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EuroDysmorpho - September 16-19 2026, University of Pavia

Wednesday 16th of September

Session Chair :

Timeslot	Session	Talk #	Speaker	Abstract Title
15:00	**Session 1**	-	Scientific Committee	*Welcome address*
15:15	Neurodevelopmental disorders	1	Jeanne Jury	HETEROZYGOUS ALTERATIONS OF GTF2I AT THE WILLIAMS-BEUREN SYNDROME'S LOCUS CAUSE A NEURODEVELOPMENTAL DISORDER
15:30		2	Manuela Priolo	AUTONOMIC DYSFUNCTION PROFILING OF MALAN SYNDROME USING PEDIATRIC AUTONOMIC SYMPTOM SCALES
15:45		3	Melissa Pauly	NUS1-RELATED DISEASE: UNCOVERING HIDDEN CLINICAL FEATURES AND NOVEL MUTATIONAL HOTSPOTS
16:00		4	Alperen Yılmaz Güler	ASPM RELATED MCPH5 IN ADULTHOOD IN A CONSANGUINEOUS FAMILY: VARIABLE NEUROCOGNITIVE AND DYSMORPHOLOGICAL FINDINGS IN TWO SISTERS
16:10		5	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		6	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 Chair :

17:00	**Invited Talk**	-	**Huw Dorkins**	
17:45	**Session 2**	7	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	NDDs	8	Gianluca Contro'	SPECIAL POPULATION REQUIRE SPECIAL APPROACH: GENETICS FINDINGS AMONG THE SINTI POPULATION IN REGGIO EMILIA, ITALY
18:05		9	Diana Carli	PHENOTYPIC MIMICRY AND DUAL DIAGNOSES IN PATIENTS EVALUATED FOR BECKWITH-WIEDEMANN SPECTRUM

18:15

Thursday 17th of September

Session Chair :

Timeslot	Session 3	Talk #	Speaker	Abstract Title
09:00	**Invited Talk**	-	**Valérie Cormier-Daire**	**TBD**
09:45	Dysmorphology	10	Giulia Bruna Marchetti	THE BURDEN OF MISDIAGNOSIS: ONE EXAMPLE FROM THE DROP BY DROP PROJECT LEADS TO EXPAND THE SPECTRUM OF SPLICEOSOMOPATHIES
10:00		11	Federica Alberti	BEYOND THE "ONE GENE-ONE DISEASE" MODEL: A COMPARATIVE PERFORMANCE ANALYSIS OF AI PHENOTYPING TOOLS IN CHROMATINOPATHIES
10:15		12	Ho-Ming Luk	FAMILIAL WHITE-SUTTON SYNDROME IN CHINESE

10:25 **Coffee break**

Session Chair :

11:00	**Session 4**	13	Paolo Fontana	UNKNOWN CASE REPORT: A MALE PATIENT WITH PROGEROID PHENOTYPE
11:10	Unknowns	14	Mari Hachmeriyan	ONE MORE UNDIAGNOSED A "MUST-BE GENETIC" CASE
11:20		15	Deema Aljeaid	AN UNRESOLVED, LETHAL NEURODEVELOPMENTAL SYNDROME CHARACTERIZED BY MICROCEPHALY, MICROTIA, PIERRE-ROBIN SEQUENCE, AND SEVERE GROWTH RESTRICTION
11:30		16	Vera Marques	AN UNRESOLVED SYNDROMIC CONDITION COMBINING CRANIOFACIAL DYSMORPHISM, BRANCHIAL ANOMALIES, NEURODEVELOPMENTAL DELAY AND NEONATAL SKIN FRAGILITY
11:40		17	Silvia Cali	AN UNDIAGNOSED SYNDROMIC PHENOTYPE COMBINING CRANIOFACIAL DYSMORPHISM, CRANIO-CERVICAL MALFORMATIONS AND BILATERAL PREAXIAL POLYDACTYLY
11:50		18	Sofia Poggi Longostrevi	UNDIAGNOSED: PATIENT WITH LARYNGOMALACIA, MULTIPLE MALFORMATIONS AND DYSMORPHIC FEATURES WITHOUT A GENETIC DIAGNOSIS
12:00		19	Stefano Benedetti	PATIENT WITH MULTIPLE CONGENITAL MALFORMATIONS AND DYSMORPHIC FEATURES WITHOUT A GENETIC DIAGNOSIS

12:10 **Lunch**

Session Chair :

13:00	**Invited Talk ONLINE**	-	**Kleoniki Roka**	**The multiple faces of telomere biology disorders: from dyskeratosis congenia and beyond**
13:45	**Session 5**	-	ESHG Young	PRESENTATION OF ESHG-YOUNG
13:55	Limb defects	20	Sally Ann Lynch	BHLHA9-RELATED SPLIT HAND AND FOOT MALFORMATION
14:05		21	Muhammed DoÄŸukan Kalenderoğlu	ALL FOUR LIMBS, ALL THE SEQUENCING, STILL NO DIAGNOSIS: A LAURIN–SANDROW-LIKE CASE OF MIRROR-IMAGE POLYDACTYLY
14:15		22	Monica-Octavia Muraru	EXPANDING THE PHENOTYPIC SPECTRUM OF GLI3-RELATED DISORDERS: ATYPICAL GREIG CEPHALOPOLYSYNDACTYLY SYNDROME ASSOCIATED WITH A TRUNCATING GLI3 VARIANT
14:25		23	Francesca Crivellaro	HALLUX MACRODATTILIA AS A HALLMARK OF CNP-PATHWAY DISRUPTION: 3 CASE REPORTS
14:35		-	Discussion	

14:45 **Coffee break**

Session Chair :

15:15	**Invited Talk**	-	**Elena Rossi & Elisa Giorgio**	**Cytogenetics in the genomic era**
16:00	**Session 6**	24	Alessandra Renieri	REVISITING RETT SYNDROME BY LONG-READ WHOLE GENOME SEQUENCING (LR-WGS)
16:15	Structural variation and chromosomal imbalances	25	Marta Calvo	TRAPPC9-RELATED INTELLECTUAL DEVELOPMENTAL DISORDER: A NOVEL CASE WITH CRYPTIC STRUCTURAL VARIANTS RESOLVED BY OPTICAL GENOME MAPPING
16:30		26	Salvatore De Francisci	EXPANDING THE PHENOTYPIC SPECTRUM OF PALLISTER-KILLIAN SYNDROME: ADOLESCENT FOLLOW-UP OF A MILD FORM
16:40		27	Pavlina Rayko	NEURODEVELOPMENTAL AND BEHAVIORAL FEATURES IN 48,XXYY SYNDROME: A CASE REPORT
16:50		28	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		29	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)
17:10	**Session 7**	30	Diana Mitter	COMPLEX GENETIC ANALYSIS IN ROUTINE PRENATAL PRACTICE – PRESENTATION OF INTERESTING CASES
17:25	Prenatal diagnosis	31	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		32	Valentina Trevisan	MOLECULAR AND CLINICOPATHOLOGIC INSIGHTS FROM A FATAL CASE OF CONGENITAL GIANT HEPATIC HEMANGIOMA HARBORING A SOMATIC GNAQ P.GLN209PRO VARIANT
17:45		33	Marianna Di Lorco Sgambati	PRENATAL DIAGNOSIS OF A PATERNALLY INHERITED FEINGOLD SYNDROME: INTRA-FAMILIAL VARIABILITY AND COMPLEX CHROMOSOMAL REARRANGEMENTS
17:55		-	**Discussion**	

Friday 18th of September

Session Chair:

Timeslot	**Session**	**Talk #**	**Speaker**	**Abstract Title**
09:00	**Session 8**	34	Teresa Neuhann	THE VALUE OF EPIGENETIC SIGNATURES AS KEY TO DIAGNOSIS IN NEURODEVELOPMENTAL DISORDERS
09:15	Epigenetics	35	Julien Van Gils	PHENOTYPIC AND DNA METHYLATION SIGNATURE VARIABILITY IN FAMILIAL RUBINSTEIN-TAYBI SYNDROME
09:30		36	Quentin Sabbagh	DNA METHYLATION SIGNATURES DRIVE MOLECULAR DIAGNOSIS IN GENETICALLY UNSOLVED MENDELIAN DISORDERS OF THE EPIGENETIC MACHINERY
09:45		37	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		38	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		39	Margherita Fatta	THE DIAGNOSTIC COMPLEXITY OF CREBBP GENE: A CLINICAL CASE AT THE PHENOTYPIC BOUNDARY BETWEEN MENKE-HENNEKAM AND RUBINSTEIN-TAYBI SYNDROMES

10:25 **Coffee break**

Session Chair:

11:00	**Invited Talk**	-	**Marco Tartaglia**	**Emerging RASopathies, novel genes and phenotypic heterogeneity**
11:45	**Session 9**	40	Giulia Severi	COMPOUND HETEROZYGOUS VARIANTS IN PAICS SUMMARIZE THE DIVERGENT PREVIOUSLY DESCRIBED PHENOTYPES
11:55	Syndrome delineation	41	Martina Bonadonna	NAMING THE ILLNESS: THE LONG ROAD FROM CLINICAL SIGNS TO MOLECULAR DIAGNOSIS
12:05		42	Gunce Basarir	ONE VARIANT, THREE PHENOTYPES: 'VARIABLE EXPRESSIVITY OF A RECURRENT NSD2 VARIANT 'IN RAUCH–STEINDL SYNDROME
12:15		43	Dorota Wicher	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED 99, SYNDROMIC (MRXS99F; OMIM#300968) AS AN EXAMPLE OF SYNDROMES WITH FEMALE-RESTRICTED PHENOTYPE.
12:25		44	Daniel Hovey	A ROBIN REDBREAST IN A CAGE: A FATAL LAMINOPATHY IN A NEWBORN
12:35		45	Marina Gingele	PROGRESSIVE ARTHROPATHY REVEALING MUCOLIPIDOSIS IIIΓ: A PHENOTYPE-BASED GENOMIC DIAGNOSIS
12:45		46	Joana da Silva Cardoso	A SEVERE NEONATAL FIBRILLINOPATHY CAUSED BY A DE NOVO FBN1 VARIANT: EXPANDING THE CLINICAL SPECTRUM

12:55 **Lunch break**

Session Chair:

14:00	Phenotypic expansion or dual diagnosis?	47	Mariya Levkova	MODIFIED FACIAL PHENOTYPE IN SYNDROMIC CHILDREN: A SOURCE OF DIAGNOSTIC UNCERTAINTY
14:10		48	Mohammad Sadegh Shams Nosrati	SNAPC4-RELATED NEURODEVELOPMENTAL DISORDER DEFINES A STAGE-DEPENDENT SPLICEOSOMOPATHY WITH HETEROGENEOUS CLINICAL TRAJECTORIES
14:20		49	Clementina Radio	DECIPHERING THE MOLECULAR AND CLINICAL SPECTRUM OF RATARS
14:30		50	Alessandra Spano	DE NOVO ZNRF3 VARIANT IN A CHILD WITH PROGRESSIVE MACROCEPHALY AND PERIVENTRICULAR NODULAR HETEROTOPIA: EXPANDIND THE NEURORADIOLOGICAL SPECTRUM.
14:40		51	Simone Carbonera	UNUSUAL PRESENTATION OF SOTOS SYNDROME WITH SPONTANEOUS PNEUMOTHORAX IN A 16-YEAR-OLD MALE
14:50		52	Martina Lattuada	EXPANDING THE DYSMORPHOLOGICAL AND CLINICAL CHARACTERIZATION OF TNRC6B-RELATED NEURODEVELOPMENTAL DISORDER: A CASE REPORT
15:00		53	Claudia Ciaccio	WHEN IMPRINTING GOES VASCULAR: MOYAMOYA ANGIOPATHY IN SILVER-RUSSELL SYNDROME
15:10		54	Federica Perino	UNMASKING HNRNPD-RELATED NEURODEVELOPMENTAL DISORDER: PHENOTYPIC EXPANSION AND INCIDENTAL DETECTION OF PARENTAL MOSAICISM
15:20		55	Giuseppe Reynolds	WDR44-RELATED CILIOPATHY WITH RENAL TUBULOPATHY AND HEPATOBILIARY INVOLVEMENT: A NOVEL DE NOVO VARIANT AND PHENOTYPIC EXPANSION
15:30		56	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?
15:40		57	Gaia Visani	EXPANDING THE MAB21L2 PHENOTYPE: A CASE WITH OCULAR ANOMALIES AND HIRSCHSPRUNG'S DISEASE
15:50		58	Maria Chiara Montalbano	A COMPLEX PULMONARY PHENOTYPE IN A PATIENT WITH FGFR2-RELATED LADD SYNDROME

16:00 Coffee break

Session Chair:

16:30	**Session 11**	59	Francesca Di Palma	DIAGNOSTIC CHALLENGES IN CHROMOSOME 15Q11-Q13 IMPRINTING DISORDERS: THE PHENOTYPIC OVERLAPPING BETWEEN ANGELMAN SYNDROME AND PRADER-WILLI SYNDROME
16:40	Syndrome delineation	60	Ariana Mendes	BEYOND FALSE LEADS: SMITH-MAGENIS SYNDROME UNMASKED BY REVERSE PHENOTYPING OVERRIDING A 22Q11.2 MICRODUPLICATION AND NON-DIAGNOSTIC SLC7A9 VARIANTS
16:50		61	Emna Koubaa	CLINICAL AND MOLECULAR CHARACTERIZATION OF KAUFMAN OCULOCEREBROFACIAL SYNDROME IN A TUNISIAN CHILD WITH A NOVEL UBE3B VARIANT
17:00		62	Elisa D'Acunto	KCNK3-RELATED CHANNELOPATHY: A NEW DISORDER CHARACTERIZED BY DEVELOPMENTAL DELAY, DYSMORPHIC FEATURES AND ORGAN MALFORMATIONS
17:10		63	Alessandro Cogorno	BILATERAL CHOANAL ATRESIA AND HEARING LOSS
17:20		64	Ngoc Anh PHAN	LATE DIAGNOSIS OF WIEDEMANN-STEINER SYNDROME IN AN ADULT: THE DIAGNOSTIC CHALLENGE
17:30		65	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		66	Anna Cattaneo	THE IMPORTANCE OF PARENTAL PHENOTYPING AND CLINICAL–LABORATORY INTEGRATION IN THE DIAGNOSIS OF AN FLNA-RELATED DISORDER: A CASE REPORT.
17:50		67	Benedetta Inzoli	MIGHT SOX4 BE AN OVERLOOKED SYNDROMIC MODY GENE?
18:00		68	Gabriela Anaya	WHEN A GENETIC DIAGNOSIS CHANGES OVER TIME: RE-EVALUATION OF AN FBN2 VARIANT PREVIOUSLY ASSOCIATED WITH BEALS SYNDROME

20:00 Restaurant dinner

Saturday 19th of September

Session Chair:

Timeslot	Session 12	Talk #	Speaker	Abstract Title
09:00	Syndrome delineation	69	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.
09:10		70	Katalin Szakszon	COPB1 GENE ASSOCIATED BARALLE-MACKEN SYNDROME – A RARE COATOPATHY. MORPHOLOGICAL OBSERVATIONS AND DIFFERENTIAL DIAGNOSTIC CONSIDERATIONS
09:20		71	Radka Kremlíková Pourová	LONG FOLLOW UP OF A GIRL WITH VISSERS-BODMER SYNDROME
09:30		72	Marcello Scala	DDX1 HAPLOINSUFFICIENCY DISRUPTS CELLULAR STRESS RESPONSES AND CAUSES A NEURODEVELOPMENTAL DISORDER
09:45		73	Rasa Traberg	VARIABLE PHENOTYPE OF PATIENTS WITH WIEDEMANN-STEINER SYNDROME: CASE SERIES FROM THE SINGLE CENTER
10:00		74	Dagmar Wieczorek	GENETIC MOSAICISM – DIAGNOSES WE ALMOST MISSED
10:15		75	Ilaria Carelli	KDM5B-RELATED NEURODEVELOPMENTAL DISORDER: VARIABLE EXPRESSIVITY AND SUBTLE DYSMORPHISM IN FOUR PATIENTS WITH NOVEL HETEROZYGOUS VARIANTS
10:30		76	Livia Garavelli	CLINICAL AND MUTATIONAL SPECTRUM OF *SEC23A*-RELATED DOMINANT CRANIO-LENTICULO-SUTURAL DYSPLASIA

10:30 Coffee break

Session Chair:

Timeslot	Session 13	Talk #	Speaker	Abstract Title
11.00	Syndrome delineation	77	Federica Francesca L'Erario	CLINICAL AND GENOMIC ENDOPHENOTYPES OF PITT-HOPKINS SYNDROME
11.15		78	Elena Cicchetti	WHAT LIES BEYOND PIERPONT SYNDROME? PHENOMIZING TBL1XR1-RELATED SPECTRUM
11.30		79	Berardo Rinaldi	"OH YES, I HAVE AN OLDER BROTHER WITH MILD INTELLECTUAL DISABILITY…"
11:45		81	Clément Sauvestre	HDR SYNDROME DUE TO GATA3 HAPLOINSUFFICIENCY: PHENOTYPIC AND MOLECULAR CHARACTERISATION OF THE LARGEST REPORTED COHORT
12:00		82	Germana Viscogliosi	THE MULTIFACETED PHENOTYPIC PROFILE OF NEUROFIBROMATOSIS TYPE 1 IN A FAMILY CARRYING THE NF1 P.GLY848GLU VARIANT: A CASE SERIES
12:10		83	Carmen Bucolo	A NOVEL CASE OF THE MRAS-RELATED DISORDER: ONE STEP CLOSER TO THE HEART OF NOONAN SYNDROME
12:20		84	Angela Gentile	PRENATAL CASE OF RIT1 MOSAIC VARIANT AND NEONATAL OUTCOME
12:30		85	Vera Giacomazzi	MOSAIC CARDIOFACIOCUTANEOUS SYNDROME DUE TO A MAP2K1 VARIANT: A MULTIDISCIPLINARY CASE REPORT WITH FUNCTIONAL EVIDENCE