ERN ITHACA

May 2020

New Initiative from ERN ITHACA!

Dear friends,

One of the missions of the ERN ITHACA is to promote clinical research and the production of cross-border collaborative work. Our field is confronted with an extremely large number of new genes or disorders, for which the clinical spectrum and/or natural history is often barely known; usually, the number of patients published is limited to a handful, their recruitment may be strongly biased towards a clinical sign, and the clinical description is often limited.
We are therefore launching a new initiative in ITHACA: collaborative calls for European series of patients carrying variants in ultra-rare but already known genes. The main challenge is to recruit cohorts of patients carrying variants in rare genes, where gathering at a single site does not yield significant series.

ITHACA can facilitate this type of study by connecting nearly 70 leading genetics departments.

The principle behind this initiative is simple:

- Any ITHACA member can submit a call for collaboration on a gene, a group of functionally related genes, a CNV...
- The call is forwarded to the entire network and remains visible for 6 months here - you have to be registered to access
- We will limit calls to 2 genes per applicant per year (can be more than 2 per HCP, of course !)
- These calls are not necessarily based on fundamental or translational research, but assume that the project leader already has at least 2 or 3 families to start the work.

- The project leader is committed to bring this work to completion (one publication), and to keep the ITHACA ExCom informed of the progress of the work.
- ITHACA's role as facilitator will be highlighted in the acknowledgements in the article.

This type of article could be furthermore entrusted to young geneticists (as part of our collaboration with the European group).

You will find the submission template on the ECP Platform, please complete it according to the established criteria and send it back to : alain.verloes@aphp.fr and coordination@ern-ithaca.eu

We are looking forward to collaborating with you!

Pr Alain VERLOES
General Coordinator

Call for collaborative clinical research on developmental disorders

ITHACA Latest News

ITHACA has recently welcomed a new project coordination team :

Anne Hugon :

Anne joined ITHACA in march /april 2020 as Project Manager & Coordination Officer in charge of supervision of the project. She has both professional and a lifelong personal Involvement in advocacy for RD patients 'rights, access to diagnosis and quality care. She is more specifically in charge of the relations with EU partners (ERNs, EJPRD…), CPMS helpdesk, ITHACA's patient-oriented commitments across WPs, and Patient Council.

SANOFI INVITÉ - ANNE

Anne has a background in Strategic Management of Health and Medical Services and a specialty in Documentary Resources Management and Databases. She has previously worked for *DéfiScience*, the French RD Network for ID and she is involved in health institutions and

EURORDIS. To contact her: anne.hugon@aphp.fr

Klea Vyshka: Project Manager

A young and dynamic professional of the EU, Klea has a background on EU law and European studies, alongside her special interest for academic research over various EU policies. Due to an early implication with the European Reference Networks, Klea has developed strong basis on EU project management and hopes to bring aboard her insight and expertise on the European Union.

She will be in charge mainly of the relations between ITHACA and the EU and the implementation of the ILIAD registry, relations with Solve RD and EJP-RD, budget supervision, monitoring of WP groups. To contact her: klea.vyshka@aphp.fr

Sarra Selatnia: Communication Manager

Sarra is in charge of the oversight of disseminated data in the open access public ERN website, relations within Ithaca Network, dissemination across the ERN network, newsletter, animation of social media networking, integration with Orphanet, and in charge of certain technical aspects of IT developments.

European Conference on Rare Diseases (ECRD) 2020 moves online!

The European Conference on Rare Diseases & Orphan Products (ECRD) is recognised globally as the largest, patient-led rare disease event. People living with a rare disease have the right to reach their highest potential of well-being. Join ECRD 2020 online to take part in discussions to inform and build the future ecosystem for rare disease policies and services. The Conference …
Read more

Fragile X Workshop - October 8-9th, 2020 - Paris, France

In collaboration with the Scientific Board and in partnership with DéfiScience (the National Network for Intellectual Disabilities), the Fragile X France association is proud to announce the next international workshop for researchers & clinicians dedicated to the Fragile X Syndrome and premutation associated conditions. This collaborative working meeting will aim to connect professionals working on Fragile X syndrome in order …
Read more

ERN ITHACA Projects

Survey Special Needs COVID pandemic

ITHACA supports this initiative by helping to desseminate a call for participation in a study targeting patents of people suffering from neurodevelopmental disorders (autism, intellectual disability) and theimpact of eviction or confinement measures Many projects are emerging in this difficult period of the COVID pandemic. ITHACA has been contacted by the University of Freiburg, Switzerland, in order to disseminate …
Read more

SOLVE RD

On the 5-6 March 2020, Solve RD held its annual meeting in Barcelona. During the DITF ITHACA Break Out Session the participants and DITF leads updated the members on the state of play.

Solve RD next data freezes for data processing of the unsolved cohort are set on the 2nd of September 2020 and on the 3rd of March 2021.
Within Solve RD, ERN ITHACA promised around 5000 unsolved exomes/genomes. But where does ITHACA stand today, in relation to the promises made? For a short update, the annual meeting showed that we are reaching the promised numbers at the start of the project, which marks good news!
If you wish to submit cases from the unsolvable cohort of Solve-RD, please use the procedure mentioned in the Call for Collaborations . These submissions will not impact the set quota for individual submissions.

Work in progress

Registries and ILIAD :

April 1st marked the official start of the ILIAD project. The registry coordination team is composed by: Alessandra Renieri (WP6 lead), Alain Verloes (ERN ITHACA coordinator), Jill Clayton-Smith (ERN ITHACA past coordinator), David Koolen (Clinical geneticist in Radboud University, Nijmegen), Sylvia Huisman (Amsterdam Expertise Center for Developmental Disorders and contact person for the Waihonapedia project), Carole Herman (ePAG). Currently, the coordination team is preparing the consortium agreement for the project implementation. Interested and thrilled by this initiative? Please contact Klea Vyshka to get involved!

Expert Recommendations

Members of WP4 on expert recommendations have been contacted through the WP's chair, Raoul Hennekam. The aim of this WP is to create expert recommendations for a series of common problems in individuals with ultrarare entities, for which it will be impossible to make syndrome-specific guidelines. For instance, reflux, constipation, seizures, scoliosis, flat feet are examples, but also transition of care, cancer treatment, aggression, and sexual education. If you think that this project sounds interesting and you would like to get involved, please contact Klea Vyshka

New Orphanet summary reviewed by ITHACA

Noonan Syndrom is a rare, highly variable, multisystemic disorder mainly characterized by short stature, distinctive facial features, congenital heart defects, cardiomyopathy and an increased risk to develop tumors in childhood This syndrome has just been codified on Orphanet (ORPHA:648)

and a summary revised by Pr Alain Verloes for the ERN ITHACA was published in April 2020. Follow the link to …

Read more

ITHACA Website :

Many thanks to all the HCPs which have responded to our call for updating their page on the website, and invite others to send us your updates using the following template: HCP Marseille with a picture of your institution, your city and the local coordinator as well.

Our network will soon be expanded with new members, and we will integrate the files of these new members on the same model.

You can send your information to coordination team and Sarra Selatnia

The ERN ITHACA website is still evolving and is updated with new pages and new sections: Eurodysmorpho

We encourage you to register to be informed of our latest publications.

News from EJP RD

EJP RD Rare Diseases Research (RDR) Challenges Call is open

This innovative call aims at solve 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

Collection date for evaluation of 2 June has been cancelled due to Covid-19 outbreak; next collection date is 1 September at 2 pm CEST. The NSS call aims to 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 usually 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 the countries involved in the EJP RD.

Training Workshops

Quality assurance, variant interpretation and data management in the NGS diagnostic era :

Registration for this course is now open, deadline is 20 July. The course will take place in Istanbul, Turkey on 12-14 October. A mitigation plan due to Covid-19 is in place. The aim is to teach participants about the evaluation of the pathogenic nature and clinical significance of genetic variants, on the criteria that have to be set to the NGS analysis pipelines and on the use of international databases. The targeted audience is: Laboratory scientists (EBMG registered), junior scientists, clinical geneticists, other medical specialists in training, policy makers and assessors for laboratory accreditation, and patient representatives, with a basic knowledge in biology or medicine. To ensure active participation and exchange with teaching staff and fellow participants, a maximum of 35 participants will be admitted to the training course. There are #2 travel fellowships available, participants from EU-13 countries are encouraged to apply. The workshop and registration are free of charge.

Implementing Biomedical Research Projects: The Complete Workflow from Concept, ELSI and Privacy Considerations to High-Quality Biobanking

Registration for this course is now open, deadline has been extended to 27 April. The course will be online, 11-14 May from 2 pm - 5.3 0pm each day. This workshop is aimed at biomedical researchers, medical professionals, and biobank managers who want to organise biomedical research projects on human biological samples. In two modules there will be used several use-cases to address the key issues in biomedical research involving human subjects, human biological samples and associated medical data. Participants from EU-13 countries and patient representatives are encouraged to apply. The workshop and registration are free of charge

EJPRD WP

In the context of the EJP RD, the WP16 (coordinated by the French Foundation for Rare Diseases) aims to provide an EU-wide education program on transversal aspects of rare diseases (RD) research. This course will be fully available online to all interested stakeholders.
Following a wide consultation and benchmark, five priority modules have been selected and will be developed with experts, in particular with representatives of ERNs.

The teaching material will focus on the cross-cutting aspects of research in all rare diseases.

The definition of the content and contributors of the first two modules (Module 1 - Diagnosis & rare diseases and Module 2 - Innovative therapies & rare diseases) has been initiated and is underway in collaboration with representatives from ERN ITHACA, GENTURIS, MetabERN and TransPlantChild.

The course is now in the development phase with the renowned platform FutureLearn (United Kingdom) French Fondation for Rare Disease

The first module to be developed on Diagnosis should be finalized by October and a first session to be launched online in early Winter. It is under the supervision of Laurence Faivre, Dijon for ITHACA, and Chrystelle Colas, Paris for GENTURIS.

Contributors are currently recruited and the content is being produced. The main topics to be addressed within this module will include : basic principles of a diagnostic consultation; analysis and interpretation of diagnostic results; current available and new tools and techniques when researching a diagnosis; current research developments in the field of RD diagnostic research; research avenues to cope with post-diagnosis situations.

Follow the link to find out more about the WP16

Upcoming Events

- European Conference on Rare Diseases (ECRD) : May 14 - 15
- ERN ITHACA virtual Board Meeting : June 8 - 9
- ERN Coordinators Meeting : Brussels, June 29 - 30

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

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