



ERN ITHACA NEWSLETTER - October 2019

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Thanks to all ITHACA HCPs who sent the data tables for 2018 and first semester 2019. – Reminder, the deadline for sending this data is **25 OCTOBER 2019**

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November 2019 perspectives

The call for new members to join ERN ITHACA is open until 30 November 2019.

For more information on the call and to access all relevant documents please visit the European Commission's ERN website:

https://ec.europa.eu/health/ern/consultations/2019_call_membership_en

For any further information, please contact us at: info@ern-ithaca.eu

I- New ERN ITHACA coordination team

The last few months have been a busy period for our network. With the coordination transfer from Manchester to Paris, we had to form a new team. We recently welcomed Sarra Selatnia as the new communication manager. She will be responsible for the development of the open access public [ERN ITHACA](#) website, meetings and congresses organization, relations with PAGs, ePAGs, EURODIS, animation of social media counts and Newsletter.



She could also provide support to the coordination officer and project manager- **Rasika Sowmylakshmi**, with her activities. The coordination team will keep growing in the coming months. We will keep you informed through our network.

II- Call for European Rare Disorders Registries

The drafting of the tender was completely finalized and submitted on last October 10th. Thanks to ExCom members who sent their comments and checked the contents of the document. As a reminder, there are 4 applicant organisations (*APHP –Paris, UMCG- Groningen, OPBG-Rome, AOUS- Siena*). Summary of the project objectives:

The general objectives of the project are:

1. To enable the building and maintenance of an interoperable registry covering the diseases and conditions covered by ERN ITHACA and placing it on the European Platform on Rare Disease registration (EU RD), thus linking and making visible patient cohorts at the European level in order to follow the natural course of diseases with sufficient patient data;
2. Based on the above link with EU RD, to contribute to a comprehensive approach for rare disease registries covering the respective ERNs and following the standards and tools provided by the EU RD Platform.

Specific Objectives:

1. Set up a central ERN ITHACA registry
2. Set up a local version of the registry software that could be used by HCPs
3. Obtain patient records from several HCPs

4. Connect with other ERN registries
5. Establish interoperability of Central ITHACA Registry with at least one professional RD registry outside of the ERN network, and one patient-driven registry
- 6- Establish connection and interoperability of Central ITHACA Registry with at least one biobank renowned in BBMRI-ERIC (Biobanking and BioMolecular Resources Research Infrastructure - European Research Infrastructure Consortium)

III- Solve RD - ERN Ithaca collaboration

Solve RD

Solving the Unsolved Rare Diseases

Solve-RD is a research project funded By European Commission for five years (2018-2022). It aims to solve a large number of rare diseases for which a molecular cause known by sophisticated combined approaches and to improve of RD patients though contribution to

and participation of A Genetic Knowledge Web based on the sharing of Knowledge on genes, genomic variants and phenotypes. Four ERN form the clinical core of Solve-RD due to their unsolved RD cohorts diagnostic research expertise and infrastructure, and ERN ITHACA is a part of this collaboration and focusses on rare congenital malformation syndromes and intellectual disability. The following ERN-ITHACA members are involved in and contributing to Solve-RD:

Collaboration of the Network is warmly expected for two cohorts:

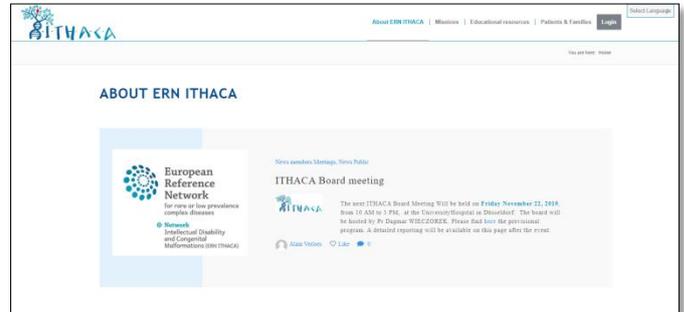
Ultrarare, undiagnosed patients. Solve-RD offers the possibility to study by WGS one trio per HCP. The patient must have a very distinctive, unique phenotype. A call has already been disseminated by mail earlier this month.

Unsolved known syndromes. ITHACA and Solve-RD have listed several disorders that are not explained by a "simple" mendelian" mechanism. The procedure will be in three steps:

1. Establish a cohort of bona fide patients through the network and evaluate them in a TC
2. Select the 3 "best" patients (new sampling in trio will be necessary) for multiomics investigation
3. Keep the reminder as a replication cohort

V- ERN – Ithaca website updating

The new version of the [ERN ITHACA](#) website is currently being developed and will be soon available. All members will be invited to login at the area provided for this purpose so that all data relating to each HCP can be updated instantly.



November 2019 perspectives:

9TH ERN Coordinators Group meeting: 14 – 15 November 2019 – European Commission
Ithaca Board Meeting – Düsseldorf : 22 November 2019 - University Hospital Düsseldorf

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