ERN ITHACA

October 2020

ITHACA Board Meeting
December 11-12 2020
PC meeting on December 10th afternoon

Ithaca Board Meeting registration here

The ITHACA Board Meeting plenary session program **(December 12, 2020 from 9:00 AM to 1:00 PM)** is almost finalized. An update will be communicated to all our members and partners during the month of November.

Please note that there are currently a few Working Groups without a finalised program, but discussions are on-going. Your referent ITHACA Project Manager will remain at the disposal of the Working Groups Chairs in order to finalize the program of your sessions during the WGs satellite meetings dedicated to the deliverables of the next two years.

This will allow us to have a better visibility on the organization of the Board and the programme to be established, registrations are now open and possible until 30 November 2020. Please Click on the Red Button above to register : At the beginning of December, we will send the connexion links to all registered attendees and WGs Members.

ERN ITHACA Projects

Latest Updates on APOGeE Project : A Practical Online Genetics e-Education

In our last newsletter, we mentioned the context in which the APOGeE project was set up ***(CEF Telecom call for proposals dedicated to e-health developments in ERNs)***, its targets and the main guidelines.

We continue to progress on the project's implementation by setting up and finalizing a Steering Committee with representatives from several European countries among our partners. The Editiorial content is under construction and will be supervised by an Editorial Board.

More concretely, the APOGeE e-learning project aims to become a reference site for medical genetics when necessary, for a specific or a more general learning purpose and an asynchronous interaction with the Editorial Board via a monitored blog. During the plenary session of the next Board meeting of ITHACA, the course of the project will be developed and explained. In the meantime, the construction of the project in its scientific and technical aspects continues with the Steering Committee finalized this month.

Eighth European Course in Clinical Dysmorphology & Eurodysmoclub : October 2020

The Eighth European Course of Dysmorphology and Eurodysmoclub is characterized by the presence of speakers who will address the question of who are the main experts in medical genetics: hence the name “What I Know Best”. The 8th edition was held on the 1st, **2nd and 3rd October 2020** and was partly sponsored by the **ERN ITHACA**, which was presented by **Pr Tjitske Kleefstra** during the first session of the course (October 2nd).
In this new edition, the latest genetic conditions were discussed, identified thanks to the support of the most recent technological advances, such as the introduction of new generation sequencing methods.
As a reminder, the eighth European Clinical Dysmorphology Course and the Eurodysmoclub are two consecutive events :

- The course “What I know best” aims to give participants an update and overview (genetic basis, diagnostic criteria, guidelines and natural history information) of selected rare genetic diseases, selected by the Scientific Committee, including newly identified conditions consisting of intellectual disabilities, epileptic encephalopathies and neuropsychiatric disorders.
- The Eurodysmoclub represents a unique opportunity to bring the discussion of scientifically interesting clinical cases to the attention of experts from all over the world.

The whole presentation will be soon available on our website ern-ithaca.eu

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Work in progress

Collaboration with Orphanet :

As a follow-up on our collaboration with Orphanet, you will find below the current month publications by ITHACA experts on the syndromes listed below:

- **Mowat-Wilson syndrome** : ITHACA Experts - Dr Stefano Giuseppe CARAFFI
- **Hypertelorism, Teebi type** : ITHACA Experts - Pr Alain VERLOES
- **Tall stature-intellectual disability-renal anomalies syndrome** : ITHACA Experts : Pr Laurence OLIVIER FAIVRE

News from ILIAD registry

The ILIAD registry organised its first open Steering Committee on October 7th. The purpose of this meeting was to invite all the relevant stakeholders to the discussion, with the main aim of presenting the legal and regulatory set-up of the national authorisation procedure and inter-institutional agreements that participating HCPs will need to fulfil. The registry prototype can be accessed on demand and members of ERN ITHACA that wish to test the prototype are kindly requested to reach out to Klea.

Some legal and regulatory points of the meeting:

- *Before the registry becomes functional*

In a nutshell, the steps that HCPs need to take in order to be able to enter patient data in ILIAD (only on a regulatory point of view) are:

Step 1: To be able to add patient data in the ILIAD Registry, a form of national authorisation is requested (the type may vary from country to country).
Step 2: The HCP must enter into an institutional agreement (usually this is a Data Transfer Agreement) with the owner of the database (UMCG).
Step 3: Start adding patients.

In the near future, the ITHACA Coordination team believes that an implementation of a “federated method” of requesting national authorisation seems as the easiest solution. For this, the “leader” of the national consortium enters into the institutional agreement with UMCG, representing its

peers. Potential bottlenecks with federal and countries organised in a regional basis (Germany, Italy), but otherwise the system will work for the rest of Europe.
Volunteer ERN ITHACA main representatives may step forward starting from this moment, if they wish to lead the national authorisation procedure. Otherwise, the Coordination Team will follow-up very soon with more details on this regard.

- *After the registry becomes functional*

The Data Access Policy will become available in the Members Area of the ITHACA website. This document covers the process that will be followed for requesting access to the data in the ILIAD Registry, the governance of the Registry and ensures compliance of the project with applicable legal and regulatory requirements and specifications on the use of Personal Data in the Action.
Data will be accessible by approved members (definitions in the DAP), or alternatively accessible after formal approval by the SC. The access to biological specimens through connected biobanks requires SC approval + local biobank approval. In every case: only anonymised data can be accessed.

Individual researchers can request data stored in the ILIAD registry for research purposes, but they and their project is vetted by the Steering Committee before being granted access: through the Data Search Form (annex to the DAP) and through the Data Sharing Protocol (annex to the DAP).

Patient Organisation

Developed Patient Journeys within ITHACA:

In collaboration with their patient communities, the ePAG advocates of ERN-ITHACA have set up five patient journeys. These journeys provide a common perspective on the needs and care that patients would like to receive throughout the various stages of their journey - from early symptoms to diagnosis, post-treatment follow-ups. Using this tool, ITHACA patient advocates and clinicians can collaborate to identify gaps in care services, adapt care pathways to more effectively deal with the needs of patients who are living with these diseases, and discuss relevant outcomes and challenges at various stages. Based on this information, the ERN will be able to develop collaborative initiatives based on a better understanding of the care needs of patients that our community is serving.

Patient journeys are also a useful reference for patients, families, and non-expert public and clinicians to understand the care needs that persons living with these diseases will encounter

throughout the various stages.

5 Patient Journeys have been developped on the following rare diseases/syndromes:

- **Rett Syndrome**
  Patient representatives, patients and their families from the United Kingdom contributed to the development of this patient journey.
- **Williams Syndrome**
  6 patient and their families representatives from Hungary, Italy, Belgium, Germany and UK have contributed to the development of this patient journey. Moreover, several patient representatives of the EU WS Federation are involved on the development of new Clinical Guideline for William's syndrome.
- **Prader Willi Syndrome**
  2 patient representatives, patients and their families from Romania, Denmark and 2 clinicians from ERN-ITHACA, have participated on the development of this patient Journey.
- **Spina Bifida**
  Several patient representatives, patients and their families from the *International Federation for Spina Bifida and Hydrocephalus* have contributed to the development of this patient journey.
- **Pitt-Hopkins Syndrome**
  Seven patient representatives, patients and their families across five EU countries (United Kingdom, Ireland, Sweden, Italy and the Netherlands) as well as ERN-ITHACA clinicians have contributed to the development of this patient journey.

Based on these individual patient journeys, ePAG advocates within ERN-ITHACA were able to identify the commonalities of these five rare diseases/syndromes. These common needs were included in a position statement which was presented at the ECRD congress on May 2020.

News from the EC

EU4Health Programme : 2021-2027

The keeping of the global european health budget at 9.4 billion for the years 2021 to 2027 is on track : Amendments contesting the initial reduction of the budget (from 9.4 billion as proposed by the Commission to 1.5 billion as voted in the Council) have been drafted by the European Parliament, which strongly supports the continued existence of ERNs.

In accordance with the following document, financial support for ERNs is encouraged by the European Parliament on the European Commission's proposals, not only by maintaining financial support for the 24 existing ERNs, but also by the creation of new ERNs covering infectious diseases, complex pregnancies and rare and complex mental health diseases.
The final vote in the European Parliament will be held on November 2020. Then, it will be up to the Council of Ministers to vote on the Parliament's proposal.

Orphanet database's financial support, which was uncertain until now, is also strengthened, which is very good news for the ERN community, in order to allow the continuity of the partnership in a sustainable way and under good conditions.

More informations on the European Parliament article

ERN Monitoring: Data Collection : 1st semester 2020

We would like to thank all our HCPs who responded to the Monitoring Data Collection Campaign for the 1st semester 2020.

We are well aware that for some HCPs it was not an easy exercice considering the sanitary situation. However it is a very important exercise to analyse and evaluate the added-value and the impact of the ERN system and we have to report to the European Commission.
Even if the submitted data is incomplete, you will have the opportunity to correct/complete them in few months via the coordination team when the platform will be re-opened by the Commission.
The Commission needs to evaluate the data collected on a first stage, in order to refine the indicators to be completed and adapted as much as possible to the HCPs activities on the field.

Submitted H2020 proposal : Positive EC evaluation on the ERICA Project

(European Rare disease research Coordination and support Action)

The ERN Research Working Group, has received a positive evaluation from The EC on the submitted ERICA Consorsium : the evaluation phase having been completed, The grant preparation phase has now been initiated.
As a reminder, the aim of the ERICA consortium, in which all 24 ERNs take part, is to build on the strength of the individual ERNs and create a platform that integrates all ERN’s 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 ERN's innovation potential; Through integration of ERN research activities, outreach to European research infrastructures to synergistically increase impact and innovation ERICA will strengthen the research and innovation capacity of the ERNs. This will result in safe, accessible and efficient access of therapies for the benefit of patients suffering from rare diseases and conditions.

News from EJP RD

The Research Mobility Fellowships call :

The call is open from the 1st October until the 13th November

The call for Research Mobility Fellowships aims to financially support PhD students and medical doctors in training affiliated to ERN Full Members or ERN Affiliated Partners to undertake short scientific visits (secondments) fostering specialist research training outside their countries of residence and within one of the ERN host institutions. Applicants who will receive fellowships for Research Mobility should acquire at their host (secondment) institution new competences and knowledge related to their research on rare diseases and with benefit to their ERN.
Fellows will be selected taking into consideration several elements such as:

- Relevance and impact of the project, for the fellow, home and host institutions and the ERN as a whole
- Quality of the research proposal
- Organization and proposed methodology of the training
- Relevance of timelines and of required resources and budget

Research mobility fellowships are meant to cover 4 weeks to 3 months. The exchange will be accomplished exclusively within member institutions of the same ERN or between member institutions of different ERNs.

Fellowship exchanges can only be facilitated between ERN full members or ERN affiliated partners. Their travel and accommodation expenses will be covered, up to fixed maximum amounts.

Applicants/Application profile:

- PhD students with a minimum of one year of research experience OR physicians having finished their first year of specialist training
- Be affiliated to an ERN Full Member or to an ERN-Affiliated Partner Institution from one of the 24 **ERNs** at the time when the application is submitted, as well as during the proposed period of the training stay
- The host (secondment) institutions must be Full or Affiliated Member of an ERN at the time when the application is submitted, as well as during the proposed period of the training stay
- Added value to ERN of the mobility stay

Please find below the relevant documents for the application:

Call text

Template-letter-of-acceptance by HOST Institution

Template-letter-of-support by HOME Institution

For further information please contact the Heidelberg and Nijmegen coordination teams :

fellowships@erknet.org or genturis@radboudumc.nl

Joint Transnational Call 2021:

The JTC 2021 will have for specific objective to promote multinational project on Social sciences and Humanities (SSH) Research to improve health care implementation and everyday life of people living with a rare disease.

Transnational research proposals must cover at least one of the following areas :

- Health & social care services research to improve patient and familial/household health outcomes
- Economic Impact of Rare diseases
- Psychological and Social Impact of Rare diseases
- Studies addressing the impact/burden of the delay in diagnosis and of the lack of therapeutic intervention.
- e-Health in rare diseases: Use of innovative technology systems for care practices in health and social services

- Development and enhancement of health outcomes research methods in rare diseases
- Effects of pandemic crisis and the global outbreak alert and response on the rare disease field, and the emergence of innovative care pathways in this regard.

The call is scheduled to open in December, with a pre-proposal submission deadline in February.

Click on the following link for more information

Upcoming Events :

Internal ERN Coordinators Group Meeting : November 4th 2020

ERN ITHACA Board Meeting :

- **Patient Council Meeting : December 10th**
- **Work Groups Satellite Meetings : December 11th**
- **ERN ITHACA Board (Plenary session) : December 12th**

Call for collaborative clinical research on developmental disorders

ERN ITHACA
Department of Genetics
APHP Nord-Paris University,
Robert DEBRE Hospital, Paris

http://robertdebre.aphp.fr/
+33.1.87.89.16.50