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E.C.A.
European
Cytogeneticists
Association

Clinical Cytogenetics **Aug. 27th to Sep. 3rd, 2016** **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.), R. Ciccone (Pavia, Italy), R. Hastings, (Oxford, U.K.), E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), N. de Leeuw (Nijmegen, The Netherlands), K. Miller (Hannover, Germany), E. Syk-Lundberg (Stockholm, Sweden), G. van Buggenhout (Leuven, Belgium), C. van Ravenswaaij (Groningen, The Netherlands), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

PROGRAM:

Saturday, August 27th – Arrival at Goldrain

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

Sunday, August 28th – Basic cytogenetics - lab & clinic

Morning Session: 09:00 – 13:25

09:00-09:35	1	C.v.Ravenswaaij	Basic concepts of chromosome aberrations and epidemiology
09:40-10:15	2	R. Hastings	Cytogenetic polymorphisms
10:20-10:55	3	E. Syk-Lundberg	Small supernumerary chromosomes
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	4	E. Syk-Lundberg	Ring chromosomes
12:05-12:40	5	A. Schinzel	Dysmorphic findings in chromosome aberrations; formation and interpretation
12:45-13:20	6	K. Miller	Chromosome aberrations in spontaneous abortions and stillborns
13:25-14:40		<i>Lunch Break</i>	

Afternoon Session: 14:45 – 18:35

14:45-15:20	7	G.v.Buggenhout	Common chromosome disorders and follow-up I
15:25-16:25	W1	N. de Leeuw/ R. Hastings	Practical exercises in cytogenetic nomenclature (half of the group, rotate with W2)
16:30-16:50		<i>Coffee Break</i>	
16:55-17:55	W2	D. Bartholdi/ G.v.Buggenhout	Clinical dysmorphic examination in cytogenetic disorders (half of the group, rotate with W1)
18:00-18:35	8	G.v.Buggenhout	Common chromosome disorders and follow-up I
19:00-20:00		<i>Dinner</i>	
20:15-21:00		Students/ Faculty	Three short student presentations with discussion
21:15-00:00		Get together	Steinkeller

Monday, August 29th – Cytogenetic techniques & reporting**Morning Session: 09:00 – 13:25**

09:00-09:35	9	A. Baumer	MLPA and QF-PCR
09:40-10:15	10	N. de Leeuw	FISH techniques and their application
10:20-10:55	11	R. Ciccone	Array: principles, technical aspects, different platforms
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	12	N. de Leeuw	SNP array analysis and interpretation in constitutional genome diagnostics
12:05-12:40	13	C.v.Ravenswaaij	Reporting array results: what the clinician needs to know
12:45-13:20	14	R. Hastings	External Quality Assessment in clinical cytogenetics
13:25-14:40		<i>Lunch Break</i>	

Afternoon Session: 14:45 – 18:35

14:45-15:20	15	R. Ciccone	Array: evaluation of the results: use of databases
15:25-16:25	W3	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results: hands-on in computer room (half of the group, rotate with W4)
16:30-16:50		<i>Coffee Break</i>	
16:55-17:55	W4	N. de Leeuw/ R. Hastings	Exercises in report writing (half of the group, rotate with W3)
18:00-18:35	16	E. Klopocki	Position effects and effects of CNVs containing no genes
19:00-20:00		<i>Dinner</i>	
20:15-21:00		Students/ Faculty	Three short student presentations with discussion
21:15-00:00		Get together	Steinkeller

Tuesday, August 30th – Prenatal diagnosis**Morning Session: 09:00 – 13:25**

09:00-09:35	17	P. Benn	Current approaches for the detection of aneuploidy in prenatal diagnosis: screening using serum and ultrasound markers
09:40-10:15	18	J. Wisser	Prenatal ultrasound findings indicative for chromosome aberrations
10:20-10:55	19	R. Hastings	Prenatal cytogenetic diagnosis I: mosaics in PND
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	20	R. Hastings	Prenatal cytogenetic diagnosis II: examples and their work-up
12:05-12:40	21	E. Syk-Lundberg	Pre-implantation genetic diagnosis
12:45-13:20	22	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I
13:25-14:40		<i>Lunch Break</i>	

Afternoon Session: 14:45 – 18:30 EXCURSION

14:45-18:30		Students/ Faculty	Excursion
19:00-20:00		<i>Dinner</i>	
20:15-21:00		Students/ Faculty	Three short student presentations with discussion
21:15-00:00		Get together	Steinkeller

Wednesday, August 31st – Mechanisms & counselling**Morning Session: 09:00 – 13:25**

09:00-09:35	23	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I
09:40-10:15	24	A. Schinzel	Karyotype-phenotype correlations in diagnostics
10:20-10:55	25	C.v.Ravenswaaij	Karyotype-phenotype correlations in research_ examples and pit falls
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	26	D. Bartholdi	Genetic counselling in case of a chromosome aberration
12:05-12:40	27	O. Zuffardi	De novo chromosome imbalances: mechanisms of origin
12:45-13:20	28	O. Zuffardi	Somatic mosaics and disease
13:25-14:40		<i>Lunch Break</i>	

Afternoon Session: 14:45 – 18:35

14:45-15:20	29	C.v.Ravenswaaij	Risk assessment in structural chromosome aberrations I
15:25-16:25	W5	K. Miller	Segregation of chromosomal translocations: practical exercises (half of the group, rotate with W6)
16:30-16:50		<i>Coffee Break</i>	
16:55-17:55	W6	N. de Leeuw/ C.v.Ravenswaaij	The ECARUCA database: practical exercises (in computer room)

18:00-18:35	30	C.v.Ravenswaaij	(half of the group, rotate with W5) Risk assessment in structural chromosome aberrations II
19:00-20:00		<i>Dinner</i>	
20:15-21:00		Students/ Faculty	Three short student presentations with discussion
21:15-00:00		Get together	Steinkeller

Thursday, September 1st – Complex clinical cytogenetics

Morning Session: 09:00 – 13:25

09:00-09:35	31	A. Baumer	Uniparental disomy and imprinting
09:40-10:15	32	G.v.Buggenhout	Microdeletion syndromes
10:20-10:55	33	O. Zuffardi	Incomplete penetrance: some light within the conundrum
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	34	T. Krones	Ethical issues with examples I
12:05-12:40	35	T. Krones	Ethical issues with examples II
12:45-14:10		<i>Lunch Break</i>	

Afternoon Session: 14:15 – 16:30

14:15-15:15	W7	T. Krones	Diagnostic decision and dilemmas (half of the group, rotate with W8)
15:20-16:20	W8	A. Baumer	Work-up of cases: strategies after the detection of CNVs (half of the group, rotate with W7)
16:30-18:45		Students/ Faculty	Free: Self-study or group walk
19:00-20:00		<i>Dinner</i>	
20:15-21:00		Students/ Faculty	Three short student presentations with discussion
21:15-00:00		Get together	Steinkeller

Friday, September 2nd - Miscellaneous

Morning Session: 09:00 – 13:25 – 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	C.v.Ravenswaaij	DSD: XX and XY sex reversal with normal SRY status
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	39	N. de Leeuw	New techniques Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome
12:05-12:40	40	E. Klopocki	CRISPR/Cas to study the effect of chromosomal imbalance
12:45-14:10		<i>Lunch Break</i>	

Afternoon Session: 14:15 – 18:35 – Reflections & evaluation

14:15-14:50	41	C.v.Ravenswaaij	Conventional versus molecular karyotyping
14:55-15:30	42	A. Schinzel	The history of errors and paradigm changes 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 Innenhof

Lectures not orally presented

Only print	45	A. Schinzel	Guidelines for cytogenetic case reports
Only print	46	A. Schinzel	When to perform fibroblast chromosome examinations?
Only print	47	K. Madan	Balanced complex chromosome rearrangements
Only print	48	J. Wisser	Issues connected with twins at prenatal diagnosis

Saturday, September 3rd – Goodbye Goldrain

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