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Universität
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E.C.A.
European
Cytogeneticists
Association

Clinical Cytogenetics Aug. 26th to Sep. 2nd, 2017 Goldrain, South Tyrol, Italy

DIRECTOR:

A. Schinzel (Zurich, Switzerland)

FACULTY:

D. Bartholdi (Berne, Switzerland), A. Baumer (Zurich, Switzerland), P. Benn (Farmington CT, U.S.A.), T.-H. Bui (Stockholm, Sweden), R. Ciccone (Pavia, Italy), E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), N. de Leeuw (Nijmegen, The Netherlands), K. Madan (Leiden, the Netherlands), K. Miller (Hannover, Germany), E. Syk-Lundberg (Stockholm, Sweden), G. van Buggenhout (Leuven, Belgium), O. Zuffardi (Pavia, Italy)

PROVISIONAL PROGRAM:

Saturday, August 26th		Arrival at Goldrain
19:00-20:30	<i>Dinner</i>	
20:30-21:00	A. Schinzel	Introduction and organizational comments
21:00-	Get together	Steinkeller
Sunday, August 27th		Basic cytogenetics - lab & clinic
Morning Session: 09:00 – 12:40		
09:00-09:35	1 K. Miller	Basic concepts of chromosome aberrations and epidemiology
09:40-10:15	2 G.v.Buggenhout	Common chromosome disorders and follow-up I
10:20-10:55	3 A. Schinzel	Dysmorphic findings in chromosome aberrations; formation and interpretation
11:00-11:20	<i>Coffee Break</i>	
11:25-12:00	4 G.v.Buggenhout	Common chromosome disorders and follow-up II
12:05-12:40	5 K. Miller	Chromosome aberrations in spontaneous abortions and stillborns
12:45-14:00	<i>Lunch Break</i>	
Afternoon Session: 14:00 – 18:10		Cytogenetics & reporting
14:00-14:35	6 D. Bartholdi	Clinical dysmorphic examination in cytogenetic disorders
14:40-15:15	7 E. Syk-Lundberg	Small supernumerary chromosomes
15:20-15:55	8 K. Miller	Ring chromosomes

16:00-16:20		<i>Coffee Break</i>	
16:25-17:10	9	N. de Leeuw	ISCN, standard and molecular
17:15-17:40	10	K. Madan	Cytogenetic polymorphisms
17:45-18:10	11	A. Schinzel	Karyotype-phenotype correlations
18:30-20:00		<i>Dinner</i>	
21:00-		Get together	Steinkeller

Monday, August 28th	Molecular cytogenetic techniques
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Morning Session: 09:00 – 12:40

09:00-09:35	12	A. Baumer	MLPA and QF-PCR
09:40-10:15	13	N. de Leeuw	FISH techniques and their application
10:20-10:55	14	R. Ciccone	Array: principles, technical aspects, different platforms
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	15	N. de Leeuw	SNP array analysis and interpretation in constitutional genome diagnostics
12:05-12:40	16	R. Ciccone	Array: evaluation of the results: use of databases
12:45-14:00		<i>Lunch Break</i>	

Afternoon Session: 14:00 – 18:40	Practical exercises
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14:00-15:10	W1/ W2	N. de Leeuw/ K. Miller	Practical exercises in cytogenetic nomenclature and report writing. Parallel with W2
15:15-16:20	W2/ W1	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results: hands-on in computer room. Parallel with W1
16:25-16:50		<i>Coffee Break</i>	
16:55-17:45	W3/ W4	N. de Leeuw	The ECARUCA database: practical exercises (in Computer room, rotate with W4)
17:50-18:40	W4/ W3	K. Miller	Segregation of chromosomal translocations: practical exercises (half of the group, rotate with W3)
19:00-20:00		<i>Dinner</i>	
21:00-		Get together	Steinkeller

Tuesday, August 29th	Prenatal diagnosis
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Morning Session: 09:00 – 12:40

09:00-09:35	17	P. Benn	Traditional approaches for the detection of aneuploidy in prenatal diagnosis: screening using serum and ultrasound markers
09:40-10:15	18	T.-H. Bui	Prenatal cytogenetic diagnosis I: mosaics in PND
10:20-10:55	19	T.-H. Bui	Prenatal cytogenetic diagnosis II: RAD (rapid aneuploidy detection) and CMA(chromosomal microarray detection)
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	20	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I
12:05-12:40	21	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects II
12:45-14:00		<i>Lunch Break</i>	

Afternoon Session: 14:10 – 18:30	EXCURSION
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19:00-20:00	<i>Dinner</i>
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20:15-21:00	22	T.-H. Bui and lecturers	NIPD versus advanced diagnostic testing. Podium discussion
21:15-		Get together	Steinkeller

Wednesday, August 30th	Mechanisms & counselling
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Morning Session: 09:00 – 12:40

09:00-09:45	23	E. Syk-Lundberg	Pre-implantation genetic diagnosis
09:50-10:25	24	T.-H. Bui	Prenatal ultrasound findings indicative for chromosome aberrations
10:30-11:00	25	O. Zuffardi	De novo chromosome imbalances: mechanisms of origin
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	26	A. Baumer	Uniparental disomy (UPD) and imprinting
12:05-12:40	27	A. Schinzel	Clinical phenotypes in case of UPD
12:45-14:00		<i>Lunch Break</i>	

Afternoon Session: 14:00 – 18:30	Practical exercises; miscellaneous
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14:00-14:40	28	O. Zuffardi	Somatic mosaics and disease
14:45-15:45	W5	K. Miller/K. Madan	Ethical issues, diagnostic dilemmas, with examples (half of the group, parallel with W6)
	W6	T. Krones	
15:50-16:10		<i>Coffee Break</i>	
16:15-17:05	W6	A. Baumer	Work-up of cases: strategies after the detection of CNVs (half of the group, parallel with W5)
	W5		
17:10-17:50	29	E. Klopocki	Position effects and effects of CNVs containing no genes
17:55-18:30	30	O. Zuffardi	Incomplete penetrance: some light within the conundrum
19:00-20:30		<i>Dinner</i>	
21:00-		Get together	Steinkeller

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Thursday, August 31st	Complex clinical cytogenetics
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Morning Session: 09:00 – 12:40

09:00-09:35	31	K. Madan	Complex chromosome rearrangements I
09:40-10:15	32	G.v. Buggenhout	Microdeletion syndromes
10:20-10:55	33	K. Madan	Mosaics and chimeras
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	34	Students	Short presentations with discussion
12:05-12:40	35	Students/Fac.	Ethical issues with examples
12:45-14:10		<i>Lunch Break</i>	

Afternoon Session: 14:15 – 18:45	Second excursion or self-study
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19:00-20:00		<i>Dinner</i>	
20:15-		Get together	Steinkeller

Friday, September 1st	Miscellaneous
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Morning Session: 09:00 – 12:40		Sex chromosomes & disorders of sex development
09:00-09:35	36 G.v.Buggenhout	The clinical effect of sex chromosomal aneuploidy
09:40-10:15	37 O. Zuffardi	Sex chromosomes: mosaics, AZF deletions, XIST, SRY
10:20-10:55	38 K. Madan	Disorders of sex development
11:00-11:20	<i>Coffee Break</i>	
		New techniques
11:25-12:00	39 N. de Leeuw	Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome
12:05-12:40	40 Eva Klopocki	An introduction to CRISPR/Cas9 and its future possibilities
12:45-14:10	<i>Lunch Break</i>	
Afternoon Session: 14:15 – 18:35		Reflections & evaluation
14:15-14:50	41 To be determ.	Conventional versus molecular karyotyping
14:55-15:30	42 A. Schinzel	The history of errors and paradigm shifts in clinical cytogenetics
15:35-16:00	<i>Coffee Break</i>	
16:05-16:35	Students / Faculty	Questions and evaluation
16:40-18:10	Students	EXAMINATION
18:15-19:00	Students / Faculty	Guided visit of the castle
19:15-21:00	<i>Dinner</i>	Steinkeller or Inner Court of the castle
Saturday, September 2nd		Goodbye Goldrain
08:00-09:00	<i>Breakfast</i>	
09:00-	<i>Checkout and departure</i>	