

## ERN ITHACA



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November 2020

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### ITHACA Board Meeting

10th to 12th of December Online

The coordination team has received many registrations via the registration form that has been made available to you on the [ern-ithaca.eu](http://ern-ithaca.eu) website.

Thank you all for your interest in our board, the form will be closed at midnight today. We are currently processing the technical aspects. To this end, we will send you the Zoom connection links during the next coming days.

Please find the final programme of the Board on the website via this [link](#), or download it [here](#).

### ERN ITHACA Projects

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

The current work on the APOGeE Project construction is still in progress: the Steering Committee met again during November, in order to identify and list a set of emblematic genetic diseases and developmental abnormalities which should be covered in this educational programme.

As APOGeE is an ITHACA's major project within the framework of the CEF TELECOM Call, it will be largely discussed during the plenary session of the Board Meeting on December 12th. A WG meeting will bring together the Steering Committee members. The Board meeting will be the moment to set up the Editorial Committee and to identify cooperation with other ERNs. The meeting of the APOGeE Working Group is scheduled for 11am on December 11th.

## News from ILIAD registry

ILIAD is moving forward with the implementation of the workplan. As the final beta version of the registry is expected to be delivered in the end of December 2020, experts of ERN ITHACA are already cooperating in order to develop extended datasets, for clearly defined subgroups of patients.

These *sub-registry* task forces will develop their own specific datasets within the core ILIAD Registry. Currently discussions are ongoing with the NSEuroNet project that targets RASopathies. As the Registry Coordination Team is conscious that different initiatives may already exist in national basis, the common feeling is that cooperation is key in order to adapt the approaches and database structures as much as possible, to ultimately facilitate interoperability and ensure the connection with ILIAD, otherwise described as the “EU node”.

**Members of ERN ITHACA that wish to test the ILIAD database currently under development are kindly asked to contact [Klea](#) for further information.**

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## Work in progress

### November Update on 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:

#### [Short stature-brachydactyly-obesity-global developmental delay syndrome](#)

**ITHACA Experts:** Dr Irene VALENZUELA

#### [Cerebellar-facial-dental syndrome](#)

**ITHACA Experts:** Dr Irene VALENZUELA

#### [Microcephalic osteodysplastic primordial dwarfism types I and III](#)

**ITHACA Experts:** Pr Patrick EDERY, Dr Audrey PUTOUX

#### [Holoprosencephaly](#)

**ITHACA Experts:** Dr Christèle DUBOURG, Dr Alinoë LAVILLAUREIX, Pr Sylvie ODENT

#### [L1 syndrome](#)

**ITHACA Experts:** Pr C.T.R.M. (Connie) STUMPEL, Dr Y.J. (Yvonne) VOS

#### [Simpson-Golabi-Behmel syndrome](#)

**ITHACA Experts:** Pr Pablo LAPUNZINA, Dr Jair Antonio TENORIO CASTAÑO

### Freeman-Sheldon syndrome

**ITHACA Experts:** Dr Valeria CAPRA, Dr Gianluca PICCOLO, Dr Marcello SCALA

### Developmental delay-facial dysmorphism syndrome due to MED13L deficiency

**ITHACA Experts:** Dr Jamal GHOU MID, Dr Thomas SMOL

### Noonan syndrome-like disorder with loose anagen hair

**ITHACA Experts:** Pr Laura MAZZANTI, Dr Annamaria PERRI, Dr Emanuela SCARANO, Dr Federica TAMBURRINO

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## Patient Organisation

### ERN ITHACA Board of the Patient Council : Election 2020

The Board of the Patient Council in ERN-ITHACA consists of **five representatives of patient organisations (ePAG advocates)** based in Europe who sit on the ERN-ITHACA Board. Among this group of five members, **two persons are designated as Chair and Co-chair** and represent the voice and the needs of the Patient Council in the ERN-ITHACA Executive Committee.

On top of their role and ePAG advocates and representatives of the Board, the chair and the co-chair are also responsible for sharing important information and for updating the Patient Council on ERN-ITHACA activities as appropriate. They consult with the Patient Council on relevant issues and feedback information to the ERN-ITHACA Network Coordinator and to the Executive Committee.

The final new composition of the Board of the Patient Council in ERN-ITHACA for the next year :

- **Chair:**

**Ammi Andersson**, International Federation for Spina Bifida and Hydrocephalus (IF), Belgium

- **Co-Chair:**

**Dorica Dan**, Romanian, Prader Willi Association, Romania

- **Board Members:**

**Gábor Pogány**, Hungarian Williams Syndrome Association (HWSA), Hungary

**Sue Routledge**, Pitt Hopkins UK, Stichting Pitt-Hopkins Pitt-Hopkins syndrome, UK and The Netherlands

# European News

## Latest Updates on 13th Coordinators Group Meeting

The 13th meeting of the Coordinators Group with the European Commission was held on Friday 27th November. Below there is a brief summary of some key points on the topics discussed during this meeting: :

1. **The 5 ERN financial priority list** : Central ERN Management issues, registries, CPMS (*simplification on the working tool*), educational and training activities, and research (*to prepare ERN reseach proposals and conduct ERN studies*).
2. **Elargement of the Networks** : The IAB review will start on Decembre 2020 until July 2021, The status of the new members will be officially known in July 2021.
3. **Activities related with training and education** : a new ERN Academy project is ongoing. We will inform our members with our next newsletters on the details that will be provided on this project.
4. **Brexit and ERNs** : In accordance with the formalization of Brexit which will be effective on *February 1st 2021*, we have been notified by the EC that the British HCPs members of the ERNs will be disconnected from all the European tools and platforms starting *January 1st 2021*, the CPMS access will also no longer be possible even as a "Guest". ***Even if UK HCPs are no more eligible to participate in ERN ITHACA activities as entities, UK citizens remain totally allowed to belong to ITHACA, individually, as Experts, and ITHACA's ExCom will totally support and encourage this mode of integration.***
5. **Affiliated Partners status** : The EC have designated several HCPs as Affiliated Partners (*without any assessment process by the ERNs*) After the final approval of Full Members to existing ERNs on July 2021, an Affiliated Partner will lose this status if the country of origin is already represented in the ERN. If the Affiliated Partner is confirmed as a Full Member, it will equally lose the Affiliated Partner status and become Full Member instead. The ERN Coordinator Group has proposed to start a new procedure for affiliated partners in the not yet covered member states.

## The ERICA Project: joining forces to integrate research and innovation capacity across all 24 ERNs

**The European Rare disease research Coordination and support Action (ERICA) Project received a positive evaluation for a H2020 grant to establish a structural framework in support of the research activities of the ERNs. ERICA will strengthen research and innovation capacity by integration of ERN research activities, outreach to European**

**research infrastructures to synergistically increase impact and Innovation. This will result in safe, accessible and efficient access of therapies for the benefit of patients suffering from rare diseases and Conditions.**

Rare diseases are defined as diseases that affect not more than 1 person per 2000 in the European population. However, while individual rare diseases affect only a small percentage of the population, they collectively affect up to 30 million people in the EU. With the objective to facilitate access to high quality cross-border healthcare and promote and foster cooperation on rare disease healthcare between member states, the [24 European Reference Networks \(ERN\) were established by the European Commission last 2017](#). The ERICA Project builds on the strength of these individual ERNs by promoting inter-ERN research activities and by establishing firm collaborative ties with existing European and international infrastructures and consortia involved in rare disease research and innovation.

The ERICA Consortium consists of 29 partners, amongst which all 24 ERNs, EURORDIS, the EJP RD, Orphanet, Mapi Trust Research, and EATRIS. The ERICA Project is coordinated by Alberto Pereira ([ENDO-ERN](#) coordinator, Leiden University Medical Center, the NL). Looking forward to develop this project together!

## ERN Mobility Programme

This [programme](#) will allow ERNs healthcare professionals (MDs) to have short stays (*maximum 5 days, with a daily allowance to cover costs*) in other centres of the ERNs system with the aim of exchange practices, share knowledge and learn from or train other colleagues in clinical and operational aspects.

The first meeting scheduled jointly with the EC, and CHAFEA's representatives have presented an overview of the programme and on its next steps. The selected contractor [ECORYS](#) have presented the working procedures.

The exchange programme will last two years **with 3 main editions** : It is envisaged that the first exchange will be held starting **March 2021**, the second on **September 2021**, and the last in **March 2022**.

A final and official meeting to present the programme conclusions and results will be held in **October 2022**.

A first meeting between ITHACA coordination team and our representative at ECORYS has been planned on December 14th, in order to prepare in practice and on a logistic aspect the first exchanges of professionals between our HCPs members .

Please note that the HCP applicants will be able to join the programme starting the second edition: *(after the officialization of their integration to the ERNs in July 2021)*. The Coordination team will keep you informed on this issue starting January 2021.

## Manifesto for a European Health Union

On 9 May 2020, Europe commemorated the 70th anniversary of the Schuman Declaration that paved the way for the European Union. The history of Europe over seven decades demonstrates that major transformations are rarely fast or easy. Let us be inspired by the words of Robert Schuman: World health *"...cannot be safeguarded without the making of creative efforts proportionate to the dangers which threaten it"*

As a health professional, make your contribution by signing up following this link : [europeanhealthunion.eu](http://europeanhealthunion.eu)



## RE(ACT) Congress and IRDiRC Conference 2021

A banner for the RE(ACT) Congress and IRDiRC Conference 2021. It features a red background with white text. The text includes: "13-15 JANUARY 2021", "International Congress Of Research On Rare And Orphan Diseases", "RE (ACT) Congress", "in a joint online event with IRDiRC Conference", and "REGISTRATIONS ARE OPEN". There are also images of a woman wearing a face mask and a man in a lab coat. Logos for Blackswan Foundation, IRDiRC, EURORDIS, and EJRD are visible at the bottom left.

The [RE\(ACT\) Congress and IRDiRC Conference](#) joint online event that will be held on **13-15 January 2021**. It aims to bring together scientific leaders, patients, and policy makers to advance

research on rare diseases. The program is available [here](#).

To obtain your discount register [here](#) before **31st of December**.

Note that in compensation to attendees who registered before the announcement of the event being moved online, 3 free extra accesses will be granted to them to invite guests.

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## Upcoming Events :

### ERN ITHACA Board Meeting :

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

**RE(ACT) Congress and IRDiRC Conference: January 13th - 16th**

Call for collaborative  
clinical research on  
developmental disorders

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