"Patient Workshop" Paris dec 1, 2026

ABSTRACTS Book 2026 – Patient Advisory Board

Learning From Each Other Workshop

Showcase Best Rare Disease Projects and Activities

Final selection nov 2026

Selected Abstracts (in no particular order)

- 3. SYNAPSE : Translating Family Caregivers' Experiential Knowledge into Professional Training Through Participatory Action Research in Angelman Syndrome
- 5. Kabuki INSIGHTS: combining electronic health record and survey data in a patient organisation-led natural history registry
- 6. Tuberous Sclerosis Complex Making the Invisible Visible
- 7. An example of a collaboration spanning more than a decade between clinicians, family members, and children with Mowat-Wilson syndrome, facilitated by the Italian Mowat-Wilson Association
- 8. ALL IN ONE: Where Relationship Meets Science in BWSp - The therapeutic alliance led by AIBWS OdV in synergy with the ERN-ITHACA community
- 9. From Parent-Led Vision to Global Consensus: Lessons in Collaboration, Trust and Shared Commitment
- 11. Partners in Research and Care: Collaboration between Patient Representatives and Professionals in Kleefstra Syndrome
- 12. Patient–Professional Collaboration in Rare Diseases: Connecting Community Care, National Expertise and European Networks in Romania
- 13. Communication Centres for Persons with Disabilities: Building a Nationwide System of Support for People with Complex Communication Needs

Abstracts Retained for Other Parts of the Programme

- 2. The Involvement of Rare Disease Patient Organisations in Research will be included in Part 1 of the programme led by Alliance Maladies Rares France.
- 4. Access to Information and Paternalism in Rare Disease Care: An Expanded European Follow-Up Analysis may be included as a short communication during the afternoon session.

Abstracts Not Selected

The following abstracts did not sufficiently match the scope of the call

- 1. From Stories to Science: AGO Syndromes as a Model for Ultrarare Disease Research
- 10. Characterization of NARS1 c.1600C>T p.Arg534* Across Patient-Reported Phenotype Surveys

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From Stories to Science: AGO Syndromes as a Model for Ultrarare Disease Research

Patient-led initiatives can accelerate ultrarare disease research by connecting lived experience with scientific expertise. The Argonaute Syndromes Conferences held in Regensburg, Copenhagen, and Prague demonstrate how patient–science meetings can transform isolated efforts into coordinated research.

Bringing families, clinicians, and researchers together helps align scientific priorities with patient needs, particularly quality of life and the most disruptive symptoms. Scientific and lay presentations, poster sessions, and networking create opportunities for collaboration while putting faces to diseases and strengthening researchers' motivation, urgency, and accountability.

The impact extends beyond the meetings themselves. Growing international participation, new collaborations, grant applications, data sharing, and joint publications demonstrate increasing research momentum. Exchange of biological resources, including patient-derived cells and animal models, supports translational research, while development of iPSC models, biobanks, and therapeutic approaches such as antisense oligonucleotides shows progress towards treatments. Virtual participation and online dissemination further extend the conferences' reach.

Patient organisations enable this ecosystem through fundraising, registries, biobanking, data sharing, and sustained family engagement. The Argonaute experience suggests that patient–science conferences can serve as research catalysts, providing a scalable model for accelerating discovery and translation across ultrarare diseases."

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The Involvement of Rare Disease Patient Organisations in Research

The involvement of French rare disease patient organisations (RDPOs) in research represents an essential yet underexplored area. This study aims to investigate the contributions, barriers, and enabling factors shaping the participation of patient organisations within the scientific landscape, in order to better understand their positioning and support their participation in research. The data presented are derived from a sample of 230 organisations. Research was identified as a high priority by 76% of surveyed organisations, alongside patient support (89%) and patient representation (78%). This strong interest is reflected in substantial engagement in research activities, with 97% of organisations either already involved or planning to become involved in the near future. Over the past three years, French RDPOs have engaged in research at multiple levels: national (53%), European (30%), and international (21%). Beyond financial support, their participation spans the entire research cycle. In conclusion, This study highlights the active and multifaceted engagement of patient organisations in rare disease research (RDPOs), extending well beyond financial contributions. It suggests that barriers to engagement are not primarily due to a lack of opportunity or willingness, but rather to insufficient structured dialogue and limited recognition of patient organisations as equal partners. Although participatory research is increasingly established, it remains imperfect. Access to and use of research findings by RDPOs remain limited, and only a minority fully leverage the knowledge generated. Strengthening collaboration, as well as the legitimacy, visibility, and dissemination of research outputs, is therefore essential to maximise the impact of RDPO engagement in research.

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SYNAPSE : Translating Family Caregivers' Experiential Knowledge into Professional Training Through Participatory Action Research in Angelman Syndrome

SYNAPSE: People with Angelman syndrome (AS) have complex lifelong support needs, including severe intellectual disability and significant communication impairments. Professional caregivers may lack the specific knowledge and skills required to provide individualized support. Family caregivers develop substantial experiential knowledge through daily interactions with their relatives, but this knowledge is not always integrated into professional education.

SYNAPSE aims to develop an innovative training programme for care professionals by collecting, analyzing, and formalizing the experiential knowledge of family caregivers, combining it with scientific and professional expertise.

Initiated in 2020 by the French AS patient's organization (AFSA), SYNAPSE follows a participatory action research approach. The first phase (2020-2023) brought together family caregivers, professionals and experts to identify support needs, analyze real-life situations, and develop shared knowledge. A second phase (2023-2024) tested the resulting methodology in three special schools, enabling the team to identify strengths, limitations, and areas for improvement. The third and current phase (2025-2027) focuses on translating the knowledge generated into a structured professional training programme, using practical case studies and participatory learning methods.

The research profess generated a solid base combining family caregivers' experiential knowledge with professional and expert perspectives. Testing the methodology in three special schools provided an opportunity to refine the approach before its wider dissemination. The current phase is translating these findings into training resources addressing the specific support needs of people with AS, with particular attention to communication, including Augmentative and Alternative Communication (AAC), understanding challenging behaviors and promoting self-determination.

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Access to Information and Paternalism in Rare Disease Care: An Expanded European Follow-Up Analysis

OBJECTIVES:
Building upon preliminary findings, this study investigates how medical paternalism, diagnostic non-disclosure, and communication barriers impact rare disease (RD) patient communities across Europe. It evaluates the transition toward patient empowerment and collaborative care, contrasting Central and Eastern European (CEE/Ex-Soviet) health systems with Western European and Pan-European frameworks.

METHODOLOGY:
An international survey was deployed across Patient Organizations (POs) representing diverse rare conditions. Data analysis of this expanded dataset (N=199) is ongoing to finalize stratification across Western Europe, CEE, and Pan-European POs. Quantitative survey data was integrated with qualitative feedback from PO leaders.

RESULTS:
Qualitative findings define paternalism as being "done to, instead of doing with". In CEE/Ex-Soviet regions, 60% of POs report their input is frequently ignored by health authorities, compared to 34% in Western Europe. Consequently, 70% of Western POs achieved advocacy success stories, versus just 5% in CEE. Crucially, patient advocates bring specialized lived experience that complements clinical expertise, highlighting the value of equal dialogue. To overcome systemic bottlenecks, POs actively build collaborative partnerships with healthcare professionals, clinical networks, and industry stakeholders.

CONCLUSIONS:
Ongoing analyses confirm a persistent "cultural lag" in CEE healthcare, where formal consent masks operational paternalism. Recommendations call for standardized diagnostic disclosure protocols, mandatory inclusion of patient advocates as partners in ERN clinical guideline drafting, and structural support to bridge regional disparities in European rare disease governance.

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Kabuki INSIGHTS: combining electronic health record and survey data in a patient organisation-led natural history registry

Background: Kabuki syndrome is a rare multisystem neurodevelopmental disorder caused mainly by KMT2D or KDM6A variants. Longitudinal natural history data are limited, which slows outcome measure development, trial readiness and evidence-based care. The Kabuki Syndrome Foundation (KSF) launched Kabuki INSIGHTS in October 2026 to address this gap.

Study design: Kabuki INSIGHTS is a prospective, longitudinal, observational natural history registry with retrospective baseline collection, open to people of any age and in any country with a clinical, genetic or molecular diagnosis. It is fully remote and runs on the Matrix platform (Across Healthcare). The design combines patient- and caregiver-reported surveys with electronic health record (EHR) data. Surveys cover development, health-related quality of life and lived experience. EHR data capture diagnoses, procedures, hospitalisations, medications and laboratory results Participants can upload genetic reports or additional medical records. Patients enrol online with electronic consent, or a parent or guardian enrols on behalf of a patient. A participant-supplied CRID, a pseudonymous research identifier, links data across sources and over time. The study was designed under a minimal-risk framework with WCG IRB review, and a Steering Committee that includes a community representative oversees registry strategy and data sharing with qualified researchers under data use agreements.

Lessons learned: Earlier registry experience shaped three design principles. The data should be owned by the patient community, the data should be portable across platforms and partners, and EHR and survey data together are more valuable than either alone. Enrolling participants in Europe under US IRB oversight adds GDPR requirements, and extending EHR contribution beyond the US remains our main open challenge.

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Tuberous Sclerosis Complex Making the Invisible Visible

Background
Tuberous Sclerosis Complex (TSC) is a rare genetic condition (approximately 1:6,000 births) with variable physical and neuropsychiatric manifestations. International consensus recommendations support personalised care coordinated by TSC experts.
In 2021, the Republic of Ireland (ROI) had no formal data on TSC care or centre of excellence; anecdotal evidence indicated fragmented care and substantial challenges for people living with TSC (PLW TSC) and their families.
Aim This study generated evidence to support implementation of internationally recognised TSC care standards in the ROI.

Methods
1.Audits in 12 epilepsy centres compared care for 83 PLW TSC with UK clinical recommendations.
2.Thirty-four semi-structured interviews explored patient and family experiences.
3.Twenty-four semi-structured interviews examined healthcare professionals' perspectives.
4.An anonymous survey of 48 PLW TSC assessed illness burden and quality of life.

Key Findings
Epilepsy and TSC-associated neuropsychiatric disorders strongly affected PLW TSC and their families. Illness burden was often high and quality of life low. Care was fragmented, with little evidence of formal surveillance or coordination beyond one adult service. Participants agreed that personalised care aligned with international recommendations required a coordinated approach.

Implications for Policy & Practice
The PhD evidence and clinical and nursing leadership have supported a growing multidisciplinary team at St James's Hospital, Dublin, providing coordinated, personalised care. Awareness, research collaboration, and international engagement have also increased.

Conclusion
Robust data can drive understanding and decision-making, while partnership is essential to complex change. Services should be co-designed with patients, families, and the TSC community. The expanding TSC service and network are improving care for PLW TSC and their families in the ROI.

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An example of a collaboration spanning more than a decade between clinicians, family members, and children with Mowat-Wilson syndrome, facilitated by the Italian Mowat-Wilson Association

Our collaboration with families of individuals with Mowat-Wilson syndrome (MWS) began after 2008, when we reported the first Italian child carrying a pathogenic variant in the ZEB2 gene. Following the establishment of the Italian Mowat-Wilson Association, we developed a close collaboration with 69 Italian individuals and their families, supported by the Association's Past-President, Diego Zotti.

We have investigated the clinical and molecular features, including genotype–phenotype correlations, and published syndrome-specific growth charts. We have also participated in the meetings of the MWS Association, with interactions between clinicians, researchers, and the patient community.

In 2018, we conducted the Telethon Exploratory Project GEP14131 on MWS neuroimaging with William B. Dobyns and Alex R. Paciorkowski and we were invited by the Portuguese Association to attend the MWS European Conference in Cascais.

In 2020, Orphanet invited us to develop the Disease Template for MWS.

We have also worked closely with the pediatric neurologists led by Duccio Maria Cordelli (Bologna), recognizing the essential role of a multidisciplinary approach in the study and management of rare diseases. The dental team led by Maria Grazia Cagetti (Milan) investigated the facial and oral characteristics. In collaboration with the Biobank at Gaslini Hospital, coordinated by Chiara Baldo, we sent samples from 29 patients to Bekim Sadikovic in Canada, who identified the methylation profile associated with MWS. A collaborative study within ERN ITHACA is currently underway, with Marco Tartaglia (Rome) and Giuseppe Merla (Naples), to investigate whether methylation profile is the same in children with milder forms of MWS.

We are also contributing to data entry for the MWS Registry of the Italian National Institute of Health (ISS). Livia Garavelli and Duccio Maria Cordelli are members of the MWSF USA Medical Advisory Board. We have published 21 indexed English-language papers on MWS.

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ALL IN ONE: Where Relationship Meets Science in BWSp - The therapeutic alliance led by AIBWS OdV in synergy with the ERN-ITHACA community (1st Revision of the International Consensus and 2nd International Congress for BWSp)

Background: Beckwith-Wiedemann Syndrome Spectrum (BWSp) requires a complex clinical journey, where heavy hospitalization and severe psychological impacts weigh deeply on children and families. As individuals are a whole ""all-in-one"" rather than fragmented pieces, professionals must unite science with conscience: a profound sensitivity to understand lived experiences beyond their specialties. Six years after the 2018 International Consensus, updating the guidelines was urgent. Out of this vital necessity, AIBWS OdV conceived ALL IN ONE to unite the global community around a shared vision of care and human closeness.

Methods : The event brought two souls to beat as one. For the Consensus update, a global group of 50 clinicians and patient representatives (many from ERN-ITHACA) collaborated online for a year, culminating in three days of intense in-person work. Parallelly, the congress embraced all stakeholders, alternating scientific rigor with conviviality and team building. This allowed clinical expertise to dialogue with lived experiences, fostering capillary, lasting collaborations within the same space.

Results: Besides delivering a crucial International Consensus update for BWSp diagnosis and management, ALL IN ONE was a deeply human experience. It served as a relational crossroads that broke down institutional barriers, catalyzed trust, and transformed clinicians and families into a single, cohesive ecosystem. Connecting clinical priorities to real needs, it proved how a small, specialized patient organization can lead an ambitious international initiative built on sharing and solidarity.

Conclusions: ALL IN ONE demonstrates that true care stems from an indissoluble alliance between science and humanity. Coordinated by AIBWS OdV, this model offers a generative, replicable experience across ERN-ITHACA networks to return patients' lives and voices to the center of decision-making.

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From Parent-Led Vision to Global Consensus: Lessons in Collaboration, Trust and Shared Commitment

Background: SATB2-associated syndrome (SAS) is a rare neurodevelopmental disorder without clinical practice guidelines. Fragmented expertise and disparities in care inspired a parent-led initiative to unite international experts and families in developing the first guidelines for SAS.

Methods: Led by a parent representative and president of SATB2 Europe, this volunteer-driven initiative brought together over 60 clinicians and researchers and 16 parent representatives, supported by ERN ITHACA methodology. Ten multidisciplinary working groups combined scientific evidence, clinical expertise and lived experience. Parents helped prioritise clinical questions and shape recommendations. Alongside The Care4SAS survey was created to address knowledge gaps, receiving 256 caregiver submissions from 37 countries.

Results: In October 2026, 70 participants from 18 countries met in Ljubljana for a hybrid consensus meeting, producing over 100 draft recommendations across ten domains. Building and sustaining this global network required motivating volunteers, coordinating across time zones and cultures, navigating diverse expectations and competing commitments, and securing resources without dedicated funding. Mutual trust, shared ownership and a common purpose helped overcome these challenges.

Conclusion: Meaningful parent–clinician collaboration goes beyond consultation to genuine partnership and shared responsibility. Respect for complementary expertise, trust and collective commitment enabled progress despite limited resources. This experience offers transferable lessons for rare disease communities seeking to build effective partnerships and co-produce person-centred clinical recommendations.

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Characterization of NARS1 c.1600C>T p.Arg534* Across Patient-Reported Phenotype Surveys

NARS1 encodes asparaginyl-tRNA synthetase 1, a cytoplasmic enzyme essential for protein synthesis. Variants in the NARS1 gene have been linked to a spectrum of early-onset neurodevelopmental conditions. The NARS1 c.1600C>T (p.Arg534*) variant, found on human chromosome 18q21.31, is a nonsense mutation inherited in an autosomal dominant pattern, arising de novo in all reported cases to date. It is currently classified as likely pathogenic. Located near the C-terminal end of NARS1, this variant results in a gain-of-function effect that disrupts normal translational activity. Developmental delay and microcephaly are prominent features, often accompanied by learning and intellectual disabilities. The known phenotypic spectrum includes neurodevelopmental impairment, behavioral comorbidities, growth failure, feeding difficulties, musculoskeletal involvement, and gastrointestinal concerns, frequently presenting within the first year of life.This poster presents phenotypic characterization of 10 individuals with NARS1 c.1600 p.Arg534* using patient-reported data collected through RARE-X — a registry designed to capture real-world phenotypic information directly from patients and caregivers. By cross-referencing data across multiple survey instruments and organ systems, we aim to build a more complete picture of what the NARS1 c.1600C>T (p.Arg534*) variant looks like in a pediatric population.

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Partners in Research and Care: Collaboration between Patient Representatives and Professionals in Kleefstra Syndrome

Background: Kleefstra syndrome involves complex care needs, scattered expertise and limited evidence. Over the past five years, a partnership between patient representatives and professionals has linked lived experience, clinical expertise and research to strengthen knowledge and care.

Methods: Patient representatives and professionals collaborated across scientific and family conferences, registry research, guideline development and accessible care resources. Activities included jointly organising Kleefstra syndrome family conferences and also the first Kleefstra syndrome scientific conference in Ljubljana in 2023 supported by an EJP RD networking support grant, alongside supporting GENIDA. The European collaboration also connects with networks of patient representatives and professionals in the USA and Australia, extending joint work beyond Europe.

Results: GENIDA collaboration produced a co-authored caregiver-reported phenotype study involving 172 individuals, using data collected during 2016–2022 (European Journal of Medical Genetics, 2024). The ERN ITHACA guideline process brought together 60 professionals and patient representatives from 17 countries across multiple continents, including an international consensus meeting at Erasmus MC, 15–17 February 2024. This resulted in an international evidence-based clinical guideline with 66 recommendations (Genetics in Medicine, 2026). Professionals and patient representatives subsequently co-authored a plain-language Practical Care Guide for Families and Professionals and a Detailed Clinical Surveillance Scheme (Zenodo, 2026).

Conclusion: Trust, mutual respect and complementary expertise sustained collective co-production across research, care and education. Transferable lessons are to involve patient representatives early, share leadership, recognise their professional and experiential expertise, and translate evidence into resources families can use.

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Patient–Professional Collaboration in Rare Diseases: Connecting Community Care, National Expertise and European Networks in Romania

Background: People living with rare diseases face fragmented care, delayed diagnosis and difficulties accessing coordinated medical and social support. Collaboration between patient organisations and healthcare professionals is essential to addressing these challenges.

Objective: To describe the collaboration between the Romanian National Alliance for Rare Diseases and the NoRo Centre in initiating RO.NMCA-ID, participating in ERN ITHACA and collaborating through NoRo and Medical Universities through ERN ITHACA and different projects: ex. ECOU, MEDI.COM.RARE, ProGeneRare, ExpertRARE and JARDIN

Approach: This descriptive overview examines a partnership combining patient advocacy, direct service delivery, multidisciplinary expertise and European cooperation. It considers how successive initiatives connect patients' needs with professional development, community support and the organisation of rare disease services.

Results: Established in 2017 with expertise centres in Timișoara, Craiova, Iași and Oradea, RO.NMCA-ID provided a collaborative platform for participation in ERN ITHACA. This connection supports knowledge exchange and links Romanian expertise with European resources. The NoRo Centre contributes practical experience in coordinating medical, social, psychological and educational support around individuals and families. Collaboration across the named projects illustrates a continuing effort to connect service development, professional learning and patient involvement. ECOU focuses on community education, patient coordination and follow-up, while ExpertRARE supports expertise centres through working tools and training for integrated care. JARDIN provides the broader European framework for integrating European Reference Networks into national health systems. Across these activities, patient and family perspectives help identify unmet needs and inform priorities for collaboration. The partnership brings together collective advocacy and experience from everyday care, offering complementary perspectives on service gaps and opportunities for improvement.

Conclusions: This experience illustrates a patient–professional partnership connecting community services, national expertise and European networks. Its contribution lies in aligning advocacy with care delivery and supporting patient participation in service development. Sustainable financing, defined responsibilities and systematic outcome evaluation are needed to consolidate these initiatives into accessible, coordinated and lasting rare disease care.

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Communication Centres for Persons with Disabilities: Building a Nationwide System of Support for People with Complex Communication Needs

People with complex communication needs face significant barriers to accessing information, expressing their needs, making decisions and participating fully in society. Ensuring their right to communicate requires accessible, professional and individualised support. This presentation focuses on the establishment of the Communication Centre for Persons with Disabilities, an initiative aimed at developing a comprehensive nationwide system of communication support in Poland.

The project is led by the Ministry of Family, Labour and Social Policy, in partnership with the Polish Association for Persons with Intellectual Disabilities (PSONI) and the Polish Association of the Deaf (PZG). Its main objective is to establish 16 Regional Communication Centres (RCCs), supported by a central Coordination Hub (Hub CK). The partnership brings together expertise in augmentative and alternative communication (AAC), Easy to Read (ETR), Polish Sign Language, communication methods for deafblind people, Braille and speech-to-text transcription.

The Regional Communication Centres will provide standardised services adapted to the diverse communication needs of persons with disabilities. A particularly important component is support for people with complex communication needs who require AAC. Services will include functional communication assessment, identification of individual communication goals, and the development, implementation and evaluation of an Individual Communication System. The approach places the individual, their preferences, abilities and active participation at the centre of the support process. Access to services will not depend on holding an official disability certificate.

An essential aspect of the initiative is the development of a qualified professional workforce and consistent service standards across all regions. This will be supported through specialist training, supervision, peer consultation, educational resources and quality monitoring coordinated by the central Hub.

The presentation will discuss the organisational model, the roles of the project leader and partners, the scope of services and the challenges of establishing a nationwide network. Ultimately, the initiative seeks to strengthen communication autonomy, promote independent living and ensure equal opportunities for people with disabilities to exercise their rights and participate in community life throughout Poland.