



**E.C.A. European Cytogeneticists Association**



**The European Society of Human Genetics**



# 16<sup>th</sup> Goldrain Course in Clinical Cytogenetics

**Aug. 22<sup>st</sup> to 28<sup>th</sup>, 2023**

**Goldrain, South Tyrol, Italy**

## PROGRAM

### **Directors**

**Albert Schinzel (Zurich, Switzerland) and Mariano Rocchi (Bari, Italy)**

### **FACULTY**

**D. Bartholdi (Bern, Switzerland), A. Baumer (Zürich, Switzerland), P. Benn (Farmington CT, U.S.A.), J.-M Dupont (Paris, France), E. Errichiello (Pavia, Italy) E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), K. Madan (Leiden, the Netherlands), K. Miller (Hannover, Germany), R. Pfundt (Nijmegen, The Netherlands), G. Van Buggenhout (Leuven, Belgium), M. Vismara (Rome, Italy), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)**

*Administrative organization: Scuola Europea di Citogenetica APS, Bari, Italy*



<http://www.biologia.uniba.it/SEC/>

<b>Monday, August 21th</b>		<b>Arrival at Goldrain</b>
19:00-20:30		Dinner
20:30-21:30	<b>A. Schinzel</b>	Introduction and organizational comments
21:30-	Get together	Steinkeller
<b>Tuesday, August 22st</b>		
<b>09:00-12:40</b>		<b>Session 1 (Basic and clinical cytogenetics)</b>
09:00-09:40	<b>S1 A. Schinzel</b>	History of clinical cytogenetics
09:40-10:20	<b>M1 K. Miller</b>	Basic concepts of chromosome aberrations and epidemiology
10:20-11:00	<b>Bt1 D. Bartholdi</b>	Genetic counselling for chromosome aberrations
11:00-11:20		Coffee break
11:20-12:00	<b>Bt2 D. Bartholdi</b>	Clinical dysmorphicologic examination in cytogenetic disorders.
12:00-12:40	<b>M2 K. Miller</b>	Quality assessment of cytogenetic laboratories
12:45-14:00		Lunch break
<b>14:00 – 18:20</b>		<b>Session 2</b>
14:00-14:40	<b>K1 T. Krones</b>	Ethical issues
14:40-15:20	<b>WS D. Bartholdi</b>	Clinical issues: difficult situations
15:20-16:00	<b>Bn1 P. Benn</b>	Traditional approaches for the detection of aneuploidy in prenatal diagnosis: screening using serum and ultrasound markers
16:00-16:20		Coffee break
16:20-17:00	<b>Bn2a P. Benn</b>	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I
17:00-17:40	<b>WS T. Krones</b>	Ethical issues: difficult situations
17:40-18:20	<b>M3 K. Miller</b>	Cytogenetics of spontaneous abortions and stillbirths
19:00-20:00		Dinner
21:00-	Get together	Steinkeller
<b>Wednesday, August 23nd</b>		
<b>09:00 – 12:40</b>		<b>Session 3</b>
09:00-09:40	<b>D1 J.-M. Dupont</b>	FISH techniques and their application
09:40-10:20	<b>Kp1 E. Klopocki</b>	Array: principles, technical aspects, different platforms
10:20-11:00	<b>P1 R. Pfundt</b>	SNP array analysis and interpretation in constitutional genome diagnostics
11:00-11:20		Coffee break
11:20-12:00	<b>WS E. Klopocki R. Pfundt</b>	Databases for the interpretation of array results
11:20-12:00	<b>WS K. Miller</b>	Segregation of chromosomal translocations: practical exercises (BRUNNENZIMMER room)
12:00-12:40	<b>Bu1 G. Van Buggenhout</b>	Common chromosome disorders and follow-up I
12:45-14:00		Lunch break
<b>14:00 – 19:00</b>		<b>Session 4</b>
14:00-14:40	<b>D2 J.-M. Dupont</b>	Optical genome mapping
14:40-15:20	<b>Bu2 G. Van Buggenhout</b>	Common chromosome disorders and follow-up II
15:20-16:00	<b>WS K. Miller E. Klopocki</b>	ISCN, standard and molecular
16:00-16:40	<b>Bu3 G. Van Buggenhout</b>	Microdeletion syndromes
16:40-17:00		Coffee break
17:00-17:40	<b>Kp2 E. Klopocki</b>	Position effects and effects of CNVs containing no genes
17:40-18:20	<b>M4 K. Miller</b>	Markers and ring chromosomes
18:20-19:00	<b>WS K. Miller</b>	Segregation of chromosomal translocations: practical exercises
18:20-19:00	<b>WS E. Klopocki R. Pfundt</b>	Databases for the interpretation of array results (BRUNNENZIMMER room)

19:00-20:00		Dinner	
21:00-		Get together	Steinkeller
<b>Thursday, August 24rd</b>			
<b>09:00 – 12:40</b>		<b>Session 5</b>	
09:00-09:40	D3	<b>J.M. Dupont</b>	Prenatal cytogenetic diagnosis: RAD (rapid aneuploidy detection) and CMA (chromosomal microarray detection)
09:40-10:20	Bu4	<b>G. Van Buggenhout</b>	The clinical effect of sex chromosomal aneuploidy
10:20-11:00	D4	<b>J.M. Dupont</b>	Prenatal cytogenetic diagnosis: mosaics in PND
11:00-11:20		Coffee break	
11:20-12:00	WS	<b>K. Miller</b>	Practical exercises in report writing
12:00-12:40	WS	<b>P. Benn J.-M. Dupont</b>	Appropriate management in prenatal screening and diagnosis
12:45-14:00		Lunch break	
<b>14:10 – 18:30</b>		<b>EXCURSION</b>	
19:00-20:00		Dinner	
21:00-22:00	R1	<b>M. Rocchi</b>	200 years after the birth of Gregor Mendel
22:00-		Get together	Steinkeller
<b>Friday, August 25th</b>			
<b>09:00 – 12:40</b>		<b>Session 6</b>	
09:00–09:40	Bn2b	<b>P. Benn</b>	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects II
09:40-10:20	Z1	<b>O. Zuffardi</b>	Complex chromosome rearrangements: chromothripsis and friends
10:20-11:00	Ba1	<b>A. Baumer</b>	MLPA and QF-PCR
11:00-11:20		Coffee break	
11:20-12:00	W1	<b>J. Wissner</b>	Prenatal ultrasound findings indicative for chromosome aberrations
12:00-12:40	Z2	<b>O. Zuffardi</b>	Genomics mosaics: something to keep in mind in genetic counseling
12:45-14:00		Lunch break	
<b>14:00 – 18:30</b>		<b>Session 7</b>	
14:00-14:40	Z3	<b>O. Zuffardi</b>	Revertant mosaics in disease and aging I
14:40-15:20	S2	<b>A. Schinzel</b>	Dysmorphic findings in chromosome aberrations: formation and interpretation
15:20-16:00	Ba2	<b>A. Baumer</b>	Uniparental disomy (UPD) and imprinting
16:00-16:20		Coffee break	
16:20-17:00	Z4	<b>O. Zuffardi</b>	From aneuploidy to structural rearrangements: a lifelong continuum
17:00-17:40	Bn3	<b>P. Benn</b>	Origin of aneuploidy
17:40-18:20	Z5	<b>O. Zuffardi</b>	Revertant mosaics in disease and aging II
18:20-19:00	Bn4	<b>P. Benn</b>	Pre-implantation genetic diagnosis
19:00-20:00		Dinner	
21:00-21:40		Students/Lecture	Case presentations
21:40-		Get together	Steinkeller
<b>Saturday, August 26th</b>			
<b>09:00 – 12:40</b>		<b>Session 8</b>	
09:00–09:40	Md1	<b>K. Madan</b>	Cytogenetic polymorphisms
09:40-10:20	V1	<b>M. Vismara</b>	Nomenclature of facial features in clinical dysmorphology
10:20-10:40		Coffee break	
10:40-11:20	Md2	<b>K. Madan</b>	Complex chromosome rearrangements
11:20-12:00	R2	<b>M. Rocchi</b>	Evolution and diseases. A trade-off
12:00-12:40	WS	<b>A. Baumer</b>	Work-up of cases: strategies after the detection of CNVs.
12:45-14:00		Lunch break	

<b>14:00 – 18:25</b>		<b>Session 9</b>	
14:00-14:40	Md3	<b>K. Madan</b>	Inversions and insertions
14:40-15:20	Bn5	<b>P. Benn</b>	Screening for single gene disorders
15:20-16:00	Md4	<b>K. Madan</b>	Sex chromosomes
16:00-16:20		<i>Coffee break</i>	
16:20-17:05	P2	<b>R. Pfundt</b>	Incidental findings during genetic testing
17:05-17:50	Md5	<b>K. Madan</b>	Disorders of sex development
17:50-18:40	P3	<b>R. Pfundt</b>	Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome
19:00-20:00		<i>Dinner</i>	
20:30-		Get together	Steinkeller
<b>Sunday, August 27th</b>			
<b>09:00 – 12:40</b>		<b>Session 10</b>	
09:00-09:40	Md6	<b>K. Madan</b>	Natural human chimeras
09:40-10:20	E1	<b>E. Errichiello</b>	breakpoint analysis of complex structural variants
10:20-11:00	WS	<b>J. Wissner A. Schinzel</b>	Twins at prenatal diagnosis
11:00-11:20		<i>Coffee break</i>	
11:20-12:40		<b>Students Lecturers</b>	Case presentations
12:45-14:10		<i>Lunch break</i>	
<b>14:15 – 18:45</b>		<b>Self-study or sight seeing</b>	
19:00-20:00		<i>Dinner</i>	
20:15-		Get together	Steinkeller
<b>Monday, August 28th</b>			
<b>09:00 – 12:40</b>		<b>Session 11</b>	
09:00–09:40	WS	<b>M. Vismara</b>	Databases for clinical genetics
9.40-10:20	WS	<b>E. Errichiello</b>	Regions of homozygosity improving the diagnostic yield in recessive disorders
10:20-11:00	R3	<b>M. Rocchi</b>	Evolution of chromosomes
11:00-11:20		<i>Coffee break</i>	
11:20-12:00		<b>Lecturers</b>	Presentation of cases raising ethical questions
12:00-12:40	S3	<b>A. Schinzel</b>	Paradigm shifts, unexpected findings and use of markers in cytogenetics
12:45-14:00		<i>Lunch break</i>	
<b>14:00 – 18:45</b>		<b>Evaluation; examination; castle tour</b>	
14:00-14:50		<b>Students</b>	Student presentations
14:50-16:10		<b>Students</b>	EXAMINATION
16:10-16:30		<i>Coffee break</i>	
16:30-17:10		<b>Faculty Students</b>	Presentation of the correct answers to exam
17:10-17:50		<b>Students Faculty</b>	Discussion of the program
18:00-18:40			Guided visit to the castle
19:30-21:00		<i>Farewell dinner</i>	Steinkeller or inner court of the castle
<b>Tuesday, September 29th</b>		<b>Goodbye Goldrain</b>	
08:00-09:00		breakfast	
09:00-		Checkout and departure 🐼	