

# 7<sup>th</sup> Goldrain Course in Clinical Cytogenetics September 15<sup>th</sup> to 21<sup>st</sup>, 2012

## LOCATION

Goldrain Castle, Goldrain, South Tyrol, Italy  
Website of the venue: [www.schloss-goldrain.it](http://www.schloss-goldrain.it)

## COURSE DESCRIPTION

The course is focused on phenotypic findings, mechanisms of origin and transmission, and correlations of clinical patterns with the chromosomal imbalance. 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 including molecular cytogenetic methods for a precise determination of segmental aneuploidy.

## TOPICS

Dysmorphic findings in chromosome aberrations: formation and interpretation – The use of measurements – 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 – Sex 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 – balanced complex chromosome aberrations – 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 – Mammalian karyotype evolution – Accreditation of cytogenetic laboratories – Neocentromeres – 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 cytogenetics findings which are difficult to interpret. The students will have the opportunity to perform a test at the end of the course.

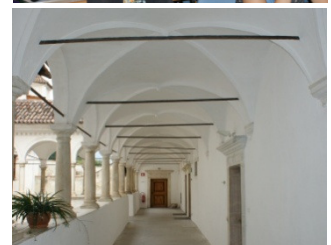
## DIRECTOR

A. Schinzel (Zurich, Switzerland)

## FACULTY

D. Bartholdi (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.), N. de Leeuw (Nijmegen, The Netherlands), G. van Buggenhout (Leuven, Belgium), C. van Ravenswaaij (Groningen, The Netherlands), M. Rocchi (Bari, Italy), J. Wisser (Zurich, Switzerland), O. Zuffardi (Pavia, Italy)

For further questions please consult the program via <http://www.goldrain.ch> or [www.e-c-a.eu/](http://www.e-c-a.eu/) or write directly to Albert Schinzel at [schinzel@medgen.uzh.ch](mailto:schinzel@medgen.uzh.ch)



*Full fee is Euro 1000 for a single room or Euro 900 (VAT included) in a 2-bed-room. It includes tuition, course material, free access to internet during the course, accommodation for 6 nights, all meals, beverages during the breaks and a ½ day excursion.*

