In this document you find the different breakout session you can participate in on Thursday and Friday. The different topics offer the chance to make deep dive from multidisciplinary view, connect the gathered knowledge in the morning sessions and define the way forward in order to further optimise our research, education and organization of care & cure.

Let's create learning waves* from a multidisciplinary view *together.

EuroNDD workshop 2026 website

Co-funded by the Health Programme of the European Union

European Reference Network for rare or low prevalence complex diseases

Breakout sessions on Thursday April 9 (14:30 – 17:30; 2 parallel rounds of 75 minutes)

Topics of the first round (14:30 -15:45)

1. Solving the undiagnosed: How recent insights translate into optimized workflows. **Katrin Ounap, Helene Cederroth, Zeynep Tümer, Tjitske Kleefstra**
2. Pain in people with intellectual disabilities: methodical and interdisciplinary collaboration (an educational session). **Leendert Sneep & Nanda de Knegt**
3. From Guideline to Practice - Implementing ERN ITHACA Guidelines (an educational session) **Mirthe Klein Haneveld, Agnies van Eeghen & co-workers cross Europe**
4. From Principles to Practice: Interprofessional Collaboration in Rare Genetic Intellectual Disability Syndromes – A European Round Table. **Mana Nasori, Kim Oostrom, Lotte Haverman, Sylvia Huisman**

Exchange break (15:45 – 16:15)

Topics of the second round (16:15 – 17:30)

1. Towards Precision Medicine for NDD. **Geatan Lesca, Beatrice Desnous, Adreas Roos, Kasia Kotulska**
2. From training skills to enabling interactions: A new paradigm in AAC for individuals with neurodevelopmental disorders and complex communication needs (an educational session). **Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek**
3. Building Patient Registries under the GDPR – The Good, the Bad and the Ugly (an educational session followed by a roundtable discussion). **Christian Gebhard, David Townend**
4. Neuropsychological insights: Cognition as a bridge between brain and behaviour in rare genetic syndromes? **Anja Bos-Roubos, Jennifer Kramer, Carmen Oldenboom, Ellen Wingbermühle, Jos Egger**
5. From Networks to Norms: Developing First European Recommendations and Care Principles for PIMD / Polyhandicap. **Sylvia Huisman, Annette van der Putten, Marie-Christine Rousseau, Ilse H. Zaal-Schuller**

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Breakout sessions on Friday April 10 (14:30 – 15:15; parallel rounds of 75 minutes)

Topics of the third round (14:15 – 15:30)

1. Measuring what matters in genetic neurodevelopmental disorders: trials and care. **Agnies van Eeghen & Ellen Elsman**
2. Transition of Care for Adults with Intellectual Disabilities in Genetic Syndromes. **Nikolinka Yordanova, Laura de Graaff, Kasia Swieczkowska**
3. Communication support for individuals with neurodevelopmental disorders and complex communication needs: sharing best practices (a round table discussion). **Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek**
4. Pain in people with intellectual disabilities: implementation and network formation (a round table discussion). **Leendert Sneep & Nanda de Knegt**

Breakout sessions schedule

Day 1 round 1				
Break-out Sessions _ 75 minutes				
1	2	3	4	
Solving the undiagnosed: "How recent insights translate into optimized workflows" Katrin Ounap, Helene Cederroth, Zeynep Tümer, Tjitske Kleefstra	Pain in people with intellectual disabilities: methodical and interdisciplinary collaboration (an educational session) Leendert Sneep & Nanda de Knegt	From Guideline to Practice - Implementing ERN ITHACA Guidelines (an educational session) Mirthe Klein Haneveld , Agnies van Eeghen & co-workers cross Europe	From Principles to Practice: Interprofessional Collaboration in Rare Genetic Intellectual Disability Syndromes – A European Round Table Mana Nasori, Kim Oostrom, Lotte Haverman, Sylvia Huisman	
Day 1 round 2				
Break-out Sessions _ 75 minutes				
1	2	3	4	5
Towards Precision Medicine for NDD Geatan Lesca, Beatrice Desnous, Adreas Roos, Kasia Kotulska (PO)	From training skills to enabling interactions: A new paradigm in AAC for individuals with neurodevelopmental disorders and complex communication needs (an educational session) Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek	Building Patient Registries under the GDPR – The Good, the Bad and the Ugly (an educational session followed by a roundtable discussion) Christian Gebhard, David Townend	Neuropsychological insights: Cognition as a bridge between brain and behavior in rare genetic syndromes? Anja Bos-Roubos, Jennifer Kramer, Carmen Oldenboom, Ellen Wingbermühle, Jos Egger	From Networks to Norms: Developing First European Recommendations and Care Principles for PIMD / Polyhandicap Sylvia Huisman , Annette van der Putten, Marie-Christine Rousseau, Ilse H. Zaal-Schuller

Day 2				
Break-out Sessions _ 75 minutes				
1	2	3	4	
Measuring what matters in genetic neurodevelopmental disorders: trials and care Agnies van Eeghen & Ellen Elsman	Transition of Care for Adults with Intellectual Disabilities in Genetic Syndromes Nikolinka Yordanova, Laura de Graaff, Kasia Swieczkowska	Communication support for individuals with neurodevelopmental disorders and complex communication needs: sharing best practices (a round table discussion) Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek	Pain in people with intellectual disabilities: implementation and network formation (a round table discussion) Leendert Sneep & Nanda de Knegt	

Co-funded by the Health Programme of the European Union

Solving the undiagnosed: How recent insights translate into optimized workflows.

Katrin Ounap, Helene Cederroth, Zeynep Tümer, Tjitske Kleefstra

Introduction: Undiagnosed rare neurodevelopmental disorders are being tackled with a mix of advanced genomic testing, "beyond the exome" technologies and deep phenotyping. The field is moving fast, in this roundtable we aim to move from the latest insights in deep geno- and pheno-typing to optimization of diagnostic workflows in daily practice that matches the different analytical abilities in the different healthcare setting across Europe.

Our Aim: In this session we invite clinicians, molecular geneticists, bioinformaticians, researchers and patient partners to reflect on and discuss what it takes to further optimize the clinical-diagnostic workflows of neurodevelopmental disorders in your national clinical settings. Topics we suggest to reflect on are the impact of Solvathons or Undiagnosed Hackathons in a national setting. Questions to address are: What is the experience on yield and added value to routine diagnostic trajectories, what would you need to organize such kind of events in your practice?

The outcomes of this workshop may help to shape the discussion on the needs and requirements of such large-scale events.

Session Structure

- **15 minutes – Plenary overview on the key lessons learned in particular by ERDERA/JARDIN initiatives**
- **45 minutes – Small-group discussions on topics that are on the top ten list of our participants.** For each selected topic participants will share experiences, discuss common challenges, and explore potential solutions for diagnostic workflow optimization across diverse healthcare settings from a family integrated perspective (in case of 25 participants > 5 small groups ideally representing different disciplines such as clinical geneticist, molecular geneticists (lab-experts), neurologist, bioinformaticians, and patient representative partners. For each table we will select two topics they will focus on.
- **15 minutes – Plenary wrap-up and feedback**
  Summary of key discussion points and reflections on the key chances and challenges in workflow optimization to come faster to final diagnose. Collective synthesis to define shared priorities and future directions for research, clinical practice, and policy.

Co-funded by the Health Programme of the European Union

Pain in people with intellectual disabilities: methodical and interdisciplinary collaboration (an educational session).

Leendert Sneep & Nanda de Knegt

Subject: In this workshop, we will explore (with real case descriptions) how healthcare professionals of people with intellectual disabilities could work together in a methodical and interdisciplinary way to identify, diagnose, and treat pain. We will especially discuss practical methods regarding two themes: personalized pain care and the biopsychosocial approach of chronic pain.

Target group: behavioural experts, physicians, nurses, paramedics (physiotherapists, occupational therapists, psychomotor therapists), Direct Support Professionals, and family of people with intellectual disabilities

Learning objectives:

1. The participant knows how to organize the methodical process and interdisciplinary collaboration around pain in people with intellectual disabilities.

2. The participant has practiced with tools for personalized pain care

3. The participant has practiced with clinical reasoning regarding the biopsychosocial approach of chronic pain in people with intellectual disabilities

Session structure

- 10 minutes - Introduction and learning objectives
- 15 minutes - Methodical work and interdisciplinary collaboration
- 20 minutes - Personalized pain care
- 20 minutes - Biopsychosocial approach of chronic pain
- 10 minutes – Wrap-up key messages and questions

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From Guideline to Practice - Implementing ERN ITHACA Guidelines (an educational session)

Mirthe Klein Haneveld, Agnies van Eeghen & co-workers cross Europe

Background: Clinical Practice Guidelines (CPGs) support evidence-based healthcare. However, for rare conditions, both guideline development and implementation are hindered by limited scientific evidence, small patient populations, limited resources, and variation across healthcare systems. ERN-ITHACA, the European Reference Network for rare congenital malformations and rare intellectual disability syndromes, addresses these challenges by developing clinical practice guidelines and advancing guideline methodology. Now many guidelines have been published or are underway, the question arises: how do we bring the guidelines to HCPs and families?

Aim of the Workshop: This workshop aims to involve healthcare professionals, researchers, and patient partners in the discussion around implementation of guidelines in care. Facilitators and barriers across care contexts, cultures, and health policies will be discussed. Insights from the workshop will directly inform ongoing and future implementation strategies within ERN ITHACA.

Session Structure

- **20 minutes – Introductory plenary presentation**
  Overview of the ERN ITHACA guideline development procedure, illustrated with examples of recently developed clinical practice guidelines.
- **45 minutes – Interactive and small-group discussions on implementation**
  Participants will share experiences, discuss common challenges, and explore potential solutions for implementing guidelines across diverse healthcare settings.
- **10 minutes – Plenary wrap-up and feedback**
  Summary of key discussion points and reflections on implications for ERN ITHACA implementation strategies.

Learning Objectives

After participating in this session, participants will be able to:

1. Describe the key steps and methodological principles underlying the development of clinical practice guidelines for rare genetic conditions.
2. Identify barriers and facilitators to implementing clinical practice guidelines for rare conditions in European daily clinical practice.
3. Reflect on and apply implementation strategies through discussion of experiences across different healthcare systems.

Co-funded by the Health Programme of the European Union

From Principles to Practice: Interprofessional Collaboration in Rare Genetic Intellectual Disability Syndromes – A European Round Table.

Mana Nasori, Kim Oostrom, Lotte Haverman, Sylvia Huisman

Subject: In this session we will present evidence-informed Principles of Care that we recently formulated in the Netherlands, to enhance interprofessional collaboration around challenging behaviour in people with rare genetic intellectual disability syndromes (RGDIS). Principles are derived from findings obtained through qualitative studies and consensus meetings. We will share key research findings on patient journey, parents' and healthcare professionals' experiences and intentions and competencies of the professionals involved, and discuss with you which principle work internationally, and explore logical next steps to operationalize them into practical tools or frameworks to ensure they can create added value in practice.

Target group:

- Healthcare and other relevant professionals across disciplines (medical, behavioural, educational, parental)
- Clinicians, researchers, policymakers, parent and other relevant representatives in rare genetic intellectual disability syndromes
- International audience interested in interprofessional collaboration, improvement of RGIDS healthcare delivery, person-centred care, and lifelong care networks

RoundTable discussion topics:

- **Research Snapshot**: Share the key findings from three studies on RGIDS care
- **Principles of Care:** Presenting the Dutch Principles of Care, aimed to enhance interprofessional collaboration around challenging behaviour
- **Discussion & international reflection**:
    - Which principles help foster interprofessional collaboration in your context?
    - Are some principles missing or need adaptation for different healthcare systems?
    - What cultural, context and differences in the organization and structure of healthcare systems affect interprofessional collaboration?
    - What are first ideas to disseminate and operationalize principles in daily practice?
    - Outcome: Further refinement of the principles and the development of actionable implementation strategies.

Session structure:

- 15 minutes - Research Snapshot
- 45 minutes - Table discussions
- 10 minutes - Plenary sharing of key insights
- 5 minutes - Reflection: take-home messages and future directions

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Towards Precision Medicine for NDD.

Geatan Lesca, Beatrice Desnous, Adreas Roos, Kasia Kotulska

Background: The development of precision medicine for neurodevelopmental disorders brings renewed hope to families seeking meaningful treatment options. By targeting the biological causes of disease, these approaches promise more personalized and potentially effective care. At the same time, scientists have a responsibility to communicate new treatment perspectives clearly, realistically, and without overstating expectations. Fair and transparent information is essential to help families make informed decisions in a rapidly evolving field. For rare diseases, progress depends on well-designed, collaborative clinical trials that ensure both scientific rigor and patient safety. Only through careful research and honest dialogue can hope be transformed into reliable, lasting therapies.

Aim of the round table: This round table invites clinicians, researchers and patients' representatives to engage in a collaborative dialogue, aiming to illustrate the role and effect of scientists, patients' association and clinicians in the implementation of precision medicine in NDD. In this roundtable discussion we will especially focus on how to communicate to the patients and families about the use of drugs and the need for drug trials. And the management of the expectations: what are the aims of the trials?

Session structure

- 4 x 5min - 'Spark' presentations by the moderators, including a selection of their recent experience on precision medicine for neurodevelopmental disorders (NDDs), to kick off and inspire the discussion with cross-syndrome themes and multidisciplinary experts perspectives.
  Candidate syndromes:
  - GRIN genes and L-serine supplementation, Gaetan Lesca (FR).
  - Bench to bedside in rare diseases, Andreas Roos (D)
  - New therapeutic perspectives for Rett syndrome, Béatrice Desnous (FR) : early access programme for trofinetide in France and in Europe.
  - mTOR inhibitors in TSC (Everolimus and sirolimus). Katarzyna Kotulska (PL)
- 15 min Guided discussion part I: Re-purposing treatment for NDD and the role of patients' associations in clinical trial design.
- 15 min Guided discussion part II What are the requirements to launch a clinical drug trials in the rare genetic NDD field?
- 15 min Guided discussion part III: What is your experience in clinical trial design, validation and dissemination to clinical practice?
- 10 min Synthesis and closing: Flash summary to secure the outcomes of the discussion and provide clear conclusions and key take-aways.

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From training skills to enabling interactions: A new paradigm in AAC for individuals with neurodevelopmental disorders and complex communication needs (an educational session).

Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek

Note: this session is linked to the posters with abstract ID 51, 52 and 55, and the oral presentation with abstract ID 66.

Introduction: In this educational session we will guide you through a transformative shift in Augmentative and Alternative Communication (AAC) practice, moving away from isolated skill training towards a model that prioritises meaningful social interaction and self-determination. We will illustrate the three pillars upon which the new framework is built:

1. Revealing hidden competence through dynamic assessment;
2. Personalising communication support aimed at self-determination and social interaction, according to the characteristics, strengths and needs of the individual and their network;
3. Fostering collaborative co-production between professionals, families and individuals with neurodevelopmental disorders (NDD) and complex communication needs (CCN).

The overall aim of this educational session is to illustrate how person-centred practice ensures that the chosen communication modes and communication goals honour the person's unique profile and innate social drives rather than following a generic clinical protocol or the typical communication development of speaking peers. Participants will learn the importance of facilitating self-determination and genuine connection. By moving beyond simple requests to embrace humour, autonomy, and relationship-building, the approach seeks to empower individuals with NND and CCN.

Who is this educational session for? This session is especially relevant for clinicians, researchers, educators, and lived-experience experts who have limited knowledge of AAC implementation. Through sharing examples from our own clinical experience, we aim to provide participants with insights into co-creation of communication tools that give a person more autonomy, promote self-confidence and provide a sense of belonging.

After this educational session participants will have gained insight into:

1. Types of AAC, including the different but complementary values of unaided AAC (gestures, body language) and aided AAC, which ranges from light-tech picture boards and communication books to high-tech devices and apps.
2. How AAC can be taught through modelling in everyday routines, and the importance of including both core words (frequently used words like "go") and fringe vocabulary (specific interests like favourite games).
3. Myth-Busting around AAC: Evidence-based reassurance that AAC does not stop verbal speech development and those children do not require "prerequisite" skills to begin using it.
4. The importance of interdisciplinary collaboration, including the individual and their network, to ensure that AAC use meets participation goals and is tailored to the wants and (access) needs and lived experiences of the AAC user and their social network.
5. Specialised Assessment Tools that may be used to support information-gathering and goal-setting when building an individual communication profile.

Educational session structure:

- 10 minutes – introduction to the 5 learning objectives
- 50 minutes – 10 minutes per learning goal
- 15 minutes – summary of key take-aways and planning for (optional) online follow-up

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Building Patient Registries under the GDPR – The Good, the Bad and the Ugly (an educational session followed by a roundtable discussion).

Christian Gebhard, David Townend

Note: This is an extended session, runs till 6 pm, and is linked to poster ID 43 + 44

This session will provide a comprehensive guide on what to consider when planning a patient registry or expanding the scope of an existing registry. It aims to help you navigate the complex landscape from initial concept to a sustainable, impactful resource for patients and researchers. The ultimate goal of our breakout is the initiation of an ongoing exchange of interested researchers, clinicians, legal and privacy experts and IT professionals.

Patient registries are powerful tools advancing rare diseases, especially in an international context. They are essential for understanding the natural history of a disease, improving patient care, and accelerating research and drug development. However, establishing a high-quality registry that generates reliable data is a significant challenge, requiring a multidisciplinary approach that balances clinical, ethical, legal, and technical considerations. Furthermore, rare disease research dictates international collaboration to collect sufficient real-world data.

A common perception is that data protection regulations, particularly the GDPR, are tedious barriers that hinder modern patient care. This workshop will challenge that myth, demonstrating that addressing these requirements from the outset is not an obstacle but a critical success factor for any registry project. At the same time, we will discuss common pitfalls, how to tackle them and problems within the current regulations where policy makers push towards data sharing and interoperability across borders without providing the legal basis.

Our workshop is divided into two parts. The first part of our session will provide a basic understanding of key aspects: primary processing of personal data, where data is gathered directly from patients or with their specific consent and secondary processing: the reuse of already-gathered personal data for new research purposes.

The second, interactive part of the workshop will delve into the complexities of translating applicable law into a usable, thriving registry that benefits patient care and research. We invite you to share the obstacles you are facing, discuss with us your positive and negative experiences “from the field” and help us sketch a path forward to advance international collaboration, research and most of all: care for patients with rare diseases.

Key topics will include:

- European GDPR vs national laws
- Patient Involvement and Empowerment
- Informed Consent, Broad Consent, “Data donation”

By the end of this 105-minute workshop, participants will have a foundational understanding of the distinction between primary and secondary data use, the necessary legal frameworks for each, and how a foresighted design of registries and consent processes can pave the way for successful and impactful research.

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Neuropsychological insights: Cognition as a bridge between brain and behaviour in rare genetic syndromes?

Anja Bos-Roubos, Jennifer Kramer, Carmen Oldenboom, Ellen Wingbermühle, Jos Egger

Introduction: Building on recent studies on cognitive and behavioral phenotyping within neurodevelopmental disorders (NDDs), this round table aims to elucidate the unique contributions of clinical neuropsychology to both scientific understanding and the provision of care for individuals with rare genetic syndromes. Clinical neuropsychology may serve as an interdisciplinary bridge, illuminating the complex interplay between genetic, neurological, somatic, and behavioral domains. These insights not only enhance scientific insights, but also guide practical approaches that benefit healthcare professionals, patients, and families alike. By investigating both syndrome-specific cognitive profiles and transdiagnostic features shared across syndromes, neuropsychological research offers a nuanced foundation for personalized care—*targeted where necessary, and generalized where appropriate*.

Methods, results & discussion: This round table invites both clinicians and researchers to engage in a collaborative dialogue, aiming to generate syndrome-transcending insights that can ultimately enhance care for individuals with rare neurodevelopmental disorders.

Content, format and schedule

15 min	Short introduction, based on 'spark' presentations by the authors/presenters, to kick off and inspire the discussion with cross-syndrome themes and expert perspectives. Candidate syndromes: 1. Koolen-De Vries syndrome (KdVS) and CAMK2-related syndromes (CAMK2); by C. Oldenboom, MSc., PhD-candidate (poster ID 34) 2. Noonan syndrome (NS), by J. Kramer, MSc., PhD-candidate (Poster ID 33) 3. HNRNPK gene variations; A.G. Bos-Roubos, MSc, PhD-candidate
45 min	Guided discussion: • Common features and challenges: identifying patterns and overlapping problems beyond single syndrome perspectives • The value of clinical neuropsychology: defining *meaningful*, patient-centric endpoints for clinical and research practice • Gaps, opportunities and collaborative action: defining a 'to-do' list and build consensus for future action
10 min	Synthesis and closing: jointly consolidating the outcomes of the discussion

Key words: NDDs, contextual neuropsychology, transdiagnostic features, personalized care

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From Networks to Norms: Developing First European Recommendations and Care Principles for PIMD / Polyhandicap.

Sylvia Huisman, Annette van der Putten, Marie-Christine Rousseau, Ilse H. Zaal-Schuller

Introduction: Individuals with profound intellectual and multiple disabilities (PIMD/polyhandicap) are among the most complex and underrepresented populations in NDD care. Despite their lifelong and complete dependence on care**, evidence-based clinical practice guidelines (CPGs) are largely lacking.**

Within ERN-ITHACA, an international interdisciplinary consortium from 14 European countries, in close collaboration with family members, developed the first European CPGs for individuals with PIMD/polyhandicap, focusing on motor functioning, sleep, and pain. Beyond domain-specific recommendations, cross-guideline analyses resulted in overarching care principles for individualized, interdisciplinary, and holistic care.

This session invites the EURO-NDD community to move **from building networks to shaping norms by** reflecting on and discussing what it takes to develop and implement CPGs for a population in which traditional evidence hierarchies fall short.

Interactive roundtable:

- Moderated interdisciplinary tables engaging researchers, clinicians, and family representatives
- Structured outcome-focused reflection prompts addressing aspects of evaluation, valorisation, and implementation of CPGs for individuals with PIMD/polyhandicap
- Collective synthesis to define shared priorities and future directions for research, clinical practice, and policy

Session structure:

15 minutes – Research Snapshot: challenges and outcomes in CPG development (plenary)
45 minutes – Table discussions: evaluation, valorisation, and implementation (small groups)
10 minutes – Key insights & cross-table synthesis of clinical and family perspectives (plenary)
05 minutes – Reflection and wrap-up: key takeaways and next steps

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Measuring what matters in genetic neurodevelopmental disorders: trials and care.
Agnies van Eeghen & Ellen Elsman

Background: In the field of genetic neurodevelopmental disorders (NDD), selecting appropriate outcomes and outcome measures for trials and care is complex. A wide variety of outcomes are used across trials and healthcare centers, ranging from clinical endpoints to patient/proxy-reported outcome measures, as well as observational tools. This heterogeneity hampers comparability across trials, complicates interpretation of trial results, and limits translation to daily clinical care.

At the same time, stakeholders, including trialists, healthcare professionals, policy makers, industry partners, and families, have different perspectives on what outcomes matter most. Ongoing international initiatives aim to harmonize outcome selection and improve methodological rigor, yet practical guidance for navigating this landscape in genetic NDD remains limited.

This interactive educational session will help participants explore the "outcome maze" in genetic NDD and jointly identify priorities and future directions for more meaningful, feasible, and harmonized outcome measurement in both trials and care.

Aim of the Workshop: This workshop aims to equip trialists, healthcare professionals, researchers, and patient partners with conceptual clarity and practical tools to critically reflect on outcomes and outcome measures in genetic NDD. Participants will exchange experiences, identify current challenges, and collaboratively define next steps toward improved outcome selection and implementation in trials and care.

Insights from this session will inform ongoing and future initiatives focused on outcome harmonization and validation in the field of rare genetic neurodevelopmental conditions.

Session Structure

- **20 minutes – Setting the scene (interactive plenary)**
  - Short introduction to the topic and objectives of the session
  - Live interactive poll to map which outcomes and outcome measures participants currently use in trials and care
  - Illustration of key bottlenecks for the use of outcome measures in trails and care
  - Collection of participants' wishes, ideas, and initiatives with respect to outcomes and outcome measures
- **25 minutes – Educational component**
  - What do we mean by outcomes and outcome measures?
  - What types of outcome measures exist?
  - What are the core psychometric properties?
  - What is the current landscape in the development, validation, and implementation of outcome measures in rare genetic ID
- **20 minutes – Interactive brainstorm in small groups**

Participants will discuss:

- Which outcomes are most important for our target populations?
- Which outcome measures are currently available?
- What are the main barriers to using these measures in trials and care?

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- **10 minutes – Plenary wrap-up and future steps**
  - Harvesting key insights from group discussions
  - Mapping stakeholders and potential collaborators
  - Identifying next steps toward harmonization and improved implementation of outcomes and outcome measures

Learning Objectives

After participating in this session, participants will be able to:

1. Differentiate between types of outcomes and outcome measures and explain their relevance in genetic NDD.
2. Recognize key psychometric properties required for good outcome measurement.
3. Identify current challenges in selecting and implementing outcome measures in trials and care for genetic NDD.
4. Contribute to defining priorities and collaborative strategies for improving outcome (measure) harmonization and implementation in the genetic NDD field.

Transition of Care for Adults with Intellectual Disabilities in Genetic Syndromes.
Nikolinka Yordanova, Laura de Graaff, Kasia Swieczkowska

Introduction: Transition of care for individuals with intellectual disabilities (ID) as part of a genetic syndrome is widely recognized as one of the most vulnerable phases in the care trajectory. Unlike many other patient groups, transition in this population is not limited to a transfer from pediatric to adult medical services. It is a complex, multidimensional process that simultaneously affects healthcare, education or day-care arrangements, legal representation, social support, and family roles.

Many adults with syndromic ID experience a sudden fragmentation of care at the moment they reach adulthood. Pediatric care is often highly coordinated, family-centered, and multidisciplinary, whereas adult care tends to be more fragmented, disease-oriented, and expects a level of autonomy that may not be realistic for this population. This gap can result in loss of follow-up, delayed recognition of health problems, behavioural crises, and avoidable health damage.

Overall aim: In this breakout session, we will explore transition as a shared responsibility that requires early planning, clear coordination, and sustained multidisciplinary collaboration. Key domains include medical follow-up (such as hormonal and metabolic issues that evolve with age), behavioural and mental health support, communication abilities, and differences in clinical presentation between paediatric and adult patients. Legal aspects—such as guardianship, decision-making capacity, and representation—are equally crucial and are often addressed too late. The role of parents changes substantially during transition, shifting from primary decision-makers to partners or legal representatives, while clinicians must adapt their communication and care models accordingly.

Session setup: We will discuss real-life cases in which transition has been problematic, highlighting common pitfalls such as lack of a coordinating professional, insufficient handover of syndrome-specific knowledge, and poor alignment between medical and social care systems. Particular attention will be paid to the differences between individuals with severe ID and those with mild ID, as their needs, autonomy, and risks during transition differ markedly and require tailored approaches.

The session will be highly interactive. Participants will be invited to reflect on fundamental questions:

- What are the core elements of good transition care?
- Who should coordinate the process, and when should it start?
- What are the respective roles of parents, caregivers, and healthcare professionals, and how are these roles supported by healthcare systems across different countries?
- And how can we prevent predictable problems before they arise?

By combining case-based discussion with audience input, this session aims to define practical, realistic principles for successful transition of care for adults with genetic syndromes and intellectual disabilities.

Co-funded by the Health Programme of the European Union

Communication support for individuals with neurodevelopmental disorders and complex communication needs: sharing best practices (a round table discussion).

Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek

Note: this session is linked to the posters with abstract ID 51, 52 and 55, and the oral presentation with abstract ID 66.

Introduction: In this roundtable session we encourage participants to share their experiences of providing communication support for individuals with neurodevelopmental disorders (NDD) and complex communication needs (CCN). We will ask questions such as:

1. What factors do you consider when meeting a new (potential) AAC user?
2. How do these factors influence your assessment of the individual and their network, the communication modes that you may recommend, and the communication goals you set?
3. How does this differ according to the individual's age and stage of their communication journey?
4. What do you do/advise/say to help the individual and their network use AAC effectively?

a. What obstacles have you encountered?

b. What solutions have you found?

5. What is your experience of working collaboratively with others in this process?
6. What do we, as researchers, need to focus on in the next couple of years?

Who is this educational session for? This session is for clinicians, researchers, educators, and lived-experience experts who already implement AAC (or actively build AAC services) and want to exchange practical strategies, case-based reasoning, and implementation lessons for individuals with NDDs and CCN.

What will participants gain from this roundtable session?

Sharing of (anonymised) cases, best practices, communication tools, challenges and potential solutions; an opportunity for networking and building collaborations that can continue online after this event.

Session structure:

- 15 minutes – introduction to the session and discussion questions
- 60 minutes – moderated discussion
- 10 minutes – summary of shared learning points and planning for (optional) online follow-up/continued networking

Co-funded by the Health Programme of the European Union

Pain in people with intellectual disabilities: implementation and network formation (a round table discussion).
Leendert Sneep & Nanda de Knegt

Topic: In this session we will show how the implementation of the guideline for pain in people with intellectual disabilities is shaped in the Netherlands. We will also tell you something about the usefulness and importance of network formation for further development and implementation. Finally, we will discuss with you the added value of and opportunities for European networking on pain and intellectual disability.

This session is linked to the educational breakout session on pain (day 1: 14:30) and the lecture of Ilse Zaal-Schulder provided during third plenary session (day 2: abstract ID 57 prestation at 11 am)

Target group: Professionals and relatives who are involved in policy on pain in people with intellectual disabilities at the organizational level or at the regional or national level: Policy professionals, professionals of knowledge or network organizations, members of parent associations or pain experts.

Discussion questions:

1. How can methodical and interdisciplinary collaboration on pain be implemented in your own workplace or within your own organization?
2. What is needed for implementation at the national level? Who should be involved? (healthcare professionals, healthcare organizations, professional associations, knowledge organizations, government)
3. How can we strengthen each other at the European level in developments around pain in people with intellectual disabilities?

Session structure

- 10 minutes introduction and goals
- 15 minutes information about implementation and networking based on developments in the Netherlands
- 35 minutes discussion
- 15 minutes summary and planning for (optional) online follow-up/continued networking

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