18th Goldrain Course in Clinical Cytogenetics

August 26-September 1, 2025

Monday, 25 August, Arrival			
20:30-21:30	**Welcome and introduction, by Albert Schinzel**		

Tuesday, 26 August			
9:00-9:40	**S1**	A.Schinzel	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
11:00-11:20	**Coffee Break**		
11:20-12:00	**V1**	M.Vismara	Introduction to three-dimensional syndromic facial morphology
12:00-12:40	**S2**	A.Schinzel	Dysmorphic findings in chromosome aberrations: formation & interpretation
12:40-14:20	**Lunch Break**		
14:20-15:00	**Bt2**	D.Bartholdi	Clinical dysmorphologic examination in cytogenetic disorders.
15:00-15:40	**WS1**	D.Bartholdi	Clinical issues: difficult situations
	WS2	K.Miller/E.Klopocki	ISCN, standard and molecular
15:40-16:20	**D1**	J.-M.Dupont	FISH techniques and their application
16:20-16:40	**Coffee Break**		
16:40-17:20	**WS1**	D.Bartholdi	Clinical issues: difficult situations
	WS2	K.Miller/E.Klopocki	ISCN, standard and molecular
17:20-18:00	**Kl1**	E.Klopocki	Arrays: principles, technical aspects, different platforms

Wednesday, 27 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	**Ba1**	A.Baumer	MLPA and QF-PCR
12:00-12:40	**Bu2**	G.v.Buggenhout	Microdeletion syndromes
12:40-14:20	**Lunch Break**		
14:20-15:00	**WS3**	T.Krones	Ethical issues: difficult situations
	WS4	K.Miller	Segregation of chromosomal translocations: practical exercises
15:00-15:40	**Ma2**	K.Madan	Sex chromosomes
15:40-16:20	**Kl2**	E.Klopocki	Position effects and effects of CNVs containing no genes
16:20-16:40	**Coffee Break**		
16:40-17:20	**WS3**	T.Krones	Ethical issues: difficult situations
	WS4	K.Miller	Segregation of chromosomal translocations: practical exercises
17:20-18:00	**Mi2**	K.Miller	Markers and ring chromosomes

Thursday, 28 August			
9:00-9:40	**Bu3**	G.v.Buggenhout	The clinical effect of sex chromosomal aneuploidy
9:40-10:20	**D2**	J.-M.Dupont	Optical genome mapping
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	**Ma3**	K.Madan	Complex chromosome rearrangements
12:40-14:20	**Lunch Break**		
Excursion			
21:00-22:00	**R1**	*M.Rocchi*	*Why we are what we are*

Friday, 29 August			
9:00-9:40	**D3**	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	**D4**	J.-M. Dupont	Prenatal cytogenetic diagnosis: RAD and CMA
11:00-11:20	**Coffee Break**		
11:20-12:00	**D5**	J.-M. Dupont	Non-invasive prenatal aneuploidy testing using cfDNA
12:00-12:40	**Mi3**	K. Miller	Cytogenetics of spontaneous abortions and stillbirths
12:40-14:20	**Lunch Break**		
14:20-15:00	**D6**	J.-M. Dupont	Prenatal cytogenetic diagnosis: mosaics in PND
15:00-15:40	**WS5**	K. Miller	Practical exercises in report writing
	WS6	J.-M. Dupont	Appropriate management in prenatal screening and diagnosis
15:40-16:20	**Ma4**	K. Madan	Inversions and insertions
16:20-16:40	**Coffee Break**		
16:40-17:20	**WS5**	K. Miller	Practical exercises in report writing
	WS6	J.-M. Dupont	Appropriate management in prenatal screening and diagnosis
17:20-18:00			**STUDENT presentations**

Saturday, 30 August			
9:00-9:40	**Ma5**	K. Madan	Natural human chimeras
9:40-10:20	**Z1**	O. Zuffardi	From trisomies to structural rearrangements (I)
10:20-11:00	**S3/W2**	A. Schinzel/J. Wisser	Twins in prenatal diagnosis
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	From trisomies to structural rearrangements (II)
12:40-14:20	**Lunch Break**		
Free afternoon			

Sunday, 31 August			
9:00-9:40	P2	R. Pfundt	Incidental findings during genetic testing
9:40-10:20	Z3	O. Zuffardi	Chromothripsis and chromoanagenesis: how frequently do they mimic a simple chromosome rearrangement?
10:20-11:00	Ma6	K. Madan	Disorders of sex development
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	**D7**	J.-M. Dupont	Origin of aneuploidy
12:40-14:20	**Lunch Break**		
14:20-15:00	**WS7**	A. Baumer	Work-up of cases: strategies after the detection of CNVs
	WS8	M. Vismara	Artificial Intelligence in medicine
15:00-15:40	**E1**	E. Errichiello	Breakpoint analysis of complex structural variants
15:40-16:20	**R2**	M. Rocchi	Chromosomes in evolution
16:20-16:40	**Coffee Break**		
16:40-17:20	**WS7**	A. Baumer	Work-up of cases: strategies after the detection of CNVs
	WS8	M. Vismara	Artificial Intelligence in medicine
17:20-18:00			**STUDENT presentations**

Monday, 1 September			
9:00 - 9:40	**R3**	M. Rocchi	Genome plasticity
9:40 - 10:20	**P3**	R. Pfundt	Next generations sequencing strategies in diagnostics
10:20 -11:00	**S4**	A. Schinzel	Paradigm shifts, unexpected findings ….
11:00-11:20	**Coffee Break**		
11:20-12:00			Cases Presentation of cases raising ethical questions
12:00-12:40			*Slot available if needed.*
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, 2 September	Departure after breakfast