"Patient Workshop" Abstract Book – PAB

"Learning from each other workshop - Best practice sharing"

Clinics and Patient care, global collaboration

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Abstract title: From Model Transfer to Sustainable Impact: The NoRo–Frambu Collaboration for Integrated Rare Disease Care in Romania.

Authors: Dorica Dan – Romanian Prader Willi Association, NoRo Center Coordinator, ePAG chair, ERN ITHACA, and Kristian Emil Kristoffersen - CEO of the Frambu Resource Centre for Rare Disorders.

Goal of the abstract: to present the impact of the collaboration between the NoRo Center (Romania) and Frambu Resource Centre for Rare Disorders (Norway) as a long-term partnership dedicated to improving care, inclusion, and quality of life for people living with rare diseases.

Methodology: Rooted in shared values of empowerment, knowledge exchange, and interdisciplinary cooperation, this collaboration has focused on developing professional competences, patient-centered services, and sustainable models of community-based care. Through joint training programs, study visits, and exchange of best practices, the partnership has strengthened the capacity of Romanian professionals and organizations to deliver integrated services aligned with European standards.

The Frambu–NoRo collaboration also contributed to the creation of the NoRo Center as a model of a multidisciplinary, patient-driven hub for rare diseases, fostering the implementation of Norway Grants projects and promoting participation in European Reference Networks (ERNs).

Together, the two centers continue to build bridges between Nordic and Eastern European practices, advancing innovation and inclusion in rare disease care and policy.

Project NoRo – "Norwegian Romanian partnership for progress in rare diseases" has offered RPWA the opportunity to develop services for patients affected by rare diseases in Romania.

Here are some example of projects developed together**: Project "Norwegian-Romanian (NoRo) Partnership for Progress in Rare Diseases",** Implemented between April 2009 and April 2011, **"NoRo-Frambu, Partnership for Future" (**March 2015 - April 2016), **Project AD AUGUSTA PER ANGUSTA, „TEAM – Together, everybody achieve more", Project NoRo Document RARE (**15th of January and 14th of March 2017)**, ECOU (**27.01.2023 – 27.01.2024, EEA Grants 2014-2021", and **MEDI.COM-RARE (**26.10.2022 – 30.04.2024, EEA Grants 2014-2021). We have also involved our partners in the **INNOVCare**- Innovative Patient- Centered Approach for Social Care Provision to Complex Conditions Project (EASI Program, Progress component 2015-2018).

Conclusions: The NoRo–Frambu collaboration (between the NoRo Center for Rare Diseases in Satu Mare, Romania, and Frambu Resource Centre for Rare Disorders in Norway) has produced several tangible and lasting results, both in terms of service development, professional training, and policy influence:

- **Establishment and Development of the NoRo Center (Romania)**
- **Capacity Building and Professional Training**
- **Institutional Development and Networking**
- **Services for Patients and Families**
- **Policy and Systemic Impact**
- **Knowledge Resources and Dissemination**

How to shift from being perceived as a burden by professionals to being recognized as symbiotic partners

Authors: Daniele Palumbo, CHAMP1 Foundation Europe/CHAMP1 Research Alliance
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Objectives

The CHAMP1 Research Alliance greatly relies on professionals and is deeply thankful for their work. In turn, professionals are willing and happy to collaborate with us.
Through practical examples, we will illustrate how to achieve this synergy, offering guidance for other patient organizations on how to strengthen their engagement with professionals—being perceived as symbionts, not parasites.

Methodology

- Describe the establishment of a structured network of families and researchers, through dedicated conferences (USA 2022, EU 2023, USA 2024, EU 2025) and quarterly scientific calls enabling global collaboration.
- Demonstrate how the direct involvement of patients and families in prioritizing research directions (e.g. financing hypotheses on mitochondrial dysfunction based on observed symptoms) can drive meaningful progress.
- Show how thinking globally, fostering collaborations, and identifying genes with potential cross-disease relevance can broaden impact.
- Provide a concrete example of cross-disease collaboration with research groups already working on POGZ, particularly the teams of Prof. Gasparini, Prof. Zollino, and Dr. Tartaglia, to assess the extension of existing trials and protocols to CHAMP1.

Outcomes

- Significant increase in the number of research groups engaged in CHAMP1 over the past three years.
- Consolidation of an international patient–scientist network with regular and transparent communication flows.
- Initial steps towards extending clinical studies initiated on POGZ to CHAMP1, with prospects for faster trials using integrated cohorts.
- Active participation in conferences traditionally reserved for professionals (e.g. ESHG, SIGU).
- Hosting of the CHAMP1 conference in the EU with the participation of ~10% of known global cases and ~20% of European cases.

Impact and potential

- Scalability: the model can be applied to other ultra-rare diseases, especially where communities are fragmented and patient numbers are limited.
- Innovation: shifts the traditional paradigm by positioning patients as catalysts of research and bridges across parallel projects.
- Collaboration: integrates families, clinicians and researchers in a shared ecosystem, fostering “horizontal” trials across related genes.
- Future perspective: potential to leverage ongoing POGZ trials to accelerate intervention timelines for CHAMP1, and the other way round, reducing duplication of efforts.

Context: AlfaSAAC Project: Enhancing communication in children with Phelan-McDermid Syndrome

The Associação Phelan-McDermid Portugal (APMP) presents an innovative initiative developed by the Asociación Síndrome Phelan-McDermid (ASPM) which has significantly improved the quality of life for PLWRD, their families, and communities by promoting greater inclusion and full participation in society.

Proponent: Rui Barbosa Guedes (rui@phelan-mcdermid.pt) and Co-authors from ASPM: Norma Alhambra - President; Constance Colin - Coordinator; Leyre de Luis Martínez de Ibarreta - Social Worker.

Objective: The AlfaSAAC project seeks to empower nonverbal individuals with IDD by providing them with a means of communication, promoting their autonomy and encouraging active participation in the community.

Why: Communication is a crucial element for the inclusion of PLWRD. Its absence often leads to significant stigma, stemming from the stereotype "if someone cannot speak, he also cannot understand". The AlfaSAAC project was designed to provide alternative forms of communication to stimulate the natural speech for children affected by Phelan-McDermid Syndrome (PMS). PMS is a genetic condition caused by the loss of genetic material on chromosome 22. The absence of this gene is associated with delayed developmental milestones, most notably in speech and language (+50% have no or few words, 30% single words and less than 20% are able to pronounce sentences), where receptive language abilities typically exceed expressive language skills.

Methodology: The project intends to fill the gap in language acquisition, ensuring PMS patients alternative ways of communicating with their families and close environment, thus reducing frustration and social isolation. It is based on the use of Alternative and Augmentative Communication (AAC) with the objective of building language. This directly impacts social relationships and cognitive development, acting as a catalyst for a child's emotional world by supporting emotional regulation. It also plays a role in literacy, which underpins all forms of interactions and enables self-determination.

The innovative approach focuses on promoting collaboration and community engagement, recognizing that a successful implementation of AAC depends on strong cooperation among all stakeholders involved in the lives of PLWRD: *"Having a AAC communication device doesn't mean knowing how to use it, just as a piano doesn't make you a pianist."*

Communication and language acquisition is learned and acquired through daily experiences and routines in familiar environments. The success of the project therefore relies on the combination of individualized AAC professional interventions with the full and meaningful engagement of those in the patient's close social environment (parents, close family members, school teachers, therapists, etc.).

The Evidence-Based Practice (EBP) is that assisted language stimulation reduces input-output asymmetry and promotes expression and understanding for individuals who use AAC, thus requiring a strong investment on the interlocutors' training – be it childhood, adolescence or adulthood. The family is the link between the PMS affected person and society.

The AlfaSAAC project is designed on a 10-months basis and developed by an AAC multidisciplinary professional team in close collaboration with the Social Worker of the Spanish PMS Association. It comprises an *individual initial assessment* of the PMS patient's language skills and his/her home and learning environment. It is followed by a *50h training program* to families and close environment and is completed with an individual guided practice where each PMS family is accompanied by a professional to adjust as much as possible the AAC therapy to their specific needs and context. It is recommended that teachers and/or therapists who regularly participate in the lives of PMS patients also accompany families in this process, and more professionals are joining.

PMS families are consulted throughout the project to capture best practices and adapt the support provided to specific needs of the PMS community. During this phase, participating families constitute a strong support group which helps identify the barriers (factors and elements that hinder) and facilitators (factors and elements that promote and facilitate) in the implementation of AAC, enabling valuable mutual support.

Challenges in implementing an AAC strategy: Despite the clear benefits of early AAC access, there is still a limited number of therapists with specialized training in AAC, and public financial support is often insufficient or lacking altogether. For AAC to be effective, strong coordination among professionals involved in the child's care is essential. However, this collaboration can in some cases be difficult as many may lack the time to fully engage. Also, integrating AAC into daily family life can be difficult due to time constraints, caregiving responsibilities, and limited training among family members. When systems are overly complex, this can lead to user apathy, unbalanced interactions, and decreased motivation. In schools, AAC can be perceived as taking time away from teaching rather than supporting it.

Evidence of Success and Best Practice Potential Impact: Thanks to the remarkable impact on beneficiaries, ASPM families steadily increased their participation, with over 50% more families joining since the first edition in 2023.

Families reported substantial progress in communication skills and behavioural regulation of the person with PMS. They noted that these changes helped the entire family overcome the frustration associated with communication difficulties, while fostering this individual's social interactions outside the home, thanks to greater understanding from others. The project not only improves the quality of life for affected individuals but also strengthens family dynamics.

Another positive outcome of the AlfaSAAC intervention model is the central role of support groups, which serve as safe spaces where families can share their experiences, learn from one another, and strengthen their sense of belonging to a united and supportive community.

Transferability and scalability: This project shows strong potential for replication in other rare genetic conditions with similar communication challenges. Although it should be personalized to each group, its robust individualized, family-centered structured practice model can be adapted to various conditions, helping reduce frustration and improving communication and quality of life for both individuals and their families.

Vergiate, 6 october 2025

CHILDREN NEED TO BE AWARE

At home, parents cuddle and spoil their children, filling them with kisses and hugs, looking after them lovingly.
There are days, though, that parents take children to the hospital or to the doctors without saying anything.
By putting ourselves in children's clothes, we can understand how much this scares and confuses them.
They are in these unknown spaces, or places were they carry bad memories; they are stripped, visited, touched, stung with needles or whatever.
Doctors don't talk to them directly, they just ask parents questions; no one explains to them why they are there and why they have to do those things.
For children, it is difficult to understand all this.

Obviously, the intent of the parents is to be quiet about these visits and checks for not scaring or agitating their kids.

Unfortunately, that's not how it works. Children perceive their parents' emotional states, even before understanding and learning words. Particularly for those who undergo major hospitalization, repeated and timed checks, we cannot think that all this has no impact on the psychological and emotional aspects of children. **Children need preparation and awareness before arriving at the doctors' offices**.

Aibws decided to try to help the children through the butterfly named Biwy. First comes “Biwy's friends”, a book created with the help of the psychologist and a pediatrician from the Scientific Committee. It’s dedicated to pre-school children, to support parents in approaching the topic of the “syndrome” with their children. In fact, in a moment of great intimacy, in which the reference adult reads and the child stands next to him to see the figures, the child receives small notions and small answers to the first questions about the syndrome and/or his own characteristics. Clearly, the moral of the book is meant to make children understand that diversity can be an opportunity, that every person has an important value, and that applies to them too.

However, wanting to help ALL children (syndromic or not), but always keeping the children's point of view, Aibws decided to change the tool and delve deeper into some specific topics. Again with the help of the psychologist and pediatrician of the Scientific Committee, a lineup of topics has been made that we’ll cover with the “cartoon” tool. This tool offers a stimulating multimedia experience that involves sounds, colors and movements, capturing attention and facilitating the understanding of complex concepts through visual immersion.

So the format of Biwy was born. In each episode (3 have already been released) Biwy is going to explain to the children what the visits and tests they’ll do in the hospital consist of. Syndromes or diseases are never mentioned, so that it can be used by anyone. **Each cartoon also comes out in English**. The first explains what the blood sampling consists of, the second explains why they must/can trust doctors/specialists, and the third recounts the ultrasound and x-ray.

It’s a project that takes on all the required characteristics as it is, clearly, adaptable to various contexts and can, also, encourage collaborations between associations. In fact, from the second episode, you’ll be able to see that they were made together with Aimp, an association that deals with the spectrum of PROS.

Making children aware and prepared means having a greater chance that they’ll be not afraid, that they’ll collaborate, that they won’t have emotional consequences and that the results of the tests are more correct, as well as performed in a shorter time.

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Building Patient-Driven Natural History Studies for Rare Neurodevelopmental Disorders: Lessons from the PATRE SYNGAP1 and PATRAS RASopathies Registries in the EURAS Project

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Objectives

The EU-funded EUropean network for neurodevelopmental RASopathies (EURAS) was initiated through collaboration between the German patient organization Syngap Elternhilfe e.V. and its medical advisory board.

One of the central aims of EURAS is to establish high-quality, interoperable registries for four neurodevelopmental RASopathies — Costello, cardiofaciocutaneous (CFC), Noonan, and SYNGAP1 syndromes — recognizing that comprehensive natural history data are critical to prepare rare disease communities for future clinical trials and evidence-based care.

Methods

The registries are co-designed and jointly operated by patient representatives and clinical experts, supported by 27 European patient organizations across 12 countries. The multilingual platform, based on REDCap technology, enables data collection through online caregiver questionnaires in the myCap app, currently available in seven languages. Patient-reported outcome measures (PROMs) are captured using harmonized Common Data Elements (CDEs) aligned with Human Phenotype Ontology (HPO) terms and the FAIR (Findable, Accessible, Interoperable, Reusable) data principles. Longitudinal follow-up allows systematic tracking of developmental, neurological, and behavioral symptoms as well as treatment responses.

Results

As of October 2025, 484 patients with SYNGAP1-related disorders are registered in the PATRE registry. The distribution by survey language illustrates the broad reach of the registry: German (114), French (68), Italian (52), Dutch (32), Polish (45), Spanish (93), and English (80).

In parallel, the PATRAS registry for RASopathies has enrolled 406 patients across Costello, CFC, and Noonan syndromes. Reported survey languages are German (150), French (69), Spanish (55), Polish (65), English (63), Italian (2), and Dutch (2).

These numbers highlight the feasibility of achieving large-scale participation in multilingual, patient-centered NHS.

Impact

High registration rates demonstrate both patient motivation and the value of direct collaboration between families and clinical experts.

The EURAS registries provide a replicable model for sustainable, patient-centered natural history studies in rare neurodevelopmental disorders. Their design supports not only clinical trial readiness but also long-term improvements in evidence-based care.

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ANGELMAN CAREGIVER SURVEY

Authors: Ms Ellen Koekoeckx, Foundation for Angelman Syndrome Therapeutics, Mr. Anthony Moisan, FAST France, Ms Karolina Pospieszyńska-Martysiuk, Ms Hanna Dianow, FAST Poland, Ms Stephanie Azout, FAST LATAM

OBJECTIVES

The AS caregiver survey aims to:

- **Collect anonymous caregiver-reported data** to understand demographics, medical care, therapies, caregiver burden, and family priorities in Angelman syndrome (AS).
- **Assess willingness to participate** in clinical research, natural history studies, and newborn screening.
- Provide evidence to **inform clinical trial readiness** and advocacy efforts at both national and international levels.

METHODOLOGY

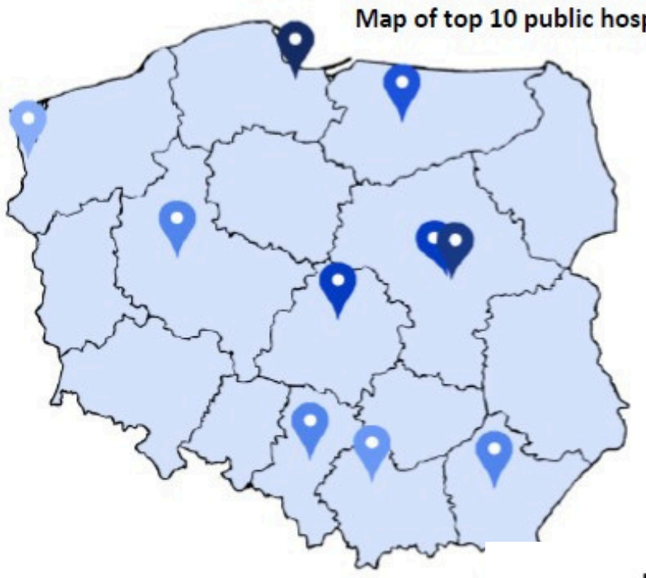
- **Developed and piloted by FAST France and refined by FAST Poland** and subsequently implemented by FAST LATAM
- Refined by FAST into a **uniform survey template** so that it can be easily replicated by other AS organizations
- Conducted using **Google forms** for accessibility and low cost
- **Disseminated** by local organization through social media channels and newsletters to maximize reach.
- **Implementation planned** by FAST UK, FAST Canada, and AS organizations in MENA and Brazil.

OUTCOMES FAST POLAND

- 148 respondents, majority children aged 0–12 years.
- Over 80% of families expressed **willingness to participate in clinical trials** and newborn screening.
- **Key challenges identified**: communication difficulties, caregiver tiredness , and long-term stress.
- The data provided robust, **country-specific evidence used to engage policymakers and industry.**
- **Direct impact:** survey outcomes supported the successful initiation of clinical trials in Poland and let to a formal study on caregiver burden in Angelman syndrome

POTENTIAL IMPACT

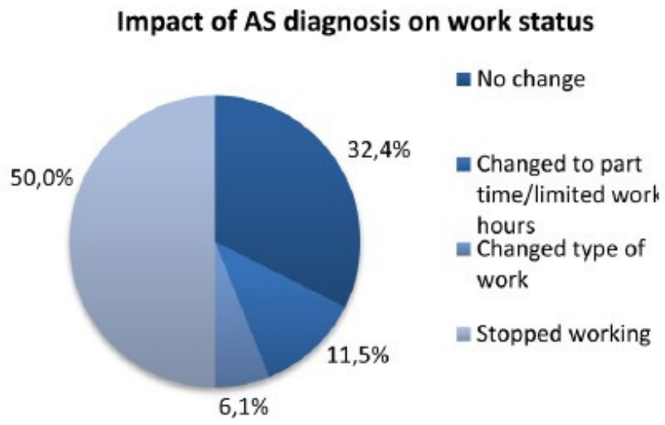
The Angelman Syndrome Caregiver Survey provides actionable insights for researchers, clinicians, regulators, and policymakers. Its replicable design ensures that other rare disease communities can adopt the model to generate evidence that accelerates clinical trial readiness and improves patient outcomes.



Map of top 10 public hospitals with specialists' care, that AS individuals were under in Poland, as stated by their parents/caregivers in the survey.

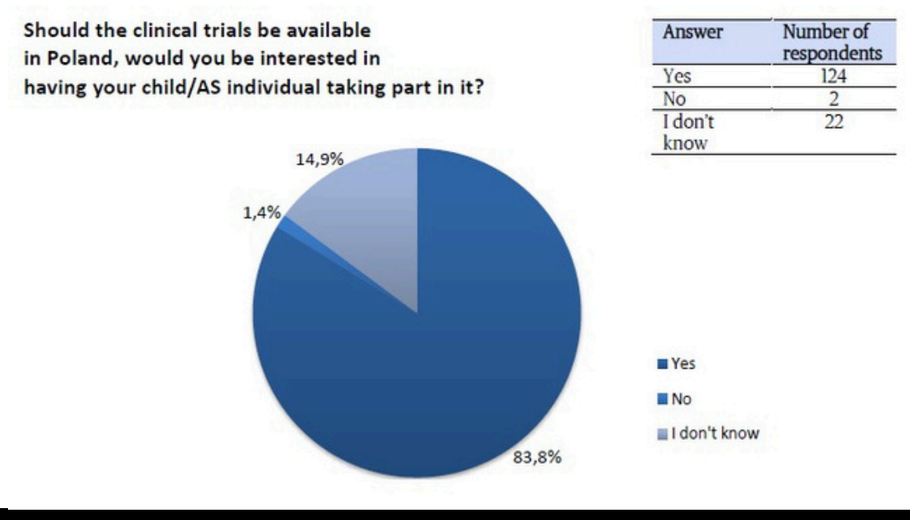
Pins of darker colour stand for more popular facilities, while pins of lighter colour - less popular.

The most popular hospitals in the survey results are facilities in Gdańsk, Warsaw and Łódź.



FUTURE PLANS

- **Expand implementation** to additional organizations worldwide.
- **Compare and analyze data** across countries to identify meaningful differences and shared patterns.



Answer	Number of respondents
Yes	124
No	2
I don't know	22

CONCLUSION

The survey model demonstrates that caregiver-led, low-cost, and scalable data collection is feasible and impactful. It empowers local patient groups, strengthens advocacy, and contributes to the global Angelman data ecosystem.

To maximize value, data collection must be consistent across regions and over time so results can be compared and pooled. That consistency starts with strong survey design: involve survey-methodology experts early to define concepts, minimize bias, pilot and validate items, and set clear update protocols.

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A Paternalistic Barrier: The Fight for Patient Information Access in Rare Disease Care in Hungary

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Introduction

Disorder of the Corpus Callosum (CCD) is a rare disease where the corpus callosum is not developed properly. The symptoms can be mild if isolated, or they can result in severe cumulative disability if other syndromes are present. Patients with rare diseases often face significant challenges in their journey, but a unique issue identified in Hungary is the systematic withholding of critical diagnostic information by medical professionals. This issue is rooted in a culture of paternalism within the healthcare system, which significantly impacts patient empowerment, family planning, and trust. This abstract presents a historical account and a survey-based analysis of this phenomenon, focusing on patients with a CCD.

Objectives

The primary objective of this project was to document the experiences of patients with DCC, who were not informed of their diagnosis despite it being present in their MRI reports. A secondary objective was to survey the extent of this paternalistic behaviour across Central and Eastern Europe and its effect on patient organizations (POs) and patient-doctor relationships.

Methodology

The project employed a two-pronged approach:

1. **Historical Account:** A personal case study and community anecdotes were used to illustrate how patients with DCC, whose condition was detected by MRI, were not informed of their diagnosis.
1. **Survey Research:** An online survey titled "Paternalism in Healthcare in Europe" was conducted to gather data from patients and POs across Europe. The survey aimed to compare experiences between Western and Central/Eastern European countries regarding access to information, patient-doctor communication, and the involvement of POs in decision-making.

Outcomes & Results

The findings reveal a significant disparity in patient information access.

- **A Paternalistic Culture:** A 2023 "non-representative research" found that while 1% of patients worldwide are not informed of their condition by doctors, this figure is 33% in the DCC community in Hungary. This is supported by the personal account of a patient who was told about their MRI results 13 years after the scan.
- **Survey Data on Paternalism:** Out of 18 survey respondents, 14 felt they were experiencing paternalism. The majority (17) believed the best model is a collaborative one where the doctor and the patient organization decide together, with 2 respondents choosing a model where the patient organization takes the initiative. However, many respondents also reported being ignored by policymakers or having their recommendations excluded even when formally included.
- **Patient Organization Influence:** Data from a Patient View survey showed that Central and Eastern European patient groups are less often seen as influential by academic/scientific bodies and government committees compared to the global average.
- **Note:** More data will be available from the "Paternalism in Healthcare in Europe" survey by the time of the conference.

Potential Impact

This research highlights a critical barrier to patient-centred care: the lack of access to fundamental health information. The systematic failure to inform patients about their diagnosis, particularly for a genetically linked condition like DCC, has severe implications for family planning and individual well-being. By exposing this paternalistic trend, the project aims to advocate for a more collaborative healthcare model. The ultimate goal is to empower patients and POs to demand their fundamental right to information, ensuring that a diagnosis on an MRI report is no longer a secret kept from the person it most affects.