EuroDysmorpho 2026

Pavia | 16-19 September

Wednesday 16 September

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15:00 Opening address by Pr. Cesare Danesino and Pr. Fabio Sirchia

Session 1: Neurodevelopmental disorders - Chair: **Dagmar Wieczorek**

15:15	***Jeanne Jury***	Heterozygous alterations of GTF2I at the Williams-Beuren syndrome's locus cause a neurodevelopmental disorder
15:30	**Manuela Priolo**	Autonomic dysfunction profiling of Malan syndrome using pediatric autonomic symptom scales
15:45	***Melissa Pauly***	NUS1-related disease: Uncovering hidden clinical features and novel mutational hotspots
16:00	***Alperen Yılmaz Güler***	ASPM-related MCPH5 in adulthood in a consanguineous family: variable neurocognitive and dysmorphological findings in two sisters
16:10	***Dilsu Dicle Erkan***	A novel missense variant in TUBB2B in a consanguineous infant: Pseudogene interference obscuring true zygosity and expanding the phenotypic spectrum of biallelic tubulinopathy
16:20	**Maria Francesca Bedeschi**	A novel homozygous intronic polrmt variant in two siblings with severe neurodevelopmental disorder: expanding the molecular spectrum of combined oxidative phosphorylation deficiency-55

16:30 **Coffee break**

Session 2: Neurodevelopmental disorders - Chair: **Silvia Kalantari**

17:00	**Huw Dorkins**	The emergence of Medical Genetics
17:45	**Federica Ruscitti**	Clinical characterisation of a cohort harbouring MAP2K1 and MAP2K2 variants: a 20-year single centre experience reflecting the phenotypic continuum between cardio-facio-cutaneous and Noonan syndrome
17:55	***Gianluca Contro'***	Special population require special approach: Genetics findings among the Sinti population in Reggio Emilia, Italy
18:05	***Diana Carli***	Phenotypic mimicry and dual diagnoses in patients evaluated for Beckwith-Wiedemann spectrum

Thursday 17 September

Session 3: Dysmorphology - Chair: **Sally Ann Lynch**

9:00	**Valérie Cormier-Daire**	The skeletal dysplasia field in 2026
9:45	***Giulia Bruna Marchetti***	The burden of misdiagnosis: One example from the drop by drop project leads to expand the spectrum of spliceosomopathies
10:00	***Federica Alberti***	Beyond the "one gene-one disease" model: A comparative performance analysis of ai phenotyping tools in chromatinopathies
10:15	**Ho-Ming Luk**	Familial White-Sutton syndrome in Chinese
10:25	**ESHG Young**	Presentation of ESHG Young

10:35 **Coffee break**

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Session 4: Unknowns - Chair: **Fabio Sirchia**

11:00	**Paolo Fontana**	Unknown case report: A male patient with progeroid phenotype
11:10	***Dinnar Yahya***	One more undiagnosed a "must-be genetic" case
11:20	**Deema Aljeaid**	An unresolved, lethal neurodevelopmental syndrome characterized by microcephaly, microtia, Pierre-Robin sequence, and severe growth restriction
11:30	***Vera Marques***	An unresolved syndromic condition combining craniofacial dysmorphism, branchial anomalies, neurodevelopmental delay and neonatal skin fragility
11:40	**Sofia Poggi Longostrevi**	Undiagnosed: patient with laryngomalacia, multiple malformations and dysmorphic features without a genetic diagnosis
11:50	**Stefano Benedetti**	Patient with multiple congenital malformations and dysmorphic features without a genetic diagnosis

Session 5: Limb defects - Chair: **Diana Carli**

12:05	***Silvia Cali***	An undiagnosed syndromic phenotype combining craniofacial dysmorphism, cranio-cervical malformations and bilateral preaxial polydactyly
12:15	**Sally Ann Lynch**	BHLHA9-related split hand and foot malformation
12:25	**Vera Giacomazzi**	Mosaic cardiofaciocutaneous syndrome due to a MAP2K1 variant: A multidisciplinary case report with functional evidence
12:35	**Monica-Octavia Muraru**	Expanding the phenotypic spectrum of GLI3-related disorders: Atypical Greig cephalopolysyndactyly syndrome associated with a truncating GLI3 variant
12:45	**Francesca Crivellaro**	Hallux macrodactilia as a hallmark of CNP-pathway disruption: 3 case reports

12:55 **Lunch**

Session 6: Structural variation and chromosomal imbalances - Chair: **Alessandra Renieri**

14:00	**Kleoniki Roka**	The multiple faces of telomere biology disorders: From dyskeratosis congenia and beyond
14:45	**Elena Rossi & Elisa Giorgio**	Cytogenetics in the genomic era

15:30 **Coffee break**

16:00	**Alessandra Renieri**	Revisiting Rett syndrome by long-read whole genome sequencing (LR-WGS)
16:15	**Marta Calvo**	TRAPPC9-related intellectual developmental disorder: A novel case with cryptic structural variants resolved by optical genome mapping
16:30	***Salvatore De Francisci***	Expanding the phenotypic spectrum of Pallister-Killian syndrome: Adolescent follow-up of a mild form
16:40	***Pavlina Rayko***	Neurodevelopmental and behavioral features in 48,XXYY syndrome: a case report
16:50	**Ozge Beyza Gundogdu Ogutlu**	De novo interstitial 3p21.31p14.3 duplication in a boy with esophageal atresia, talipes equinovarus, hypotonia, and global developmental delay
17:00	**Ieva Snieckute**	Intrafamilial phenotypic variability in two sisters with partial trisomy 13q31.1–q34 and terminal 3p26.3 deletion due to maternal balanced T(3;13)

Session 7: Prenatal diagnosis - Chair: **Mathys Wéber**

17:10	**Diana Mitter**	Complex genetic analysis in routine prenatal practice – Presentation of interesting cases
17:25	***Marta Melfa***	Prenatal diagnosis of a de novo 7q11.21q21.3 interstitial deletion in a fetus with a severe skeletal dysplasia-like phenotype and multiple congenital malformations
17:35	***Valentina Trevisan***	Molecular and clinicopathologic insights from a fatal case of congenital giant hepatic hemangioma harboring a somatic GNAQ p.Gln209pro variant
17:45	***Marianna Di Lorco Sgambati***	Prenatal diagnosis of a paternally inherited Feingold syndrome: Intra-familial variability and complex chromosomal rearrangements

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Session 8: Epigenetics - Chair: **Francesca Clementina Radio**

9:00	**Teresa Neuhann**	The value of epigenetic signatures as key to diagnosis in neurodevelopmental disorders
9:15	**Julien Van Gils**	Phenotypic and DNA methylation signature variability in familial Rubinstein-Taybi syndrome
9:30	***Quentin Sabbagh***	DNA methylation signatures drive molecular diagnosis in genetically unsolved Mendelian disorders of the epigenetic machinery
9:45	***Cord-Christian Becker***	Utility of episignature analysis in clinical genetics and a potential novel association of TLK2 gain-of-function with syndromic neurodevelopmental disorder
10:00	***Alma Kuechler***	The utility of epigenetic signature analysis in the evaluation of variants of uncertain significance (VUS) and for the molecular confirmation of genetic diagnoses
10:15	**Margherita Fatta**	The diagnostic complexity of CREBBP gene: a clinical case at the phenotypic boundary between Menke-Hennekam and Rubinstein-Taybi syndromes

10:30 **Coffee break**

Session 9: Syndrome delineation - Chair: **Julien Van Gils**

11:00	**Marco Tartaglia**	Emerging RASopathies, novel genes and phenotypic heterogeneity
11:45	***Giulia Severi***	Compound heterozygous variants in PAICS summarize the divergent previously described phenotypes
11:55	**Martina Bonadonna**	Naming the illness: the long road from clinical signs to molecular diagnosis
12:05	***Gunce Basarir***	One variant, three phenotypes: variable expressivity of a recurrent NSD2 variant in Rauch–Steindl syndrome
12:15	**Dorota Wicher**	Intellectual developmental disorder, x-linked 99, syndromic (MRXS99F) as an example of syndromes with female-restricted phenotype.
12:25	**Daniel Hovey**	A robin redbreast in a cage: a fatal laminopathy in a newborn
12:35	***Marina Gingele***	Progressive arthropathy revealing mucolipidosis IIIγ: A phenotype-based genomic diagnosis
12:45	**Joana da Silva Cardoso**	A severe neonatal fibrillinopathy caused by a de novo FBN1 variant: Expanding the clinical spectrum

12:55 **Lunch**

Session 10: Dual diagnosis or phenotypic expansion? - Chair: **Manuela Priolo**

14:00	**Mariya Levkova**	Modified facial phenotype in syndromic children: A source of diagnostic uncertainty
14:10	**Natalia Braun-Walicka**	Skin pigmentation disorders as a dominant trait in patients with a phenotype suggesting genetic predisposition – an attempt to elucidate the molecular basis: A case study.
14:20	**Clementina Radio**	Deciphering the molecular and clinical spectrum of ratars
14:30	**Alessandra Spano**	De novo ZNRF3 variant in a child with progressive macrocephaly and periventricular nodular heterotopia: expandind the neuroradiological spectrum.
14:40	***Simone Carbonera***	Unusual presentation of sotos syndrome with spontaneous pneumothorax in a 16-year-old male
14:50	***Claudia Ciaccio***	When imprinting goes vascular: moyamoya angiopathy in Silver-Russell syndrome
15:00	***Federica Perino***	Unmasking HNRNPD-related neurodevelopmental disorder: phenotypic expansion and incidental detection of parental mosaicism
15:10	***Giuseppe Reynolds***	WDR44-related ciliopathy with renal tubulopathy and hepatobiliary involvement: a novel de novo variant and phenotypic expansion
15:20	**Beatrice Rosa**	Isolated complex craniosynostosis associated with a de novo pathogenic CHD8 variant: incidental finding or expansion of the phenotypic spectrum of CHD8-related disorder?
15:30	**Gaia Visani**	Expanding the MAB21L2 phenotype: a case with ocular anomalies and Hirschsprung's disease
15:40	**Maria Chiara Montalbano**	A complex pulmonary phenotype in a patient with FGFR2-related LADD syndrome

10:30 **Coffee break**

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Session 11: Syndrome delination - Chair: **Livia Garavelli**

16:30	**Francesca Di Palma**	Diagnostic challenges in chromosome 15q11-q13 imprinting disorders: the phenotypic overlapping between Angelman syndrome and Prader-Willi syndrome
16:40	***Ariana Mendes***	Beyond false leads: Smith-Magenis syndrome unmasked by reverse phenotyping overriding a 22q11.2 microduplication and non-diagnostic SLC7A9 variants
16:50	***Emna Koubaa***	Clinical and molecular characterization of Kaufman oculocerebrofacial syndrome in a Tunisian child with a novel UBE3B variant
17:00	***Elisa D'Acunto***	KCNK3-related channelopathy: a new disorder characterized by developmental delay, dysmorphic features and organ malformations
17:10	**Alessandro Cogorno**	Bilateral choanal atresia and hearing loss
17:20	***Ngoc Anh PHAN***	Late diagnosis of Wiedemann-Steiner syndrome in an adult: the diagnostic challenge
17:30	***Eleonora Orlandini***	A case of RNU4ATAC-opathy complicated by chilblain-like lesions and severe lung disease associated with type i interferon pathway disregulation
17:40	***Anna Cattaneo***	The importance of parental phenotyping and clinical–laboratory integration in the diagnosis of an FLNA-related disorder: a case report.
17:50	***Benedetta Inzoli***	Might SOX4 be an overlooked syndromic MODY gene?
18:00	***Gabriela Anaya***	When a genetic diagnosis changes over time: re-evaluation of an FBN2 variant previously associated with Beals syndrome

20:00 **Dinner at Collegio Ghislieri** (By registration only)

Saturday 19 September

Session 12: Syndrome delination - Chair: **Maria Francesca Bedeschi**

9:00	**Katalin Szakszon**	COPB1 gene associated baralle-macken syndrome – a rare coatopathy. Morphological observations and differential diagnostic considerations
9:10	**Radka Kremlíková Pourová**	Long follow up of a girl with Vissers-Bodmer syndrome
9:20	***Marcello Scala***	DDX1 haploinsufficiency disrupts cellular stress responses and causes a neurodevelopmental disorder
9:35	***Rasa Traberg***	Variable phenotype of patients with Wiedemann-Steiner syndrome: case series from the single center
9:50	**Dagmar Wieczorek**	Genetic mosaicism – Diagnoses we almost missed
10:05	***Ilaria Carelli***	KDM5B-related neurodevelopmental disorder: variable expressivity and subtle dysmorphism in four patients with novel heterozygous variants
10:20	**Livia Garavelli**	Clinical and mutational spectrum of SEC23A-related dominant cranio-lenticulo-sutural dysplasia

Session 13: Syndrome delination - Chair: **Alain Verloes**

10:35	***Federica Francesca L'Erario***	Clinical and genomic endophenotypes of Pitt-Hopkins syndrome
10:50	***Elena Cicchetti***	What lies beyond Pierpont syndrome? Phenomizing TBL1XR1-related spectrum
11:05	***Berardo Rinaldi***	"Oh yes, I have an older brother with mild intellectual disability..."
11:20	***Clément Sauvestre***	HDR syndrome due to GATA3 haploinsufficiency: Phenotypic and molecular characterisation of the largest reported cohort
11:35	**Germana Viscogliosi**	The multifaceted phenotypic profile of neurofibromatosis type 1 in a family carrying the NF1 p.Gly848glu variant: A case series
11:45	***Angela Gentile***	Prenatal case of RIT1 mosaic variant and neonatal outcome
11:55	***Mohammad Sadegh Shams Nosrati***	SNAPC4-related neurodevelopmental disorder defines a stage-dependent spliceosomopathy with heterogeneous clinical trajectories

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