

ERN ITHACA



May 2021

ERN ITHACA Projects

Next ERN ITHACA Webinars

Webinar #1 on fetal phenotyping and genotyping: June 8, 2.00-3.00 PM (CEST):

All interested HCP members and other partners are invited to join the **fetal phenotyping and genotyping webinar**.

Gijs Santen (LUMC, Clinical Genetics Leiden) and **Peter Robinson** (The Jackson Laboratory for Genomic Medicine, Farmington) will present and invite to their [two collaborative projects](#).

We would appreciate if you could fill out this [FORM](#) if you plan to participate in this webinar and leave your contact details in the space provided if you wish to become an active part of the prenatal working group. Further specialists from your centre, such as ultrasonologists, echocardiologists, fetal MRI experts, perinatal pathologists, are welcome.

Connection details:

ERN-ITHACA Webinar on fetal phenotyping and genotyping

June 8, 2:00-3:00 PM CEST

<https://u-paris.zoom.us/j/85421629698?pwd=Rmh1V2ZYV1JCNU00MINtaEFiWGJBdz09>

Meeting ID: **854 2162 9698**

Passcode (if required): **074342**

Webinar #2 "Care for the old": June 11, 5:00 to 6:30 PM (CEST):

This webinar is dedicated to adult and aging within **Intellectual Disability, standardized geriatric assessment and care**. The program includes new experts' attendance and aims to

enrich the ERN ITHACA Working Group on Intellectual Disability projects development. Your expert contribution is an asset for improving the care of older adults with intellectual disabilities. The webinar is intended for professionals, medical students, medico-social professionals, paramedical specialists. We will also be pleased to welcome ePAGs and Patient Council representatives. [Please click on the following link to get the agenda.](#)

Connection details:

[Free registration before June 6th, 2021 following this link](#)

ERN-ITHACA Webinar "Care for the old", 5:00 to 6:30 PM CEST

<https://u-paris.zoom.us/j/86512922405?pwd=M1ZDZ2NuSE1CbFVDWVpscEM2N1ILUT09>

Meeting ID: **865 1292 2405**

Access code: **860191**

31st EuroDysmorpho Meeting: Registrations are ongoing!

Registrations for the [31st European Dysmorphology](#) meeting have started on **May 1st** and will stay open until **June 18th**:

- We remind you that young geneticists will be eligible for a special registration rate (**50€**) after submitting a proof of trainee document: *the template to be completed and submitted is available for download on [the registration & abstract submission web page](#).*
- Unsolved cases for submission to [SOLVE RD](#):
Only one abstract is mandatory per participant, and optionally, **you may submit (in addition to your first abstract or as a single submission) an unsolved case for submission to Solve RD**: Unsolved cases will be presented and discussed on **Thursday 23 September**.
If your unsolved case for submission to Solve RD is accepted and discussed during the Workshop, we will provide you with a submission guide at the end of the meeting.

Do not hesitate to contact the [coordination team for any further question](#).

Work in progress

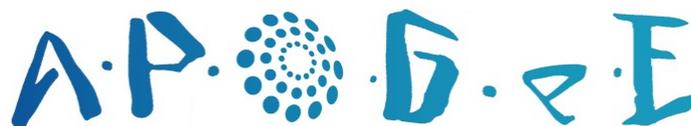
Transdiagnostic guideline on the transition of Care

We are launching a call for interest to join the consortium on the development of the [transdiagnostic guideline on Transition of Care](#), chaired by Agnies van Eeghen. The general purpose of this group will be to propose EU-wide recommendations on a purposeful and planned process of supporting young people with diagnosed and undiagnosed rare diseases linked to intellectual disabilities to move from children to adults' services. We would be very pleased if you

would come forward to the coordination team if you are willing to consider this initiative. Also, please do not hesitate to spread the word to colleagues that might be interested. We are especially in need of disciplines such as **social workers, internists, child and youth psychiatrists, psychologists, teachers, neurologists, and general practitioners** coming from all the European Union or beyond. For the moment, Italy, the Netherlands and Poland are sufficiently represented.

If you are personally interested in contributing to this initiative and want to integrate the working group or are aware of colleagues that are knowledgeable on the topic, please [email Klea](#) to indicate your willingness and/or transfer this invitation to your potentially interested colleagues.

APOGeE Online e-learning tool



A Practical **O**nline **G**enetics **e**-**E**ducation

The development of the APOGeE project has well-evolved and increased over the last weeks: it concerns the platform's future content and presentation. The ITHACA team is currently working on a test moodle platform with our French partner for consulting, editing, technical integration, and the hosting service.

Updates on APOGeE will be discussed at the next ITHACA Board meeting on **18 June**.

European News

Public consultation for the revision of the EU regulations on medicines for children and rare diseases 07 May - 30 July

The Rare disease research Team in DG Research & Innovation, and DG SANTE representatives would like to inform you about the Open **Public Consultation** to receive input from the relevant stakeholders in the context of the discussions for the revision of the EU regulations [on medicines for rare diseases](#) and [medicines for children](#) which were respectively adopted in 2000 and 2006.

This revision will explore several options to address the shortcomings identified in the [evaluation of the Regulations of medicines for children and rare diseases](#). With this public consultation, citizens and stakeholders are invited to share their views and experiences on the main obstacles they are facing concerning treatments for rare diseases and children, on possible ways to

overcome these obstacles and on how to make the current legislation future-proof.

Access to the public consultation:

https://ec.europa.eu/info/law/better-regulation/have-your-say/initiatives/12767-Medicines-for-children-&-rare-diseases-updated-rules/public-consultation_en

The **deadline** is **30 July 2021**.

European Health Union: Commission publishes open public consultation on the European Health Data Space

The Commission has published an open [public consultation](#) on the European Health Data Space (EHDS) - an important building block of the European Health Union. The EHDS aims to make full use of digital health to provide high-quality healthcare and reduce inequalities. It will promote access to health data for prevention, diagnosis and treatment, research and innovation, as well as for policy-making and legislation. The EHDS will place individuals' rights to control their own personal health data at its core. The consultation will remain open for responses until **26 July 2021**.

A press release is available [online](#). (For more information: Stefan De Keersmaecker – Tel.: +32 229 84680; Darragh Cassidy – Tel.: +32 229 83978)

https://twitter.com/EU_Health/status/1389190155597139969



European Commission: Online consultation on patients' rights in cross-border healthcare

The Commission launches an [online public consultation](#) on patients' rights in cross-border healthcare to assist its evaluation of the EU directive a decade on since its adoption. Stakeholders are invited to share views on how the Directive has improved patients' rights to healthcare abroad and its role to strengthen European cooperation in the area of rare and complex diseases. This online consultation remains open for responses until **27 July 2021**.



News from EJP RD



The ERN Research Mobility Fellowship

In the context of [EJP RD](#)'s ERN Workshops, a **face-to-face workshop** on **clinical epidemiological research for ERNs** titled "**Clinical Research: The Basics & Beyond**" (consisting of **presentations by experts** in the field of epidemiology and a **feedback**

session on the research projects of participants) is being organised by **Prof. Olaf Dekkers and Prof. Frits Rosendaal** of the Leiden University Medical Center.

The **in-person event** will take place over two days on **30th September and 1st October 2021** at the **Hotel Golden Tulip Leiden Centre**, Schipholweg 3, 2316 XB **Leiden, the Netherlands**.

The workshop is open by prior registration and selection to **medical specialists** affiliated to an ERN Full Member or Affiliated Partner institution and who **have a special interest in clinical research with databases**. **Registration closes on 18th June 2021**, and those selected to participate from among the applicants will be **informed by 9th July 2021 of their selection**.

More information and registration here: <https://www.ejprarediseases.org/event/clinical-research-with-databases-the-basics-beyond-workshop-in-clinical-epidemiological-research-for-erns/>

ITHACA Upcoming Events :

- **June 8 : Webinar #1** on fetal phenotyping and genotyping: **2.00 - 3.00 PM CEST**
 - **June 11: Webinar #2** "Care for the old": **5:00 - 6:30 PM CEST**
 - **June 15:** Second 2021 ExCOM Meeting: **6:00 - 7:30 PM CEST**
 - **June 18:** ITHACA intermediate Board meeting: **1:00 - 4:00 PM CEST**
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Registration for the
next EuroDsismorpho
Meeting

Call for collaborative
clinical research on
developmental disorders

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