



September 2020

Next ITHACA Board Meeting : Additional Information

December 11-12 2020

PC meeting on December 10th afternoon

As we informed you in the last Newsletter, the ITHACA Board meeting will remain in virtual format this year. However, this still requires careful organisation in order to have high-quality interactions. The program will be refined over the weeks, **and it will be available in its entirety on the website ern-ithaca.eu.**

On December 11th, the parallel sessions of each WP will be organised before the plenary session scheduled on December 12th. The coordination team remains at your disposal to help each chair to organize a working session with its WP in order to:

- **Assess actions in progress**
- **Prepare ITHACA's Proposal for the renewal of the ERN system**
- **Prepare action plan for the last two years of the ERN**

For this purpose, please contact our PMs ([Anne](#), [Klea](#) and [Sarrah](#)) to define your topics of discussion, and the definition of the action plan of your WPs for the next two years.

We remain at the disposal of the WP chairs to organize preparatory meetings on Zoom starting September. We also would like to ask each HCP coordinator to inform us about his representative during the Board meeting. The list of attendees will be needed to organize the plenary session and the satellite meetings of the working groups.

The coordination team provides you below with a registration link for the next ERN Board meeting, the link is accessible and open to all HCP members, to the representatives of applicants HCPs, affiliated partners, and HUBs - and to our partners (Solve RD, EJP RD...).

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 free until 15 November 2020. The link will be communicated to you again in the October 2020 Newsletter and disseminated to all our network by mailing.

**Ithaca Board Meeting
registration here**

ERN ITHACA Projects

Moodle E-learning Project : Building APOGeE within a Moodle platform

Overall, there is a need to improve knowledge and understanding of the genetic content related to RD. A study carried out through an online survey to define the needs of the genetics research community and to inform them about online genetics courses: Out of a total of 251 participants, 95.6% were very interested in genetics education, while 87% of them had already done research prior to their participation. Among the results of the survey were reported difficulties in finding information on genetic testing, disease pathogenesis, gene function and scientific progress, as well as psychological needs (e.g. to reduce emotional burden) [The study is available Here](#) (*Orphanet Newsletter : August 2020*)

In this way, and within the framework of a **new CEF Telecom call for proposals dedicated to e-health developments in ERNs**, we have the opportunity to finance the development of one or more websites built with the open-access platform Moodle moodle.org.

The call for proposals covers very specifically interactive e-learning projects.

For this purpose, ITHACA is launching the APOGeE (A Practical Online Genetics e-Education) project. This project will involve the whole ITHACA network. APOGeE could be ITHACA's main contribution to one of the ERN development objectives, i.e. to develop and contribute to a structured programme of post-graduate education and training for ERN health doctors and affiliated partners in the service of ERN-specific knowledge generation.

The aim of this project is to establish a free and open access interactive training source in medical genetics: learners will be able to access an interface offering them different blocks of e-learning.

The content will be coordinated by an editorial team, and will be able to benefit from documents and courses already available in our network.

A few details on the target audience of this project:

1. Doctors in training in genetics and those in other specialities who are interested in certain chapters of genetics.
2. Candidates for the European examination in medical genetics and genomics of the Medical Genetics Section (MGS) of the [European Union of Medical Specialists \(UEMS\)](#) and [the European Board of Medical Genetics EBMG](#). This examination, which issues a European certificate, is aimed at European countries (and for some of them it is one of the obligations of the national curriculum) and doctors from third countries.
3. Doctors in training in genetics in Asia, Africa or South America, who would have access to a free university teaching tool.

The APOGeE project is one of ITHACA's major projects for the year 2020 : The coordination team will keep you informed of its progress in the next Newsletter. You will also be regularly informed via the information mailings that we send out at each new stage of the project.

31th European Dysmorphology Meeting - September 2021

Following the general consultation that has been launched with the Organising Committee and the Scientific Coordination team on the proposed changes to the logistics and content of the annual Eurodysmorphic meeting, we can now announce , with the agreement of all participants, that a final session will take place at the Bischenberg Conference Centre (near Strasbourg).

This annual meeting will take place from **22-25 September 2021** (*departure on 26 September*).

Starting 2022, the organisation of this meeting will be integrated within the framework of ITHACA's education and training objectives. Indeed, from the year 2022, the meeting will be held annually on September in a different location to be chosen among the ITHACA members, with the following pre-established criteria :

- 1- Easy access to the city centre and the conference centre (near an international airport).
- 2- Attractive accommodation and organisational costs, with the collaboration of ITHACA members.

Concerning the conclusions about the new meeting format that has been decided:

Day 1 (Thursday): Educational sessions with distinguished guest speakers (sponsored by ITHACA)

Day 2 and 3 (Friday and Saturday): usual oral sessions with communication from each participant. All practical information and the program will be available on our website: <https://ern-ithaca.eu/eurodysmorpho/> . The logistical organisation for next year meeting will start in January 2021, the abstracts will be submitted between January and the end of June 2021.





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:

- [22q11.2 duplication syndrome](#) : ITHACA Experts : Dr Tiffany BUSA and Pr Nicole PHILIP
- [22q11.2 deletion syndrome](#) : ITHACA Experts : Dr Tiffany BUSA and Pr Nicole PHILIP
- [Kabuki syndrome](#) : ITHACA Expert : Pr David Geneviève (*English and French versions*)
- [Nasopalpebral lipoma-coloboma syndrome](#) : ITHACA Expert : Pr Laurence OLIVIER - FAIVRE
- [KBG syndrome](#) : ITHACA Experts : Pr Nicole PHILIP and Dr. Florence RICCARDI

Collaboration with European Medical Education Initiative on Noonan Syndrome

We are pleased to announce our collaboration with the [European Medical Education Initiative on Noonan Syndrome](#). This Initiative is comprised of a group of experts from different specialities across Europe and aims to raise awareness of this rare disorder, as well as related conditions, with the ultimate aim of improving patient care. The Initiative was supported by an unrestricted grant from Novo Nordisk Europe A/S; Novo Nordisk will have no influence on any of the scientific content or material that will be generated as part of this project. In order to assess disease management across Europe and identify gaps in current clinical practice, the Initiative has compiled a clinical practice survey on the diagnosis and management of diseases within the Noonan Syndrome phenotypic spectrum targeted at clinical geneticists, paediatric endocrinologists and paediatric cardiologists.

To contribute to this Initiative, please complete the **clinical practice survey** at the following link:

https://www.surveymonkey.de/r/EMEI_NoonanSyndromeSurvey.

The survey should take ~15–20 min to complete. Please note that this survey will be closed on 25 Oct 2020. Thank you for supporting this important Initiative!

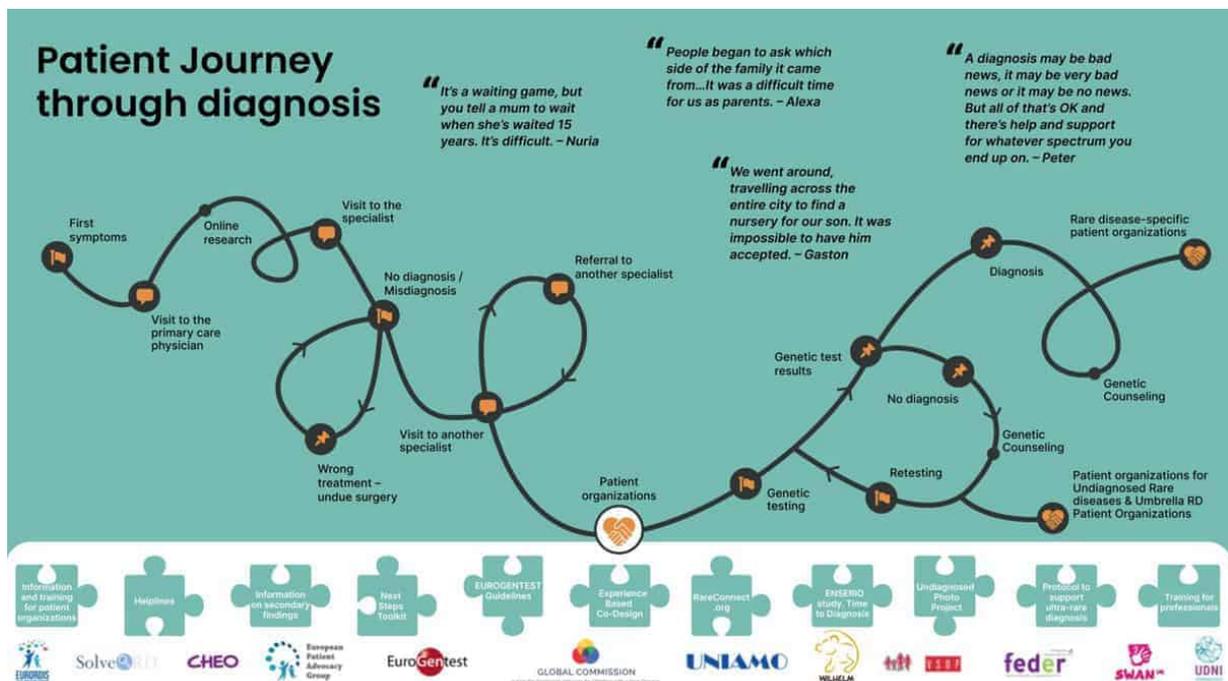
Reminder : Call for Collaborative Clinical Research on Developmental Disorders

We remind our partners that the coordination team is still available to relay calls for collaboration for European series of patients carrying ultra rare but already known gene variants. ITHACA can facilitate this type of study **by connecting nearly 70 leading genetics departments**. All information and condition are available by clicking All information are available at the end of this newsletter by clicking on the button: **Call for collaborative Clinical Research on developmental Disorders**.

Patient Organisation

Solve-RD infographic on the patient journey to diagnosis :

A research project funded by the European Commission, aiming to solve large numbers of rare diseases for which a molecular cause is not known yet. To date, the [Solve-RD Project](#) is analysing 8,463 datasets (8,152 whole exome sequencing and 311 whole genome sequencing datasets) including datasets from 5,205 individuals from 4,862 families.



The project has already solved 130 rare disease cases for which a molecular cause was not previously known.

Within the Solve-RD project, EURORDIS initiated the Community Engagement Task Force (CETF - a multi-stakeholder community of patients, scientists and clinicians to support the needs of undiagnosed and recently diagnosed patients and leave a legacy of a strengthened undiagnosed community. The EURORDIS-led CETF has created an infographic setting out the patient journey to diagnosis. The infographic demonstrates the diagnostic odyssey many people experience on a daily basis and presents existing resources from CETF member organisations to support patients on this journey. **The infographic is now also available in French, German, Dutch, Turkish, Czech, Spanish, Italian, Portuguese and Swedish**

News from the EC

ERN - COI Document and Disclosure Form

The European Working Group on "**Legal & Ethical issues and relations with Stakeholders**" has developed 2 documents that may interest each HCP main representative :

- ERN Policy Document Managing conflict of Interest [here](#)
- ERN Disclosure Form [Here](#)

Follow-up on the application procedure to join ITHACA

The examination of applications to join the existing European reference networks has been considerably delayed due to the emergence of the COVID-19 pandemic and only restarted on 1 September 2020. The ERN ITHACA Board has therefore finally made its final decision on all membership applications.

Applications were subject to a double peer review by the ITHACA Executive Committee (by members from outside the applicant's country) and their revisions were then approved at a meeting to which all members of the ITHACA Board (who had access to the applications) were invited, including representatives of the Patients' Council.

Applicants who unfortunately received an unfavourable opinion on their applications have until **15 October to amend and modify their applications accordingly**, for a final re-submission. For this purpose, the European Commission has made available an [applicant user manual](#) , and the procedure for re-submission of applications is specified on pages 17-21.

All decisions adopted by the ITHACA Board are duly justified and can be consulted on the individual evaluation form for each applicant, which is available on the HEALTH ERN HCP Applications platform.

News from EJP RD



Joint Transnational Calls 2021

The general aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear future benefit for patients.

Topic: 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, which are equal in relevance for this call:

- **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. There will be a **two-stage submission procedure** for joint applications: pre-proposals and full proposals. The call is scheduled to open in December, with a pre-proposal submission deadline in February.

For general questions regarding the joint call please contact the Joint Call Secretariat at :

JTC2021@ejprarediseases.org

Or to the following 2 individuals contacts :

Diana Désir-Parseille : diana.desir-parseille@fondation-maladiesrares.com

Laura Benkemoun : laura.benkemoun@fondation-maladiesrares.com

Patient Partnerships



[The EJP RD Short guide on patient partnerships in rare diseases research projects](#) aims to encourage fruitful, sustainable and enduring partnerships between scientists and patient organisations, co-leading the way for systematic patient-centered research.

“Collaboration requires an effort from both sides. The more we understood each other’s goals, the better the communication and the collaboration became.”

Veronica Popa, MCT8-AHDS Foundation, Romania

Providing definition, examples, testimonials of patient partnerships, describing its benefits, preventing common pitfalls and accompanying applicants, this guide will **support applicants to describe the role and added value of patient partnerships in research proposals**.

This guide was developed with the help of a working group led by [EURORDIS](#) comprising of patient representatives and research funders and reviewed by independent academic researchers. Ultimately, the guide will foster a partnership culture and contribute to an improved understanding of the added value of patient engagement and involvement in basic, pre-clinical, translational and social research for the Rare Disease Community in Europe and beyond. The book will be presented during our next General Assembly.

Upcoming Events :

- Virtual Eighth European Course in Clinical Dysmorphology : **2020 October 1-2-3**
- Next ITHACA Board Meeting : **2020 December 11 - 12**

Call for collaborative clinical research on developmental disorders

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