13th Goldrain Course in
Clinical Cytogenetics
August 25 to September 02, 2018

LOCATION

Goldrain Castle, Goldrain, South Tyrol, Italy
Website of the venue: **www.schloss-goldrain.it**

COURSE DESCRIPTION

The course is focused on phenotypic findings, mechanisms of origin and transmission, correlations of clinical patterns with chromosomal imbalance and modern ways of diagnosis of the latter. Special attention is paid to an understanding how deletions and/or duplications of chromosomal segments cause developmental defects. The course also addresses the optimal application of the diagnostic possibilities, both pre- and postnatally and including molecular cytogenetic methods for a precise determination of segmental aneuploidy.

TOPICS

Dysmorphic findings in chromosome aberrations: formation and interpretation – The adult and elderly patient with a chromosome aberration – Follow-up studies in patients with chromosome aberrations – Clinical findings associated with chromosome aberrations – Microdeletion syndromes: clinical pictures – ISCN – Practical exercises in cytogenetic nomenclature – The ECARUCA database: Introduction and practical exercises – Students presentation of cases with difficult-to-interpret chromosome aberrations– prenatal cytogenetic diagnosis – Mosaics and chimeras – imprinting and uniparental disomy – FISH techniques and their interpretation – MLPA – Array-CGH: principles, technical aspects; evaluation of the results – SNP arrays – QF-PCR – Epidemiology of chromosome aberrations – Chromosome aberrations in spontaneous abortions and stillborns –Harmless chromosome aberrations – Risk assessment in structural chromosome aberrations – Optimal use of available techniques in clinical cytogenetics – Extra small supernumerary chromosomes – Genomic variation: a continuum from SNPs to chromosome aneuploidy – Use of genomic databases – Pre-implantation cytogenetic diagnosis – Ultrasound findings indicative of chromosome aberrations– Accreditation of cytogenetic laboratories –Ethical issues in the context of cytogenetic diagnosis – Non-invasive prenatal cytogenetic diagnosis.

Practical exercises will be offered with the ISCN system for chromosome aberrations and with cytogenetic and genomic databases. Students will have the opportunity to present their own observations and cytogenetic findings which are difficult to interpret. The students will have the opportunity to perform a test at the end of the course.

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), 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)

For further questions please write directly to Albert Schinzel at **schinzel@medgen.uzh.ch**

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Full fee is Euro 1300 for a single room or Euro 1150 (VAT included) in a 2-bed-room. It includes tuition, course material, free access to internet during the course, accommodation for 7 nights, all meals, beverages during the breaks and a ½ day excursion.