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Universität
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E.C.A.
European
Cytogeneticists
Association

Clinical Cytogenetics September 14th – 20st, 2014 Goldrain, South Tyrol, Italy

DIRECTOR:

A. Schinzel (Zurich, Switzerland)

FACULTY:

D. Bartholdi (Zurich, Switzerland), A. Baumer (Zurich, Switzerland), P. Benn (Farmington CT, U.S.A.), E. Blennow (Stockholm, Sweden), R. Ciccone (Pavia, Italy), R. Hastings, (Oxford, U.K.), T. Krones (Zurich, Switzerland), N. de Leeuw (Nijmegen, The Netherlands), G. van Buggenhout (Leuven, Belgium), C. van Ravenswaaij (Groningen, The Netherlands), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

PROGRAM:

Sunday, September 14th – Arrival at Goldrain

19:00-20:30	<i>Dinner</i>	
20:30-21:00	A. Schinzel	Introduction and organisational comments
21:00-00:00	Get together	Steinkeller

Monday, September 15th – Clinical findings in chromosome aberrations

Morning Session: 09:00 – 12:45

09:00-09:30	1	A. Schinzel	Dysmorphic findings in chromosome aberrations: Formation and interpretation
09:35-10:30	2	D. Bartholdi	Clinical dysmorphic examinations with the help of measurements and computer programs
10:35-11:00	3	A. Schinzel	Common and uncommon malformations in patients with chromosome aberrations
11:05-11:30		<i>Coffee Break</i>	
11:30-12:05	4	P. Benn	Current Approaches for the Aneuploidy: Screening Using Serum and Ultrasound Markers
12:10-12:45	5	J. Wisser	Prenatal ultrasound findings indicative for chromosome aberrations
12:50-14:30		<i>Lunch Break</i>	

**Afternoon Session: 14:30 – 18:30 - Clinical findings in chromosome aberrations/
Ethical issues**

Nomenclature and databases

14:30-15:05	6	C.v.Ravenswaaij	Conventional vs molecular karyotyping
15:10-15:45	7	C.v.Ravenswaaij for K. Miller	Chromosome aberrations in spontaneous abortions and stillborns
15:50-16:15		<i>Coffee Break</i>	
16:15-16:40	8	J. Wisser & A. Schinzel	Issues connecting with twins at prenatal diagnosis
16:45-17:15	9	C.v.Ravenswaaij	Epidemiology of chromosome aberrations
17:20-18:15	10	T.Krones	Ethical issues in pre- and postnatal genetic diagnosis, with examples
18:20-18:55	11	R. Hastings for J. Barber	Microscopically visible copy number variation
19:00-20:30		<i>Dinner</i>	
20:30-21:30	12	Students/Faculty	Short presentations with discussions
21:30-00:00		Get together	Steinkeller

Tuesday, September 16th – ISCN / Prenatal diagnosis

Morning Session: 09:00 – 12:55

09:00-09:40	13	N. de Leeuw	ISCN
09:45-10:20	14	N. de Leeuw	Workshops: Practical exercises in cytogenetic nomenclature. Report writing.
10:25-11:00	15	C.v.Ravenswaaij	Risk assessment in structural chromosome aberrations
11:00-11:20		<i>Coffee Break</i>	Group picture
11:20-11:55	16	R. Hastings	Prenatal Cytogenetic Diagnosis I
12:00-12:55	17	R. Hastings	Prenatal Cytogenetic Diagnosis II
13:00-14:30		<i>Lunch Break</i>	

Afternoon Session: 14:30 – 18:30 – Miscellaneous

14:30-15:05	18	N. de Leeuw	FISH techniques and their application
15:10-15:45	19	R. Hastings & N. de Leeuw	Exercises in report writing
15:50-16:15		<i>Coffee Break</i>	
16:15-16:40	20	N. de Leeuw	The ECARUCA database. Introduction
16:45-17:25	21	N. de Leeuw & C. v.Ravenswaaij	Workshops: The ECARUCA database. Practical exercises. First group, alternating with 24
17:30-18:05	22	C.v.Ravenswaaij	Reporting array results: what the clinician needs to know
18:10-18:45	23	R. Hastings	Accreditation of cytogenetic laboratories
18:45-19:55		<i>Dinner</i>	
20:00-21:00	24	Students/Faculty	Case presentations and workshop in small groups: Diagnostic decisions and dilemmas. Second group, alternating with 21
21:00-00:00		Get together	Steinkeller

Wednesday, September 17th - Clinical phenotype; Cytogenetic techniques; Databases**Morning Session: 09:00 – 12:45**

09:00-09:35	25	G. van Buggenhout	The adult and elderly patient with a chromosome aberration;
09:40-10:15	26	G. van Buggenhout	Follow-up studies in patients with chromosome aberrations
10:20-10:55	27	O. Zuffardi	Within the genome: the new medicine
11:00-11:20		<i>Coffee Break</i>	
11:20-11:55	28	R. Ciccone	Array-CGH: principals, technical aspects
12:00-12:45	29	G. van Buggenhout	Microdeletion syndromes
12:50-14:00		<i>Lunch Break</i>	

Afternoon

14:00-14:45	30	R. Ciccone	Array-CGH: evaluation of the results; use of databases
14:50-15:15	31	A. Baumer	MLPA and QF-PCR
15:15-15:30		<i>Coffee Break</i>	
15:50-20:00			Excursion and dinner
21:00-00:00		Get together	Steinkeller

Thursday, September 18th - From microscope to array**Morning Session: 9:00 – 12:45**

09:00-09:35	32	N. de Leeuw / A. Baumer	Optimal use of available techniques in clinical cytogenetics / strategies after detection of CNVs
09:40-10:15	33	A. Schinzel / C. van Ravenswaaij	Karyotype-phenotype correlations
10:20-10:55	34	N. de Leeuw	Comparison of array platforms used in constitutional genome diagnostics; SNP arrays; Best test for a given indication
11:00-11:20		<i>Coffee Break</i>	
11:20-11:55	35	O. Zuffardi	Two-hit model for severe developmental delay- understanding variable expressivity in microdeletion/microduplication syndromes
12:00-12:45	36	N. de Leeuw	Beyond cytogenetics: Next generation sequencing / exome sequencing
12:50-14:30		<i>Lunch</i>	

Afternoon Session: 14:00 – 19:00 – Free for individual excursions

19:00-20:30		<i>Dinner</i>	
20:30-21:30	37	Faculty & Students	Case presentations / workshops
20:30-00:00		Get together	Steinkeller

Friday, September 19th – Miscellaneous

Morning Session: 09:00 – 12:55

09:00-09:35	38	E. Blennow	Extra small supernumerary chromosomes
09:40-10:15	39	O. Zuffardi	Cytogenetics, microarrays, next generation sequencing: OI and new acquisitions
10:20-11:05	40	E. Blennow	Pre-implantation cytogenetic diagnosis
11:10-11:30		<i>Coffee Break</i>	Second group picture
11:30-12:15	41	O. Zuffardi	Microarrays in prenatal cytogenetic diagnosis
12:20-13:00	42	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects.I.
13:00-14:30		<i>Lunch Break</i>	

Afternoon Session: 14:30 – 18:15

14:30-15:05	43	P. Benn	Non-invasive prenatal testing using cfDNA: current status and future prospects.II
15:10-15:45	44	A. Baumer	Uniparental disomy and imprinting
15:50-16:15		<i>Coffee Break</i>	
16:15-17:00	45	Faculty	Workshops: Student presentations of difficult-to-interpret chromosome aberrations
17:00-17:30		Students and Faculty	Questions;
17:30-18:15		Students	Discussion of the program
18:15-19:00		Students & Faculty	EXAMINATION / Questionnaire
19:00-20:30		<i>Dinner</i>	Guided tour through the castle
20:30-00:00		Get together	Steinkeller

Lectures not orally presented

Only print	46	A. Schinzel	Guidelines for cytogenetic case reports
Only print	47	A. Schinzel	Mechanisms of formation and origin of chromosome aberrations
		A. Baumer	
Only print	48	A. Schinzel	When to perform fibroblast chromosome examinations?
Only print	49	K. Madan	Balanced complex chromosome rearrangements

Saturday, September 20th – Goodby Goldrain

08:00-09:00	<i>Breakfast</i>
09:00-	<i>Checkout and departure</i>