

ERN ITHACA



June 2020

ITHACA Latest News

ITHACA Virtual Board Meeting : June 8th 2020

Pr Alain Verloes and The Coordination Team would like to warmly thank our speakers, all the HCPs that were represented, and welcome all our guests among the HCPs applicant. Several important topics on the progress of the work of ITHACA and its collaborations have been covered.



Daria Julkowska coordinator of the EJP-RD, presented the International Rare Diseases Consortium, which the main goal is to provide recommendations and subsequently implement them at national/international level to achieve shared objectives. The EJP RD Pillars and initiatives for a collaborative work with the ERNs are detailed in the meeting minutes.

Morris Swetz, of the MOLGENIS team updated the Board on the advancement of the ILIAD meta registry / ILIAD will be build using MOLGENIS open source software platform for 'scientific' data, a web based service. The official starting date of ILIAD as a project is the 1st of April 2020.

Raoul Hennekam, chair of WP4 of ERN ITHACA presented the Board briefly with news from the Guidelines Development WG. in ITHACA there are 3,000-5,000 ultrarare entities with insufficient data. The WG is going to look at the need for general guidelines on different topics. WP4 members will meet shortly after the Board to continue with the choice of topic and the creation of subgroups.

Julien Thevenon, the founder of **BIG** (*Bio Informatique pour la Genetique Medicale*) **MOOC** (*Massive Open Online Course*) has presented to the Board this project. The MOOC is aimed as an introduction to medical genomics, online. Next steps for this MOOC, supported by ITHACA would be building a second version, with content improvement, as well as a translation in English.

Klea Vyshka reminded the importance of the continuous monitoring procedure (in to which the participation of the HCPs is required) and its updates mentioned during the European meeting of the Monitoring WP, she also reported on the integration procedure of the new applications for ITHACA.

Alain Verloes mentioned the new communication routines for ITHACA and the procedure for disseminating calls for research collaboration, and the partnership with SOLVE RD and the collaboration with Orphanet.

You can access and download the Board presentation as well as the detailed Minutes while being connected to your Member Area by following this link: [ITHACA Board Meeting 2020](#)

Next ITHACA Board Meeting : December 11-12 2020

Our next Board will take place in Budapest, at the kind invitation of Kinga Hadzsiev. We have set up a 3-day provisional agenda, which will be finalised by July. If you are an HCP coordinator, or a Patient Council representative, you can already confirm your participation by email to the coordination team : we cover one representative per HCP and by HCP applicant.

We will get back to you in our next communications with more details on the logistics of the event.

The deadline to register with the coordination team is September 11th.

COVID-19 special Survey still ongoing up to end of June 2020



special needs COVID

“How Families with Children with Special Needs are coping with the COVID-19 Pandemic: An International Online Study”

- The survey is currently available in 13 languages,
- Participants have replied in 29 countries from Europe, Asia, North and South America, Australasia
- The total number of participants to have completed the survey is now 2597

International Online Study : specialneedscovid.org

"ePAG Impact Assessment Framework " Pilot



The ePAG Impact Assessment Framework has been developed by the ePAG Steering Committee and EURORDIS with the following objectives:

1. Develop together a clear understanding of the ePAG advocates role and the objectives for their involvement in the ERNs (learning and focus)
2. Identify the key activities that will help progress towards the identified objectives (learning and focus)
3. Explain the value of ePAG advocates involvement in the ERNs and provide measures to assess the impact of this involvement overtime (accountability)

The ePAG Steering Committee members identified [3 main objectives](#) that drive their involvement in the Networks, [10 measures](#) were developed to assess the extent in which the ePAG advocates were contributing to these objectives with their activities.

Currently 3 ePAGs are piloting the framework (ITHACA, ReConnet and RITA). The pilot is being led by the ePAG advocates in each group with the support of EURORDIS and it will run from May 2020 until December 2020. In addition to the 10 measures, each ePAG will draft an accompanying report describing their involvement and impact in the ERN meetings and calls.

The call will be held on **June 29th from 11H to 12H**. You are still welcome to register with [Dorica Dan](#) if you wish to join the call

Find all the objectives and details of the established measures by following this [link](#)

IRDiRC Nomination Call: Diagnostic Scientific Committee (DSC)

Share your expertise with IRDiRC The Consortium is seeking experts to join its Diagnostics Scientific Committee to work together to the achievement of IRDiRC Goal 1 for 2027. I- Nomination Call: Diagnostic Scientific Committee (DSC) IRDiRC has three Scientific Committees, one each for Diagnostics, Therapies, and Interdisciplinary aspects of rare diseases research. The Scientific Committees identify scientific issues common to many or all members that limit the achievement of IRDiRC goals. Among ...

[Read more](#)

Transversal cross-ERNs Study Group on Pregnancy and Family planning in Rare, low prevalence and complex diseases

The Coordinators Group of ERNs has proposed the creation of a transversal cross-ERNs Study Group on Pregnancy and Family planning in Rare, low prevalence and complex diseases. Starting from the many proposals of the patients' communities, we hope that the ERNs can address together this important topic in the agenda. For this reason, the creation of a transversal cross-ERNs Study Group ...

[Read more](#)

ERN ITHACA Projects

Advisory Ethics Committee

ITHACA is actively seeking candidates to establish the Ethics Committee of the ILIAD registry. The Advisory Ethics Committee (AEC) will give ethical advice and will oversee the registry development. It is in charge of ensuring compliance with all international and European relevant rules and ethical norms: fundamental and patients' rights, data management, protection, confidentiality.

Practically the AEC operates as follows:

1. There are 3-5 Members, invited to participate in periodical TCs.
2. The Members can be part of the ERN ITHACA HCPs or not.
3. The Members participate in annual face to-face meetings with ITHACA's ExCom. Additional meetings may be scheduled upon request of the majority of the AEC members in order to address possible specific topics.
4. The AEC prepares periodical reports.
5. The AEC revises documents and deliverables with reference to regulatory and ethical aspects upon request by Project Leaders and provides specific feedback to possible emerging issues.

At this stage, we welcome your nominations. For this purpose, please fill this form:

[Advisory Ethics Committee Proposal Form](#)

We are grateful for any suggestions that you may provide us with.

Work in progress

Collaboration with Orphanet

ERN ITHACA has a collaboration ongoing with Orphanet and hence, expertise is needed in producing Orphanet summaries.

The Coordination Team will circulate in the coming days an Excel sheet, filled with around 300 entries of the prioritized rare syndromes, which are currently outdated. You will be asked to choose from these 300 entries. ERN ITHACA will then help in identifying the working group and the leader of the writing process for these summaries.

Our Goal is to produce over 100 summaries of 3500 words long in 2020, 2 per HCP.

This should not be a very time-consuming process, due to the limited number of words required for an Orphanet summary.

News from EC :

Termination Procedure Of The HCP Membership



The coordination team has been informed by the European Commission of a termination procedure for non-cooperative HCPs initiative. This procedure will concern HCPs that do not maintain the level of expertise required by the ERN and that do not keep to their commitments by providing the data requested during Monitoring Data operations, and that are totally inactive in the network.

This procedure, which will have several stages, will initially involve mediation between the ERN Coordinator and the HCP Coordinator in order to explain the relevant points and find solutions.

If, despite this stage, the HCP continues to fail to respect its commitments and to be inactive within the network, its exclusion from the ERN may then be acted upon.

The coordination team will inform you of the follow-up given to this issue.

News from EJP RD



OPEN CALLS

EJP RD Rare Diseases Research (RDR)

Challenges Call is [open](#)

This innovative call aims at solving specific research challenges in the field of therapeutic development for rare diseases as well as at facilitate and fund collaborative projects between industry, academia, SMEs, and patient organizations. The total budget is 1.5 Mio €. The call is open to academia, clinical/public health sector, SMEs, and rare disease patient representatives from eligible countries involved in the EJP RD; deadline is 30 June at 2pm CEST. Industry partners have been involved in the identification of four challenges that are the topics of the call and that were validated by the EJP RD partners including patients. It is still possible to establish consortia via a B2B tool.

EJP RD Networking Support

Scheme (NSS) Call is [open](#)

Next applications' collection date is 1 September at 2 pm CEST. The NSS call aims to encourage sharing of knowledge on rare diseases and rare cancers of health care

professionals, researchers and patients. The NSS also aims to enable or increase the participation of underrepresented countries in Europe in new and in existing research networks on rare disease(s) or rare cancer(s). Eligible applicants are health care professionals, researchers and patient advocacy organizations from countries involved in the EJP RD.

Upcoming Events :

- **Signing and return of the ITHACA Network Agreement : 2020 July 31th**
- **Registration with the coordination team to attend the next ITHACA Board meeting in Budapest : 2020 September 11th**
- **Next ITHACA Board Meeting : 2020 December 11 - 12**

Call for collaborative clinical
research on developmental
disorders



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