EuroNDD workshop 2023 I FINAL PROGRAMME POSTER PRESENTATIONS I Thursday 20 April

#	ABSTRACT TITLE	PRESENTER
	Delphinus **Theme 1 - Applied &Emerging Therapies**	
9	Syndromic intellectual disability in Tunisian patients: contribution of next generation sequencing	Cyrine Adhoum
10	Reanalysis of SNP analysis data in patients with neurodevelopmental delay	Emilia Bijlsma
11	Clinical practice guidelines for rare genetic neurodevelopmental disorders: a review and systematic quality appraisal	Mirthe Klein Haneveld
12	miRNAs and isomiRs: serum-based biomarkers for the development of Intellectual Disability and Autism Spectrum Disorder in Tuberous Sclerosis Complex	Mirthe Scheper
13	Cannabidiol (EPIDYOLEX) for behavioral problems in patients with Tuberous Sclerosis Complex, Sanfilippo Disease and Fragile X Syndrome: protocol for a series of randomized placebo-controlled N-of-1 trials; can we make personalized care for individuals with an intellectual disability happen? Insights from a large intellectual disability registry	Annelieke Müller
	Delphinus **Theme 2 – Data collection, database, registries, use of AI**	
14	Can we make personalized care for individuals with an intellectual disability happen? Insights from a large intellectual disability registry	Annelieke Müller
15	GenIDA, an international participatory database to better characterise comorbidities of genetic forms of intellectual disability: novel findings on Koolen-de Vries syndrome	Pauline Burger
16	“Dysmorphology Meeting” – An MDT Approach To Improve Access To The Clinical Genetics Services	John Coleman
17	Parents as partners: data consistency and data availability of parent-reported phenotypes	Aafke Engwerda
	Orion 2 **Theme 3 – Profound and multiple learning disability**	
29	Clinical pathways in rare diseases - cancer screening in PTEN hamartoma tumor syndrome in Portugal	Celia Azevedo Soares
30	Irritability in children with rare neurodevelopmental copy number variants (ND-CNVS)	Jessica Hall
31	Autism symptom profiles in children and young adults with Fragile X Syndrome, Neurofibromatosis type 1, Tubereus Sclerosis Complex and Angelman Syndrome	Kamil Hiralal
32	Don't forget about me: Dementia in rare genetic neurodevelopmental disorders, a systematic review	Hadassa Kwestie
33	Recognizing collagenopathy disorders in neuropaediatrics: a case series	Karen O'Neil
34	Psychiatric findings in adults with Triple X syndrome	Maarten Otter
35	Disabling Fatigue in adults with Neurofibromatosis type 1: a multidisciplinary approach	Anna Rosenberg
36	Analysis of neurodevelopmental, behavioral and social status of Polish adult population of Cornelia de Lange Syndrome	Karolina Śledzińska
37	Case report of Potocki-Lupski Syndrome diagnosis in adulthood	Rasa Traberg
38	Improving detection of rare overgrowth syndromes among patients referred to the endocrinology ward for treatment of acromegaly	Trui van Essen
39	Healthcare needs of people with NDD PROM4RARE: Giving a voice to individuals with a rare genetic disorder associated with intellectual disability	Nadia van Silfhout
40	Parkinsonism in individuals with genetic neurodevelopmental disorders: a systemic review	Emma von Scheibler

#	ABSTRACT TITLE	PRESENTER
	Orion 1 **Theme 4 – Mechanisms of diseases, model systems & translational pre-clinical work**	
19	Pontocerebellar hypoplasia genetic diagnosis: experience of our reference center	Lydie Burglen
20	DNA Methylation episignature in Gabriele-de Vries Syndrome	Florian Cherik
21	Understanding the effect of ANKRD11 haploinsufficiency on early brain development	Jet Coenen-van der Spek
22	7-TESLA IN-VIVO 1H-Magnetic Resonance Spectroscopy of glutamate & GABA in 22Q11.2 copy number variants	Claudia Vingerhoets
23	Genetic abnormalities in neurodevelopmental disorders with multidimensional impairment	Cyril Hanin
24	Interest of exome sequencing in non-syndromic specific learning disorders: a French pilot study	Eléonore Viora
	Orion 1 **Theme 5 – Ethical, legal and Psycho-social aspects**	
25	Parents of a child with a rare neurological as stakeholders of his social & education process - a case study analysis	Katazyna Świeczkowska
28	Family centered approach – ICF based partnership between families and professionals	Katazyna Świeczkowska
26	Contributing factors to parental stress in neurodevelopmental disorders; An international survey among 587 PMS families worldwide	Annemiek Landlust
27	Neurodevelopmental disorders in Wiedemann-Steiner syndrome	Heidi Elisabeth Nag
	Orion 3 **Theme 6 – Genes and pathways**	
41	A novel FZR1 variant causing developmental and epileptic encephalopathy	Boglarka Bansagi
42	New NDD candidate genes discovered and followed-up through the Norwegian ERN-ITHACA network	Sofia Douzgou Houge
43	LIG4Syndrome: phenotypic variability in a consanguineous family	Diogo Fernandes da Rocha
44	Reanalysis of whole-exome sequencing data of previously WES-negative children with intellectual disability and/or developmental delay: what are the outcomes of reanalysis in a standard patient care context?	Saskia Koene
46	Two affected brothers with Tsukahara Syndrome present with a homozygous deletion in PUS7	Stéphanie Moortgat
47	Triplications of chromosome 1P36.3, including the genes GABRD and SKI, are associated with a developmental disorder and recurrent facial features	Elise Pelgrims
48	Two case report of a new recognizable syndrome - DEGCAGS (developmental delay with gastrointestinal, cardiovascular, genitourinary and skeletal abnormalities)	Mafalda Santos
49	Homozygous variant c.226C>T p.(Arg76*) in the TRMT10A GENE – a Portuguese case	Isabel Silva
50	Global developmental delay, dysmorphia and severe oropharyngeal dysfunction in a girl with a nonsense mutation in CUX1 gene	Dorota Wicher
	Walking Bridge **Theme 7 – Update on most frequent syndromes**	
1	Interpreting genetic variants in the context of dual diagnosis in a patient with developmental delay	Luka Abashishvili
2	Creatine transporter deficiency: importance of clinical, biochemical and genotype-phenotype correlations	Kakha Bregvadze
3	Pathogenic variations of gene encoding subunits of the SWI/SNF complex in for 4 patients presenting complex neurodevelopmental disorders without intellectual disability	Roseline Caumes
4	Recurrence of infant death from severe epileptic encephalopathy without molecular diagnosis	Mafalda Melo
5	Syntelencephaly (middle interhemispheric variant): an holoprosencephaly like the others?	Sylvie Odent
6	Could severe microcephaly (< 6 SD) be a good clinical indicator of disease severity and functionality of sequential variants in patients with KIF-11-related congenital microcephaly?	Malgorzata Pawlowicz
7	"TAF2 related to the transcription factor TFIID: a new family and review of the literature"	Catarina Silva Rosas
8	Generalized lipodystrophy due to diencephalic syndrome as IHPRF1 syndrome's clinical onset in a 10-month-old male: patient report and 14 months of follow-up	Davide Vecchio

Scan me for detailed program