ERN ITHACA

December 2020

ERN ITHACA Projects

Overview on the ITHACA Board Meeting 2020

A special thank you goes to all the attendees among the representatives of our full member HCPs, the working groups Chairs and Co- chairs, the applicant HCPs representatives, and the Patient Council's representatives.

Many thanks also to all the external guests of the ERN who accepted our invitation.

The presentations will be made available on our website on the Member Area section. We will also send you the slides used during the meeting by e-mail at the beginning of January.
If you wish to access the scientific bibliography on 100 rare diseases syndromes provided by Prof. Raoul Hennekam, and available on our website on the Member Area, please follow this link (after registration / login).

The Board's recordings videos will also be available at the beginning of the year in a dedicated private section on ern-ithaca.eu.

We are delighted to welcome among our Working Groups:

- **Prof. Zeynep Tümer** as Co-Chair of the Work Group 7 *(Research)* with the current Chair, **Prof Marco Tartaglia.**
- **Prof Giovanni Zampino** as Co-chair of the Work Group 8 *(Teaching & Training)* with the current Chair, **Prof Laurence Faivre**

WG9 ID – A new subgroup SNW PIMD has been launched

Led by Dr Sylvia Huisman, this group's aim is to produce good practice recommendations for the management (healthcare, daily care and support) of children to young adults with profound intellectual impairment and multiple disabilities. Part of the work will be based on a new publication:

French National Diagnostic and Care Protocols (NDCPs) : A Consensus statements endorsed by ERN ITHACA and translated in English version.

We have retained the linguistic terminology of the French word “Polyhandicap”. Its equivalent, with some differences, can be found in the Anglo-Saxon literature under the term "Profound Intellectual and Multiple Disabilities" (PIMD) (Nakken 2007).This term defines the clinical features of the most highly disabled children, who require very specific human and material assistance. [Bourg, V. 2008] The prevalence of polyhandicap is 0.50 per 1,000, so it is not uncommon. However, the etiologies of polyhandicap are by and large rare diseases, diagnosed or not. Multiple brain lesions and their consequences on growth and secondary repercussions on various organs are sufficiently characteristic for this situation of severe disability, from birth or early childhood, to be declared as a true pathological entity in a national summary document. The implementation of the NDCPs dedicated to Polyhandicap has been entrusted to the Rare Cause ID Reference Centres within the framework of the DéfiScience French National Network *(Original version in May 2020).*
You can access this document online following this **link**

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:

- **Treacher-Collins syndrome**

ITHACA Experts: Pr Corinne COLLET

- **Carpenter syndrome**

ITHACA Experts: Pr Corinne COLLET

- **Smith-Magenis syndrome**

ITHACA Experts: Dr Laurence PERRIN

- **Sotos syndrome**

ITHACA Experts: Dr Alessandro MUSSA

- **Fraser syndrome**

ITHACA Experts: Dr Maria Francesca BEDESCHI, Dr Max LIEBAU (ERKNet) - Dr A.M. [Albertien] VAN EERDE (ERKNet)

- **Oculocerebrofacial syndrome, Kaufman type**

ITHACA Experts: Dr Katalin SZAKSZON

- **THOC6-related developmental delay-microcephaly-facial dysmorphism syndrome**

ITHACA Experts: Dr Andrea ACCOGLI, Dr Valeria CAPRA, Dr Gianluca PICCOLO

- **Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome**

ITHACA Experts: Pr Laurence OLIVIER-FAIVRE

- **Malan overgrowth syndrome**

ITHACA Experts: Pr Valérie CORMIER-DAIRE, Dr Valérie MALAN

- **Marshall-Smith syndrome**

ITHACA Experts: Pr Valérie CORMIER-DAIRE, Dr Valérie MALAN

- **Noonan syndrome with multiple lentigines**

ITHACA Experts: Dr Maria Cristina DIGILIO

- **Postaxial acrofacial dysostosis**

ITHACA Experts: Pr Dagmar WIECZOREK

- **Mandibulofacial dysostosis-microcephaly syndrome**

ITHACA Experts: Pr Dagmar WIECZOREK

- **Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome**

ITHACA Experts: Pr Dagmar WIECZOREK

News from ILIAD registry

The ILIAD Coordination team took part at the ERNs Conference on “Registry Informed Consent Facilitation; Domain Specific Common Data Elements & Registries FAIRification Implementation Choices”, an initiative proposed by our EJP-RD partners. Views and best practices were exchanged on the important topic of the Registry Informed Consent facilitation, where an EJP working group has been adapting and drafting an EU-wide informed consent form, based on the national ethics committees’ feedbacks during the authorisation process of another ERN registry

(ERKReg).

Moreover, the issues of the selection and definition of the common Domain Specific Common Data Elements of transversal relevance were discussed with the representatives of the ERNs, as well as FAIRification implementation choices. This collaboration and knowledge exchange will continue and in the beginning of January, it will formalise with a series of workshops dedicated to ERN registries.

Trans-ERN Working Group for Spina Bifida Training: Workshop for future research on innovative diagnostic and interdisciplinary treatment

The workshop will be organised by Giovanni Mosiello, MD, FEAPU, FEBPS, Bambino Gesu Pediatric and Research Hospital, Rome, Italy Department of Urology: Neuro-Urology and Andrea Manunta, MD, FRCS, FEBU, Department of Urology and Neuro-Urology, Centre Hospitalier Universitaire de Rennes, eUROGEN and ITHACA EJP-RD beneficiary.
Spina Bifida (SB) is a congenital malformation syndrome that requires multidisciplinary expertise. The trans-ERN Spina Bifida Workgroup has been created to produce care documents and guidelines for all ERNs involved with the main aim of harmonization of clinical practices for SB patients with EU-wide transnational recommendations for all people with SB. The Workshop will examine how to consider innovative diagnostic tools, new treatments and address future research. The objectives of this 2 days' workshop are to train researchers and clinicians from different ERNs on:

* Genetic neural tube defects, correlation with Orphacode and post-natal outcome
* TRASCET: transamniotic stem cell therapy
* Prenatal diagnosis correlating fetal MRI and Ultrasound, with post-natal outcome
* Urinary marker on renal and bladder function
* Fetal Surgery (different approaches), results on bladder function etc.
* Innovative urodynamic investigation on neurogenic bladder related to spina bifida (Radiomics)
* Long-term sequelae of continence treatment considering the specific spina bifida patient.

Patients' representatives will be present during the presentation and the discussion of every single topic in order to lead future research considering the special needs of spina bifida patients, their expectations, and then helping researchers to define the correct modalities for research.
ERNs ITHACA, eUROGEN, ERKnet are the main beneficiaries. The workshop is aimed at researchers and clinicians involved in spina bifida management: neurologist, neurosurgeon, obstetrician, neonatologist, urologist, pediatric urologist, fetal and pediatric surgeon, nephrologist, geneticist, epidemiologist, immunologist, etc.

The workshop is open to all ERN researchers: junior or senior researcher or clinician. Junior researchers should be well motivated, involved in an ERN, very active in research and transERN activities are warmly welcome as well as senior clinical researchers (Team Coordinator).

The workshop will consist of both presentations by experts, as well as hands-on sessions, discussing clinical cases or theoretical cross-ERN scientific studies or projects, all of which will train participants and gain practical insights. At the end of the workshop all participants will have gained more knowledge on new promising diagnostic procedures, new treatments defining their feasibility, safety and efficacy for spina bifida patients with a view to developing a trans-ERN multicentric clinical trial based in Europe, according to EU rules respecting ethical and legal requirements, in order to make research findable, accessible, interoperable and reusable for all.

Patient Organisation

Eurordis : Rare Barometer survey

To ePAG Advocates, the new **Rare Barometer survey** on the future of your rare disease is now live.

You can access the online survey here

It should take no more than 15 minutes to complete and closes on **3 January 2021**. This survey will enable to gather your opinion on the future of your rare disease and give the possibility to communicate facts and figures to decision-makers, so that your opinion is taken into account when shaping the future of rare diseases.

Through asking questions on **your needs and preferences** regarding patients' **access to health care**, **priorities for medical and social research or early diagnosis for children**, we will be able to better understand how to shape the future of rare diseases.

You can find more information about the survey here.

This survey is open to people living with a rare disease and their family members from any country in the world. It is translated in 23 languages. All responses are anonymous and will be kept in secure storage only accessible to the Rare Barometer research team.

We need your help to ensure as many people as possible from the rare disease community complete the survey. The more people will take the survey, the stronger our voice will be!

Social innovation: inspirational practices supporting people throughout their lives

Recently, the European Commission has published a brochure that showcases 27 examples of social innovation initiatives across the European Union, one from each Member State. For Romania, an ERN ITHACA member was taken as an example. **"A holistic and centralized approach to supporting patients with rare diseases: The NoRo Centre"** was as such described in the brochure. This publication highlights the diversity and success of social innovations in Europe, showcasing initiatives that facilitate transitions, integrate disadvantaged groups, redesign business models, empower people, build partnerships and deliver public policies, in new and creative ways. Innovators include NGOs, social enterprises, local, regional and national authorities, social partners and the private sector, often working in partnership to improve people's lives.
Contact information: Dorica Dan
View more following this **link**

European News

Brexit: deactivation of access to ERN IT systems (ITHACA)

As announced in the last meeting of the Coordinators Group on 27 November 2020, as of 1 January 2021, the EC have to discontinue access to the ERN IT systems to users affiliated to UK healthcare providers and UK institutions.
Following that announcement it will be proceeded as follows:

1. The few still open panels in CPMS led by UK healthcare providers will be closed on 1 January 2021
2. All the UK Healthcare providers registered on the EC database will be deactivated and will cease to have access to the CPMS as of 1 January 2021.

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.

EJP RD News

Internal call for innovation project in clinical trials methodology

The Internal Call for Innovation Project in Clinical Trials Methodology in Limited Populations has been launched on December 7, 2020. The call is open to EJP RD beneficiaries and their linked third parties.
The innovation methodologies topics particularly include (but are not limited to):

- Development of a disease progression model from a natural history cohort or other observational studies.
- Development and validation of a disease specific clinically meaningful outcome with special interest in PCOMs, or composite endpoints.
- Development of a design and analysis procedure for a pharmacometric model and/or bridging study.
- Development of a randomization-based model as an alternative analysis strategy and explore the level of evidence.

The call aims to encourage collaborations among groups of experts consisting of different stakeholders including methodological experts, clinicians, patients and industry (when relevant) who will jointly develop innovative ready-to-use methods to enhance RD clinical trial methodologies.
TIMELINE

- **11 January 2021**: A networking meeting will be held to consolidate the collaborative networks.
- **3 March 2021**: Foreseen call closing deadline

To get more information click here: https://www.ejprarediseases.org/index.php/ongoing-calls/innovation-project/

The 3rd EJP RD – JTC 2021 is OPEN

The JTC 2021 will have for specific objective to promote multinational project on 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 :

- 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.

Click on the following link for more information

Y4&5 ITHACA Upcoming Events :

I- Next teleconferences:

- **ExCom** [2h call, Bi-monthly on Friday]
- **Feb 19th; Apr 16th; Sept 3rd; Oct 8th or 15th** (depending on ASHG) Foresee a special meeting focused on the new application (1 representative/HCPs + WG leaders, When call available)
- **WGs :** please fix your own calendars for 2021/2022 *(Proposed: every 3 months : we must fill and check all our objectives)*
- **Special Coordinator's meeting for New Members:** July 2021 *(after confirmation)*

II- Next ITHACA Board Meetings

- **2021:** *Virtual Meeting.* June 11th *(day before ESHG)* or 25th *(1 week after)*: Please let us now about your opinion
- **2021:** Madrid - December 2-4 (Thursday to Saturday)
- **2022:** Budapest - Probably on December
- **2023:** Please contact us if you wish to be the host of the annual Board

See you soon !

The ERN ITHACA coordination team will be taking a short break. We will meet again starting from the 4th of January to start a new year full of challenging projects.

We thank you for your daily involvement by our side and we wish you all a happy holiday season!

Call for collaborative clinical research on developmental disorders

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

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