



19th Goldrain Course in Clinical Cytogenetics

August 25-31, 2026



Monday, 24 August, Arrival

20:30-21:30	Welcome and introduction, by Mariano Rocchi		
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Tuesday, 25 August

9:00-9:40	D1	J.-M. Dupont	History of clinical cytogenetics
9:40-10:20	Mi1	K. Miller	Basic concepts of chromosome aberrations and epidemiology
10:20-11:00	Bt1	D. Bartholdi	Genetic counselling for chromosome aberrations (I)
11:00-11:20	Coffee break		
11:20-12:00	Bt2	D. Bartholdi	Genetic counselling for chromosome aberrations (II)
12:00-12:40	D2	J.-M. Dupont	FISH techniques and their application
12:40-14:20	Lunch break		
14:20-15:00	Bt3	D. Bartholdi	Clinical dysmorphic examination in cytogenetic disorders.
15:05-15:45 (A)	WS1	D. Bartholdi	Clinical issues: difficult situations
15:05-15:45 (B)	WS2	K. Miller/E. Klopocki	ISCN, standard and molecular
15:50-16:30	K1	E. Klopocki	Array: principles, technical aspects, different platforms and their application
16:30-16:50	Coffee break		
16:50-17:30 (B)	WS1	D. Bartholdi	Clinical issues: difficult situations
16:50-17:30 (A)	WS2	K. Miller/E. Klopocki	ISCN, standard and molecular
17:35-18:15	Mi2	K. Miller	Markers and ring chromosomes

Wednesday, 26 August

9:00-9:40	Bu1	G.v. Buggenhout	Common chromosome disorders (in adults and elderly patients)
9:40-10:20	Kr1	T. Krones	Ethical issues
10:20-11:00	Ma1	K. Madan	Cytogenetic polymorphisms
11:00-11:20	Coffee break		
11:20-12:00	D3	J.-M Dupont	Optical genome mapping
12:00-12:40	Bu2	G.v. Buggenhout	Microdeletion syndromes
12:40-14:20	Lunch break		
14:20-15:00 (A)	WS3	T. Krones & D. Battisti	Ethical issues: difficult situations
14:20-15:00 (B)	WS4	K. Miller	Segregation of chromosomal translocations: practical exercises
15:05-15:45	K12	E. Klopocki	Position effects and effects of CNVs containing no genes
15:45-16:25	Ma2	K. Madan	Sex chromosomes
16:25-16:45	Coffee break		
16:45-17:25 (B)	WS3	T. Krones & D. Battisti	Ethical issues: difficult situations
16:45-17:25 (A)	WS4	K. Miller	Segregation of chromosomal translocations: practical exercises
17:30-18:10	Ba1	A. Baumer	MLPA and QF-PCR

Thursday, 27 August

9:00-9:40	Bu3	G.v. Buggenhout	The clinical effect of sex chromosomal aneuploidy
9:40-10:20	Ma3	K. Madan	Disorders of Sex Development
10:20-11:00	Ba2a	A. Baumer	Uniparental disomy (UPD) and imprinting (part I)
11:00-11:20	Coffee Break		
11:20-12:00	Ba2b	A. Baumer	Uniparental disomy (UPD) and imprinting (part II)
12:00-12:40	Ma4	K. Madan	Complex chromosome rearrangements
12:40-14:20	Lunch Break		
Excursion			
21:00-22:00	R1	<i>M. Rocchi</i>	<i>Why we are what we are</i>

Friday, 28 August			
9:00-9:40	D4	J.-M. Dupont	Introduction to prenatal screening
9:40-10:20	W1	J. Wisser	Prenatal ultrasound findings indicative for chromosome aberrations
10:20-11:00	D5	J.-M. Dupont	Non-invasive prenatal aneuploidy testing using cfDNA
11:00-11:20	Coffee break		
11:20-12:00	D6	J.-M. Dupont	DNA Prenatal cytogenetic diagnosis: RAD and CMA
12:00-12:40	Mi3	K. Miller	Cytogenetics of spontaneous abortions and stillbirths
12:40-14:20	Lunch break		
14:20-15:00 (A)	WS5	K. Miller	Practical exercises in report writing
14:20-15:00 (B)	WS6	J.-M. Dupont	Appropriate management in prenatal screening and diagnosis
15:05-15:45	D7	J.-M. Dupont	Prenatal cytogenetic diagnosis: mosaics in PND
15:45-16:25	Ma5	K. Madan	Inversions and insertions
16:25-16:45	Coffee break		
16:45-17:25 (B)	WS5	K. Miller	Practical exercises in report writing
16:45-17:25 (A)	WS6	J.-M. Dupont	Appropriate management in prenatal screening and diagnosis
17:30-18:10	Student presentations		

Saturday, 29 August			
9:00-9:40	Z1	O. Zuffardi	The partial rescue of unfavourable zygotic imbalances (I)
9:40-10:20	W2	J. Wisser	Twins in prenatal diagnosis
10:20-11:00	Ma6	K. Madan	Natural human chimeras
11:00-11:20	Coffee break		
11:20-12:00	P1	R. Pfundt	SNP array analysis and interpretation in constitutional genome diagnostics
12:00-12:40	Z2	O. Zuffardi	The partial rescue of unfavourable zygotic imbalances (II)
12:40-14:20	Lunch break		
Free afternoon			

Sunday, 30 August			
9:00-9:40	P2	R. Pfundt	Incidental findings during genetic testing
9:40-10:20	Z3	O. Zuffardi	How hypomorphic variants modulate expression variability
10:20-11:00	D8	J.-M. Dupont	Origin of aneuploidy
11:00-11:20	Coffee break		
11:20-12:00	Z4	O. Zuffardi	Constitutional chromosome rearrangements and susceptibility to their correction by somatic recombination
12:00-12:40	P1	M. Pinelli	AI in clinical genetics
12:40-14:20	Lunch break		
14:20-15:00 (A)	WS7	A. Baumer	Work-up of cases: strategies after the detection of CNVs
14:20-15:00 (B)	WS8	M. Pinelli	AI in clinical genetics
15:00-15:35	R2	M. Rocchi	Chromosomes in evolution
15:40-16:20 (B)	WS7	A. Baumer	Work-up of cases: strategies after the detection of CNVs
15:40-16:20 (A)	WS8	M. Pinelli	AI in clinical genetics
16:20-16:40	Coffee break		
16:45-17:25	Lecturers		Students presentations
17:25-18:05	Lecturers		Cases raising ethical questions

Monday, 31 August			
9:00-9:40	E1	E. Errichiello	Breakpoint analysis of complex structural variants (I)
9:40-10:20	P3	R. Pfundt	Next generations sequencing strategies in diagnostics
10:20-11:00	R3	M. Rocchi	Genome plasticity
11:00-11:20	Coffee Break		
11:20-12:00	E2	E. Errichiello	Breakpoint analysis of complex structural variants (II)
12:00-12:40	R4	M. Rocchi	Health and disease through an evolutionary lens
12:40-14:20	Lunch Break		
14:30-15:50			Examination
15:50-16:10	Coffee Break		
16:10-17:00			Presentation of correct exam results
17:00-17:40			Discussion of the programme
18:00-18:40			Castle tour
19:30-21:00			Farwell dinner

Tuesday, 1 September		Departure after breakfast	
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