malformation with associated anomalies	3	2016-01-25	Link
A Randomised, Placebo Controlled, Double Blind, Multicentre Proof of Concept Study to Assess the Safety and Efficacy of Two Doses of VAD044 in Patients With Hereditary Hemorrhagic Telangiectasia (HHT)	VASCERN	Clinical trial	Ongoing	774	- Hereditary Hemorrhagic Telangiectasia	1	2022-07-18	Link
Effectiveness of Somatostatin Analogues in Patients with hereditary hemorrhagic telangiectasia and symptomatic gastrointestinal bleeding, the SAIPAN-trial: a multicenter, randomized, open-label, parallelgroup, superiority trial.	VASCERN	Clinical trial	Ongoing	774	- Hereditary hemorrhagic telangiectasia	3	2020-11-26	Link
NF1-EXCEL: The Effect of Lamotrigine on Cognitive Deficits Associated With Neurofibromatosis Type 1: a Phase II Randomized Controlled Multi-centre Trial	GENTURIS	Clinical trial	Ongoing	636	- Neurofibromatosis type 1	2 3	2014-10-01	Link
A Phase 2, Randomized, Double-blind, Placebo-controlled Evaluation of the Safety and Efficacy of BMS-986165 With Background Treatment in Subjects With Lupus Nephritis	ReCONNET	Clinical trial	Ongoing	536	- Systemic lupus erythematosus	2	2019-10-02	Link
SENSCISTM- ON: An open-label extension trial to assess the long term safety of nintedanib in patients with Systemic Sclerosis associated Interstitial Lung Disease (SSc-ILD)	ReCONNET	Clinical trial	Ongoing	90291	- Systemic sclerosis	3	2018-08-01	Link

Once Weekly Dosing of
With Growth Hormone

https://erica-rd.eu/work-packages/integration-outreach-dissemination/clinical-trials/

Added benefit to ERNs:

- A central platform resource for clinicians (ERN and non-ERN), to find in one place relevant ongoing RD CTs that that their patient could benefit from/ they can contribute research or participate to
- A resource for researchers to keep track of ongoing ERN research
- A single place for rare disease patients to source information on relevant research/ Clinical trials pertaining to ERNs
- Advertising & increased awareness of ERNs via the Orphanet website – ERN trials are marked as such in the Orphanet database
- Means to check data/ validate data submitted for the continuous monitoring program.

Menti time!

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https://www.mentimeter.com/app/presentation/al1mjqvukaan6euraa42wxmxq3ka64mb/edit?question=6fujdvvatm6s

Conclusions & Future Prospectives

programme