ERN ITHACA 2024 Patient Workshop

Abstracts Book : Learning From Each Other Workshop

Thursday 12 of December, Bucharest

Showcase Best Rare Disease Projects and Activities

ERN ITHACA 2024 Patient Workshop is dedicated to strengthening collaboration between healthcare providers and patient advocacy groups to improve support and care for people affected by rare genetic diseases. This interactive event combines presentations, abstract showcases and in-depth discussions to encourage meaningful engagement among all participants.

Clinics and Patient care

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Abstract title: NoRo in community or how to upscale your resources?

Co-authors: Dorica Dan – president RONARD, RPWA, ARCrare, coordinator NoRo Center, Prof. Dr. Maria Puiu, Spitalul de Copii Louis Țurcanu Timisoara, Dan Alexandru Tiberiu, NoRo Center - Director of Operations, Prof Dr. Emilia Severin, UMF Carol Davila București, Dan Alexandra Loredana, psychologist NoRo Center, Director of social services, Conf. Dr. Ioana Streața, UMF Craiova, Prof. Dr. Adela Chiriță Emandi, Children Hospital Timișoara, members of ERN ITHACA through RO.NMCA-ID.

Background: Patients need to be partners in shaping the RD ecosystem because they bring their everyday experiences in living with RDs, they can significantly contribute to improve information, care, research and treatment development and, they do advocate for system change.

Aim: To generate a translational knowledge agenda, which identifies and prioritizes patients need and system change.

Methodology:

Since the establishment of NoRo Center in 2011 we have created a multidisciplinary team at the center, composed of 30 professionals from different fields (doctors, social workers, special education teachers, speech therapists, kinetotherapists, psychologists, art therapist, nurses, etc.) that provide everyday therapies and life skills for our 85 children and adults from the center. We do also counseling and trainings with families and organized more than 150 patients' groups, face to face for 1 week or online for the same period (to create connections, support groups, sharing experience and connect them with different experts and services that they need).

For the most isolated patients, since 2016 we are training community nurses in case management for rare diseases, in order to reduce the waiting time for patients to be connected with the professionals that they need.

We exchange experience with schools and kindergartens in the community, where our children are integrated to ensure continuity of care and ensure their inclusion in society.

Patient involvement at every stage ensured that their perspectives were integrated, solidifying patient-centeredness and integration of these perspectives in the local, regional and national strategies as part of the NCRD. Together with 4 other centers of expertise from Timișoara, Craiova, Iași and Oradea, we initiated in 2017 RO.NMCA-ID, and became full member of ERN ITHACA.

Conclusions: This collective endeavor reflects the collaborative spirit needed for rare disease care. This knowledge agenda will not only guide where we need to bridge the gaps but will also boost interdisciplinary collaboration to push the field of rare diseases consortium and change the system of care in Romania. Patient engagement, transparency, and a comprehensive approach make this knowledge agenda a pivotal step toward addressing the pressing integrated care needs and priorities in this domain.

MULTIDISCIPLINARY CLINIC FOR PATIENTS WITH MACROGLOSSIA IN BWS

Project made by AIBWS ODV – Italy

What is BWS: BWS is a cellular overgrowth syndrome caused by few types of imprinting defects.
The symptoms are about thirty, so our children are very heterogeneous.
The symptoms that most affect the daily life of children and families are macroglossia, the tongue overgrown that doesn't fall into the oral cavity, and the hemiipertrophy, when one or more parts of the body overgrow compared to the mirror (for example they can have a leg lenght discrepancy).
What all BWS children have in common is a greater predisposition to the development of cancers from internal organs.
The management of the syndrome, therefore, involves a strong hospitalization in the first years of life but also a strong multidisciplinary approach.

Who is AIBWS: AIBWS is the Italian association for Beckwith-Wiedemann syndrome and related diseases (eg Wilms tumor, hepatoblastoma and isolated hemipertrophy) that has been involved in supporting affected people and their families for 20 years, support research and disseminate knowledge.

The problem and the need: As I said at the beginning, multidisciplinary approach is fundamental and one of the major symptoms is macroglossia. Obviously, not all macroglossia are surgical needing and not all will require orthodontic care. Each child must have a personal clinical pathway. Unfortunately, there are not many specialists in this pathology and, above all, they are not all present at the same place and at the same time.

Today's good practices: bringing together all the specialists needed in one hospital, in one morning a month, to visit only BWS patients.

The project is in collaboration with San Paolo Hospital in Milan (member of ERN CRANIO) since 2020, and with Regina Margherita Hospital in Turin since last october.

What AIBWS do:

- takes care of the agenda of the clinic, and it's the pivot between the families and the structure;
- finances the presence of specialists to ensure continuity and the possibility of developing new experts.
- a representative of the association is present at each meeting to facilitate activities, support families and give concrete help (for example if parents need to talk with doctors or go to the bathroom, our representative takes care of entertaining children)

What the specialists do:

- They take care of the bureaucratic part for access to the clinics, so families only have to show up in the waiting room.
- Each individual specialist visits, on a rotating basis, each individual patient. This way, the visits are not too hectic and allow children to acclimatize and show their best.
- At the end of all visits, specialists meet to discuss each individual case and issue a coordinated and shared report

The specialist involved are:
- Maxillofacial surgeon
- orthodontist
- speech therapist
- otorhinolaryngologist
- neurologist expert in sleep problems

RESULTS:

Families don't have to travel around and waste time in booking, and they can find experienced specialists to be trusted.

Specialists are gaining more and more experience and gathering useful data; the first scientific paper will be published shortly.

Children find a peaceful atmosphere, friends to play with and always the same friendly doctors.

Thank you.

Monica Bertoletti
monica@aibws.org

Title: PKS Clinic: A Multidimensional Approach to Pallister-Killian Syndrome Care
Authors: Samantha Carletti, Duccio Maria Cordelli, Chiara Locatelli, Giacomo Sperti, Alessandro Rocca, Anna Fetta
Contact Email: info@pksitalia.org

Abstract:
The PKS Clinic project was developed to meet the complex needs of patients with Pallister-Killian Syndrome (PKS), a rare genetic disorder characterized by multi-organ anomalies, epilepsy, neuromotor developmental delays, and intellectual disability. Due to the mosaicism of PKS, clinical manifestations differ widely among patients, requiring personalized care that presents emotional and logistical challenges for families.

Located at the IRCCS Azienda Ospedaliero-Universitaria of Bologna and the IRCCS Institute of Neurological Sciences of Bologna, the PKS Clinic serves as a center of excellence on a national and European scale. Its mission includes delivering multidisciplinary care, promoting research, and supporting families of children with PKS. Patients undergo a comprehensive 3-day hospitalization where they receive all necessary evaluations, including neurology, cardiology, pulmonology, gastroenterology and orthopedics. During the stay, a parent stays with the child in the hospital room, while other family members are housed free of charge by Fondazione S.Orsola and the Bimbo Tu Association.

Key innovations include the creation of a European PKS Registry to collect and centralize clinical and genetic data, enhancing diagnostic accuracy and treatment approaches. Additionally, the clinic provides telemedicine services, virtual support groups, and specialized training for healthcare professionals to advance care for PKS and other rare diseases.

By integrating clinical care, research, and psychosocial support, the PKS Clinic provides a holistic and scalable model that improves patient outcomes and offers a replicable framework for other rare diseases. This innovative project bridges gaps in care while enhancing collaboration and knowledge sharing across Europe.

Abstract for ERN ITHACA – Workshop Meeting Bucharest December

Presentation by Anne Lawlor and Lyndsey Walsh

Title - Care coordination for all rare conditions in Ireland – A comparison between 22q11 Deletion Syndrome and 2q24 Deletion Syndrome

Introduction – This presentation will highlight the commonalities between 22q11 Deletion Syndrome and 2q24 Deletion Syndrome. It will highlight the commonalities between a relatively common rare condition and an ultra-rare condition. A paediatric 22q11 Clinic was established in Ireland in 2017 and this presentation will show how this clinic could serve as a blueprint for a model of care for all those living with a rare condition and their families.

Objective – The objective of the presentation is to consider the existing 22q11 Paediatric Clinic as a blueprint for a model of care for all children and adults living with a rare disease. It will introduce the clinic, outline its formation, and what it does. A comparison between a person living with 22q11 Deletion Syndrome and a person living with 2q24 Deletion Syndrome will be made to highlight the commonalities and how both individuals' needs could be met in a broader rare clinic. It will briefly consider further examples of persons living with a rare disease and further highlight the commonalities. It will discuss the importance of care coordination and how it has made such a meaningful difference for the 22q11 community.

Conclusion – In summary, the presentation will argue that in the context of complex care coordination for all rare conditions, there is a transferable model of care in the current paediatric 22q11 Clinic. This inevitably needs to be expanded to include all rare conditions but also extended to provide lifelong care.

Arabella, 2q24 Deletion

Aine, 22q11Deletion

Abstract: The Value of WaihonaPedia for your community, guide for community leaders

This presentation showcases the opportunities for family communities dealing with rare conditions to be empowered through the sharing of knowledge and experiences. It highlights how communities can benefit from collective wisdom, drawing on the lived experiences of families, caregivers, educators, doctors, and researchers. By creating a central, international, multilingual resource of practical information, participants can improve their understanding of conditions, enhance care, and reduce uncertainty, fostering a stronger sense of confidence and support.

As a European collaboration project, this initiative returns digital sovereignty to families. In an age where families are often dependent on big tech solutions with potential risks—such as privacy concerns or the power of companies like Elon Musk's enterprises or META to shut down services in Europe—this platform offers a more secure, community-driven alternative. It ensures that sensitive information is handled with care, while empowering families to retain control over their data and services.

The presentation also emphasizes the personal and collective impact this knowledge-sharing process has on families, caregivers, and professionals. Families feel more empowered and supported, caregivers develop better strategies, and medical professionals adopt a more collaborative, multidisciplinary approach. The functional benefits extend to improved information management, better communication, and reduced stress, resulting in stronger communities and lower healthcare costs.

Additionally, the presentation provides an implementation roadmap for community leaders to integrate their existing knowledge into a centralized platform, facilitating global collaboration and ensuring that communities are better equipped to manage the complex, long-term care associated with rare conditions. Examples of successful implementations are included to demonstrate the transformative potential of this collective, secure approach.

Abstract submission for **Learning From Each Other Workshop**
in Bucharest, December 2024

author: **Erika Stariha**, ePAG at ERN ITHACA, president of SATB2 EUROPE
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I would like to share our innovative, **parent-led initiative in creating the first global Clinical Practice Guideline (CPG) for a rare neurodevelopmental disorder called SATB2-Associated Syndrome (SAS).**

In this presentation, I will cover:

- How we started with no clinical network in Europe or globally, and within just two years, gathered a network of 60 clinicians and experts familiar with SAS. The aim is to empower patient advocates by showing that it is possible to achieve this kind of network, even starting from scratch, and how they can do it if they find themselves in a similar situation.
- How we began advocating within the European Reference Network (ERN) ITHACA to have our syndrome considered as a candidate for future CPGs from a very early stage.
- How to organise a project without an obvious European clinician to chair or lead it, demonstrating a pioneering approach where a patient advocate took on the role of a chair.
- How we involved all existing SAS patient organisations worldwide as part of the guideline consortium.
- Our efforts to genuinely incorporate the voices of patients and parents in selecting the clinical topics addressed in the guidelines:
    - Initial input for relevant clinical questions was collected from 20 parents across 13 countries, totalling in 376 identified questions.
    - From that the final list of clinical questions to be rated for relevance was reviewed and approved by those parents (besides co-chairing clinicians).
    - The survey was translated into 8 languages and distributed globally through all SAS organisations.
    - We are also exploring ways to involve patients with mild intellectual disabilities in the guideline project.

Additionally, I will discuss the potential impactful side projects arising from this initiative, including:

- Creating a list of basic and clinical research projects for the future.
- Establishing national contact points for clinical collaboration on SAS-related topics (including biobank network, natural history study and similar).
- Facilitating the formation of a European Center of Expertise for SAS within ERN.
- Enabling an informal platform for clinicians treating SAS patients to exchange clinical dilemmas and experiences.

I believe our journey in improving clinical care for SAS patients, both in Europe and beyond, is unique. I hope this journey can inspire others to take those first bold steps, even when no leading clinician is there to pave the way.

GLOBAL ANGELMAN SYNDROME REGISTRY

Authors: Ms Ellen Koekoeckx[1], Dr Honey Heussler[2], Dr Megan Tones[3] ,Ms Niki Armstrong[1], Ms Meagan Cross[4], Ms Chloe Simons[4], Prof Matthew Bellgard[3,5]
1. Foundation for Angelman Syndrome Therapeutics United States of America 2. Children's Health Queensland, 3. eResearch, Queensland University of Technology, 4. Foundation for Angelman Syndrome Therapeutics Australia, 5 University of East London

OBJECTIVES

The Global Angelman Syndrome Registry aims to:

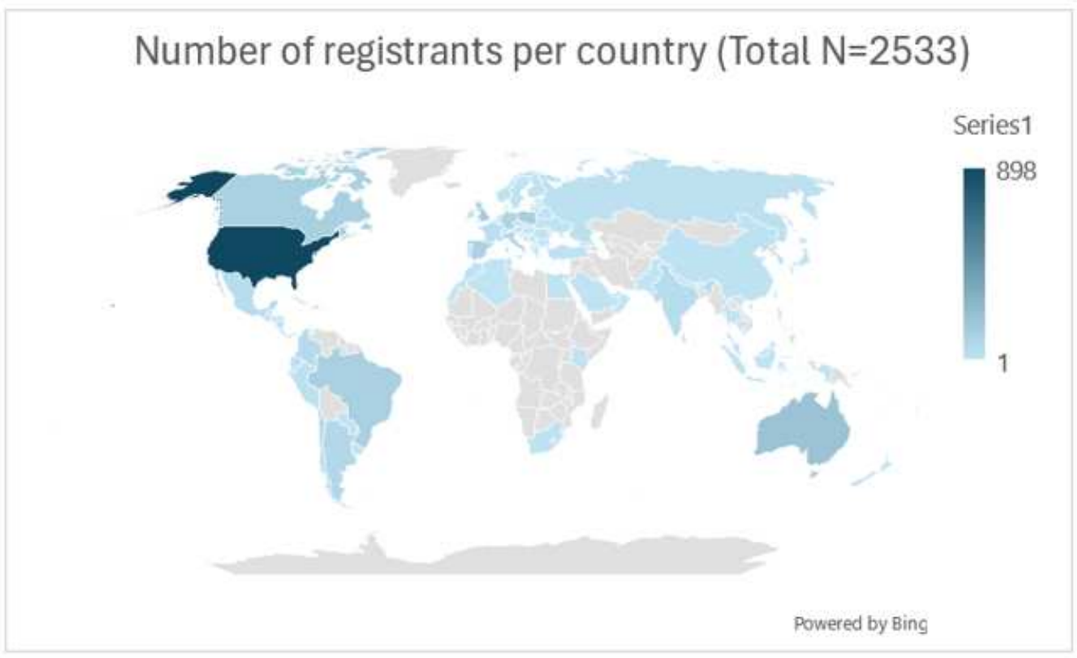
- **Collect comprehensive, global data** on individuals with Angelman syndrome (AS) to better understand its natural history and variability.
- **Facilitate research** into the progression, symptoms, and genetic factors associated with AS.
- **Support the development of new treatments** and therapies by providing a resource for clinical trial design and patient recruitment.

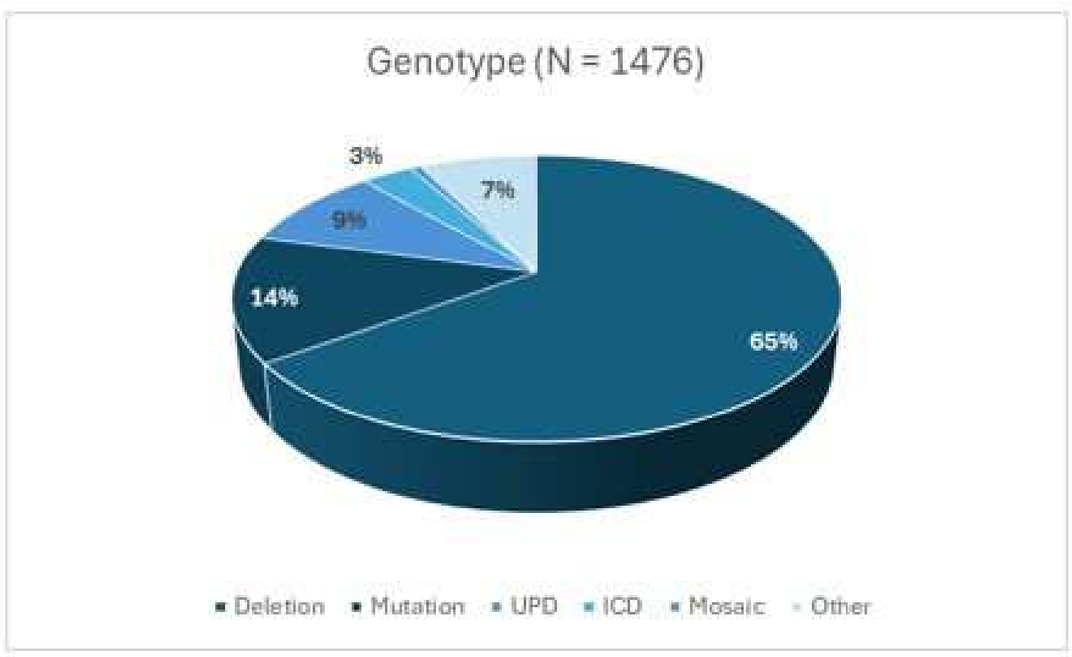
METHODOLOGY

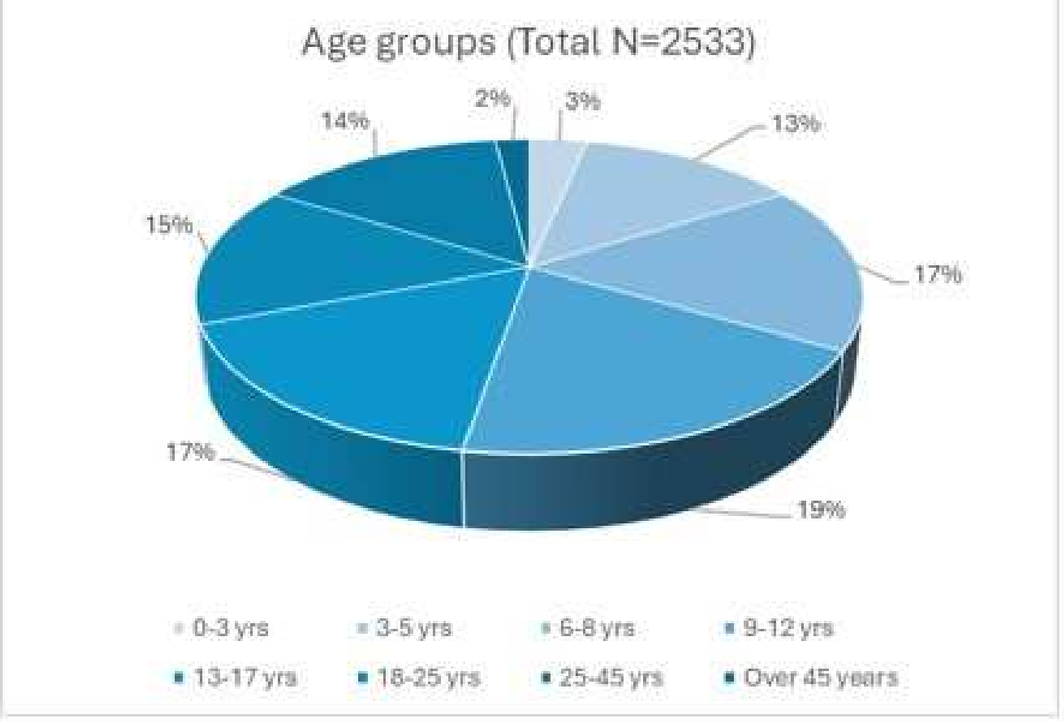
- **Initiated** by Dr Honey Heussler and FAST Australia members (FAST – Foundation for Angelman Syndrome Therapeutics).
- **Data Ownership and Access**: FAST Australia owns the data, but access is controlled through a governance Data Access Committee, with data curators administering access to ensure data privacy and compliance.
- **Developed** with the Rare Disease Registry Framework (RDRF) platform at Murdoch University and migrated to the Trial Ready Registry Framework (TRRF) platform hosted by the division of eResearch at Queensland University of Technology in 2020.
- **Available in 7 languages**: English, Spanish, Hindi, Italian, traditional Chinese, Brazilian Portuguese and Polish.
- **Registration modules** are completed by caregivers including demography & parent/carer consent, newborn and infancy history, history of diagnosis, medical history, epilepsy/seizures, medications/interventions, test results/clinical trials/clinics, illness or medical problems, communication, behavior and development, sleep, data collection for other studies.

OUTCOMES

- **2533 participants** worldwide from 95 countries
- GASR has published **8 research publications**, advancing the knowledge in the field of Angelman syndrome.







POTENTIAL IMPACT

- **Accelerated Research:** By providing robust, patient-centered data, the registry is expected to speed up the development of targeted therapies and treatments for AS.
- **Informed Clinical Practices**: Insights from the data may lead to improved clinical care guidelines and personalized treatment approaches for individuals with Angelman syndrome.
- **Global Collaboration:** The registry fosters collaboration between researchers, clinicians, and the AS community worldwide, driving innovation and shared progress toward a cure.

FUTURE PLANS

- **Streamline the dataset** to keep it minimal but impactful, reducing the participant burden.
- Addition of **new outcome measures** such as ORCA and ABC.
- **Expand translations** to make the platform accessible to more families worldwide.

CONCLUSION

The Global Angelman Syndrome Registry serves as a vital resource for researchers, clinicians, and the community. Its ongoing data collection and analysis aim to transform our understanding of Angelman syndrome, support the development of new therapies, and ultimately improve the lives of individuals with AS and their families.

For general information about FAST: ellen.koekoeckx@cureangelman.org
For information about the global registry: curator@angelmanregistry.info

Title: Building a Research Portal for Rare Disease Collaboration: CHAMP1 Approach

Authors: Daniele Palumbo, CHAMP1 Foundation - Europe / CHAMP1 Alliance
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Reference: https://research.champ1foundation.org

Objective:
The CHAMP1 Foundation developed a research portal (see reference above) to address the disorganization of information related to the CHAMP1 gene mutation. The portal was designed to centralize research papers, connect researchers, and make information easily accessible for patients, caregivers, clinicians, and patient organizations.
By doing so, the portal helps advance collaboration in the rare disease community, especially in disorders related to congenital malformations and intellectual disabilities.

Methodology:
The portal was built using no-code platforms (Softr for the frontend and Airtable as the database).
This approach allows for easy updates and contributions from the community.
By using external databases SaaS and a low-code environment SaaS, the platform dramatically lowers the barrier to who can manage and contribute data.
It can also be integrated with registries through APIs to further expand its use in the research and patient care ecosystem.

Outcomes:
The portal has enabled patient organizations to track research progress, quickly identify relevant studies (e.g., for CHAMP1 and related genes like POGZ/White-Sutton), and make this information accessible to patients and clinicians.
Clinicians, for example, can quickly access all necessary research related to rare conditions like cyclic vomiting syndrome, which is part of CHAMP1 mutations, significantly improving patient care.

Potential Impact:
The portal fosters collaboration between researchers and helps patient organizations manage research projects, including sensitive materials under NDAs.
The project has received positive feedback from all stakeholders and has proven to be a cost-effective solution (currently at zero cost).
The model is scalable and can be replicated by other organizations, potentially using open-source platforms for broader implementation.

Conclusion:
This research portal represents an innovative approach to addressing the challenges of information management in the rare disease community.
By providing centralized, easy access to research data, it empowers patients, caregivers, and clinicians alike, fostering collaboration and improving the lives of those affected by rare diseases.

Author: Tanja Zdolšek Draksler, PhD, IDefine Europe Foundation

Title: Solving Rare Diseases: How Global Collaboration and Data are Advancing Kleefstra Syndrome Research

This presentation gives an in-depth exploration on Kleefstra syndrome (KS) global community, highlighting its global nature and the pivotal role of collaboration in advancing research and data collection. The Kleefstra Syndrome Global Community—a diverse network spanning Europe, USA, Canada, Australia, South America and even Asia. Several data collection initiatives that are crucial: 1. Kleefstra Syndrome Worldwide Map, 2. GENIDA registry (caregiver-reported data), 3. RARE-X (caregiver-reported data), 4. Citizen Health (clinical data from e-health records to deliver natural history study). Alongside these long-term data collection activities, there are also short-term activities, collecting specific data. The data collection initiatives contribute to a more accurate understanding of KS clinical symptoms and prevalence. There is an increased need for advocacy activities for heighten awareness and increase efforts in promoting the importance of data collection to both families and clinicians.

Objectives: The primary objective is to highlight the advancements in research on Kleefstra syndrome, a rare genetic disorder, through global collaboration and comprehensive data collection initiatives. Specifically, it aims to:
1. Emphasize the importance of international cooperation and networks.
2. Showcase the role of patient organizations in data collection.

Methodology:
1. **Global Collaboration**: extensive collaboration between organizations and research teams across Europe, USA, Australia, Asia, and other regions.
2. **Community Engagement**: patient organizations and parents actively contribute to data collection and research.
3. **Data Collection Initiatives**: Various platforms are used to gather caregiver-reported data and clinical information.

Outcomes:
1. **Enhanced Understanding of Kleefstra syndrome**: The collaborative efforts and data collection have significantly contributed to a deeper understanding of KS clinical manifestations, prevalence, and genetic underpinnings. Till today 793 identified individuals with KS globally.
2. **Improved Research and Clinical Networks**: The establishment of a global community and international collaborations has facilitated knowledge sharing among researchers, clinicians, and families, improving diagnostic and care standards across regions (e.g., Kleefstra clinic in Boston).
3. **Increased Advocacy and Awareness**: Through patient organizations and global conferences, awareness about the importance of data collection and sharing has been heightened among both families and clinicians. Advocacy efforts have expanded, resulting in more proactive engagement and research initiatives.

Impacts:
1. **Advancing Rare Disease Research**: The global collaboration and data-driven approach in Kleefstra syndrome is creating a best practice and model for the study of other rare diseases.
2. **Global Health Outcomes**: The research is contributing to better health outcomes by facilitating early diagnosis, personalized care, and more effective interventions for individuals affected by KS.
3. **Future-Oriented Approach**: The integration of cutting-edge technologies like data science in research signals a forward-looking approach, paving the way for novel discoveries in the realm of rare diseases.