ERN-ITHACA

PATIENT ADVISORY BOARD

Funded by the European Union

European Reference Network

European Patient Advocacy Group

ITHACA

Who we are and what we do

ERN-ITHACA is a **patient-centered network** that gathers more that seventy medical expert centres **HCP** across 25 European Member States, **to further patient care, research and access to information about rare congenital malformations and intellectual disability at the European level.**

The name **ITHACA** (acronym for Intellectual disability, TeleHealth, Autism and Congenital Anomalies) is a reference to the odyssey that, for many families, the diagnosis of a child with a rare developmental disease represents.

Indeed, the heterogeneity of the different clinical pictures and the enormous variation in the first symptoms can, as early as the neonatal period, lead to misdiagnoses, and therefore to the absence of early care. And even once the disease is diagnosed, it's not uncommon for specialists to find themselves at a loss when faced with a syndrome they've never heard of, and about which the scientific literature is scattered and difficult to access. Imagine that there are over 5,000 rare conditions!

We cover many rare malformative or developmental syndromes, here are some of them:

22q11.2	Acrodysostosis	ADNP	Angelman	Beckwith-Wiedemann	Chromosomal disorders
Chromosomal abnormalities	Corpus Callosum	Costello and cardio-facio-cutaneous syndrome	Cowden (mental health)	Coffin-Lowry	Cdls Cornelia Delange
CDKL5	Dravet	Fragile X	Goldenhar	Kleefstra	MED13L
Mowat-Wilson	Noonan	PACS1	Pallister-Killian	Pitt Hopkins	Prader Willi
Phelan Mc Dermid	Rett	SAT2B	Smith-Lemli-Opitz	Smith-Magenis	Spina Bifida
Tuberous Sclerosis Complex	Williams	White-Sutton	Brain malformations, genetic on NDD	ID rare diseases	Ultra rare condition
	Undiagnosed no names	RD Borys the Hero Foundation	RD Alliance IT,ES,CZ,NL,FR,RO	**Etc.** (Specific conditions)	

The ITHACA network's mission is to help patients find the right specialist, and help specialists access all existing medical resources, in order to give them every chance of receiving **the right diagnosis and the best possible care,** as early as possible. To achieve this, the involvement of representatives of European patient associations (ePAGs, European Patient Advocacy Groups) is essential. Their mission is **to represent the voice and interests of patients and their families in all the work carried out by the network.** ePAG and specialists, whether doctors or researchers, work in partnership towards the same goal: **improving the lives of patients with rare malformative syndromes.**

We are constantly looking for new organisations to enrich the network's activities with **as many different life courses, care trajectories, lived-by-expertise, possible,** and to further build a strong ePAGs community. As you maybe already know, a rich and efficient network of associations is a highly valuable tool for patients and their families!

Are you part of an association representing patients with a rare malformation or a rare intellectual/developmental disorder, and would you like to work with us? Are you a patient yourself or a member of their families, and would like to know more about what we can offer? Reach out to us by contacting Anne Hugon (anne.hugon@aphp.fr)

WHO CAN REACH US ?

Why get involved in the ERN-ITHACA ?

- To represent your organisation and rare diseases patients at the European level;
- To put your expertise and experience in good use, by integrating one or more of our Working Groups;
- To share your association's successes and struggles, and to come up with new ideas and projects;
- To build bridges with us: bridges between patient associations, between patients and medical experts, and between European borders!

Some examples of projects you can help us develop by integrating our different working groups :

- Writing of European Clinical guidelines on rare and complex syndromes. Patients are needed as guideline experts!
- Disseminating knowledge by creating workshops or webinars on patients' care trajectories, transition of care, on a certain syndrome, on how to access expertise in the field of rare diseases, etc - any topics can be brought on the table;
- Representing the ERN-ITHACA network in international or European congresses or meetings.
- Pushing forward a new underway tool that could change the medical care for European patients called the Clinical Patient Management System (CPMS): a secure internet platform used to discuss complex and rare patient cases between specialists from different countries and different fields of expertise
- And more...

Come and join us!
Together, we go further!
https://ern-ithaca.eu/contact/

FOCUS on the ePAGs

In 2016, EURORDIS and the European rare disease community established 24 European Patient Advocacy Groups (ePAGs) aligned to the clinical scope of each ERN. Today, there are over 280 ePAGs to represent the wider patient community in the development of ERNs.
The role of an ePAG is to represent the voice and interests of their patient community and act as the bridge between their community and the ERN, notably by participating in the Patient Board (see below) and ITHACA activities.
Membership of ePAGs is open to all rare disease patient organizations (EURORDIS members and non-members based in the European Union). Recruitment is ongoing. To candidate as an ePAG please contact Anne Hugon ERN ITHACA Project Manager anne.hugon@aphp.fr

FOCUS on the ERN ITHACA Patient Advisory Board

The Patient Advisory Board is part and parcel of ERN-ITHACA's governance and thus participates in each Workgroup or transversal activities and is represented in both the Executive Committee (2 representatives: chair and co-chair) and the Steering Committee (5 representatives *ToR). Through this organization, ITHACA aims to maintain a high degree of cooperation between ePAGs and other National or European lay public or patients' organizations at all levels of decision making and to ensure that all the ERN activities include patients and their families.