ERN ITHACA

March 2021

ERN ITHACA Projects

31st European Dysmorphology Online Meeting pre-announcement

ERN ITHACA is pleased to announce the forthcoming registration opening for the **31st European Dysmorphology** meeting which will start on **May 1st**: the meeting will exceptionally be held online this year due to the sanitary situation.

With the aim to bring young clinical geneticists and trained dysmorphologists together to share their professional experiences and present their clinical challenges; Eurodysmorpho is open to any presentation in the field of human development: large series of patients and single, illustrative case reports are equally welcome.

In this regard, young geneticists will be eligible for a special registration rate after submitting a proof of trainee document *(the template to be completed and submitted is as well available for downloading on our website).* The meeting will offer great exchanges & discussion opportunities - This is facilitated by the appreciated friendly atmosphere:

- Everyone coming to the meeting is committed to present a communication and/or an « unknown » case report.
- Abstract submission template is already available to download on the dedicated section.
- The official announcement of the registration opening and the programme will be disseminated at the end of April via our networks, and newsletters

The organising committee would also like **to warmly thank the YGNs for their inspiration and involvement** within the preparation meetings of this event.

For any further question, do not hesitate to contact the coordination team

Calls for collaborative clinical research on developmental disorders

We remind you that ITHACA coordination is still committed to share and promote clinical research and the production of cross-border collaborative work. **Through this section**, we share collaborative calls for European series of patients carrying variants in ultra-rare but already known genes. ITHACA can facilitate this type of study by connecting nearly 70 leading genetics departments.

These calls for collaborative projects typically aim to rapidly build up consistent clinical series for rare monogenic disorders, in order to better delineate the clinical spectrum and natural history of recently identified entities in the field of ITHACA. The call will be disseminated to the mailing list of ITHACA and will be posted on the web site.

If you are interested in submitting a new call for collaboration, please download the **following template**, and send it back to **Prof. Alain Verloes**, and to the **ITHACA Coordination Team.**

Work in progress

March 2021: Follow-up on our collaboration with Orphanet

We have shared a **new list of disorders** which need a newly drafted or an updated summary in the framework of our collaboration with Orphanet. If you wish to participate, please contact your institution's main representative to ERN ITHACA. Declarations of interest should be sent to the Orphanet editorial team before **April 15th**

Please find **here** this month's updates by ITHACA experts.

ILIAD registry update

The ILIAD registry project will be present at the ESHG 2021 conference with an ePoster!

ERN ITHACA is developing a “meta-registry” called ILIAD, connecting 37 HCPs, databases, and biobanks in 13 countries for patients with dysmorphic/MCA syndromes and/or intellectual disability. Through the ERN-ITHACA’s expert and patient participation network, ILIAD is able to provide an infrastructure for diagnosis, highly specialised multidisciplinary healthcare, evidence-based management, and collection of secure patient data. The registry is built on MOLGENIS open-source software, providing flexible rich data structures, user friendly data import and querying, and FAIR interfaces for programmatic data exchange. To support the interoperability, the registry is connected to the European Rare Disease Registry Infrastructure ERDRI), uses the minimal dataset of variables from JRC and uses the common Pseudonymisation Tool (EUPID) to allow the linking of RD patient cohorts. ILIAD consists of 2 components: a central, web-based registry and a network of linked satellite/client registries forming the ERN-ITHACA registry federation. Data is modelled adhering to international interoperability standards from JRC and EJP-RD. In addition to the core registry, ILIAD will include thematic sub-registries of patients with biologically proven monogenic or genomic (chromosomal) diagnoses, under the supervision of ERN-based curation teams. ILIAD has adopted a data access policy, for requesting access to the data, Governance of the Registry, compliance with applicable legal and regulatory requirements on the use of Personal Data. We are well underway to share ERN-ITHACA patient data, yielding high-quality epidemiological insights and expert consensus statements, informing policy decisions that impact RD patients in general and care for ERN-ITHACA patients in particular *(EU Health Programme Grant 947617)*.

Educational resources : Teaching & Training related video series

We have expanded the "Educational resources" section on our website with the kind support of the French health networks, AnDDI-RARES, Défiscience, and FAI²R, as well Prof. Laurence Faivre and Prof. Sally-Ann Lynch: you will be able to access the teaching & training videos on the genetics related field which are available in English here.
The section will continue to be enriched in the coming weeks, in order to structure the educational resources and categorize them by topic.

If you are interested to contribute by allowing us to relay your video content, please contact the coordination team

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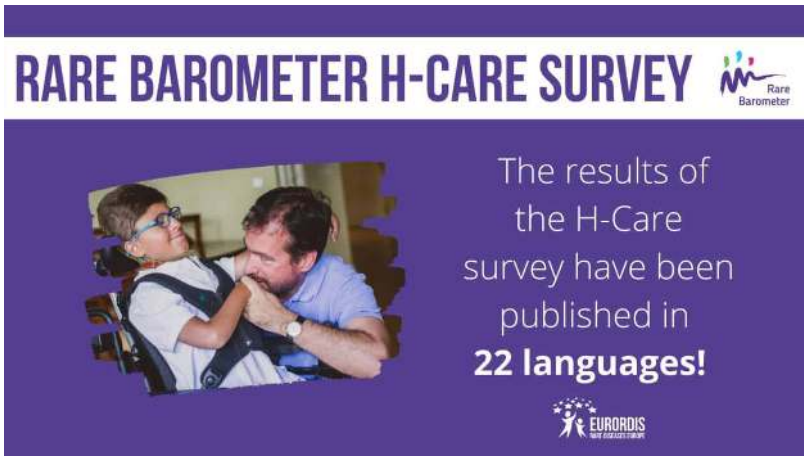
Patient Organisation

The H-Care #RareBarometer Survey

The report is now available in 22 different languages!
Check it out to learn more about the healthcare experience of people with rare diseases and their carers as well as our recommendations to improve it!



Learning from the Pandemic to Improve Care for Vulnerable Communities: The Perspectives and Recommendations from the Rare Disease Community

The This editorial touches upon the situation of people living with a rare disease during the pandemic, recommendations for holistic integrated care, and rare diseases community's perspectives on virtual care. figures and testimonials from Rare Barometer surveys have been used as well as the recommendations of Eurordis Holistic Care position paper.

Castro R, Berjonneau E, Courbier S. Learning from the Pandemic to Improve Care for Vulnerable Communities: The Perspectives and Recommendations from the Rare Disease Community. International Journal of Integrated Care. 2021;21(1):12. DOI: http://doi.org/10.5334/ijic.5812

European News

Assessment of applicants to the ERNs - Independent Assessment Body (ACSA)

The Independent Assessment Body that has been contracted by CHAFEA is Andalusian Agency for Healthcare Quality (ACSA), and they have started with performing the technical assessment of the new member applications. .

The assessment process of the applicants will take part in 3 stages:

1. **29th March – 16th April:** Uploading documentation in the ERN-Assessment Tool. During this period, you will have to upload all the additional documentation that the IAB will ask you.
2. **19th April – 30th April:** Documentation review. The assessors' teams of the IAB will perform desktop review of the information provided.
3. **12th May-09th June:** audits of selected number of applicants.

The final BoMS validation for membership is scheduled to take place in July 2021. For any question please contact the Andalusian Agency for Healthcare Quality's ERN assessment team at:

ern-assessmenttool.acsa@juntadeandalucia.es

Publication by BoMS member and 8 ERN coordinators: "To a sustainable future for ERNs"

The new article **"European Reference Networks: challenges and opportunities"** written by Birute Tumiene, Holm Graessner (ERN-RND), Irene MJ Mathijssen (ERN CRANIO), Alberto M Pereira (Endo-ERN), Franz Schaefer (ERKNet), Maurizio Scarpa (MetabERN), Jean-Yves Blay (ERN EURACAN), Helene Dollfus (ERN EYE) and Nicoline Hoogerbrugge (ERN GENTURIS) is now available.

Tumiene, B., Graessner, H., Mathijssen, I.M. et al. European Reference Networks: challenges and opportunities. J Community Genet (2021). https://doi.org/10.1007/s12687-021-00521-8

Official start of the ERICA project

The **European Rare disease research Coordination and support Action Consortium (ERICA)** project kicked off on 1 March 2021 with the aim of coordinating the clinical research activities of the European Reference Networks (ERNs).

By uniting the expertise of the 24 ERNs, ERICA will create a collaborative platform to share knowledge and good practices through the assembly of transdisciplinary research groups across the different medical areas.

ERICA is composed of 29 partners and is coordinated by Alberto Pereira, coordinator of Endo-ERN. It will run until February 2025. The first **ERICA Kick-off meeting** that will be held on **Thursday 27th – Friday 28th May 2021**

Solve-RD and the Rare Diseases Models & Mechanisms – Europe (RDMM-Europe) Network were prominently featured in the latest issue of *Lab*Animal, an imprint of *Nature Research*

The article, “Model matchmaking”, discusses the efforts to connect clinicial scientists with model organism researchers, and describes the "powerful potential" that lies in platforms for making these connections.

The article also picks up examples of scientists that have been matched by RDMM-Europe and that have received Seeding Grant funding by Solve-RD.

Please find here the link to the article.

Official AMEQUIS Project starting

The AMEQUIS project aims to develop an integrated Assessment, Monitoring, Evaluation and Quality Improvement system (AMEQUIS). The project contactor (Avedis Donabedian Research Institute) needs ERNs input to identify the strengths and weaknesses of current procedures and possible areas of improvement of the quality improvement system for ERNs.

Two forthcoming stages must be highlighted:

1. An EU survey has been disseminated among ERN coordinators and a selection of HCPs, to collect their input on the assessment, monitoring and evaluation process: 20% of our HCPs were selected by the contractor. A second EU-survey will be sent to a randomly 5 selected patient representatives.
2. On **Monday 12 and Tuesday 13th of April**, AMEQUIS research team will host an online stakeholder conference with one representative per ERN to provide input to the different steps of the quality improvement system for ERNs.

We will keep you informed about the further AMEQUIS project development through our weekly newsletters.

News from EJP RD

The ERN Research Mobility Fellowship

EJP RD has announced the **opening of the call for Research Mobility Fellowships on the 15th of March**, which aims to support PhD students, Postdocs and medical doctors in training to undertake scientific visits fostering specialist research training outside their countries of residence.
The exchange can be carried out **within the same ERN (Full Members and Affiliated Partners), between different ERNs (Full Members and Affiliated Partners) and between ERN Full Members / Affiliated Partners and non-ERN institutions.**
Either home or host (secondment) institution must be a Full Member or Affiliated Partner of an ERN at the time when the application is submitted, as well as during the proposed period of the training stay.
Successful applicants should acquire new competences and knowledge related to their research on rare diseases, with a defined research plan and demonstrable benefit to the ERN of the home and/or host institution.
The research mobility fellowships are meant to cover stays of 4 weeks to 6 months duration.
More information HERE: https://www.ejprarediseases.org/index.php/training-and-empowerment/ern-trainings/

Training on RD Biobanks: Roles in Networks and International Collaborations : **10-11 May 2021**

Registration is now open for the EJP RD supported training workshop **"Training on RD Biobanks: Roles in Networks and International Collaborations"**, online **10-11 May 2021**.
This is a part of the capacity building programs from Pillar 3, and this edition is co-organized by Hacettepe University Biobank for Rare Diseases (HUGEN), Turkey and Fondazione Telethon, Italy.
The aim of the present training workshop is to emphasize the need for rare disease biobank networks to overcome sample scarcity and to draw attention to some of the **challenges for collaborative research** but at the same time also give examples of platforms, infrastructures and national activities that are catalysing national and international collaborations. It is hoped that at

the end of this training course, lectures will highlight a checklist for the attendants as a guideline for biobank networks and research.

This beginning level training course is open to the international research community, clinicians, medical specialists, RD biobank managers, healthcare professionals and RD patient representatives.

- Preliminary program and link to registration page are available on the dedicated section.

ITHACA Upcoming Events :

- April 2th: EU Monitoring Working Group Meeting

- April 7th: EU Knowledge Generation Working Group Meeting

- April 12-13th: Project AMEQUIS (Stakeholder Conference)

- April 14th: EU Integration Working Group Meeting

- April 19- 21th: Solve RD Annual Meeting

- April 29th: ERN Coordinators Group Meeting

Call for collaborative clinical research on developmental disorders

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