EuroNDD workshop 2026 I FINAL PROGRAMME POSTER PRESENTATIONS

ID #	ABSTRACT TITLE	PRESENTER
	Theme 1 - Understanding the Neurobiology	
1	One of the Eight Known Cases Worldwide: A 7-Year-Old Girl with a De Novo RYBP Variant and Syndromic Neurodevelopmental Disorder	Agata Cieślikowska
2	Molecular and Clinical Insights from Seven Patients with Crisponi Syndrome from Southeastern Turkey	Akçahan Akalın
3	From Clinical Phenotypes to Mechanisms: Linking Human and Mouse Studies in Dup15q Syndrome	Amy van Hattem
85	UBE3A-neighbouring genes on chromosome 15q11.2-13.1 contribute to Angelman syndrome phenotype	Amy van Hattem
4	DUP15q syndrome : Clinical and genetic description of a Spanish cohort	Ana Roche
5	Decoding CAMK2A: Deep Phenotyping of a Unique Cohort Sheds Light on a Crucial Learning and Memory Pathway	Anjuli Dijkmans
7	Familial Chromosomal Translocation Reveals Novel Brain-Specific Transcripts Associated with Mild Cerebellar	Asli Silahtaroglu
8	Integrative Genomic Approaches Reveal Novel Etiologies in Cerebral Palsy and Related Neurodevelopmental	Ayca Yigit
10	A truncating variant in ADORA3 encoding Adenosine Receptor A3, is associated with autosomal dominant Tourette Syndrome.	Ginevra Zanni
13	Integrating Radiogenomics into Neurogenetic Diagnostics	Alfi Aran Shukur
14	Expanding the evaluation of skeletal anomalies in patients with KBG syndrome ; recommendations for clinical	Marit van der Leij
15	Novel microdeletion syndrome identified on chromosome 7?	Kerstin Kamolane
16	Is the Autism Phenotype in Phelan-McDermid Syndrome (PHMDS) more Friendly?	Kristin A Bakke
17	Whole Exome Sequencing in a Tunisian cohort of 100 children with Neurodevelopemental Disorder	Lamia Ben Jemaa
18	A Systematic Review Illustrates the Expanding Clinical and Molecular Landscape of Helsmoortel-Van der Aa	Lusine Harutyunyan
19	Severe epilepsy, prominent myoclonus and dystonia in patient with AGO1 p.Phe180del de novo variant (Case	Maria Giertlova
20	Neurodevelopmental phenotype caused by complex structural chromosomal rearrangement – small cohort of patients with 8p inverted duplication/deletion syndrome	Monika Kowalczyk-Rusak
23	Neurodevelopmental disorders and genotype-phenotype correlation in Smith Magenis Syndrome: systematic review and French cohort results	Pauline Boiroux
25	MED13L syndrome : Contribution of the GENIDA database to patient phenotyping	Roseline Caumes
26	Multilevel approach to unravel neuropathogenic mechanisms of SIN3A haploinsufficiency in Witteveen-Kolk	Sharon Kolk
80	The ProMiSe of integrating knowledge and tailored intervention strategies to improve NDD care	Sharon Kolk
27	X-linked disorders associated with RAB39B: a phenotype similar to FMR1-related disorders?	Sylvie Odent
28	Widening the IHPRF1's clinical and molecular spectrum through NALCN in silico structural analysis	Lorenzo Sinibaldi, Vito Luigi Colona
29	New Pathogenic RAI1 Variant in a Patient Presenting with Severe Behavioral Dysregulation in a Multidimensional Neurodevelopmental Profile: A Case Report	Zeineb Ghattassi
33	Neuropsychological insights: Executive and social functioning in children and adults with Noonan syndrome	Jennifer Kramer
34	Neuropsychological insights: Koolen-de Vries syndrome and CAMK2-related syndromes	Carmen Oldenboom
36	A Novel TTI2 Variant: Expanding the Clinical Spectrum of Triple T Complex Disorder	Hatice Mutlu
37	Long-Term Follow-Up in Two Siblings with CODAS Syndrome	Hatice Mutlu
38	Beyond The Classical Triad: Rare Features And Multidisciplinary Insights From Five Sotos Syndrome Patients	Ieva Snieckute
83	A Genetically Unresolved Neurodevelopmental Disorder with Unusual Cutaneous Manifestations in a Consanguineous Palestinian Family	Zeynep Tümer as representative of Yaqoub Ashhab
	Theme 2 – From Molecular Diagnostic to Intervention	
32	A case of 17p13.3 microduplication with global developmental delay, dysgenesis of the corpus callosum, and multiple congenital anomalies: diagnostic challenges and genetic insights	Anzhela Yervandyan
35	Allelic Missense Variants in FGF13A: Same Site, Different Syndrome, and a Caffeine Fix	Georgia Vasileiou
40	DNA methylation episignatures in NDDs: expanding diagnostics, validating with nanopore, and future functional applications	Liselot van der Laan
	Theme 3 – Health Information System and Data Availability	
43	FindMe2care: a contact platform for patients with confirmed genetic diagnoses	Christian Gebhard
44	Privacy and Data Protection Issues in Creating a Rare Disease Patient Registry : Experiences from RettX	David Townend

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	Theme 4 – Multidisciplinary Diagnostics and Interventions	
47	Diagnostic of FMR1 premutation in a cohort of children with neurodevelopmental disorders: definition of a clinical phenotype for pediatric population	Ana Roche, Carme Torrents
48	Natural History Study of a cohort of Spanish patients: interim analysis focused on epilepsy and sleep	Ana Roche
50	Follow-up of a 16-months old baby assessed with ADNP syndrome: does a very early genetic assessment allow an effective preventive multidisciplinary work?	Catherine Saint-Georges
51	The use of the Computer-Based instrument for Low motor Language Testing (C-BiLLT) in children with complex communication needs due to rare genetic disorders	Cindy Navis
52	From Gene to Communication: Translating Neurogenetic Profiles into AAC-Based Communication Interventions in Rare Neurodevelopmental Syndromes	Elzbieta Dawidek, Paulina Rutka
55	Placing the person with a genetic neurodevelopmental disorder at the heart of the holistic care network: the experience of running focus groups with people with Rett syndrome in order to co-produce training and educational resources for multi-disciplinary teams.	Gillian Townend
53	Botulinum Toxin Treatments in Children and Adults with Profound Intellectual and Multiple Disabilities (PIMD) in the Netherlands: Sense or Nonsense?	Esther Calame
54	Autosomal Recessive Intellectual Developmental Disorder Type 67 (MRT67) Associated with EIF3F c.694T>G (p.Phe232Val): Case Series and Perspectives for Translational Research	Ewelina Preizner-Rzucidło
56	Adult Emotional and Behavioural Patterns in Prader-Willis syndrome and Williams syndrome	Heidi Elisabeth Nag
58	A rare variant of the ETF1 gene: Broadening the phenotypic spectrum and possible influence of early multidisciplinary management	Joana Matos
60	“Mind the gap” –ERN ITACHA guidelines on transition from paediatric to adult healthcare system.	Katarzyna Åświeczkowska
61	Environmental enrichment for children with developmental disabilities and behavioral phenotypes: bridge from theory into practice…. yes we can!	Katleen Ballon
62	Establishing a multidisciplinary clinic for children with Smith Magenis Syndrome: the Leuven experience	Katleen Ballon
64	Interdisciplinary networks targeting pain in people with intellectual disabilities: recommendations on micro (care center), meso (national), and macro (European) levels	Leendert Sneep, Nanda de Knegt
65	Blood Biomarkers for Neurodegeneration and Alzheimer's Disease in Individuals with Intellectual Disabilities: A Systematic Review	Lindsey Koster
70	A therapeutic education programme for patients with PIMD/polyhandicap in France: a multidisciplinary and network-based approach.	Marie Hully
67	The need for interprofessional collaboration in the care of individuals with rare genetic intellectual disability: A patient journey mapping study (Project 1)	Mana Nasori
68	Who Takes the Lead? Dispersed Responsibility in Interprofessional Collaboration around Challenging Behaviour in Rare Genetic Intellectual Disability Syndromes (Project 2)	Mana Nasori
69	Healthcare professionals’ intentions and competencies towards interprofessional collaboration around challenging behaviour in people with rare genetic intellectual disabilities syndromes (Project 3)	Mana Nasori
81	From building an International and Interdisciplinary Network towards Care Principles for Individualized, Interdisciplinary, and Holistic Care for Individuals with PIMD/polyhandicap	Sylvia A. Huisman
82	Lessons Learned from the Development of Clinical Practice Guidelines for Individuals with PIMD/polyhandicap: Methodological Challenges and Future Directions	Sylvia A. Huisman
72	Health and care characteristics of aging adults with Profound Intellectual and Multiple Disabilities: a cross-sectional study in a national cohort	Marie-Christine Rousseau
73	Factors Associated with the Quality of Life of Parents of Individuals with Profound Intellectual and Multiple Disabilities/Polyhandicap: A 5-Year Follow-Up	Marie-Christine Rousseau
74	Social representations and parenthood: a qualitative study among parents of persons living with Profound Intellectual and Multiple Disabilities/Polyhandicap	Marie-Anastasie Aim
75	Impact of Communication and Feeding Disorders on the Quality of Life of Patients with Polyhandicap	Marie-Christine Rousseau
76	Factors Associated with Family Functioning Among Parents and Siblings of persons with PIMD/Polyhandicap	Marie-Christine Rousseau
77	Thyroid Hormone Transporter Monocarboxylate Transporter 8 (MCT8) Deficiency : A Case Report	Leah Loughlin

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