EuroNDD workshop 2024 I FINAL PROGRAMME POSTER PRESENTATIONS I Thursday 4 April

ID #	ABSTRACT TITLE	PRESENTER
	Theme 1 - Genes & Pathways	
11	GenIDA, an international participatory database to better characterise comorbidities of genetic forms of intellectual disability: Novel findings on KBG, SETD5 and DDX3X syndromes	Pauline Burger
39	Csmd3 as a potential candidate for neurodevelopmental disorders	Kaisa Teele Oja
58	New insights in 9q21.13 microdeletion syndrome: Genotype–phenotype correlation of 28 patients	Alessandro De Falco
70	Rare copy number variations in a romanian pediatric cohort with autism spectrum disorders	Aurora Arghir
77	X-linked intellectual disability syndrome in female patient with mutation in HUWE1 gene	Agnieszka Madej-Pilarczyk
	Theme 2 – Ethical, legal and Psycho-social aspects	
14	A new French network for challenging behaviors with genetic origin: the GenoPsy Network	Caroline Demily
35	A longitudinal study of challenging behavour and autistic symptoms in smith-magenis' syndrome	Monica Stolen Dønnum
36	Psyciatric symptoms in the norwegian smith-magenis' and potocki-lupski syndrome population	Monica Stolen Dønnum
47	Prioritizing for impact: Establishing criteria to select new ERN-ITHACA clinical practice guideline topics	Mirthe Klein Haneveld
48	'We are the engine': Patient advocate perspectives on clinical practice guideline development for rare congenital malformations and/or intellectual disability	Mirthe Klein Haneveld
51	What matters most? A mixed-method study to develop a core patient reported outcome set for individuals with genetic intellectual disability	Nadia van Silfhout
79	Integrated care for patients with NDD and rare diseases in Romania	Dorica Dan
84	Navigating neurodevelopmental disorders: Insights from a Romanian cohort and empowering patients through education	Alexandra Dumitra
	Theme 3 – Applied & Emerging Therapies	
17	Towards precision medicine: Challenges and advancements	Arianne Bouman
37	Eating behaviour and issues in rare genetic disorders	Heidi Elisabeth Nag
45	Individualized antisense oligonucleotide therapies for patients with rare neurological disorders	Marlen Lauffer
56	Cognition and emotion in noonan syndrome: Current insights into diagnostics and treatment	Ellen Wingbermuhle
96	A 16-months old baby with ADNP syndrome: when a very early assessment allows a preventive transdisciplinary work	Catherine Saint Georges
	Theme 4 – Diagnostics	
1	De novo variant in TLK2: Clinical evaluation and genotype-phenotype of a neurodevelopmental disorder	Teresa Carrion
2	A recurrent de novo mutation in zmynd11 associated with global developmental delay genocopy the 10p15.3 deletion syndrome: A case report	Teresa Carrion
3	Exploring the mild phenotype of KCNC1 variants beyond myoclonic epilepsy: A case report	Diogo Fernandes da Rocha
26	Developing a novel genotype-to-phenotype prediction tool for chromosome deletions – the chromosome 6 project	Eleana Rraku
29	The association between autism and genetic syndromes	Kristin Andersen Bakke
30	Rare genetic syndromes with specific and ordinary clinical challenges; examplified by phelan mcdermid syndrome	Sissel Berge Helverschou
33	From craniofacial development to psychosis: Deciphering the impact of a rare EFTUD2 variant in a case early onset schizophrenia with dysostosis mandibulofaciale and microcephaly. A case report	Cyril Hanin
31	Fragile X syndrome – should it still be considered as a first line test in children with developmental delay?	Karolina Śledzińska
41	Identifying novel candidate genes and variants in a turkish cerebral palsy cohort: Utilizing whole and clinical exome sequencing data for improved diagnostics	Ayça Yiğit
42	Multidimensional impairment: Exploring the diagnostic concept through two case presentations with rare genetic variants	Joana Matos
66	Confirmation and expansion of the tceal1 related neurodevelopmental disorder fenotype	Marjon van Slegtenhorst
72	Compound heterozygous variants in the non-coding RNU12 gene in two siblings with neurodevelopmental and movement disorder	Kerstin Alt
73	Reanalysis of whole exome data lead to diagnosis in unsolved patient with intellectual disability, spastic paraparesis, retinitis pigmentosa, hearing loss and dysmorphic facies	Markéta Vlčková
74	Pallister-killian syndrome: neonatal phenotype and key elements for timely diagnosis	Giacomo Sperti

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	Theme 4 – Diagnostics	
75	Mapping the trajectory of syt1-associated neurodevelopmental disorder (baker-gordon syndrome)	Sam Norwitz
76	Genome sequencing supports the role of scn1a and pcdh19 in patients with undiagnosed dravet syndrome and related disorders	Myriam Essid
81	Clinical and molecular characterization of a patient presenting with neurodevelopmental disorder and multiple congenital abnormalities, carrying homozygous pathogenic variant in GZF1	Alessandro De Falco
85	Coffin-Lowry Syndrome : Phenotypic spectrum in affected females	Anna Gerasimenko
86	Molecular diagnosis of Tay-Sachs disease: A Moroccan case report	Sabrine Bouressas
88	Beaulieu-Boycott-Innes Syndrome as a rare genetic neurodevelopmental disorder caused by a novel homozygous variant in THOC6 gene in a sibling	Syrine Hizem
89	Microcephaly, Epilepsy, and Diabetes Syndrome 1: A Moroccan case report of compound Heterozygous IER3IP1 mutations	Houda Jelti
91	Interest of High-Throughput Sequencing in adult patients with intellectual disability	Perrine Charles
92	Psychomotor regression and mouvement disorder retalted to sucla2: A Moroccan case report	Abdelhaq Lamaibdel
97	Grange syndrome, characterized by early-onset hypertension: preliminary finding of intellectual disability due to compound heterozygous YY1AP1 gene frameshift variants	Hilmi Bolat
98	Cohen syndrome: Can early-onset neutropenia and hypotonia provide early diagnosis and intervention for intellectual disability?	Gül Ünsel-Bolat
	Theme 5 – Mechanisms of diseases, model systems & translational pre-clinical work	
23	Validation of a preclinical model for the evaluation and development of new therapeutic approaches in duplication 15q disease	Verdiana Pullano
32	Neuronal phenotypes associated with FBXO11-deficiency can be alleviated with chemical activation of the proteasome	Anne Gregor
40	Integrating biological and neuropsychiatric underpinnings of neurodevelopmental disorders in order to design novel treatment strategies	Sharon Kolk
44	Mitotic defects in human ASPM microcephaly	Sandrine Passemard
49	Patients with Kabuki Syndrome type-1 and Kleefstra Syndromes present altered immune cell responses	Burcu Al
55	Identification of a cryptic splicing site in MED13L Intron 7 leading to truncated protein in patients with MED13L Syndrome	Jade Fauqueux
61	Building meta-cohorts from copy number variant (CNV) carriers and their family members	Adrian Harwood
65	Functional characterization of variants in CACNA1A causing developmental epileptic encephalopathy, hemiplegic migraine, and ataxia	Amanda Levy
94	Alterations in cortical differentiation and neuronal network activity in 22q11.2 deletion syndrome	Gemma Wilkinson
	Theme 6 – Polyhandicap	
5	Health care of persons with complex developmental disabilities from 3 european experiences: France, Italy, and Norway	Marie-Christine Rousseau
6	Persons with polyhandicap: Being cared for in a pediatric structure for young people over 18: is it really a problem?	Marie-Christine Rousseau
7	Persons with polyhandicap: Could the presence of behavioral disorders be the cause of a greater isolation?	Any Beltran
8	Development and initial validation of the polyhandicap severity scale	Karine Baumstarck
15	Persons with polyhandicap, their families, and the institutional caregivers: the French EVAL-PLH cohort	Ilyes Hamouda
21	How can we improve outpatient care for children with PIMD? Insight into experiences and preferences of parents and healthcare professionals	Catelijne Coppens
22	Parents' experiences of parenting a child with profound intellectual and multiple disabilities in France: a qualitative study	Marie-Anastasie Aim
34	Influencing factors on quality of transitional care for adolescents with profound intellectual and multiple disabilities in The Netherlands	Ilse Ooms
43	Focusing on what is important in severe neurological impairment- a prioritisation survey of parents and professional caregivers	Siobhan McCormack
80	Tacit knowledge in the care for persons with profound intellectual and multiple disabilities	Sylvia Huisman

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