ERN ITHACA

January 2021

ERN ITHACA Projects

ERN Exchange Programme (ITHACA)

The European Commission in cooperation with ECORYS has launched a Clinical Exchange Programme to share knowledge and stimulate collaborations between healthcare professionals within the ERNs. The aim of the ERN Exchange Programme is to harmonize specific knowledge and reduce gaps in expertise. The basic idea is to transmit expert knowledge either by experts visiting other sites or by on-site training of junior professionals at experienced ITHACA's …

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2020 ITHACA Board Meeting: video recordings available on our website

ERN ITHACA coordination team is pleased to inform you that the recorded and downloadable presentations are available on our website (presentations for which the authors have agreed to publication), via the dedicated page (Board presentations) in the Members' Area:
Please note that in order to access this page, you must have created an account on our website. In addition, we have set-up a restricted access through the YouTube platform: the presentations being intended for a specialized audience, they are not visible via a search by speaker name or keywords.
We also would appreciate your participation to this short & anonymous survey, in order to improve our organization for the next events. It will remain available until February 15th.

Eurodysmorpho Meeting 2021

Due to the ongoing uncertainties about the sanitary situation, the Organising Committee and the Scientific Coordination have decided that the 31th European Dysmorphology Meeting, which was originally planned to take place at the Bischenberg Centre, will instead be organised online from 22 to 25 September 2021. The coordination team is currently working on the registration arrangements and the programme. Next month we will inform you about the official announcements concerning the organisation, registration and abstracts submission. ERN ITHACA will also be an official partner of this event.

Work in progress

Latest updates on collaboration with Orphanet : 63 updated abstracts

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

Witteveen-Kolk syndrome A rare genetic neurodevelopmental syndrome characterized by mild intellectual disability, developmental delay, dysmorphic facial features, growth- and feeding problems, hypotonia, epilepsy, behavioral problems and a variety of congenital abnormalities. The summary has been updated on Orphanet (ORPHA:500163) - December 2020 and reviewed by Dr Meena Balasubramanian, Pr Tjitske Kleefstra (ITHACA) and Dr Jet Van Der Spek.

Blepharophimosis-intellectual disability …

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Syndrome de Basel-Vanagaite-Smirin-Yosef: A rare, genetic intellectual disability syndrome characterized by severe global developmental delay with intellectual disability, microcephaly, growth retardation, ocular defects such as congenital cataract, and nevus flammeus simplex on the forehead. Cardiac, urogenital, and skeletal abnormalities, as well as seizures are present in most patients. Dysmorphic craniofacial features include sparse hair, downslanting palpebral fissures, hypertelorism, broad and …

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News from ILIAD registry

The ILIAD core database has undergone some recent developments by the Molgenis team that differentiate between items pertaining to the Joint Research Centre (JRC) minimal dataset

implemented for use in the core registry and the non-mandatory CPMS-related fields that once appeared in the first version of the database. The developers are currently thinking about possible interactions with sub-registries. The functionalities of formatting and search will also be changed within the Molgenis system.

A new way of coding the genetic diagnosis has been proposed: in the genetic diagnosis field users will normally be able to add variants (each allele has a separate field) and in the chromosomal diagnosis fields, users will be able to add genomic variants and switch between diagnosis. They can currently code up to two genetic diagnosis for a patient.

Patient Organisation

EURORDIS 10th Black Pearl Awards & Rare2030 Final Conference

ERN ITHACA is one of the official outreach partners of the EURORDIS LIVE at the **Rare2030 Final Conference** and the **10th annual Black Pearl Award.** Click on the **following link** to get more information and register.
The **Rare 2030 Final Event** marks the end of this two-year foresight study. It will be the occasion to present the Rare 2030 policy recommendations for a new policy framework in the presence of high-level speakers, such as Frédérique Ries, Member of the European Parliament, and Stella Kyriakides, European Commissioner for Health and Food Safety, to name a few. The event will be held virtually –and for **free – on 23 February 2021 at 13:30 – 18:30 CET**.

Taking place in February to mark the occasion of Rare Disease Day, the EURORDIS Black Pearl Awards celebrate the inspirational qualities of people living with a rare disease along with those who go that extra mile to make a difference to their lives. **24 February 2021 at 17:00 CET**.

Rare Diseases week: 22 - 25 February 2021

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European News

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

This year marks the tenth anniversary of the entry into force of the Directive on patient rights on cross-border healthcare. This piece of legislation aims to establish the general framework for efficient and accessible cross-border healthcare, also backed up by a reimbursement scheme by the Member State of affiliation of the healthcare obtained abroad. Most importantly for us, the Directive …

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Eurordis: Key principles for Newborn Screening

EURORDIS, alongside its Council of National Alliances, Council of European Federations and its members, **have set out 11 Key Principles** to support an harmonised European approach to Newborn Screening. The vast inequalities across Europe, coupled with technological and scientific advances highlight the urgent need to move forward from the status quo.

Advisory Body of the ERNs Clinical Guidelines Programme

The Advisory Body (AB) on ERN Guidelines Programme has been formally constituted on the 14 of January 2021. Find here the information on the profile and affiliation of the Members of the AB. Download here the Rules of Procedure of the Advisory Body where the roles and tasks of the AB are detailed. The AB members will be invited to participate as observers in the general meetings of the ERN CPG&CDST Programme. The first meeting they will be invited to will be the ERN - Expert Panel meeting on 12th February.

EJP RD News

Information Webinar for the Joint Transnational Call 2021 interested applicants

The EJP RD is glad to invite you to the information webinar organised for potential applicants to the **Joint Transnational Call 2021 on "Social Sciences and Humanities Research to Improve Health Care Implementation and Everyday Life of People Living with a Rare Disease".**
The webinar will take place **online on February 2nd, 2021 3:00pm – 4:30pm CET.**
All interested applicants are invited to register and participate to this event. **The registration to this event is mandatory and will close on January 29th, 2021**.
The objective of this information webinar is to give you details on the objectives, topics and administrative rules for this call for projects. The general presentation will be followed by a Q&A session with the participants.
You can already submit your question in the **registration form**.

3rd Call for Research Training Workshops (27January - 7 March 2021)

The goal of the workshops is to train researchers and clinicians affiliated to ERN- Full Members or – Affiliated Partners in relevant topics on research in rare diseases. Training themes may include innovative research methodologies, diagnostic research methodologies, interdisciplinary treatment approaches, such as gene therapy and transplantation, etc. Moreover, the workshops will be aiming to provide a cross-ERN added value.
Applicants/Application profile:
The applicant submitting workshop topics must fulfil one of the following conditions:

- Affiliated to any EJP RD beneficiary institution.
- Affiliated to an ERN Full Member. The list of full ERN members per country and per network can be found here
- Affiliated to an ERN Affiliated Partner institution at the time when the application is submitted, as well as during the period of the execution of the workshop.

Please follow this link to get more details. Or contact sanja.hermanns@ejprd-project.eu

ITHACA Upcoming Events :

- **February 5th : WG7 (Reseach) online meeting - To discuss your visit proposals on ERN (ITHACA) Exchange Programme**
- **February 11th**: ERN & Screen4Rare Meeting
- **February 11th**: ERN exchange program - Follow-up meeting with ECORYS
- **February 19th:** First 2021 ExCom Meeting
- **February 24th:** EURORDIS Black Pearl Awards

February 28th: Rare Disease Day

Call for collaborative clinical research on developmental disorders

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