ERN ITHACA Webinars: Advancing Knowledge and Collaboration in Rare Malformative and Neurodevelopmental Disorders

Anne Hugon First author / i.e., presenting author
Cheffe de Projets ERN ITHACA & CLAD - Team WG Teaching & Training ERN ITHACA
ITHACA, Department of Genetics, APHP Paris Nord Université Robert DEBRE, Paris, France
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ERN ITHACA Webinars: Advancing Knowledge and Collaboration in Rare Malformative and Neurodevelopmental Disorders

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Introduction

The Teaching & Training Workgroup of ERN ITHACA has initiated a webinar programme with the aim of providing highly specialised knowledge on rare congenital malformations and neurodevelopmental disorders. The programme is aimed at healthcare professionals, patients and their families and covers topics such as fetal surgery, newborn screening, care of older patients and guideline methodology. The webinars are also made relevant and inclusive through input from the ITHACA Patient Advisory Board.

ERN ITHACA currently links 71 expert clinical centres and over 49 European Patient Advocacy Groups (EPAGs) in 26 countries. Its mission is to improve patient care, research and access to information through collaboration, and the webinar programme exemplifies this mission by facilitating knowledge sharing and engagement without travel, promoting inclusivity and convenience.

Webinar format

Each webinar, hosted on Microsoft Teams, includes a one-hour presentation by an expert, followed by a question-and-answer session for interaction between participants and presenters. Approximately ten webinars are held each year, with topics suggested by ERN ITHACA experts and stakeholders, including patient representatives. Collaboration with entities such as the European Society of Paediatric Neurology (EPNS); Young Geneticists Committee (ESHG-Y) and other ERNs to ensure diverse perspectives and produce joint webinars. Sessions are recorded and made available on the ITHACA website for wide access. Impact is measured through upstream and downstream surveys, which are regularly analysed. Within the network, each HCP is committed to producing at least one webinar during the grant period.

Participants can sign up and receive a TEAMS link to the webinar.

2025 webinars

Webinars currently planned for 2025:

JAN: Rho-GTPase in intellectual disability and neuro-developmental disorders

JAN: Ukraine webinar on spinal dysraphisms: Accessing European expertise for rare urological cases (CPMS -cross-border collaboration.)

MAR: ARID1B associated Coffin-Siris Syndrome – Diagnosis and management

APR: With the patient in the middle - focus on the whole family, A Norwegian model

JUN: Diagnostic guidelines for cutis laxa

SEP: Non coding genome and human disease

OCT: Reproductive Genetic Carrier Screening

NOV: Endocrine aspects of RASopathies

The list will be updated throughout the year:
https://ern-ithaca.eu/events-news/upcoming-events/

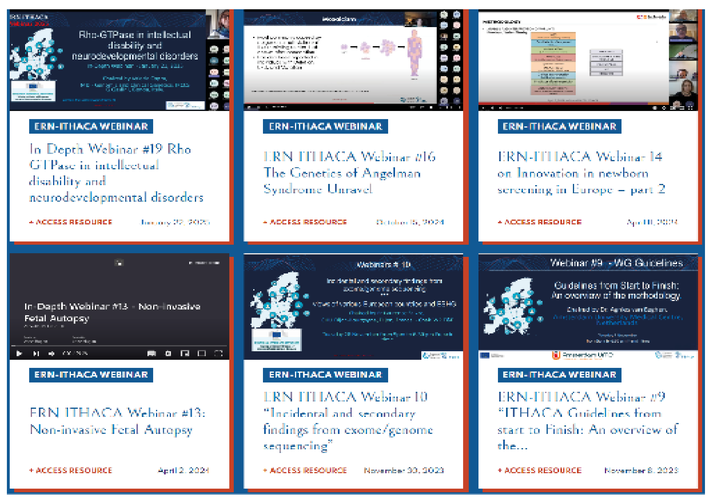
Access to past webinars

The Teaching & Training workgroup has set up a webinar program to help healthcare professionals and patients to acquire highly specialized knowledge in the field of rare congenital malformations or neurodevelopmental disorders, without the need to travel, at their convenience. Available via this link: https://ern-ithaca.eu/webinars

About ten webinars are organised per year by ITHACA experts, on various disorders or aspects of care, such as fetal surgery for omen dysraphisms, newborn screening, guidelines methodology, Down syndrome or care for old.

To elaborate each webinar, this taskforce pays special attention to the ITHACA Patient Advisory Board's inputs, so that the productions remain at least partially accessible for patients and their families, when they are not designed for them.

The taskforce also collaborates with other research or care entities, such as the European Paediatric Neurology Society EPNS), or other ERNs, to produce joint webinars



The webinars are recorded and uploaded to the ERN-ITHACA website for access without registration.

Audience survey

Registered participants:

>600

in 2024

Audience satisfaction:

94%

In general, the audience was satisfied with the webinars. Nevertheless, some participants expected more interaction between the speakers and the floor. Whilst the online interactive experience might be limited due to its nature, new technology and tools might improve the responsiveness of any spontaneous chats. In response to feedback regarding pre-record Webinar EU-Ukraine series on spinal dysraphisms (Trans ERN EUROGEN/ ITHACA / ERKNET) more effort will be put in reaching out to target audience in Ukraine, such that Ukrainian families and healthcare professionals will be interested in the webinars.

18 March 2025 (Tues) Webinar

ARID1B Associated Coffin-Siris Syndrome – Diagnosis and Management

Coffin-Siris syndrome (CSS) is a relatively frequent syndrome with developmental or cognitive delay, classically characterized by small or missing nails,and distinctive facial appearance, among several other signs. Since the discovery of mutations in ARID1B, a component of the BAF chromatin remodelling complex, as the main cause of the syndrome, our knowledge on diagnosis, natural history and management have greatly improved. In this webinar we aim to explore these recent developments. **Register now:**

Programme:

Clinical and molecular diagnosis in children
Georgia Vasileiou - University Hospital Erlangen, Erlangen, Germany

ARID1B-related disorder in adults
Gijs Santen - Leiden University Medical Center, Leiden, The Netherlands

CARE4ARID1B
Dagmar Wieczorek - Medical Faculty and University Hospital, Düsseldorf, Germany

The patient's perspective
Gal Lazarus - Department of Psychology, The Hebrew University of Jerusalem, Israel

Chair: André Reis - University Hospital Erlangen, Erlangen, Germany

Past webinars

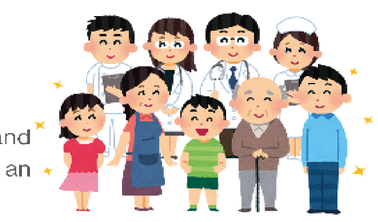
Selected past webinars for different target audiences:

Healthcare professionals:

- Non-invasive Fetal Autopsy
- Innovation in Newborn Screening across Europe
- Guidelines from Start to Finish: An overview of the methodology. EU Guidelines for the ITHACA ERN

General public:

- The Genetics of Angelman Syndrome Unraveled
- Patient Engagement "How and why get involve in ITHACA has an EPAG"



Specialist topics:

- The new SBoD Orphanet classification in practice

Conclusion

The programme has successfully delivered webinars on topics such as ageing with intellectual disability, innovation in newborn screening and integrated care for rare diseases, fostering cross-border collaboration, improving patient advocacy and providing professional development opportunities. The programme includes tailored sessions, from open-access webinars for the public to technical sessions for professionals, to increase accessibility and impact. This programme demonstrates how virtual education can bridge knowledge and collaboration gaps across Europe, integrating patient voices and leveraging technology.

Anne Hugon anne.hugon@aphp.fr
ERN-ITHACA, University Hospital Robert Debré - AP-HP, Clinical Genetics Department, Paris, France
WG Teaching & Training, Pr Faivre, Pr G. Zampino, ERN ITHACA Consortium, Pr A Verloes

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